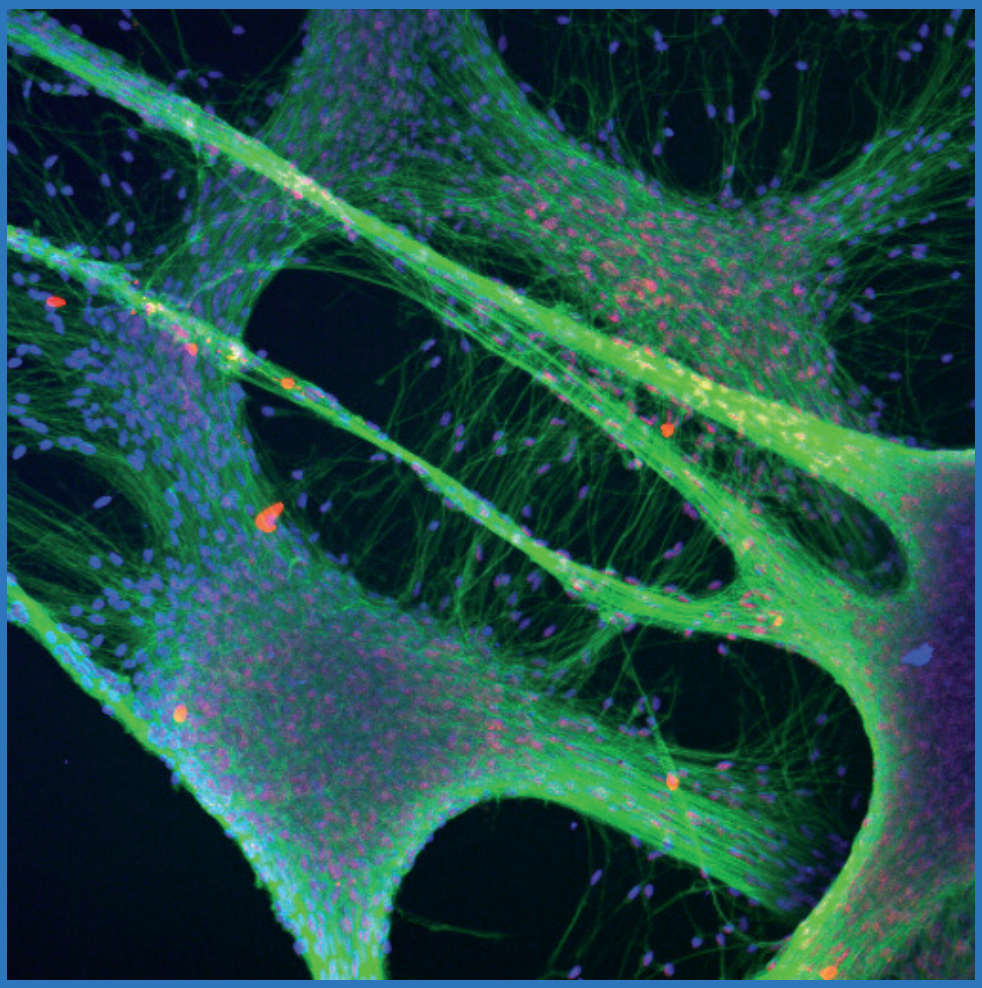
Fondazione Telethon

XVIII Scientific Convention

March 9-11, 2015

Riva del Garda (TN) - Palazzo dei Congressi



Provincia Autonoma di Trento

Telethon

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PROVINCIA
AUTONOMA
DI TRENTO

Front cover - *Immagine di copertina*

Using human neural stem cells to model the neurologic aspects of Cockayne syndrome

The picture illustrates neurons (green) and oligodendrocytes (red) differentiated from human neural stem cells. Nuclei are stained blue.

Cellule staminali neurali come modello per lo studio degli aspetti neurologici della sindrome di Cockayne

L'immagine rappresenta neuroni (in verde) ed oligodendrociti (in rosso) differenziati da cellule staminali neurali umane. I nuclei sono marcati in blu.

Courtesy of dr. Luca Proietti De Santis, Università degli Studi della Tuscia, Viterbo, Italy

(Telethon grant GGP11176, talk 22, abstract 160)

Back cover - *Retro di copertina*

We put our hearts in the research on SCA28

Mitochondrial network in Mouse Embryonic Fibroblasts of spinocerebellar ataxia SCA28 Knockin Mouse model, labeled with Mitored fluorescent probe.

Nella ricerca sulla SCA28 noi mettiamo il cuore

Network mitocondriale delle cellule fibroblasti embrionali di topo del modello murino knockin di atassia spinocerebellare SCA28, in cui i mitocondri sono stati marcati con la sonda fluorescente Mitored.

Courtesy of drs. Cecilia Mancini and Alfredo Brusco, University of Torino, Italy

(Telethon grant GGP12217, abstract 85)

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RINGRAZIAMENTI

La Fondazione Telethon desidera esprimere la propria gratitudine a coloro che, con la loro generosità, hanno contribuito a rendere possibile la XVIII Convention Scientifica

Provincia Autonoma di Trento

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SCIENTIFIC PROGRAM
(Sala 1000)

Monday, 09th March 2015

10.00 – 14.00 *Registration and poster setting up*

14.00 – 14.30 **Welcome and Opening address**
Lucia Monaco (Fondazione Telethon)

14.30 – 15.00 **OPENING LECTURE**
Role of the advocacy organization in rare disease research
Sharon Terry (Genetic Alliance, Washington DC, USA)

15.00 – 17.00 **PLENARY SESSION 1** – **Motorneuron diseases: from molecular targets to trial design**
Chairpersons: Francesco Muntoni, Maria Pennuto
Pathogenesis and therapeutic window in chromosome 5 spinal muscular atrophy
Francesco Muntoni (UCL, London, UK) (*Talk 1)*
From standards of care to natural history: recipe for trial readiness
Eugenio Mercuri (Università Cattolica del Sacro Cuore, Rome) (*Talk 2)*
SMN2 splicing regulation: from disease-linked mechanism to therapeutic opportunity in spinal muscular atrophy (*Talk 3)*
Claudio Sette (University of Rome Tor Vergata, Fondazione Santa Lucia, Rome)
Skeletal muscle degeneration in spinal and bulbar muscular atrophy
Maria Pennuto (DTI, Università degli Studi di Trento, Trento) (*Talk 4)*
The protein quality control system in motor neuron degeneration in spinal and bulbar muscular atrophy
Angelo Poletti (Università degli Studi di Milano, Milan) (*Talk 5)*

17.00 – 17.30 *Coffee break*

17.30 – 19.00 **POSTER SESSION 1**

19.00 – 20.00 ***SCIENCE AND SOCIETY*** **- Animal experimentation**
Moderator: Anna Maria Zaccheddu (Fondazione Telethon)
Speakers: Elena Cattaneo (Università degli Studi di Milano and Italian senator for life)
Giuliano Grignaschi (Mario Negri Institute, Milan)
Kirk Leech (European Animal Research Association, London, UK)

20.00 – 21.00 *Welcome cocktail*

Tuesday, 10th March 2015

08.30 – 09.00 *Registration and poster setting up*

09.00 – 10.00 **ROUND TABLE – From the lab to the clinic, yet remaining a basic scientist**

Moderators: Lana Skirboll, Lucia Faccio

Speakers: Lana Skirboll (Sanofi Washington DC, USA)
Lucia Faccio (Fondazione Telethon)
Andrea Ballabio (TIGEM, Naples)
Luigi Naldini (OSR-TIGET, Milan)

10.00 – 11.00 **PLENARY SESSION 2 – Updates on the Telethon clinical trials**

Chairperson: Lana Skirboll

Gene therapy of Mucopolisaccharidosis VI

Alberto Auricchio (TIGEM, Naples) (*Talk 6)*

Hematopoietic stem cell gene therapy for inherited disorders: 15 years of experience at TIGET

Alessandro Aiuti (OSR-TIGET, Milan) (*Talk 7)*

Gene therapy for beta thalassemia: towards clinical application

Giuliana Ferrari and Fabio Ciceri (OSR-TIGET, Milan) (*Talk 8)*

11.00 – 11.30 *Coffee break*

11.30 – 13.00 **POSTER SESSION 2**

13.00 – 14.00 *Buffet Lunch*

14.00 – 16.30 **PLENARY SESSION 3 – Hereditary ataxias: convergences and divergences**

Chairpersons: Laura Ranum, Enrico Bertini, Joel Gottesfeld

Introduction on hereditary ataxias

Enrico Bertini (Ospedale Pediatrico Bambino Gesù, Rome) (*Talk 9)*

Domain-specific regulation of cerebellar neurogenesis by Zfp423, a gene implicated in Joubert Syndrome and cerebellar vermis hypoplasia

Gian Giacomo Consalez (Università Vita-Salute San Raffaele, San Raffaele Scientific Institute, Milan) (*Talk 10)*

Molecular determinants of Marinesco-Sjogren syndrome: cell stress, and alterations of proteostasis

Michele Sallese (Fondazione Mario Negri Sud, Santa Maria Imbaro, Chieti) (*Talk 11)*

Epigenetic therapy for Friedreich's ataxia

Joel Gottesfeld (The Scripps Research Institute, La Jolla, USA) (*Talk 12)*

Preventing Frataxin degradation as a therapeutic strategy for Friedreich's ataxia

Roberto Testi (Università degli Studi di Roma Tor Vergata, Rome) (*Talk 13)*

Repeat associated non-ATG (RAN) translation in spinocerebellar ataxia type 8

Laura Ranum (University of Florida, Gainesville, USA) (*Talk 14)*

Genetic and pharmacological rescues of spinocerebellar ataxia in the SCA28 model open to human therapy (*Talk 15)*

Giorgio Casari (Università Vita-Salute San Raffaele, San Raffaele Scientific Institute, Milan)

16.30 – 17.00 *Coffee break*

17.00 – 18.30 **POSTER SESSION 3**

18.30 – 19.30 ***SCIENCE POLICIES - Research integrity***

Michael Caplan (Yale School of Medicine, New Haven, USA)

Wednesday, 11th March 2015

09.00 – 11.00 **PLENARY SESSION 4 - The central role of the skeleton in human health**

Chairpersons: Anna Maria Teti, Marta Serafini

Can we treat systemic autosomal dominant diseases? An example from osteopetrosis
Anna Maria Teti (Università degli Studi dell'Aquila, L'Aquila) (*Talk 16)*

Neonatal bone marrow transplantation for mucopolysaccharidosis type I: the earlier the better
Marta Serafini (DTI, San Gerardo Hospital/Fondazione MBBM, Università di Milano-Bicocca, Monza) (*Talk 17)*

Congenital malformations of the limbs reveal complex genetic interactions between the p63 and Dlx5 genes: towards identification of exploitable targets
Luisa Guerrini (Università degli Studi di Milano, Milan) (*Talk 18)*

In vivo models of chondrodysplasias caused by defects in proteoglycan biosynthesis: phenotyping and pharmacological approaches
Antonio Rossi (Università degli Studi di Pavia, Pavia) (*Talk 19)*

New roles of cellular catabolism during skeletal development
Carmine Settembre (DTI, TIGEM, Naples) (*Talk 20)*

11.00 – 11.30 *Coffee break*

11.30 – 13.00 **PLENARY SESSION 5 - Neurological disorders and DNA damage accumulation**

Chairpersons: Fabrizio d'Adda di Fagagna, Domenico Delia

DNA damage response activation in the brain: causes and consequences
Fabrizio d'Adda di Fagagna (IFOM Foundation Milan, CNR Pavia) (*Talk 21)*

Dissecting the neurogenic role of CSB protein and its implication in Cockayne syndrome
Luca Proietti de Santis (Università degli Studi della Tuscia, Viterbo) (*Talk 22)*

Links between DNA repair and chromosome structural pathways revealed by cellular models of the Warsaw breakage syndrome cohesinopathy disorder
Takuya Abe (IFOM, the FIRC institute of Molecular Oncology, Milan) (*Talk 23)*

Mechanisms of neurodegeneration in ATM-deficient ataxia-telangiectasia
Domenico Delia (Fondazione IRCCS Istituto Nazionale Tumori, Milan) (*Talk 24)*

13.00 – 13.30 **LATE BREAKING NEWS**

The Intellectual disability protein RAB39B regulates selectively GluA2 trafficking determining synaptic AMPAR composition
Patrizia D'Adamo (DTI, San Raffaele Scientific Institute, Milan)

***COQ4* mutations cause a broad spectrum of mitochondrial disorders associated with CoQ10 deficiency**
Daniele Ghezzi (Foundation of the Carlo Besta Neurological Institute, Milan)

Liver-directed lentiviral gene therapy in a dog model of hemophilia B
Alessio Cantore (OSR-TIGET, Milan)

13.30 – 14.00 **POSTER PRIZE AWARD AND CLOSING REMARKS**

IV CONVEGNO DELLE ASSOCIAZIONI DI PAZIENTI

9 MARZO 2015

10.00 – 13.30 *Registrazione dei partecipanti e ritiro cuffie per traduzione simultanea*

14.00 – 15.00 **SESSIONE PLENARIA (Sala 1000, traduzione simultanea)**

Benvenuto e apertura dei lavori
Lucia Monaco, Direttore Scientifico, Fondazione Telethon

OPENING LECTURE – Role of the advocacy organization in Rare Disease research
Sharon Terry, Presidente Genetic Alliance, Washington DC, USA

15.00 - 15.30 *Coffee break*

15.30 -18.00 **WORKSHOP 1 (Sala 300) – Percorsi di empowerment nella ricerca scientifica**

Moderatore: Alessia Daturi, Fondazione Telethon

L'esperienza di Determinazione Rara
Renza Galluppi, Presidente Uniamo FIMR
Sara Casati, bioeticista, Università degli Studi di Milano Bicocca
Anna Ambrosini, Fondazione Telethon

Processi partecipativi in uno studio clinico
Tommasina Iorno, Associazione Talassemici e Drepanocitici Lombardi
Giuliana Ferrari, Istituto Telethon per la terapia genica, Milano
Sarah Marktel, Ospedale San Raffaele, Milano
Sara Casati, bioeticista, Università degli Studi di Milano Bicocca
Alessia Daturi**,** Fondazione Telethon

Discussione con la platea

18.00 - 19.00 **SESSIONE POSTER 1: Incontro con i Ricercatori**

19.00 - 20.00 **SESSIONE PLENARIA (Sala 1000, traduzione simultanea)**

SCIENZA E SOCIETÀ: Sperimentazione animale
Elena Cattaneo, Università degli Studi di Milano
Giuliano Grignaschi, Istituto Mario Negri, Milano
Kirk Leech, Executive Director, European Animal Research Association (EARA), Londra, UK

20.00 – 21.00 *Cocktail di benvenuto*

10 MARZO 2015

9.00 - 11.30 **WORKSHOP 2 (sala 300) – Strategie per promuovere la ricerca sulle malattie rare**

Moderatore: Alessia Daturi, Fondazione Telethon

Prepararsi per uno studio clinico: trial readiness per la SMA
Daniela Lauro, Presidente Famiglie Sma, Palermo
Eugenio Mercuri, Policlinico Gemelli, Roma
Anna Ambrosini, Fondazione Telethon, Milano

I progetti esplorativi Telethon
Anna Ambrosini, Fondazione Telethon

Discussione con la platea

11.30 - 12.00 *Coffee break*

12.00 – 13.00 **SESSIONE POSTER 2: Incontro con i Ricercatori**

13.00 – 14.00 *Pranzo*

14.30 *Partenza navetta per Verona aeroporto e stazione FS*

ORAL PRESENTATIONS

OPENING LECTURE

ROLE OF THE ADVOCACY ORGANIZATION IN RARE DISEASE RESEARCH

Sharon Terry (Genetic Alliance, Washington DC, USA)

MOTORNEURON DISEASES: FROM MOLECULAR TARGETS TO TRIAL DESIGN

Talk 1

PATHOGENESIS AND THERAPEUTIC WINDOW IN CHROMOSOME 5 SPINAL MUSCULAR ATROPHY

Francesco Muntoni

Dubowitz Neuromuscular Centre, Institute of Child Health, University College London, UK

The majority of patients affected by spinal muscular atrophy (SMA) are due to homozygous deletions of Survival Motor Neuron gene 1 (SMN1) which leads to the absence of functional SMN protein, and results in motor neurons degeneration and muscle weakness. SMN is a ubiquitously expressed protein, although the main target of disease appears to be motorneurons. A second gene, SMN2, encodes a related protein, but a single nucleotide difference from SMN1 prevents efficient splicing of exon 7, leading to a truncated transcript and less functional and unstable protein.
The understanding of the genetic basis of SMA is being exploited for developing SMN gene specific therapies, which for the first time could address the primary defect in SMA patients. Indeed improving SMN2 exon 7 to splice more efficiently is being pursued as a major therapeutic strategy for SMA.
Three different but related strategies are being developed; one is the direct replacement of SMN gene by gene therapy; the second one relates to the use of antisense oligonucleotides (AONs) technology to redirect the exon 7 splicing in SMN2; the 3rd strategy takes advantage of a high throughput study which identified a orally bioavailable small molecule which appears to be very efficient and selective in allowing SMN2 exon 7 incorporation.
During my presentation I will provide an update on where these experimental programs are in their clinical development. I will also discuss our understanding on the central nervous system vs peripheral requirements for SMN protein, and issues related to the window for therapeutic intervention in this condition.

Talk 2

FROM STANDARDS OF CARE TO NATURAL HISTORY: RECIPE FOR TRIAL READINESS

Eugenio Mercuri

Università Cattolica del Sacro Cuore, Rome, Italy

With clinical trials currently underway, improvement in our ability to evaluate standards of care and clinical benefit using functional outcome measures in spinal muscular atrophy (SMA) is timely. Over the last years there has been an effort to implement standards of care and identify different outcome measures that can be chosen according to the requirements in individual studies. Meetings with regulatory agencies and family advocacy groups have confirmed that outcome measures should be statistically robust, validated and suitable for multicentric studies, and able to reflect clinically meaningful changes.
Regarding SMA, preferences were given to measures that had already been validated in SMA for which natural history longitudinal data were available and had already been successfully used in previous trials. For patients with type I SMA, survival and need for respiratory support at generally the preferred outcome measures, together with a clinical measure of function with the CHOP INTEND. The Hammersmith Motor Function Scale (HFMS), and thee Motor Function measure (MFM) have recently been proposed for the non-ambulant SMA and the 6 minute walk test for the ambulant SMA.
These studies have allowed following possible changes over time and to help to power the study and establish how a treatment would be different from what is expected. As a quite wide variability has been found in these measures, several studies have focused on staging of the disease and description of how several variables, such as age or values at baseline can affect the longitudinal performance.

Talk 3

SMN2 SPLICING REGULATION: FROM DISEASE-LINKED MECHANISM TO THERAPEUTIC OPPORTUNITY IN SPINAL MUSCULAR ATROPHY

Claudio Sette

Department of Biomedicine and Prevention, University of Rome Tor Vergata and Laboratory of Neuro-embryology, Fondazione Santa Lucia, Rome, Italy

Spinal Muscular Atrophy (SMA) represents the primary genetic cause of infant mortality. SMA is determined by loss of function mutations in the SMN1 gene, which cause deficiency in the Survival Motor Neuron (SMN) protein and degeneration of alpha-motor neurons in the spinal cord. SMN2, a nearly identical gene present in humans, does not complement SMN1 loss because it mostly yields a truncated and unstable SMN protein, due to skipping of exon 7 during the processing of the pre-mRNA. Nevertheless, the possibility of rescuing exon 7 splicing in SMN2 offers a therapeutic opportunity to restore the expression of full length SMN protein and to ameliorate the symptoms of SMA.
Alternative splicing is a complex process regulated at multiple layers, from epigenetic modification of the chromatin to binding of specific splicing factors to sequences in the regulated exons or flanking introns. In the case of SMA, histone deacetylase (HDAC) inhibitors were found to promote splicing of SMN2 exon 7, even though the mechanism involved was not elucidated. Similarly, a large number of splicing factors were shown to modulate SMN2 exon 7 in cultured cells, but whether or not they participate to regulation of this regulation in vivo was not shown for any of them.
Our laboratory is investigating the mechanism of action of SMN2 splicing regulation by LBH589, a novel generation HDAC inhibitor with promising therapeutic potential. We found that LBH589 affects the deposition of selected histone marks near the exon 7 region of the SMN2 locus, thereby affecting the processivity of the RNA polymerase and the recognition of the exon. Moreover, we have identified SAM68 as the first splicing factor that regulates exon 7 splicing in the brain and peripheral tissues of a mouse model of SMA, thus influencing the severity of the disease.
These studies highlight the potential of using the modulation of SMN2 exon 7 splicing as therapeutic approach for SMA and shed light on key players in the regulation of this disease-causing splicing event.

Talk 4

SKELETAL MUSCLE DEGENERATION IN SPINAL AND BULBAR MUSCULAR ATROPHY

Maria Pennuto

Dulbecco Telethon Institute (DTI), Università di Trento, Italy

Spinal and bulbar muscular atrophy (SBMA), or Kennedy disease, is a neuromuscular disease that manifests in adult males. SBMA is caused by expansion of a CAG trinucleotide tandem repeat in the gene coding for androgen receptor (AR). SBMA symptoms develop only in males because the disease is triggered by the binding of androgens to mutant AR. Others and we have provided evidence that skeletal muscle is a primary target of mutant AR toxicity. Expression of mutant AR in all tissues except muscle prevented disease manifestations in transgenic mice. Moreover, selective overexpression of the insulin-like growth factor 1 in muscle attenuated disease manifestations in transgenic SBMA mice, providing further evidence that targeting muscle is sufficient to delay disease progression in vivo. To elucidate the mechanism through which mutant AR causes muscle atrophy, we used knock in SBMA mice. Expression of mutant AR leads to a metabolic switch in muscle, which was modified by a specific high calorie diet regime. We propose that SBMA is a metabolic disease.

Talk 5

THE PROTEIN QUALITY CONTROL SYSTEM IN MOTOR NEURON DEGENERATION IN SPINAL AND BULBAR MUSCULAR ATROPHY

Paola Rusmini[1], Elisa Giorgetti[1], Valeria Crippa[2], Riccardo Cristofani[1], Maria E. Cicardi[1], Mariarita Galbiati[1], Maria Pennuto[3], Serena Carra[3], Angelo Poletti[1]

[1] Dipartimento di Scienze Farmacologiche e Biomolecolari, and Centre of Excellence on Neurodegenerative Diseases, Università degli studi di Milano, Italy
[2] IRCCS, Istituto Neurologico C. Mondino, Pavia, Italy
[3] CIBIO, Università degli studi di Trento, Italy
[4] Dipartimento di Scienze Biomediche, Metaboliche e Neuroscienze, Università degli studi di Modena e Reggio Emilia, Italy

Spinal and bulbar muscular atrophy (SBMA) is an X-linked motorneuron disease caused by an abnormal expansion of a CAG triplet repeat in exon 1 of the androgen receptor (AR) gene. This expansion is translated into an abnormally long polyglutamine tract (polyQ) in the AR protein. The AR is a transcription factor that mediated the biological activities of male sexual hormones. Several evidence suggests that the elongated polyQ tract confers a toxic gain-of-function to the mutant AR. This ARpolyQ toxicity is activated by the AR ligand testosterone (or dihydrotestosterone). Testosterone-dependent toxicity is triggers by the generation of ARpolyQ misfolded species, that in some conditions, tend to aggregate in spinal cord motorneurons and muscle cells. ARpolyQ aggregates are formed in the cytoplasm and nuclei of affected cells, but they toxic role is still largely debated. Instead, there is general agreement on the fact that ARpolyQ toxicity is associated with the nuclear localization of an altered (misfolded) fraction of the protein. Therefore, the prevention of nuclear localization and/or the facilitation of ARpolyQ misfolded fraction degradation may have great beneficial effects against ARpolyQ toxicity.
Recently, we demonstrated that the ubiquitin proteasome system (UPS) degrades a large fraction of misfolded ARpolyQ. However, an excess of ARpolyQ may overwhelm the UPS, escaping the degradation and thus tend to accumulate in aggregates. These aggregates are useful to confine the toxic protein in a physically defined subcellular compartment, thus reducing the potential toxicity of un-degraded misfolded ARpolyQ. Generally, the aggregates are transported to the microtubule organization center (MTOC) to be engulfed into autophagosomes and destroyed by the autophagic system. When an insufficient pool of specific chaperones is present, the autophagic flux may be blocked and this lead to the formation of insoluble inclusions. In fact, we found that the silencing of HspB8, a member of the small heat shock protein family, correlated with a massive accumulation of misfolded ARpolyQ in immortalized motorneuronal cells. Conversely, HspB8 overexpression facilitates the autophagic removal of misfolded aggregating species of ARpolyQ. HspB8 is highly induced in surviving motorneurons of patients affected by motorneuron diseases and HspB8 participates in the stress response aimed at cell protection. HspB8 does not induce autophagy (p62 and LC3 expression, two key autophagic molecules), but it prevents p62 bodies formation, restoring a fully functional autophagic flux. Notably, we found that, trehalose, a natural disaccharide know to be a potent autophagy stimulator, induces HspB8 expression suggesting that HspB8 could be one of the molecular mediators of the pro-autophagic activity of trehalose.
Therefore, based on the evidence that testosterone triggers nuclear toxicity by inducing AR nuclear translocation, and misfolded species accumulate because of an impaired autophagic flux, we hypothesize that prevention of ARpolyQ nuclear localization, combined with an increased ARpolyQ cytoplasmic clearance should reduce its toxicity. We thus used the antiandrogen Bicalutamide (Casodex®), which slows down AR activation and nuclear translocation, in combination with trehalose, and we found that in motorneurons the two compounds synergically reduced ARpolyQ insoluble forms, with an efficiency higher than the one observed in single treatments. This effect was also present on insoluble species of AR with a very long polyQ (Q112) tract, capable to generate nuclear aggregates into the cell nuclei. Therefore, the combinatory use of Bicalutamide and trehalose greatly facilitate ARpolyQ clearance, and might be a promising approach to be tested in vivo in animal models of SBMA.

UPDATES ON THE TELETHON CLINICAL TRIALS

Talk 6

GENE THERAPY OF MUCOPOLISACCHARIDOSIS VI

Alberto Auricchio

Telethon Institute of Genetics and Medicine (TIGEM), Naples, Italy

Mucopolysaccharidosis type VI (MPS VI) or Maroteaux-Lamy syndrome is a rare lysosomal storage disorder caused by arylsulfatase B (ARSB) deficiency which results in widespread accumulation and urinary excretion of dermatan sulfate (DS). MPS VI is characterized by hepatomegaly, dysostosis multiplex, heart valve engulfment and corneal opacities without central nervous system involvement. Enzyme replacement therapy (ERT) which requires weekly infusions of costly enzyme reduces DS urinary excretion, liver size and improves endurance however ERT has little to no effect on heart, skeletal and ocular disease. We have recently shown that a single systemic injection of an adeno-associated viral (AAV) vector 8 expressing ARSB from a liver-specific promoter results in significant biochemical, pathological and functional improvement in MPS VI cats. ARSB expression and DS clearance is sustained for at least 6 years after vector administration in cats. In addition, we have shown in a mouse model of MPS VI that a single high-dose vector administration of AAV8 results in therapeutic efficacy simi-

lar to weekly infusions of recombinant enzyme. Interestingly, similar results are achieved combining low doses of AAV8 with monthly ERT. Based on our pre-clinical data we have recently received EU funding to develop a clinical trial to test the safety and efficacy of a single administration of AAV8 in MPS VI patients.

Talk 7

HEMATOPOIETIC STEM CELL GENE THERAPY FOR INHERITED DISORDERS: 15 YEARS OF EXPERIENCE AT TIGET

Alessandro Aiuti [1,2,3], Alessandra Biffi [1,2], Luigi Naldini [1,3]

[1] San Raffaele Telethon Institute for Gene Therapy (TIGET), San Raffaele Scientific Institute, Milan, Italy
[2] Pediatric Immuno-hematology and Bone Marrow Transplant, San Raffaele Scientific Institute, Milan, Italy
[3] Vita-Salute San Raffaele University, Milan, Italy

Gene therapy (GT) with hematopoietic stem cells (HSC) is an attractive therapeutic strategy for inherited genetic disorders. In the past 15 years TIGET has pioneered the clinical application of HSC GT for two primary immunodeficiency disorders (ADA-SCID and Wiskott-Aldrich Syndrome, WAS) and Metachromatic Leukodystropy (MLD), a severe lysosomal storage disorder. Collectively, more than 40 patients have received autologous CD34+ cells transduced with a gammaretroviral vector (ADA-SCID) or lentiviral vector (LV) (WAS and MLD) encoding the therapeutic gene. Prior to GT patients received a chemotherapy conditioning to favour HSC engraftment. The intensity and type of conditioning were adapted to take in account the disease biology (i.e. selective advantage for immunodeficiencies and the need to replace microglia in MLD) as well as the degree of chimerism needed. Sustained HSC engraftment was observed in all trials but higher levels (20-80%) were achieved in the LV-based studies. The efficiency of gene transfer in CD34+ cells, the dose of cells, and the intensity of conditioning were key parameters in determining the level of in vivo HSC engraftment. Genetically modified cells expressed the therapeutic gene to levels that were sufficient to achieve biological activity and clinical benefit (see specific TIGET abstracts for detailed information). All treated patients are alive and no leukemic events have been observed, indicating that GT has a favourable safety profile. The clinical development is now continuing in the context of the alliance with GSK, which has in-licensed GT for the three diseases. These results will pave the way for the application of HSC GT in a wider spectrum of genetic disorders with unmet medical need.

Selected references

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A. Aiuti at al. Gene therapy for immunodeficiency due to adenosine deaminase deficiency. N. Engl. J. Med. 360, 447–458 (2009)
A. Aiuti et al. Lentivirus based gene therapy of hematopoietic stem cell in Wiskott-Aldrich Syndrome. Science 341(6148):1233151. Epub 2013 Jul 11 (2013)
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Talk 8

GENE THERAPY FOR BETA THALASSEMIA: TOWARDS CLINICAL APPLICATION

Giuliana Ferrari[1,2] and Fabio Ciceri[3]

[1] Telethon Institute for Gene Therapy (TIGET), San Raffaele Scientific Institute
[2] "Vita-Salute" San Raffaele University Medical School
[3] Hematology and BMT Unit , IRCCS San Raffaele Scientific Institute, Milan, Italy.

Beta-thalassemia is a severe congenital anemia caused by reduced or absent beta-globin chain production of the adult hemoglobin tetramer. More than 300 mutations leading to the disease have been described, affecting all the steps related to the expression of the b-globin gene. It represents the most common autosomal recessive syndrome to cause a major health problem worldwide with an estimated annual birth incidence of 40.000/year.
Profound anemia, rapid expansion of erythroid bone marrow mass and secondary organ damage due to iron overload are the most frequent clinical manifestation of the severe form of beta-thalassemia major.
Treatment of beta-thalassemia is essentially supportive. Patients require a lifelong transfusion regimen combined with iron chelation therapy to reduce hemosiderosis that is ultimately fatal if not continuously treated. At present, the only curative approach is represented by allogeneic hematopoietic stem cell transplantation (HSCT), which, however, is limited by HLA compatibility and toxicity due to graft versus host disease, graft rejection and immunosuppressive regimens required. Medical management and HSCT are both therapeutic options that could improve the quality of life and survival of thalassemic patients. Nevertheless, they are both burden by complications and limitations, outlining the need for testing innovative curative approaches.
Gene therapy for beta-thalassemia, as an alternative cure to allogeneic HSCT, is based on the autologous transplantation of hematopoietic stem cells (HSCs) engineered to express a transcriptionally regulated human beta-globin gene. Due to complexity of vector design and requirement of high level of gene transfer and expression, several decades were needed to find that the vector of choice in this pathology is an HIV-derived lentiviral vector. The target cell of gene transfer by globin lentiviral vectors are the HSC and in humans they can be collected as CD34+ cells by different sources, such as cord blood, bone marrow or peripheral blood following mobilization with different agents, like G-CSF and/or Plerixafor. The development and the production of globin lentiviral vectors and the optimization of gene transfer protocols in human CD34+ cells, aimed to achieve high efficiency of transduction without impairing the "stemness" and hematopoietic reconstitution potential, have progressed this field to the pioneering clinical trials in France and in U.S.A. The results so far available are encouraging and will help also to better understand current limitations and way for improvement.
Our contribution to this field was devoted to the clinical development of a safe gene therapy approach, relying on the high-titer globin vector GLOBE (Miccio et al, PNAS 2008), new source of HSCs and an innovative clinical protocol favoring efficient engraftment of genetically modified cells with reduced toxicity.
Starting from the demonstration of proof of efficacy of gene therapy in thalassemic mutant mice and in hematopoietic cells from thalassemic patients (Miccio et al, PNAS 2008 and PLoSOne 2011, Roselli et al, EMBO MolMed 2010; Milsom and Williams, 2010), we moved towards the clinical development by assessing the risk/benefit ratio prior to administration in humans, in comprehensive in vivo pre-clinical studies. Evaluating the biosafety of gene therapy medicinal products following EMA and ICH guidelines, in the GLPs (good laboratory practices) framework, provides results of scientific significance within regulatory standards, and paves the way towards future market registration. Recently, we have obtained certification for a GLP Test Facility within the academic HSR-TIGET, including two BSL2 cell and molecular biology

laboratories, a flow cytometry lab, four BSL2 labs in the SPF animal facility fully equipped for necropsy and data monitoring/recording, a pathology lab and an archive room.
In order to support the documentation for a clinical trial application (CTA) of gene therapy for beta-thalassemia two main GLP studies were performed: a toxicology and tumorigenicity study in a thalassemic mouse strain, and a biodistribution study in the hematochimera model of the NSG mice.
In both GLP studies, the safety of the use of GLOBE vector was demonstrated. Overall, the preclinical data provided the rationale and the basis for the proposal of a first-in-human clinical protocol.
The proposal of a clinical trial has been anticipated for Scientific Advice to EMA, and all the documents for CTA, including IMPD, IB and the clinical protocol, have been submitted to the San Raffaele Hospital Ethical Committee and to the Italian regulatory authorities (AIFA/ISS) for evaluation and approval.
The clinical protocol of gene therapy for transfusion dependent beta-thalassemia, using HSCs genetically corrected with the GLOBE lentiviral vector, which is currently under final review for approval, will be presented.

HEREDITARY ATAXIAS: CONVERGENCES AND DIVERGENCES

Talk 9

INTRODUCTION ON HEREDITARY ATAXIAS

Enrico Bertini

Ospedale Pediatrico Bambino Gesù, Unit of Neuromuscular and Neurodegenerative Disorders, Laboratory of Molecular Medicine, Rome, Italy

Inherited ataxias are a large, heterogeneous group of either non-progressive (malformative) or progressive (neurodegenerative) conditions with unknown overall prevalence due to the lack of reliable epidemiological data. The clinical association of ataxia with a wide spectrum of neurological and non-neurological signs makes these conditions highly disabling.
Several are the criteria for classification of inherited ataxias: mode of inheritance, age at onset (congenital versus later onset ataxias), pathogenetic mechanisms, association with other neurologic and non-neurologic symptoms (pure cerebellar ataxia, spinal cerebellar ataxia, ataxia and mental retardation, etc), neuroimaging features (Joubert syndrome, cerebellar atrophy, PCH, etc). Clinical and neuroimaging features may remain unspecific, while genetic diagnosis is often impaired by the scarce access to testing, high costs, and limited knowledge of the genetic basis of many Inherited Ataxias. As a result, diagnosis is delayed or remains merely descriptive in many patients, leading to uncertain prognosis and wrong counselling of families. To date, therapies for most PAs (especially non-progressive) are lacking, and effective motor and cognitive rehabilitation remains a key strategy to improve patients' quality of life and favour their inclusion in the social context.

Talk 10

DOMAIN-SPECIFIC REGULATION OF CEREBELLAR NEUROGENESIS BY *ZFP423*, A GENE IMPLICATED IN JOUBERT SYNDROME AND CEREBELLAR VERMIS HYPOPLASIA

Gian Giacomo Consalez [1], Enrico Bertini[2], Enza Maria Valente[3]
[1] Università Vita-Salute San Raffaele, and Division of Neuroscience, San Raffaele Scientific Institute, Milan, Italy
[2] Laboratory of Molecular Medicine, Unit of Neuromuscular Disorders, Dept of Neurosciences, IRCCS Bambino Gesù Children's Hospital, Rome, Italy
[3] IRCCS Casa Sollievo della Sofferenza, Mendel Institute, Rome, Italy

Aims - Neurogenesis is a tightly regulated process. Its success depends on the ability to balance the expansion/maintenance of an undifferentiated neural progenitor pool with the production of sequential generations of neurons and glia. The *Zfp423* gene encodes a 30 Zn-finger transcription factor (TF) that acts as a nuclear scaffold for the assembly of complex transcriptional machineries regulating neural development. Mutations of this gene have been identified in patients carrying cerebellar vermis hypoplasia (CVH) or Joubert Syndrome (JS), a classical ciliopathy.
Methods - To characterize the role of ZFP423 in cerebellar neurogenesis, we have performed morphological and molecular studies of two mouse mutant lines carrying allelic in-frame deletions of *Zfp423*. Both mutants are profoundly ataxic. One of them lacks Zn-finger domains 9-20 (Δ9-20), implicated in BMP and Notch signal transduction, while the other one lacks a C-terminal domain (Δ28-30), which interacts with EBF TFs, involved in neuronal differentiation.
Results - While both mutants exhibit cerebellar hypoplasia, GABAergic neurogenesis is more severely impaired in $Zfp423^{\Delta 9\text{-}20/\Delta 9\text{-}20}$ mutants, which feature a premature decline of the Purkinje cell progenitor pool in the ventricular zone (VZ). In this mutant, dividing VZ progenitors feature obvious changes in mitotic spindle orientation and cilium turnover. Conversely, $Zfp423^{\Delta 28\text{-}30/\Delta 28\text{-}30}$ mutants exhibit a selective impairment in the differentiation and survival of glutamatergic progenitors, and especially a sharp decrease in the number of nucleofugal neuron precursors in the medial cerebellar nuclei primordium.
Conclusions - The results of this *in vivo* and *in vitro* structure-function analysis shed light on the domain- and context-specific roles played by ZFP423 in different aspects of cerebellar neurogenesis, and on its potential involvement as a cofactor/modifier gene in several, nosologically distinct, cerebellar malformations.

Talk 11

MOLECULAR DETERMINANTS OF MARINESCO-SJOGREN SYNDROME: CELL STRESS, AND ALTERATIONS OF PROTEOSTASIS

Vanessa Capone[1] Giorgia Fragassi[1], Francesca Ornaghi[2], Roberto Chiesa[2] and Michele Sallese[1]

[1]Unit of Genomic Approaches to Membrane Traffic, Fondazione Mario Negri Sud, Santa Maria Imbaro (Chieti), Italy
[2]Department of Neuroscience, IRCCS - Mario Negri Institute for Pharmacological Research, Milan, Italy

The Marinesco-Sjogren syndrome (MSS) is a rare, early-onset, autosomal recessive genetic disease caused by mutations in the SIL1 gene. One of the main symptoms of MSS is ataxia due to degeneration of Purkinje cells (PC). There is no treatment for MSS and the medical care of the patients is essentially symptomatic. SIL1 encodes an ADP exchange factor for GRP78, the master operator of endoplasmic reticulum (ER) functions.
To elucidate the basis of PC degeneration we investigated SIL1/GRP78-dependent cell processes, including the activating of ER stress pathways (unfolded protein response, UPR), and the protein folding/transport in MSS cell models in which SIL1 expression was knocked down (KD).
SIL1-KD cells underwent spontaneous apoptosis and were more sensitive to stress factors than controls. We observed activation of the ER stress sensors IRE1, PERK and ATF6 that was boosted by a

challenge with tunicamycin (glycosylation inhibitor), indicating that a secondary stress might contribute to trigger neurodegeneration. Importantly, the inhibition of IRE1, PERK or ATF6 signalling pathways prevented the cell apoptosis. Since PERK was strongly activated also in the *woozy* mouse model of MSS, we planned to investigate the potential therapeutic effect of the PERK inhibitor GSK2606414. Preliminary to this, we carried out an accurate analysis of disease onset and progression in these animals. The motor deficit was periodically assessed by the rotarod test. Motor dysfunction arose at 11 weeks of age, progressively worsened up to17 weeks then stabilized. UPR activation and degeneration of Purkinje cells forerun the motor dysfunction, indicating that treatment should be initiated before the onset of the symptoms or, at the latest, in the early symptomatic phase.
The analysis of membrane trafficking detected a strong delay of ER to Golgi transport of cargoes in SIL-1 KD cells, with accumulation of the endogenous Na/K ATPase in the Golgi complex. The impairment of Golgi transport might be triggered by a shortage of structural Golgi proteins consequent to the deficient folding machinery. The Na/K ATPase is essential for important neuronal functions, including the maintenance of resting membrane potential, the regulation of cell volume and the intrinsic firing of Purkinje cells, and there is evidence that mutation or functional inhibition of Na/K ATPase causes cerebellar ataxia and dystonia.
Our results confirm UPR activation in MSS and indicate that SIL1 deficiency affects the trafficking and the distribution of vital neuronal proteins, potentially contributing to PC dysfunction and degeneration.

Talk 12

EPIGENETIC THERAPY FOR FRIEDREICH'S ATAXIA

Joel Gottesfeld

Department of Cell and Molecular Biology and Department of Chemistry, The Scripps Research Institute, La Jolla CA USA 92037, USA

The genetic defect in Friedreich's ataxia (FRDA) is the hyperexpansion of a GAA·TCC triplet in the first intron of the FXN gene, encoding the essential mitochondrial protein frataxin. Histone post-translational modifications near the expanded repeats are consistent with heterochromatin formation and consequent FXN gene silencing. Using a newly developed human neuronal cell model, based on patient-derived induced pluripotent stem (iPS) cells, we find that 2-aminobenzamide histone deacetylase (HDAC) inhibitors increase FXN mRNA levels and frataxin protein in FRDA neuronal cells. Preclinical studies of safety and toxicity have been completed and a phase I clinical trial in FRDA patients has been performed at the University of Turin. Drug treatment increases FXN mRNA and histone acetylation in peripheral blood mononuclear cells, with no drug-related adverse effects, providing a proof of concept for this therapeutic approach. Moreover, similar drug concentrations were required in serum and in cell culture to increase FXN mRNA levels. To gain further understanding of the mechanism of action of these small molecules, we analyzed their effect on eleven histone posttranslational modifications in three cell types that respond differently to HDAC inhibitors: patient fibroblasts and iPS cells, that do not show increased FXN transcription upon treatment with HDAC inhibitors and patient neuronal cells. This comparison shows that the difference between the ratio of acetylated histone H3 lysine 9 (H3K9) to tri-methylated H3K9 in FRDA and unaffected cells is lowest in neuronal cells compared to fibroblasts and iPS cells. In addition, our molecules increase this ratio by the highest amount in neuronal cells. Effects of compounds with different HDAC selectivities were also analyzed and compounds that are most effective in restoring FXN transcription produced the highest H3K9 acetylation/tri-methylation ratio. We propose that histone H3K9 is an important residue in the activation of the FXN gene and that since methylation of H3K9 is involved in heterochromatin formation, the ratio of acetylation versus methylation for this residue has to reach a threshold in order to be able to reverse FXN silencing. Our findings provide a valuable cellular model for predicting effective drug dosing in the clinical setting and suggest that epigenetic drugs hold promise for the treatment of Friedreich's ataxia.

Talk 13

PREVENTING FRATAXIN DEGRADATION AS A THERAPEUTIC STRATEGY FOR FRIEDREICH'S ATAXIA

Roberto Testi

Department of Biomedicine and Prevention, University of Rome "Tor Vergata", Italy

Friedreich ataxia (FRDA) is caused by insufficient levels of the mitochondrial protein frataxin. This is due to GAA triplet expansions, combined with a serie of epigenetic changes, that impair the transcription of the *FRX* gene. Insufficient frataxin causes neurodegeneration that progressively leads to disability and premature death. There is no approved cure for FRDA. Disease-specific therapeutic efforts aim at elevating frataxin levels in patients.
Our approach builds on the finding that a substantial fraction of neo-synthesized frataxin is ubiquitinated and targeted to destruction by the proteasome. We are developing novel small molecules that bind frataxin and prevent frataxin ubiquitination and destruction, thus resulting in frataxin accumulation in cells. The most promising of these potential drug candidates have been functionally validated in cells derived from FRDA patients.
We also search for approved drugs that could be repurposed for FRDA. One of these is interferon gamma, a cytokine involved in immunity and iron metabolism, that we showed effective in a FRDA animal model. Interferon gamma increases frataxin levels in sensory neurons and ameliorates sensorimotor performance of FRDA mice. Phase II clinical trials with FRDA patients, that provided encouraging results, have been performed.

Talk 14

REPEAT ASSOCIATED NON-ATG (RAN) TRANSLATION IN SPINOCEREBELLAR ATAXIA TYPE 8

Laura P.W. Ranum, Yuanjing Liu, Fatma Ayhan, Monica Banez, John D. Cleary, Tammy Reid, Tao Zu

Department of Molecular Genetics and Microbiology, Center for Neuro Genetics and the Genetics Institute, College of Medicine, University of Florida, Gainesville, Florida, USA
Microsatellite expansion mutations cause more than 30 neurologic diseases. Typically, research for these disorders has focused on understanding the effects of the loss- or gain-of-function of a single mutant protein or RNA. In 2006, we demonstrated that the SCA8 CTG•CAG expansion produces RNAs in both directions. In 2011, we discovered that one of the basic rules scientists use to predict if a gene can make a protein does not apply for hairpin forming microsatellite expansions and that CAG and CUG expansion RNAs can express homopolymeric expansion proteins in all three reading frames without an AUG start codon. We showed this repeat associated non-ATG (RAN) translation, is hairpin-dependent, occurs without frameshifting or RNA editing and is observed in SCA8 and DM1 mouse and human tissues. We and others

have also shown RAN translation occurs in *C9orf72* amyotrophic lateral sclerosis/frontotemporal dementia (ALS/FTD9), and other microsatellite expansion diseases. Similar to SCA8, the C9orf72 ALS/FTD hexanucleotide expansion mutation is expressed in both the sense GGGGCC and antisense GGCCCC directions and that that these RNAs accumulate as nuclear and cytoplasmic foci in patient tissues. Additionally both sense GGGGCC and antisense GGCCCC expansion mutations produce dipeptide expansion proteins with Gly-Ala, Gly-Pro, Gly-Arg, Pro-Arg, Pro-Ala expansion motifs. We will discuss recent work on the mechanisms of RAN translation and insights from mouse models. The discovery of RAN translation has implications for understanding fundamental mechanisms of protein synthesis and neurologic disease. Additionally, our new understanding that some expansion mutations can express two mutant RNAs and up to six mutant proteins highlights the need to explore therapies that will block the effects of both sense and antisense RNAs and RAN proteins.

Talk 15

GENETIC AND PHARMACOLOGICAL RESCUES OF SPINOCEREBELLAR ATAXIA IN THE SCA28 MODEL OPEN TO HUMAN THERAPY

Giorgio Casari

Università Vita-Salute San Raffaele, San Raffaele Scientific Institute, Milan, Italy

Spinocerebellar ataxia type 28 (SCA28) is a neurodegenerative disease caused by mutations of the AFG3L2 gene. The encoded protein assembles into multimeric complexes (the m-AAA proteases), which exert protein quality control in the inner mitochondrial membrane and participate to the regulation of mitochondrial morphology. The Afg3l2 haploinsufficient mouse recapitulates the symptoms of SCA28 patients, presenting a progressive decline in motor skills caused by dark degeneration of Purkinje cells (PC-DCD) of mitochondrial origin. In this work, we define the pathogenetic mechanism of SCA28 and provide the first evidence of a pre-clinical treatment of this disease. We demonstrated in cultured PCs that an inefficient buffering of stimulus-evoked Ca^{2+} peaks by Afg3l2-deficient mitochondria provokes an increase in cytoplasmic Ca^{2+} concentration, thus triggering PC-DCD. Proving this mechanism, we completely recover the ataxic phenotype of SCA28 mice by genetically reducing the metabotropic glutamate receptors mGluR1, and thus decreased Ca^{2+} influx in PCs. The same result has been successfully replicated by administration of an off-label therapy with ceftriaxone that favors the synaptic glutamate clearance. This treatment is effective when applied at both presymptomatic and after the ataxia onset in the preclinical model, thus representing a safe and immediately accessible therapy for presymptomatic carriers of AFG3L2 mutations and also SCA28 patients with overt symptoms.

THE CENTRAL ROLE OF THE SKELETON IN HUMAN HEALTH

Talk 16

CAN WE TREAT SYSTEMIC AUTOSOMAL DOMINANT DISEASES? AN EXAMPLE FROM OSTEOPETROSIS

Anna Maria Teti

Dipartimento di Scienze Cliniche Applicate e Biotecnologiche, Università dell'Aquila, L'Aquila, Italy

Typical feature of autosomal dominant (AD) diseases is the single allele missense nature of the gene mutations involved. These diseases often affect multiple organs or impair systems that are widespread in the organism, including the musculoskeletal tissues. They are frequently incurable, and current treatments are largely palliative. Therefore, there is an unmet medical need to innovate this research field and identify therapeutic strategies aimed at eradicating the mutations rather that treat the symptoms. When the genes involved in AD diseases are haplosufficient, any treatment leading to the specific suppression of the mutant mRNA would theoretically rescue a nearly normal phenotype. In nature, RNA interfering has this potentiality and could represent a new strategy to downregulate the mutant mRNA, leaving the normal mRNA intact, a condition made possible by its high specificity, also aided by chemical modifications. Between the two known RNA interfering machineries, namely small interfering RNA (siRNA) and small hairpin RNA (shRNA), the former would be more suitable for systemic treatments because it could rely on a pharmacological-like approach. We applied this strategy to the AD osteopetrosis type 2 (ADO2), caused by the G213R mutation of the Clcn7 gene encoding the chloride/proton antiporter type 7, which is characterised by impaired osteoclast activity, increased bone mass, bone fragility and secondary haematological and neural failures. A $Clcn7^{G213R}$-specific siRNA, identified in vitro through a systematic mutation-driven strategy, reduced the $Clcn7^{G213R}$ mRNA without affecting the normal mRNA, thus mimicking a condition of haplosufficiency. This siRNA, conjugated to a commercially available vehicle, was effective at rescuing a normal bone phenotype in a $Clcn7^{G213/WT}$ knock-in mouse model that recapitulates the human ADO2. After 4 weeks of treatment bone volume, trabecular number and separation returned to normal, with a full rescue of the bone phenotype. In the rescued ADO2 mice, serum biomarkers of bone resorption, osteoclast number and erosion surface were normalized vs. scrambled-siRNA-treated ADO2 mice, while osteoblast and dynamic parameters were unremarkable. Treatment was well tolerated, with no overt adverse events, suggesting that our strategy could be translated to the clinic. To advance our discovery, we patented the $Clcn7^{G213R}$-siRNAs and are working closely to Fondazione Telethon on the development of this approach, including activities to promote industrial partnerships. We believe that our strategy could have high chances of bringing a benefit to ADO2 patients, as it would be the first actual treatment available for this therapeutically neglected form of osteopetrosis.

Talk 17

NEONATAL BONE MARROW TRANSPLANTATION FOR MUCOPOLYSACCHARIDOSIS TYPE I: THE EARLIER THE BETTER

Marta Serafini

Dulbecco Telethon Institute (DTI) and Centro Ricerca M. Tettamanti, Department of Paediatrics, San Gerardo Hospital/Fondazione MBBM, University of Milano-Bicocca, Monza, Italy

Bone marrow transplantation (BMT) is an efficacious therapeutic modality for a number of nonmalignant conditions, both acquired and genetically determined. In the treatment of genetic disorders, the hematopoiesis transplanted from a healthy allogeneic donor can produce the missing protein at levels sufficient to correct the underlying abnormalities. However, the current clinical experience is only partially successful. One of the reasons for this could be the limited efficacy of the protein provided through the hematopoietic cell secretome. Alternatively, the time at which BMT is currently performed could be too late to prevent organ damage. This fundamental question is well exemplified by the case of mucopolysac-

charidosis type I-Hurler syndrome (MPS IH), a lysosomal storage disease caused by deficiency of α-L-iduronidase (IDUA), in which allogeneic BMT, although considered firstline treatment, has a limited effect on the skeletal abnormalities. The incomplete correction of the skeletal phenotype produced by BMT may also be because bone abnormalities are irreversible at the time of the transplant. Therefore, we hypothesize that the first months of life represent the best window of opportunity for preventing bone deformities in Hurler children. In our study, we have evaluated whether neonatal transplantation of syngeneic normal bone marrow could rescue the severe skeletal phenotype in the mouse model of MPS I. First, neonatal BMT was effective at restoring a-L-iduronidase activity and clearing elevated glycosaminoglycans in blood and multiple organs. At 37 weeks of age, we observed an almost complete normalization of all bone tissue parameters, using radiographic, microcomputed tomography, biochemical, and histological analyses. Moreover, improvements in bone parameters correlated with high levels of bone marrow-derived cell engraftment in multiple hematopoietic compartments, suggesting the early and complete restoration of normal hematopoiesis can have a significant effect on the bone development of newborn MPS I mice. Overall, our study supports the notion that hematopoietic transplantation from a healthy donor can be an effective strategy to restore the missing or defective protein underlying the genetic disorder. However, the clinical efficacy is strictly dependent on providing the missing protein at a very early stage, before symptoms become apparent.
This is a proof-of-concept study that advocates neonatal BMT as a highly effective therapeutic approach for Hurler syndrome, demonstrating that an early treatment may further affect the clinical outcome of these patients, and strongly supports the importance of the implementation of newborn screening procedures to allow an early diagnosis and immediate treatment of affected children

Talk 18

CONGENITAL MALFORMATIONS OF THE LIMBS REVEAL COMPLEX GENETIC INTERACTIONS BETWEEN THE P63 AND DLX5 GENES: TOWARDS IDENTIFICATION OF EXPLOITABLE TARGETS

Giorgio R. Merlo[1], Antonio Costanzo[2], Luisa Guerrini[3]

[1] Department of Molecular Biotechnology, University of Torino, Italy
[2] Department of Dermatology, University of Rome "Tor Vergata", Roma, Italy
[3] Department of Biosciences, University of Milano, Italy

The p63 transcription factor plays a crucial role in proliferation/differentiation/stratification of embryonic and adult ectodermal cells: its mutation in human leads to congenital syndromes characterized by skin dysplasia, craniofacial (cleft palate) and limb (Split Hand/Foot, SHFM) malformations, collectively known as EEC. Our team investigates on the upstream and downstream regulations centered on p63, with the hope to identify therapeutic molecules.
Upstream - 1) The stability of the ΔNp63α protein is controlled by several post-translation modifications. The prolyl-isomerase Pin1 interacts with ΔNp63α and acts as negative regulator of its stability via proteasome-mediated degradation. Interestingly, p63 mutant proteins associated to SHFM or EEC syndromes are resistant to Pin1 action. We also show that FGF8, a morphogenetic signaling molecule essential for limb extension and morphogenesis, counteracts the interaction of Pin1 with ΔNp63α, leading to ΔNp63α stabilization. Notably the *FGF8* locus is a downstream target of DLX5, a ΔNp63α target gene causing SHFM type-I when mutated (Restelli et al., 2014).
2) The p300 acetyl transferase physically interacts with ΔNp63α, catalyzes its acetylation on lysine K193 and induces its stabilization and transcriptional activation. A pre-requisite for p300-ΔNp63α interaction is ΔNp63α phosphorylation by the tyrosine kinase c-Abl. Furthermore, we show that FGF8 increases the level of p300-dependent ΔNp63α acetylation. Notably, the natural mutant ΔNp63α-K193E, associated to the SHFM-type IV, cannot be acetylated by this pathway and displays promoter-specific loss of DNA binding activity and altered expression of development-associated ΔNp63α targets.
Our results link FGF8, c-Abl and p300 in a regulatory pathway that controls DNp63a protein stability and transcriptional activity. Hence, limb malformation-causing p63 mutations, such as the K193E mutation, are likely to result in aberrant limb development via the combined action of altered protein stability and altered promoter occupancy (Restelli et al, submitted).
Downstream - ΔNp63α regulates the transcription of the *Dlx5;Dlx6* transcription factors, disease genes for SHFM-type I syndrome, which in turn regulate the *FGF8* locus (Lo Iacono et al. 2008). Thus, FGF8 participates in multiple non-cell autonomous loops that promote ΔNp63α protein stability and transcriptional activity. Our more recent results indicate that Wnt5a, a diffusible factor of the non-canonical Wnt family, also participates in this loop.
To exploit these results, we are adopting *ex-vivo* cultures of embryonic limb buds to attempt correction of the malformation defect. The exposure of cultured embryonic limbs to FGF8 and Wnt5a peptides partly restores AER molecular marker expression and the pattern of *Sox9*-expressing mesenchymal condensation. These results suggest the possibility that the SHFM limb malformation could be corrected by administration of exogenous FGF8 and Wnt5a, or via gene-transfer to the embryonic ectoderm, in the mouse model.
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Talk 19

IN VIVO MODELS OF CHONDRODYSPLASIAS CAUSED BY DEFECTS IN PROTEOGLYCAN BIOSYNTHESIS: PHENOTYPING AND PHARMACOLOGICAL APPROACHES

Antonio Rossi

Department of Molecular Medicine, Unit of Biochemistry, University of Pavia, Pavia, Italy

Different chondrodysplasias with defects in GAG biosynthesis lead to disorders with similar clinical features. In this respect Desbuquois Dysplasia (DBQD) and Diastrophic Dysplasia (DTD) are paradigmatic: they both show antenatal and postnatal growth retardation, precocious carpal ossification, short metacarpals and joint dislocations that may lead to an erroneous diagnosis.
Due to their common features we have focused on both disorders. DBQD type 1 is caused by mutations in the Calcium-Activated Nucleotidase 1 a Golgi/ER resident enzyme that preferentially hy-

drolyzes UDP. In fibroblasts from DBQD type 1 patients we demonstrated that GAGs synthesis was almost normal under basal conditions, but it was significantly reduced in presence of beta-D-xyloside, an enhancer of chondroitin and dermatan sulfate synthesis, suggesting that CANT1 plays a role in proteoglycan metabolism. The defect in proteoglycan metabolism was further confirmed by gel filtration chromatography: GAG chains in the patient cells showed a lower molecular mass compared to the controls. To define the role of CANT1 in the etiology of DBQD, we generated a Cant1 knock-in mouse reproducing the R300H mutation detected in patients with DBQD type 1. Mutant mice are smaller in size compared to wild type littermates reproducing the growth defects observed in patients. Morphometric analyses demonstrated significantly shorter and thinner tibiae, femurs and ilia compared to wild-type animals. Dbqd mice at P14 and P21 reproduced also the typical anomalies of extremities described in DBQD type 1 patients: additional carpal ossification centers, bone dislocations and the delta phalanx of the great toe. In conclusion mutant mice develops a skeletal phenotype very reminiscent of DBQD type 1, thus validating the animal model.
Nowadays animal models are useful tools to elucidate the molecular mechanisms underlying the genetic diseases, but also to develop therapeutic strategies. The dtd mouse is a murine model of Diastrophic Dysplasia (DTD) a recessive chondrodysplasia caused by mutations in the sulfate-chloride antiporter of the cell membrane (SLC26A2), crucial for sulfate uptake and GAG sulfation. Deep phenotyping of the model has suggested that N-acetyl-L-cysteine (NAC), a well-known drug with antioxidant properties, might play a role as an intracellular sulfate source for macromolecular sulfation. Because of the important prenatal phase of skeletal development and growth we administered NAC in the drinking water to pregnant mice to explore a possible transplacental effect on the fetuses. A marked increase of proteoglycan sulfation was observed in dtd newborns from NAC treated pregnancies compared to the placebo group. Morphometric studies of the femur, tibia and ilium after skeletal staining with alcian blue and alizarin red indicated a partial rescue of the abnormal bone morphology in dtd newborns from treated females compared to pups from untreated females. These data provide proof of principle that NAC is a useful source of sulfate for macromolecular sulfation *in vivo* when extracellular sulfate supply is reduced, confirming the potential of therapeutic approaches of DTD with thiol compounds.

Talk 20

NEW ROLES OF CELLULAR CATABOLISM DURING SKELETAL DEVELOPMENT

Carmine Settembre

Dulbecco Telethon Institute (DTI), Telethon Institute of Genetics and Medicine (TIGEM), Naples, Italy

Skeletal growth relies on both biosynthetic and catabolic processes. While the role of the former is clearly established, how the latter contributes to growth-promoting pathways is less understood. Autophagy is a lysosomal pathway deputed to the degradation of a wide variety of structurally diverse substances. Clinical studies indicate that 10% of all genes associated to skeletal disorders encode for lysosomal proteins, suggesting that cellular catabolism plays an important, yet unexplored, role during bone development. We studied the role of lysosome and autophagy during longitudinal bone growth, which is mediated by chondrocyte rate of proliferation, hypertrophic differentiation and extracellular matrix (ECM) deposition in the growth plates. We found that autophagy is induced in growth plate chondrocytes during postnatal development and regulates bone length by controlling the composition of the cartilage ECM. By combining high content microscopy and mouse genetic approaches, we demonstrated that the post-natal induction of autophagy in chondrocytes is mediated by the FGF signaling. These data demonstrate that autophagy is a developmentally regulated process, which is necessary for bone growth, and identify FGF signaling as a crucial regulator of autophagy in chondrocytes. Most importantly, these data indicate that modulation of lysosomal and autophagy by the use of signaling molecules may represent a novel therapeutic approach for the treatment.

NEUROLOGICAL DISORDERS AND DNA DAMAGE ACCUMULATION

Talk 21

DNA DAMAGE RESPONSE ACTIVATION IN THE BRAIN: CAUSES AND CONSEQUENCES

Fabrizio d'Adda di Fagagna

IFOM Foundation – FIRC Institute of Molecular Oncology Foundation, 20139 Milan, Italy; Istituto di Genetica Molecolare, Consiglio Nazionale delle Ricerche, 27100 Pavia, Italy

Nuclear DNA damage triggers a prompt signaling cascade known as the DNA damage response (DDR) that enforces a cell-cycle arrest (checkpoint function) and coordinates DNA repair efforts (DNA repair function).
We will present our recent results suggesting that genomes are not uniformly repairable and portions of our chromosomes show reduced rates of DNA repair and thus accumulate irreparable DNA lesions. We will discuss how this may impact on a number of physiological and pathological conditions, including ageing.
In addition, we will discuss how these events may be relevant in the context of neurobiology studies, and their impact on the biology of neural stem cells.
Finally, we will highlight the emerging evidence linking DDR activation and neurodegenerative disorders.

Talk 22

DISSECTING THE NEUROGENIC ROLE OF CSB PROTEIN AND ITS IMPLICATION IN COCKAYNE SYNDROME

Luca Proietti de Santis

Laboratorio di Genetica Molecolare dell'Invecchiamento, Dipartimento di Scienze Ecologiche e Biologiche, Università degli Studi della Tuscia, Viterbo, Italy

Cockayne syndrome (CS) is a progressive developmental and neurodegenerative disorder resulting in premature death at childhood and cells derived from CS patients display DNA repair and transcriptional defects. CS is caused by mutations in csa and csb genes and patients with csb mutation are more prevalent. A hallmark feature of CS-B patients is neurodegeneration but the precise molecular cause for this defect remains enigmatic. Further, it is not clear whether the neurodegenerative condition is due to loss of CSB mediated functions in adult neurogenesis. In this study, we examined the role of CSB in neurogenesis by using the human neural progenitor cells that have self-renewal and differentiation capabilities. In this model system, stable CSB knock-down dramatically reduced the differentiation potential of human neural progenitor cells revealing a key role for CSB in neurogenesis. Neurite out-

growth, a characteristic feature of differentiated neurons, was also greatly abolished in CSB suppressed cells. In corroboration with this, expression of MAP2 (microtubule-associated protein 2), a crucial player in neuritogenesis, was also impaired in CSB suppressed cells. Consistent with reduced MAP2 expression in CSB depleted neural cells, Tandem Affinity Purification (TAP) and Chromatin immunoprecipitation (ChIP) studies revealed a potential role for CSB in the assembly of transcription complex on MAP2 promoter. Altogether our data led us to conclude that CSB plays a crucial role in coordinated regulation of transcription and chromatin remodeling activities that are required during neurogenesis.

Talk 23

LINKS BETWEEN DNA REPAIR AND CHROMOSOME STRUCTURAL PATHWAYS REVEALED BY CELLULAR MODELS OF THE WARSAW BREAKAGE SYNDROME COHESINOPATHY DISORDER

Takuya Abe, Marco Fumasoni, and Dana Branzei

IFOM, the FIRC institute of Molecular Oncology, Milan, Italy

The Warsaw Breakage syndrome (WABS) cohesinopathy is caused by a mutation in the *DDX11* gene (*CHL1* in *Saccharomyces cerevisiae*), encoding an iron-sulfur family of DNA helicase. Cells derived from WABS patients are distinguished by the rare co-existence of a DNA repair defect and chromosome morphology anomalies that include sister chromatid cohesion defects. However, the molecular mechanisms underlying these defects and whether they are linked remain unclear. Here we used genetically amenable chicken DT40 cells and *Saccharomyces cerevisiae* to establish cellular models of WABS and related replication pathways. We found that inactivation of DDX11 or functionally related factors lead to chromosome arm cohesion and repair defects. In both organisms the repair defects are due to suboptimal ability of cells to undergo a replication-associated recombination-mediated DNA damage tolerance process known as template switching. We further find that cohesin plays a structural role in facilitating both sister chromatid proximity and template switch recombination. On the other hand, both the recombination and cohesion defects associated with mutations in DDX11 are likely to derive instead from altered single stranded DNA metabolism and abnormal replication fork topology. We propose a replication-associated DNA damage tolerance defect associated with fork reversal and sister chromatid proximity alterations as an underlying source of chromosome lesions.

Talk 24

MECHANISMS OF NEURODEGENERATION IN ATM-DEFICIENT ATAXIA-TELANGIECTASIA

Domenico Delia

Fondazione IRCCS Istituto Nazionale Tumori, DOSMM, Milan, Italy

The DNA damage response/repair (DDR) pathway is crucial for preventing neurodegeneration, as attested by the several inherited neuropathological disorders arising from mutations in genes disrupting this pathway. Ataxia-Telangiectasia (A-T), the most prominent among these inherited disorders, results from inactivation of ATM kinase, a central player in DDR signalling and downstream events like cell cycle arrest, DNA processing and repair, chromatin remodeling, transcription, apoptosis. Cerebellar ataxia in A-T is due to Purkinje cell death. Albeit neurodegeneration in A-T has initially been attributed to defective DDR signalling in pre- and post-mitotic neurons, oxidative stress and reduced anti-oxidant defence may equally play a role, given the recently established function of ATM in redox signalling. An underlying mechanism in neurodegeneration is transcriptional decline and downregulation of genes that play a role in synaptic vesicle trafficking and release. Quite notably, ATM deficiency in post-mitotic neurons results in the accumulation of DNA-Topoisomerase 1 covalent complexes (Top1-cc), and this abnormality likely impairs RNA processing and elongation, especially of long transcripts. Overall, while the biochemical function of ATM has been extensively elucidated, it is still unclear what makes neurons, especially Purkinje cells, extremely vulnerable in A-T. A summary of latest findings also obtained from the study of A-T iPSCs-derived pre- and post-mitotic neurons will highlight key neurodegenerative factors linked to faulty DDR.

POSTERS

TELETHON NETWORK OF BIOBANKS

ABSTRACT N. 1

Telethon Research Services		
Principal Investigator	**FILOCAMO MIRELLA**	
Telethon grant N.	**GTB12001**	
Total budget €	**2.500.000**	
Centres: **11**	*Duration (yrs):* **5**	*Starting year:* **2013**
Partners	CHIARA BALDO, STEFANO GOLDWURM, ALESSANDRA RENIERI, ELENA PEGORARO, MAURIZIO MOGGIO, MARINA MORA, GIUSEPPE MERLA, LUISA POLITANO, BARBARA GARAVAGLIA, LUCA SANGIORGI	

TELETHON NETWORK OF GENETIC BIOBANKS (TNGB)

RETE TELETHON DI BIOBANCHE GENETICHE

Baldo Chiara (2), Goldwurm Stefano (3), Renieri Alessandra (4), Pegoraro Elena (5), Moggio Maurizio (6), Mora Marina (7), Merla Giuseppe (8), Politano Luisa (9), Garavaglia Barbara (10), Filocamo Mirella (1)

(1) UOSD Centro di Diagnostica Genetica e Biochimica delle Malattie Metaboliche, Istituto G. Gaslini - Via G. Gaslini 5, 16147 Genova. Tel. 01056362792 - Fax 010383983 - email: mirellafilocamo@ospedale-gaslini.ge.it; info@biobanknetwork.org
(2) SC Laboratorio di Genetica Umana, E.O. Ospedali Galliera, Genova
(3) Parkinson Institute, Istituti Clinici di Perfezionamento, Milano
(4) Medical Genetics, University of Siena; Genetica Medica, Azienda Ospedaliera Universitaria Senese, Siena
(5) Università di Padova, Azienda Ospedaliera Universitaria, Padova
(6) UOD Malattie Neuromuscolari e Rare, Fondazione IRCCS Ca' Granda Ospedale Maggiore Policlinico, Centro Dino Ferrari, Università Studi Milano, Milano
(7) Laboratorio di Biologia Cellulare, UO Malattie Neuromuscolari e Neuroimmunologia, Fond. IRCCS Istituto Neurologico C. Besta, Milano
(8) Medical Genetics Unit, IRCCS Casa Sollievo della Sofferenza, S. Giovanni Rotondo (FG)
(9) Cardiomiologia e Genetica Medica, Dipartimento di Medicina Sperimentale, Seconda Università di Napoli e Azienda Ospedaliera Universitaria SUN, Napoli
(10) UO Neurogenetica Molecolare, Fondazione IRCCS Istituto Neurologico Carlo Besta, Milano

NEUROMUSCULAR DISEASES

ABSTRACT N. 2

Telethon Research Projects - Neuromuscular Diseases		
Principal Investigator	**GAZZERRO ELISABETTA**	
Telethon grant N.	**GEP12046**	
Total budget €	**50.000**	
Centres: **1**	*Duration (yrs):* **1**	*Starting year:* **2012**

DRUG DISCOVERY FOR DYSTROGLYCANOPATHIES VIA LARGE PROMOTER ACTIVATION SCREENING

RICERCA DI NUOVI FARMACI PER LE DISTROGLICANOPATIE TRAMITE UNO SCREENING BASATO SULL'ATTIVAZIONE DEL PROMOTORE DEL GENE LARGE

Assereto Stefania (1), Baratto Serena (1), Massacesi Manuela (1), Galietta Luis (2), Zara Federico (1), Bruno Claudio (3), Minetti Carlo (1), Gazzerro Elisabetta (1)

(1) Unit of Paediatric Neurology, G. Gaslini Institute, L.go Gaslini 5, 16147 Genova, phone: 010-5636803, 010-3538653, elisabettagazzerro@ospedale-gaslini.ge.it
(2) Laboratory of Molecular Genetics, G. Gaslini Institute
(3) Center of Myology and Neurodegenerative Disorders , G. Gaslini Institute

ABSTRACT N. 3

Telethon Research Projects - Neuromuscular Diseases		
Principal Investigator	**CUBELLIS MARIA VITTORIA**	
Telethon grant N.	**GGP12108**	
Total budget €	**191.700**	
Centres: **2**	*Duration (yrs):* **3**	*Starting year:* **2012**
Partners	GIUSEPPINA ANDREOTTI	

PHARMACOLOGICAL CHAPERONES TO CURE GENETIC DISEASES: DEVELOPMENT OF DRUGS AND IDENTIFICATION OF NEW TARGETS

CHAPERONE FARMACOLOGICI PER LA CURA DI MALATTIE GENETICHE: SVILUPPO DI NUOVI FARMACI E INDIVIDUAZIONE DI BERSAGLI

Andreotti Giuseppina (2), Citro Valentina (1), Poziello Angelita (1,2), Cubellis Maria Vittoria (1)

(1) Dipartimento di Biologia, Università "Federico II" - via Cinthia, 80126, Napoli, tel-081679152, fax-081679233 cubellis@unina.it
(2) Istituto di Chimica Biomolecolare – CNR, Pozzuoli-Napoli

ABSTRACT N. 4

Telethon Research Projects - Neuromuscular Diseases		
Principal Investigator	**SALVATORE DOMENICO**	
Telethon grant N.	**GGP11185**	
Total budget €	**213.900**	
Centres: **1**	*Duration (yrs):* **3**	*Starting year:* **2011**

THE INTRACELLULAR CONTROL OF THYROID HORMONE SIGNALING IN MUSCLE STEM CELLS AND IN DUCHENNE MUSCULAR DYSTROPHY

IL CONTROLLO INTRACELLULARE DELL'AZIONE DELL'ORMONE TIROIDEO NELLE CELLULE STAMINALI DEL MUSCOLO E NELLA DISTROFIA MUSCOLARE DI DUCHENNE

Dentice Monica, Ambrosio Raffaele, Di Girolamo Daniela, De Stefano Angela, Porcelli Tommaso, Salvatore Domenico

University of Naples "Federico II" Department of Clinical Medicine and Surgery, Via Pansini 5, 80131 Napoli Italy. Tel +39 081 7463780

ABSTRACT N. 5

Telethon Research Projects - Neuromuscular Diseases		
Principal Investigator	**MUSARÒ ANTONIO**	
Telethon grant N.	**GGP13013**	
Total budget €	**209.400**	
Centres: **1**	*Duration (yrs):* **3**	*Starting year:* **2013**

MODULATION OF DYSTROPHIC MICROENVIRONMENT TO IMPROVE STEM CELL-MEDIATED THERAPY

MODULAZIONE DEL MICROAMBIENTE DISTROFICO PER MIGLIORARE LA TERAPIA CELLULARE MEDIATA DA CELLULE STAMINALI

Pelosi Laura (1), Berardinelli Maria Grazia (1), Nicoletti Carmine (1), Forcina Laura (1), Vizzaccaro Elisa (1), De Benedetti Fabrizio (2), Musarò Antonio (1)

(1) Unità di Istologia ed Embriologia Medica- Sapienza Università di Roma; Via A. Scarpa, 14 Roma 00161. Tel: 06 49766956; Fax: 06 4462854; email: antonio.musaro@uniroma1.it
(2) Ospedale Pediatrico Bambino Gesù, Roma

ABSTRACT N. 6

Telethon Research Projects - Neuromuscular Diseases		
Principal Investigator	**CARETTI GIUSEPPINA**	
Telethon grant N.	**GGP13165**	
Total budget €	**2103.900**	
Centres: **1**	*Duration (yrs):* **1**	*Starting year:* **2013**

ROLE OF THE BROMODOMAIN PROTEIN BRD4 IN THE TRANSCRIPTIONAL REGULATION OF PRO-ATROPHIC GENES, IN THE MOUSE MODEL OF DUCHENNE MUSCULAR DYSTROPHY

RUOLO DELLA BROMODOMAIN PROTEIN BRD4 NELLA REGOLAZIONE TRASCRIZIONALE DI PROTEINE PRO-ATROFICHE NELLA DISTROFIA MUSCOLARE DI DUCHENNE

Segatto Marco, Fittipaldi Raffaella, Fenizia Claudio, Camilli Giulia, Caretti Giuseppina

Dipartimento di Bioscienze, Univ. degli Studi di Milano, Via Celoria 26, tel: 02-50315002, fax: 02-50315044, giuseppina.caretti@unimi.it

ABSTRACT N. 7

Telethon Research Projects - Neuromuscular Diseases		
Principal Investigator	**BOUCHÈ MARINA**	
Telethon grant N.	**GGP13233**	
Total budget €	**237.600**	
Centres: **1**	*Duration (yrs):* **3**	*Starting year:* **2013**

PROTEIN KINASE C THETA AS A NOVEL MOLECULAR TARGET TO COUNTERACT INFLAMMATION IN MUSCULAR DYSTROPHY

LA PROTEINCHINASI C TETA COME NUOVO POSSIBILE BERSAGLIO TERAPEUTICO PER CONTRASTARE LA RISPOSTA INFIAMMATORIA NELLA DISTROFIA MUSCOLARE

Lozanoska-Lochser Biliana (1), Marrocco Valeria (1), Fiore Piera (1), Crupi Annunziata (1), Madaro Luca (2), Bouche Marina (1)

(1) Unit of Histology and Med. Embr., DAHFMO, Sapienza Univ. of Rome, - Via A. Scarpa 14, 00161, Rome, +39 06 49766755, +39 06 4462854, marina.bouche@uniroma1.it
(2) Santa Lucia Foundation, Rome

ABSTRACT N. 8

DTI - Neuromuscular Diseases		
Principal Investigator	**BIRESSI STEFANO AUGUSTO MARIA**	
Telethon grant N.	**TCP13007**	
Total budget €	**517.000**	
Centres: **1**	*Duration (yrs):* **5**	*Starting year:* **2014**

MECHANISMS OF FIBROSIS IN MUSCULAR DYSTROPHIES

MECCANISMI CHE CONTROLLANO LA FIBROSI NELLE DISTROFIE MUSCOLARI

Miyabara Elen H. (2,3), Gopinath Suchitra D. (2), Carlig Poppy M. (2), Rando Thomas A. (2,4), Biressi Stefano (1,2)

(1) Dulbecco Telethon Institute, Centre for Integrative Biology-CIBIO, University of Trento - Via Sommarive 9, Povo, TN 38123, Italy; sbiressi@stanford.edu
(2) Paul F. Glenn Laboratories for the Biology of Aging and Department of Neurology and Neurological Sciences, Stanford University School of Medicine, Stanford, CA, USA
(3) Department of Anatomy, Institute of Biomedical Sciences, University of São Paulo, 2415 Lineu Prestes Av., São Paulo, São Paulo, Brazil
(4) Neurology Service, VA Palo Alto Health Care System, 3801 Miranda Ave, Palo Alto, CA, USA

ABSTRACT N. 9

DTI - Neuromuscular Diseases		
Principal Investigator	**SANDRI MARCO**	
Telethon grant N.	**TCR09003**	
Total budget €	**512.125**	
Centres: **1**	*Duration (yrs):* **5**	*Starting year:* **2010**

DEFINING THE MOLECULAR SIGNATURE OF MUSCLE WASTING. IDENTIFICATION OF THERAPEUTIC TARGETS TO COUNTERACT MUSCLE DEGENERATION

DEFINIZIONE DEI MECCANISMI MOLECOLARI DELLA PERDITA DI MASSA MUSCOLARE. IDENTIFICAZIONE DI NUOVI TARGET TERAPEUTICI PER BLOCCARE LA DEGENERAZIONE MUSCOLARE

Sandri Marco

University of Padova, Venetian Institute of Molecular Medicine, via Orus 2, 35129 Padova, phone 0497923258, fax 0497923250, email: marco.sandri@unipd.it

ABSTRACT N. 10

Telethon Research Projects - Neuromuscular Diseases		
Principal Investigator	**BOZZONI IRENE**	
Telethon grant N.	**GGP11149**	
Total budget €	**305.700**	
Centres: **1**	*Duration (yrs):* **3**	*Starting year:* **2011**

RNA-BASED GENE THERAPY OF DUCHENNE MUSCULAR DYSTROPHY

TERAPIA GENICA DELLA DISTROFIA MUSCOLARE DI DUCHENNE BASATA SULL'USO DI RNA

Martone Julie (1), Ballarino Monica (1), Briganti Francesca (2), Legnini Ivano (1), Cipriano Andrea (1), Bisceglie Lavinia (1), Shamloo Sama (1), Bozzoni Irene (1)

(1) Prof. Irene Bozzoni Dept. of Biology and Biotechnology "Charles Darwin" Sapienza - University of Rome P.le A. Moro 5 00185 Rome Italy (0039) tel 06-49912202, fax (0039) 06-49912500, irene.bozzoni@uniroma1.it
(2) EMBL Heidelberg Meyerhofstraße 1, 69117 Heidelberg, Germany

ABSTRACT N. 11

Telethon Research Projects - Neuromuscular Diseases		
Principal Investigator	**CORBI NICOLETTA**	
Telethon grant N.	**GGP14073**	
Total budget €	**183.500**	
Centres: **2**	*Duration (yrs):* **2**	*Starting year:* **2014**
Partners	ELISABETTA MATTEI	

INNOVATIVE THERAPEUTIC STRATEGY FOR DUCHENNE MUSCULAR DYSTROPHY BY AAV MEDIATED DELIVERY OF ARTIFICIAL TRANSCRIPTION FACTOR GENES

STRATEGIA TERAPEUTICA INNOVATIVA PER LA CURA DELLA DISTROFIA MUSCOLARE DI DUCHENNE, BASATA SULL'USO DI FATTORI TRASCRIZIONALI ARTIFICIALI VEICOLATI NEL MUSCOLO DA VETTORI VIRALI ADENO-ASSOCIATI

Passananti Claudio (1), Di Certo Maria Grazia (2), Strimpakos Georgios (2), Pisani Cinzia (1), Onori Annalisa (1), Luvisetto Siro (2), Se-

verini Cinzia (2), Gabanella Francesca (2), Monaco Lucia (3), Mattei Elisabetta (2), Corbi Nicoletta (1)

(1) Institute of Molecular Biology and Pathology CNR, c/o Department of Molecular Medicine, Sapienza University, Viale Regina Elena 291, 00161 Rome, Italy. Tel. +0039.06.49255125. Email: nicoletta.corbi@uniroma1.it
(2) Institute of Cell Biology and Neurobiology CNR, Rome
(3) Department of Physiology and Pharmacology, Sapienza University, Rome

ABSTRACT N. 12

Telethon Research Projects - Neuromuscular Diseases		
Principal Investigator	**COSSU GIULIO**	
Telethon grant N.	**GSP11002**	
Total budget €	**822.000**	
Centres: **1**	*Duration (yrs):* **2**	*Starting year:* **2011**

CELL THERAPY OF DUCHENNE MUSCULAR DYSTROPHY BY INTRA-ARTERIAL DELIVERY OF HLA-IDENTICAL ALLOGENEIC MESOANGIOBLASTS

ALLO-TRAPIANTO DI MESOANGIOBLASTI DA DONATORE HLA-IDENTICO PER LA TERAPIA CELLULARE DELLA DISTROFIA MUSCOLARE DI DUCHENNE

Previtali Stefano C. (1), Sara Napolitano (1), Maria Pia Cicalese (1), F. Saverio Tedesco (1,2), Francesca Nicastro (1), Maddalena Noviello (1), Urmas Roostalu (3), Maria Grazia Natali Sora (1), Marina Scarlato (1), Maurizio De Pellegrin (1), Claudia Godi (1), Serena Giuliani (1), Francesca Ciotti (1), Rossana Tonlorenzi (1), Isabella Lorenzetti (1), Cristina Rivellini (1), Alessandro Ambrosi (1), Sara Benedetti (2), Roberto Gatti (1), Sarah Marktel (1), Andrea Tettamanti (1), Martina Ragazzi (2), Rossana Fiori (1), Maria Pia Sormani (4), Chiara Bonini (1), Massimo Venturini (1), Letterio S Politi (1), Yvan Torrente (5), Fabio Ciceri (1), Giulio Cossu (1,2,3)

(1) Ospedale San Raffaele - Via Olgettina 58. 20132 Milano 3358444282; cossu.giulio@hsr.it
(2) University College London
(3) University of Manchester
(4) University of Genoa
(5) University of Milan

ABSTRACT N. 13

Telethon Research Projects - Neuromuscular Diseases		
Principal Investigator	**BIANCHI MARIA LUISA**	
Telethon grant N.	**GUP11011**	
Total budget €	**380.900**	
Centres: **1**	*Duration (yrs):* **3**	*Starting year:* **2012**

EVALUATION OF BONE TURNOVER, BONE METABOLISM, BONE DENSITY, AND FRACTURES IN CHILDREN WITH DUCHENNE MUSCULAR DYSTROPHY AND POSSIBLE SIDE EFFECTS OF LONG-TERM CORTICOSTEROID THERAPY (BON-DMD)

VALUTAZIONE DI TURNOVER OSSEO, METABOLISMO OSSEO, DENSITÀ OSSEA E FRATTURE NEI BAMBINI AFFETTI DA DISTROFIA MUSCOLARE DI DUCHENNE E DEI POSSIBILI EFFETTI DI UNA TERAPIA STEROIDEA CRONICA

Bianchi Maria Luisa, Vai Silvia, Broggi Francesca

Laboratorio sperimentale di ricerche sul metabolismo osseo infantile, Centro Malattie Metaboliche Ossee - Istituto Auxologico Italiano IRCCS, via L. Ariosto 13, 20145 Milano, tel 02619112505, fax 02619112519; e-mail ml.bianchi@auxologico.it

ABSTRACT N. 14

Telethon Research Projects - Neuromuscular Diseases		
Principal Investigator	**MESSINA SONIA**	
Telethon grant N.	**GUP09010**	
Total budget €	**189.000**	
Centres: **10**	*Duration (yrs):* **2**	*Starting year:* **2010**
Partners	GIUSEPPE VITA, ENRICO SILVIO BERTINI, ANGELA LUCIA BERARDINELLI, CARLO MINETTI, LUISA POLITANO, TIZIANA MONGINI, ELENA PEGORARO, LUCIA OVIDIA MORANDI, MARIA GRAZIA D'ANGELO	

OUTCOME MEASURES IN DUCHENNE MUSCULAR DYSTROPHY: VALIDATION OF THE PEDIATRIC QUALITY OF LIFE INVENTORY TM NEUROMUSCULAR MODULE IN THE ITALIAN POPULATION AND CORRELATION WITH OTHER FUNCTIONAL ASSESSMENTS

MISURE DI OUTCOME NELLA DISTROFIA MUSCOLARE DI DUCHENNE: VALIDAZIONE DEL PEDIATRIC QUALITY OF LIFE INVENTORY TM NEUROMUSCULAR MODULE NELLA POPOLAZIONE ITALIANA E CORRELAZIONI CON ALTRE VALUTAZIONI FUNZIONALI

Mercuri Eugenio (1), Bertini Enrico (2), Vita Giuseppe (3), Berardinelli Angela Lucia (4), Minetti Carlo (5), Politano Luisa (6), Mongini Tiziana (7), Pegoraro Elena (8), Morandi Lucia (9), D'Angelo Maria Grazia (10), Messina Sonia (1,3)

(1) Department of Paediatrics Policlinico Gemelli Università Cattolica Sacro Cuore Roma - Department of Neurosciences University of Messina, Via Consolare Valeria N.1, Pad. B 2°piano. tel. 0902217191, fax: 0902217065; smessina@unime.it
(2) Dept of Laboratories, Bambino Gesu' Children's Hospital, Unit of Molecular Medicine for Neuromuscular and Neurodegenerative Disorders Rome
(3) Dipartimento di Neuroscienze, Scienze Psichiatriche e Anestesiologiche, Università di Messina, Messina
(4) IRCCS "C. Mondino" Institute, Unit of Child Neuropsychiatry, University of Pavia, Pavia
(5) Laboratory of Muscle Pathology, Muscular and Neurodegenerative Disease Unit, Department of Neuroscience and Rehabilitation, Giannina Gaslini Institute; Department of Pediatrics, University of Genova, Genova
(6) Dipartimento di Medicina Sperimentale, Seconda Università di Napoli, Napoli
(7) Centro per le Malattie Neuromuscolari Paolo Peirolo, Dipartimento di Neuroscienze, Torino
(8) Department of Neurosciences/University of Padova, Padova
(9) Muscle Pathology and Neuroimmunology Neurological Institute "Carlo Besta"
(10) IRCCS E Medea Bosisio Parini NeuroMuscular Unit Department of Neuro Rehabilitation Bosisio Parini

ABSTRACT N. 15

Telethon Research Projects - Neuromuscular Diseases		
Principal Investigator	**PANE MARIKA**	
Telethon grant N.	**GUP11002**	
Total budget €	**296.601**	
Centres: **12**	*Duration (yrs):* **3**	*Starting year:* **2012**
Partners	ENRICO SILVIO BERTINI, TIZIANA MONGINI, ANGELA LUCIA BERARDINELLI, GIUSEPPE VITA, GIOVANNI BARANELLO, MARINA PEDEMONTE, ANTONELLA PINI, MARIA GRAZIA D'ANGELO, CHIARA FIORILLO, ELENA PEGORARO, LUISA POLITANO	

ASSESSMENT OF UPPER LIMB FUNCTION IN NON AMBULANT DUCHENNE MUSCULAR DYSTROPHY

VALUTAZIONE DELLA FUNZIONE DEGLI ARTI SUPERIORI IN PAZIENTI NON DEAMBULANTI AFFETTI DA DISTROFIA MUSCOLARE DI DUCHENNE

Pane Marika (1), Mazzone Elena (1), Fanelli Lavinia(1), De Sanctis Roberto (1), Palermo Concetta (1), Sivo Serena (1), D'Amido Adele (2), Messina Sonia (3), Politano Luisa (4), Battini Roberta (5), Pedemonte Marina (6), Tiziana Mongini (7), Pegoraro Elena (8) Berardinelli Angela (9), D'Angelo Maria Grazia (10), Pini Antonella (11), Baranello Giovanni (12)

(1) Child Neurology and Psychiatry Unit, Rome; - Policlinico Gemelli Roma 347/9347864 marika.pane@rm.unicatt.it, 06/30154363
(2) Department of Laboratory Medicine, Unit of Molecular Medicine, Bambino Gesù Hospital, Rome;
(3) Department of Neurosciences, Psychiatry and Anaesthesiology, University of Messina
(4) Department of experimental Medicine, University of Naples;
(5) Department of Developmental Neuroscience, Stella Maris Institute, University of Pisa
(6) Neuromuscular Disease Unit, G. Gaslini Institute, Genoa;
(7) Neuromuscular Center, SG. Battista Hospital, University of Turin
(8) Department of Neurosciences, University of Padua
(9) IRCCS "C. Mondino" Foundation, University of Pavia
(10) IRCCS Eugenio Medea, Bosisio Parini
(11) Child Neurology and Psychiatry Unit, Maggiore Hospital, Bologna
(12) Pediatric Neurology and Neuroradiology Units, Neurological Institute C. Besta, Milan;

ABSTRACT N. 16

Telethon Research Projects - Neuromuscular Diseases		
Principal Investigator	**MAGLIANO LORENZA**	
Telethon grant N.	**GUP10002**	
Total budget €	**145.000**	
Centres: **9**	*Duration (yrs):* **2**	*Starting year:* **2011**
Partners	LUISA POLITANO, UMBERTO BALOTTIN, GIUSEPPE VITA, MARIKA PANE, ADELE D'AMICO, CORRADO ANGELINI, ROBERTA BATTINI, MARIA GRAZIA D'ANGELO	

THE FAMILIES OF CHILDREN WITH MUSCULAR DYSTROPHIES: BURDEN, SOCIAL NETWORK AND PROFESSIONAL SUPPORT

LA FAMIGLIA DEI PAZIENTI AFFETTI DA DISTROFIE MUSCOLARI: CARICO, RETE SOCIALE E SUPPORTO PROFESSIONALE

Politano Luisa (2), Scutifero Marianna (2), Zaccaro Antonella (2), Balottin Umberto (3), Berardinelli Angela (3), Camia Michela (3), Motta Maria Chiara (3), Vita Giuseppe (4), Messina Sonia (4), Sframeli Maria (4), Vita Gian Luca (4), Pane Marika (5), Lombardo Maria Elena (5), Scalise Roberta (5), D'Amico Adele (6), Catteruccia Michela (6), Colia Giulia (6), Angelini Corrado (7), Gaiani Alessandra (7), Semplicini Claudio (7), Battini Roberta (8), Astrea Guja (8), D'Angelo Maria Grazia (9), Brighina Erika (9), Civati Federica (9), Patalano Melania (10), Sagliocchi Alessandra (10), Magliano Lorenza (1)

(1) Psychology Department, Second University of Naples (SUN) - Viale Ellittico, 31, 81100 Caserta; phone/fax 0823275358 lorenza.magliano@unina2.it
(2) Cardiomyology and Medical Genetics, Experimental Medicine Dept., Second University of Naples (SUN)
(3) Child Neuropsychiatry Dept., IRCCS "Mondino" Pavia
(4) Neurosciences, Psychiatry and Anaesthesiology Dept., Messina University
(5) Paediatric Neurology Dept., Catholic University, Rome
(6) Neuromuscular and Neurodegenerative Disorders Unit, Bambino Gesù Children Hospital, Rome
(7) Neurosciences Dept., Padova University
(8) Neurology Dept., Pisa University
(9) NeuroMuscular Unit-NeuroRehabilitation Dept., IRCCS E. Medea, Bosisio Parini (Lc)
(10) Psychology Department, Second University of Naples (SUN)

ABSTRACT N. 17

Telethon Research Projects - Neuromuscular Diseases		
Principal Investigator	**TUPLER ROSSELLA GINEVRA**	
Telethon grant N.	**GUP13012**	
Total budget €	**598.550**	
Centres: **15**	*Duration (yrs):* **3**	*Starting year:* **2014**
Partners	EMILIANO GIARDINA, TIZIANA MONGINI, MAURIZIO MOGGIO, CORRADO ANGELINI, LUCIO SANTORO, GABRIELE SICILIANO, LUCIA OVIDIA MORANDI, ENZO RICCI, CARMELO RODOLICO, ANTONIO DI MUZIO, ANGELA LUCIA BERARDINELLI, MASSIMILIANO FILOSTO, GAETANO NICOLA VATTEMI, GIOVANNI ANTONINI	

PHENOTYPIC AND MOLECULAR CHARACTERIZATION OF FSHD FAMILIES: A SYSTEMATIC APPROACH TOWARDS TRIAL READINESS

CARATTERIZZAZIONE CLINICA E MOLECOLARE DI FAMIGLIE FSHD COME PRESUPPOSTO PER VALUTARE L'EFFICACIA DI TERAPIE

Ricci Giulia (1,3), Nikolic Ana (1), Sera Francesco (4), Govi Monica (1), Mele Fabiano (1), Daolio Jessica (1), Ruggiero Lucia (5), Vercelli Liliana (6), D'Amico Roberto (23), Berardinelli Angela (7), Angelini Corrado (8), Antonini Giovanni (9), Bucci Elisabetta (9), Filosto Massimiliano (10), Cao Michelangelo (11), Giardina Emiliano (12), Pegoraro Elena (11), Di Muzio Antonio (13), D'Amico Maria Chiara (13), Maggi Lorenzo (14), Morandi Lucia (14), Ricci Enzo (15), Portaro Simona (16), Rodolico Carmelo (16), Villa Luisa (17), Testolin Silvia (17), Siciliano Gabriele (3), Santoro Lucio (5), Tomelleri Giuliano (18), Enzo Ricci (19), Grazia D'Angelo (20), Bruno Claudio (21), Maioli Maria Antonietta (22), Mongini Tiziana (6), Moggio Maurizio (17), Rossella Tupler (1,2)

(1) Department of Life Sciences, University of Modena and Reggio Emilia, Modena - Via G. Campi 287, 41124 Modena, tel: 059-2055440-5171, fax: 059-2055426. rossella.tupler@unimore.it
(2) Program in Gene Function and Expression, University of Massachusetts Medical School, Worcester, MA 01605, USA
(3) University of Pisa, Pisa, Italy
(4) Department of Population Health Sciences, Faculty of Population Health Sciences, London UK
(5) University Federico II, Naples, Italy
(6) University of Turin, Turin, Italy
(7) IRCCS "C. Mondino" Foundation, Pavia, Italy
(8) IRCCS San Camillo Venice, Italy
(9) University of Rome "Sapienza", Rome, Italy
(10) University of Brescia, Italy
(11) University of Padua, Padua, Italy
(12) IRCCS Santa Lucia Foundation, Rome, Italy
(13) University "G. d'Annunzio", Chieti, Italy
(14) IRCCS Foundation, C. Besta Neurological Institute, Milano, Italy
(15) Università Cattolica Policlinico A. Gemelli, Rome, Italy
(16) University of Messina, Messina, Italy
(17) IRCCS Fondazione Ospedale Maggiore Policlinico, University of Milan, Milan, Italy
(18) University of Verona, Verona, Italy
(19) Universita` Cattolica of Rome, Rome, Italy

(20) IRCCS E. Medea, Bosisio Parini, Italy
(21) Gaslini Hospital, Genova, Italy
(22) University of Cagliari, Cagliari, Italy
(23) Unit of Statistics, University of Modena and Reggio Emilia, Modena, Italy

ABSTRACT N. 18

DTI - Neuromuscular Diseases		
Principal Investigator	**GABELLINI DAVIDE**	
Telethon grant N.	**TCR13001**	
Total budget €	**750.000**	
Centres: **1**	*Duration (yrs):* **3**	*Starting year:* **2014**

FSHD MUSCULAR DYSTROPHY PROVIDES A MOLECULAR UNDERSTANDING OF THE REPETITIVE (EPI)GENOME

LA DISTROFIA MUSCOLARE FSHD FORNISCE UNA COMPRENSIONE MOLECOLARE DELL'(EPI)GENOMA RIPETUTO

Giambruno Roberto (1,2), Caccia Valentina (1,2,3), Ferri Giulia (1,2,3), Micheloni Stefano (1,2), Mathivanan Jothi (1,2), Cabianca Daphne (1,2), Huber Johannes (1,2), Warner Stephanie (1,2), Gabellini Davide (1)

(1) Dulbecco Telethon Institute - via Olgettina 58, 20132, Milano; tel 0226435934; fax 0226435544; email gabellini.davide@hsr.it
(2) Division of Regenerative Medicine, San Raffaele Scientific Institute, Milano
(3) Cellular and Molecular Biology PhD Program, Vita-Salute San Raffaele University, Milano

ABSTRACT N. 19

Telethon Research Projects - Neuromuscular Diseases		
Principal Investigator	**FIORILLO CHIARA**	
Telethon grant N.	**GEP12019**	
Total budget €	**35.300**	
Centres: **1**	*Duration (yrs):* **1**	*Starting year:* **2012**

EXPLORING MITOCHONDRIAL DYSFUNCTION IN CALPAIN-3 RELATED MUSCULAR DYSTROPHY

ANALISI DELLA FUNZIONE MITOCONDRIALE NELLE DISTROFIE MUSCOLARI DA DEFICIT DI CALPAINA-3

Fiorillo Chiara (1), Baldacci Jacopo (2), Nesti Claudia (2), Doccini Stefano (2), Meschini Chiara (2), Mora Marina (3), Santorelli Filippo (2)

(1) Paediatric Neurology and Neuromuscular Disorders, G. Gaslini Paediatric Hospital, Genoa, Italy - Largo Gaslini 5, 16147, Genoa, Italy, 01056363566, 0103538265, patologiamuscolare@ospedale-gaslini.ge.it
(2) Molecular Medicine and Neuromuscular Disorders, Irccs Stella Maris, Pisa, Italy
(3) Department of Diagnostic And Experimental Research, Istituto Nazionale Neurologico "C. Besta", Milan, Italy

ABSTRACT N. 20

Telethon Research Projects - Neuromuscular Diseases		
Principal Investigator	**BRUNO CLAUDIO**	
Telethon grant N.	**GEP12053**	
Total budget €	**50.000**	
Centres: **1**	*Duration (yrs):* **1**	*Starting year:* **2012**

EXTRACELLULAR ADENOSINE-TRIPHOSPHATE (E-ATP) AND PURINERGIC SIGNALLING IN THE PATHOGENESIS OF ALPHA-SARCOGLYCAN DEFICIENT MUSCULAR DYSTROPHY (LGMD2D)

RUOLO DELL'ADENOSINA TRIFOSFATO (E-ATP) E DEI RECETTORI PURINERGICI NELLA PATOGENESI DELLA DISTROFIA MUSCOLARE DA DEFICIT DI ALFA-SARCOGLICANO (LGMD2D)

Gazzerro Elisabetta (2), Baldassari Simona (2), Assereto Stefania (2), Panicucci Chiara (2), Fiorillo Chiara (2), Minetti Carlo (2), Traggiai Elisabetta (2), Grassi Fabio (4), Claudio Bruno (1,2)

(1) Center of Myology and Neurodegenerative Disorders-Dept. of Neuroscience and Rehabilitation, Istituto Giannina Gaslini - Via G. Gaslini, 5- 16147 Genova. Phone +39-010-56362756; Fax: +39-010-8612070; email: claudio2246@gmail.com
(2) Pediatric Neurology and Muscle Disease Unit, Istituto Giannina Gaslini, Genova
(3) Novartis Institute for Research in Biomedicine, Basel, Switzerland
(4) Department of Medical Biotechnologies & Translational Medicine, University of Milan, Milan, Italy

ABSTRACT N. 21

Telethon Research Projects - Neuromuscular Diseases		
Principal Investigator	**SANDONÀ DORIANNA**	
Telethon grant N.	**GEP12058**	
Total budget €	**49.400**	
Centres: **1**	*Duration (yrs):* **1**	*Starting year:* **2012**

PHARMACOLOGICAL RESCUE OF MISFOLDED PROTEINS: INNOVATIVE APPROACHES FOR THE CURE OF THREE MUSCULAR DISEASES

RECUPERO FARMACOLOGICO DI PROTEINE MAL RIPIEGATE: APPROCCI INNOVATIVI PER LA CURA DI TRE PATOLOGIE MUSCOLARI

Bianchini Elisa (1), Gomiero Chiara (1,2), Dorotea Tiziano (2), Valle Giorgia (1), Mascarello Francesco (2), Betto Romeo (1,3), Volpe Pompeo (1), Sandonà Dorianna (1)

(1) Pathophysiology of Striated Muscle, Department of Biomedical Sciences; University of Padova - Via U. Bassi 58/B, 35123 Padova, Tel. +39 049 8276028, Fax +39 049 8276040, E-mail: dorianna.sandona@unipd.it
(2) Department of Comparative Biomedicine and Food Science, University of Padova
(3) Institute of Neurosciences, Padova, National Research Council of Italy

ABSTRACT N. 22

Telethon Research Projects - Neuromuscular Diseases		
Principal Investigator	**COMI GIACOMO PIETRO**	
Telethon grant N.	**GUP10006**	
Total budget €	**163.600**	
Centres: **8**	*Duration (yrs):* **2**	*Starting year:* **2011**
Partners	VINCENZO NIGRO, CORRADO ANGELINI, TIZIANA MONGINI, MARINA MORA, ANTONIO TOSCANO, GIULIANO TOMELLERI, GABRIELE SICILIANO	

CLINICAL AND LABORATORY NETWORK FOR LGMD DIAGNOSIS, IN VIEW OF A NATIONAL REGISTRY

RETE CLINICA E DI LABORATORIO DELLE DISTROFIE DEI CINGOLI PER STABILIRE UN REGISTRO NAZIONALE

Magri Francesca (1), Nigro Vincenzo (2,3), Angelini Corrado (4), Mongini Tiziana (5), Mora Marina (6), Moroni Isabella (6), Toscano Antonio (7), D'Angelo Maria Grazia (8), Brusa Roberta (1), Tomelleri Giuliano (9), Siciliano Gabriele (10), Comi Giacomo Pietro (1)
(1) Dipartimento di Scienze Neurologiche, Università degli Studi di Milano, Milano - Dipartimento di Scienze Neurologiche, Università degli Studi di Milano, Fondazione IRCCS Ca' Granda Ospedale Maggiore Policlinico, via F. Sforza 35, 20122 Milano. Tel +390255033817, FAX: +390255033800; e-mail: giacomo.comi@unimi.it

(2) Dipartimento di Patologia Generale, Seconda Università degli Studi di Napoli, Napoli
(3) Telethon Institute of Genetics and Medicine (TIGEM), Napoli
(4) Dipartimento di Neuroscienze, Università di Padova, Padova
(5) Dipartimento di Neuroscienze, AOU S. Giovanni Battista di Torino, Torino
(6) Divisione di Malattie Neuromuscolari e Neuroimmunologia, Istituto Neurologico C. Besta, Milano
(7) Dipartimento di Neuroscienze, Università di Messina, Messina
(8) IRCCS E. Medea Bosisio Parini, Lecco
(9) Dipartimento di Scienze Neurologiche, Verona
(10) Dipartimento di Neuroscienze, Università di Pisa, Pisa

ABSTRACT N. 23

Telethon Research Projects - Neuromuscular Diseases		
Principal Investigator	**NIGRO VINCENZO**	
Telethon grant N.	**GUP11006**	
Total budget €	**240.800**	
Centres: **1**	*Duration (yrs):* **3**	*Starting year:* **2012**

GENETIC DIAGNOSIS OF ITALIAN LGMD PATIENTS BY NGS TECHNOLOGY

DIAGNOSI GENETICA DEI PAZIENTI ITALIANI CON DISTROFIA MUSCOLARE DEI CINGOLI BASATA SU SEQUENZIAMENTO DI PROSSIMA GENERAZIONE (NGS)

Marco Savarese (1,2), Annalaura Torella (1,2), Giuseppina Di Fruscio (1,2), Margherita Mutarelli (2), Marina Fanin (3), Chiara Fiorillo (4), Lucia Ruggiero (5), Arca Garofalo (1,2), Teresa Giugliano (1,2), Manuela Dionisi (2), Ombretta De Concilio (1,2), Francesca Del Vecchio Blanco (1,2), Giulio Piluso (1,2), Olimpia Musumeci (6), Gabriele Siciliano (7), Marina Mora (8), Giorgio Tasca (9), Adele D'Amico (10), Lucia Morandi (8), Enrico Bertini (10), Claudio Minetti (4), Giulia Ricci (7), Filippo Maria Santorelli (11), Sabrina Sacconi (12), Massimo Santoro (5), Sandra Janssens (13), Kathleen Claes (13), Tiziana Mongini (14), Corrado Angelini (3), Luisa Politano (15), Claudio Bruno (6), Giacomo Pietro Comi (16), Vincenzo Nigro (1,2)

(1) Dipartimento di Biochimica, Biofisica e Patologia Generale, Seconda Università di Napoli, via Luigi de Crecchio, 7 80138 Napoli, tel/fax 0815665704 vinnigro@gmail.com
(2) Telethon Institute of Genetics and Medicine, Pozzuoli
(3) Dipartimento di Neuroscienze, Università di Padova, Padova
(4) U.O.C. Neurologia Pediatrica e Malattie Muscolari, IRCCS Istituto Giannina Gaslini, Genova
(5) Dipartimento di Neuroscienze e Scienze riproduttive ed odontostomatologiche, Università degli Studi di Napoli "Federico II", Napoli
(6) Dipartimento di Neuroscienze, Università degli Studi di Messina, Messina
(7) Dipartimento di Medicina Clinica e Sperimentale, Università degli Studi di Pisa, Pisa
(8) Dipartimento di Neuroscienze, Istituto Besta, Milano
(9) Istituto di Neurologia, Università Cattolica del Sacro Cuore, Roma
(10) Dipartimento di Neuroscienze, IRCCS Ospedale Pediatrico Bambino Gesù, Roma
(11) Medicina Molecolare, IRCCS Fondazione Stella Maris, Pisa
(12) Neuromuscular Diseases Specialized Center, Archet 1 Hospital, Nice, France
(13) Center for Medical Genetics, Ghent University Hospital, Ghent, Belgium
(14) S.S. Malattie Neuromuscolari, Università degli Studi di Torino, Torino
(15) Dipartimento di Medicina Sperimentale, Seconda Università di Napoli, Napoli
(16) Dipartimento di Fisiopatologia Medico-Chirurgica e dei Trapianti, Università degli Studi di Milano, Milano

ABSTRACT N. 24

Telethon Research Projects - Neuromuscular Diseases		
Principal Investigator	**PETRUZZELLA VITTORIA**	
Telethon grant N.	**GEP12025**	
Total budget €	**41.840**	
Centres: **1**	*Duration (yrs):* **1**	*Starting year:* **2012**

IDENTIFICATION OF THE GENE DETERMINING LIMB GIRDLE MUSCULAR DYSTROPHY TYPE 1H

IDENTIFICAZIONE DEL GENE RESPONSABILE DELLA DISTROFIA MUSCOLARE DEI CINGOLI 1H

Bianco Angelica (1), Bisceglia Luigi (2), Santorelli M. Filippo (3), De Bonis Patrizia (2), Amati Angela (4), Zoccolella Stefano (4), Petruzzella Vittoria (1)

(1) Dipartimento di Scienze Mediche di Base, Neuroscienze e Organi di Senso - Università degli Studi "Aldo Moro", Piazza G. Cesare, 70124, Bari - Tel.: 080 5448529 Fax: 080 5448538 Email: vittoria.petruzzella@uniba.it
(2) Ospedale Casa Sollievo della Sofferenza IRCCS, UOC Genetica Medica, San Giovanni Rotondo, Italia
(3) IRCCS Fondazione "Stella Maris", Pisa
(4) Azienda Ospedaliera-Universitaria Policlinico Consorziale, Raggruppamento di Scienze Neurologiche, Policlinico di Bari

ABSTRACT N. 25

Telethon Research Projects - Neuromuscular Diseases		
Principal Investigator	**FIMIA GIAN MARIA**	
Telethon grant N.	**GEP12072**	
Total budget €	**50.000**	
Centres: **1**	*Duration (yrs):* **1**	*Starting year:* **2012**

IS THE LIMB-GIRDLE MUSCULAR DYSTROPHY TYPE 2H A DEFECTIVE AUTOPHAGY DISEASE?

È UN DIFETTIVO PROCESSO AUTOFAGICO ALLA BASE DELLA DISTROFIA MUSCOLARE DEI CINGOLI 2H?

Di Rienzo Martina (1,2), Romagnoli Alessandra (1), Fusco Carmela (3), Antonioli Manuela (1,2), Piacentini Mauro (1,2), Merla Giuseppe (3), Fimia Gian Maria (1,4)

(1) Department of Epidemiology and Preclinical Research, National Institute for Infectious Diseases IRCCS "L. Spallanzani", Rome, Italy - Via Portuense 292, Rome, Italy. Tel: +390655170908. Fax: +39065582825 email: gianmaria.fimia@inmi.it
(2) Department of Biology, University of Rome "Tor Vergata", Rome, Italy
(3) Medical Genetics Unit, IRCCS "Casa Sollievo della Sofferenza", San Giovanni Rotondo, Italy
(4) Department of Biological and Environmental Sciences and Technologies, University of Salento, Lecce, Italy

ABSTRACT N. 26

Telethon Research Projects - Neuromuscular Diseases		
Principal Investigator	**MERCURI EUGENIO**	
Telethon grant N.	**GUP11001**	
Total budget €	**203.300**	
Centres: **13**	*Duration (yrs):* **2**	*Starting year:* **2012**
Partners	ADELE D'AMICO, SONIA MESSINA, LUISA POLITANO, FILIPPO M. SANTORELLI , CLAUDIO BRUNO, PATRIZIA BOFFI, MASSIMO CORBO, ELENA PEGORARO, ANGELA LUCIA BERARDINELLI, GIACOMO PIETRO COMI, ANTONELLA PINI, ISABELLA MORONI	

DEVELOPMENT OF A REGISTRY AND A DATABASE FOR A NATION-WIDE ITALIAN COLLABORATIVE NETWORK ON CONGENITAL MUSCULAR DYSTROPHY

SVILUPPO DI UN DATA BASE SULLE DISTROFIE MUSCOLARI CONGENITE NEL CONTESTO DI UN NETWORK COLLABORATIVO NAZIONALE PER RICOSTRUIRE ELEMENTI DI STORIA NATURALE DI QUESTE MALATTIE

Mercuri Eugenio (1), Graziano Alessandra (1), Bianco Flaviana (1), D'Amico Adele (2), Moroni Isabella (3), Messina Sonia (4), Bruno Claudio (5), Pegoraro Elena (6), Mora Marina (3), Magri Francesca (8), Battini Roberta (7), Berardinelli Angela (9), Moggio Maurizio (8), Morandi Lucia (3), Pini Antonella (10), Minetti Carlo (5), Mongini Tiziana (12), Tasca Giorgio (2), Gornji Ksenia (13), Comi Giacomo (8), Villanova Marcello (14), Politano Luisa (11), Gualandi Francesca (15), Ferlini Alessandra (15), Muntoni Francesco (16), Santorelli Filippo (7), Bertini Enrico (3), Pane Marika (1)

(1) Departments of Paediatrics and Neurology, Catholic University, Rome, Italy - Department of Child Neurology, Policlinico Gemelli, Largo Gemelli 00168, Roma, Italy. Tel.: +39 06 30155340; fax: +39 06 30154363. E-mail address: mercuri@rm.unicatt.it
(2) Unit of Neuromuscular and Neurodegenerative Disorders. Dept of Neurosciences, Bambino Gesù Children's Hospital
(3) Pediatric Neurology and Neuromuscular Disease and Immunology Unit Istituto Neurologico "Besta" Milan Italy
(4) Department of Neurosciences, Psychiatry and Anaesthesiology, University of Messina, Messina, Italy
(5) Neuromuscular Disease Unit, G. Gaslini Institute, Genoa, Italy
(6) Department of Neurosciences, University of Padua, Padua, Italy
(7) Department of Developmental Neuroscience and Molecular Medicine Neuromuscular Unit, Stella Maris Institute, Pisa, Italy
(8) Department of Neurological Sciences, IRCCS Ospedale Maggiore Policlinico, University of Milan, Italy
(9) Child Neurology and Psychiatry Unit, IRCCS "C. Mondino" Foundation, University of Pavia, Italy
(10) Child Neurology and Psychiatry Unit, IRCCS Istituto delle Scienze Neurologiche di Bologna, Bellaria Hospital, Bologna, Italy
(11) Cardiomiologia e Genetica Medica, Dipartimento di Medicina Sperimentale, Seconda Università di Napoli, Italy
(12) Neuromuscular Center, S.G. Battista Hospital, University of Turin
(13) Centro Nemo, Milan, Italy
(14) Neuromuscular Unit, Nigrisoli Hospital, Bologna, Italy
(15) Section of Medical Genetics, Department of Experimental and Diagnostic Medicine, Ferrara, Italy
(16) Dubowitz Neuromuscular Centre, UCL Institute of Child Health & Great Ormond Street Hospital for Children, London, UK

ABSTRACT N. 27

Telethon Research Projects - Neuromuscular Diseases		
Principal Investigator	**D'AMICO ADELE**	
Telethon grant N.	**GUP13004**	
Total budget €	**267.100**	
Centres: **12**	*Duration (yrs):* **2**	*Starting year:* **2014**
Partners	EUGENIO MERCURI, GUJA ASTREA, SONIA MESSINA, CLAUDIO BRUNO, ELENA PEGORARO, ANGELA LUCIA BERARDINELLI, GIACOMO PIETRO COMI, MARINA MORA, ANTONELLA PINI, LUISA POLITANO, FEDERICA RICCI	

COMPLETE MOLECULAR CHARACTERIZATION OF PATIENTS AFFECTED BY CONGENITAL MUSCULAR DYSTROPHIES WITH ALPHA-DYSTROGLYCAN DEFECT USING NEXT GENERATION SEQUENCING STRATEGIES

COMPLETA CARATTERIZZAZIONE GENETICA DI PAZIENTI AFFETTI DA DISTROFIA MUSCOLARE CONGENITA CON DIFETTO DI GLICOSILAZIONE DELL'ALFA-DISTROGLICANO APPLICANDO STRATEGIE DI NEXT-GENERATION SEQUENCING

Mercuri Eugenio (2), Astra Guja (3), Messina Sonia (4), Bruno Claudio (5), Pegoraro Elena (6), Berardinelli Angela (7), Comi Pietro Giacomo (8), Mora Marina (9), Pini Antonella (10), Politano Luisa (11), Ricci Federica (12), D'Amico Adele (1)

(1) Unit of Neuromuscular and Neurodegenerative, Dep.of neurosciences, IRCCS, Bambino Gesù Children's Hospital - Viale S. Paolo 5, 00146 Rome, Italy, phone: +390668592105, fax +390668592024, adele2.damico@opbg.net
(2) Dep. of Child Neuology, Università Cattolica del Sacro Cuore, Rome
(3) Molecular Medicine and Neurogenetics- IRCCS Fondazione, Stella Maris, Calambrone
(4) Department of Neurosciences, University of Messina
(5) Center of Myology and Neurodegenerative Disease-Istituto Giannina Gaslini, Genova
(6) Neuromuscular Center, Department of Neurosciences, University of Padova
(7) Unit of Child Neurology and Psychiatry, Istitute "C. Mondino", Pavia
(8) Dino Ferrari Centre, Ospedale Maggiore Policlinico, Milan
(9) Neuromuscular Disease Unit, Neurological Institute Carlo Besta, Milan
(10) Child Neurology Unit, IRCCS Institute of Neurological Sciences, Bologna
(11) Department of Experimental Medicine, Cardiomyology and Medical Genetics, Second University, Napoli
(12) Department of Neuroscience Rita Levi Montalcini, University of Torino

ABSTRACT N. 28

Telethon Research Projects - Neuromuscular Diseases		
Principal Investigator	**PREVITALI STEFANO CARLO**	
Telethon grant N.	**GGP12024**	
Total budget €	**290.900**	
Centres: **1**	*Duration (yrs):* **3**	*Starting year:* **2012**

ROLE OF JAB1 IN THE CONTROL OF NERVE DEVELOPMENT AND REPAIR: IMPLICATION IN THE PATHOGENESIS OF MEROSIN DEFICIENT CONGENITAL MUSCULAR DYSTROPHY (MDC1A)-ASSOCIATED HEREDITARY NEUROPATHIES

RUOLO DI JAB1 NEL CONTROLLO DELLO SVILUPPO E RIGENERAZIONE DEL NERVO PERIFERICO: IMPLICAZIONE NELLA PATOGENESI DELLE NEUROPATIE EREDITARIE ASSOCIATE ALLA DISTROFIA MUSCOLARE CONGENITA (MDC1A)

Porrello Emanuela, Rivellini Cristina, Previtali Stefano Carlo

Inspe and Division of Neuroscience, IRCCS San Raffaele Scientific Institute - Via Olgettina 60, 20132 Milano. Tel 0226433036. Fax 0226435093. Email previtali.stefano@hsr.it

ABSTRACT N. 29

Telethon Research Projects - Neuromuscular Diseases		
Principal Investigator	**FIUMARA FERDINANDO**	
Telethon grant N.	**GEP12087**	
Total budget €	**44.100**	
Centres: **1**	*Duration (yrs):* **1**	*Starting year:* **2012**

STRUCTURE-GUIDED THERAPEUTIC APPROACHES FOR OCULOPHARYNGEAL MUSCULAR DYSTROPHY (OPMD) AND RELATED DISEASES

NUOVI APPROCCI TERAPEUTICI GUIDATI DALLA STRUTTURE PROTEICA PER LA DISTROFIA MUSCOLARE OCULOFARINGEA E PATOLOGIE CORRELATE

Cibelli Marica, Pelassa Ilaria, Fiumara Ferdinando
Dipartimento di Neuroscienze, Università degli Studi di Torino - ferdinando.fiumara@unito.it

ABSTRACT N. 30

Telethon Research Projects - Neuromuscular Diseases		
Principal Investigator	**MERLINI LUCIANO**	
Telethon grant N.	**GUP11007**	
Total budget €	**77.300**	
Centres: **1**	*Duration (yrs):* **2**	*Starting year:* **2012**

LOW-PROTEIN DIET TO CORRECT DEFECTIVE AUTOPHAGY IN PATIENTS WITH COLLAGEN VI RELATED MYOPATHIES

LA DIETA IPOPROTEICA PER CORREGGERE IL DIFETTO AUTOFAGICO NEI PAZIENTI CON MIOPATIE DA DEFICIT DI COLLAGENE TIPO VI

Bonaldo Paolo (6), Cocchi Daniela (7), Pellegrini Massimo (3), Sabatelli Patrizia (2,1), Sandri Marco (4,5,8), Merlini Luciano (1)

(1) SC Laboratory of Musculoskeletal Cell Biology, Rizzoli Orthopedic Institute , Bologna - Istituto Ortopedico Rizzoli, via di Barbiano 1/10, 40136 Bologna, T: 051-6366755, F: 051-5835937, luciano.merlini@ior.it
(2) Institute of Molecular Genetics, CNR-National Research Council of Italy , Bologna
(3) Cattedra di Scienze Tecniche Dietetiche Applicate, Università di Modena e Reggio Emilia, Modena
(4) Department of Biomedical Science, University of Padova, Padova
(5) Dulbecco Telethon Institute, Venetian Institute of Molecular Medicine, Padova
(6) Department of Molecular Medicine, University of Padova, Padova
(7) Dipartimento di Scienze Statistiche Paolo Fortunati, Università di Bologna, Bologna
(8) Telethon Institute of Genetics and Medicine (TIGEM), Napoli

ABSTRACT N. 31

Telethon Research Projects - Neuromuscular Diseases		
Principal Investigator	**BERNARDI PAOLO**	
Telethon grant N.	**GGP14037**	
Total budget €	**293.100**	
Centres: **1**	*Duration (yrs):* **2**	*Starting year:* **2014**

A MITOCHONDRIAL THERAPY FOR MUSCULAR DYSTROPHIES

UNA TERAPIA MITOCONDRIALE PER LE DISTROFIE MUSCOLARI

Bernardi Paolo

Dipartimento di Scienze Biomediche, Università di Padova - Via Ugo Bassi 58/B, I-35131 Padova, tel 049 827 6365, fax 049 827 6049, bernardi@bio.unipd.it

ABSTRACT N. 32

Telethon Research Projects - Neuromuscular Diseases		
Principal Investigator	**CECCONI FRANCESCO**	
Telethon grant N.	**GGP14202**	
Total budget €	**378.400**	
Centres: **2**	*Duration (yrs):* **2**	*Starting year:* **2014**
Partners	PAOLO BONALDO	

MANIPULATING AUTOPHAGY IN MUSCLE DISEASES

MODULAZIONE DELL'AUTOFAGIA NELLE MALATTIE MUSCOLARI

Bonaldo Paolo (2), Di Bartolomeo Sabrina (1), Grumati Paolo (2), Castagnaro Silvia (2), Nazio Francesca (1,3), Chrisam Martina (2), Cianfanelli Valentina (1,5), Skobo Tatjana (4), Dalla Valle Luisa (4), Cecconi Francesco (1,3,5)
(1) Department of Biology, University of Rome Tor Vergata, Rome, Italy - Via della Ricerca Scientifica, 00133 Rome, Italy. Tel. +39 06 72594230, +39 06 501703092; Fx. +39 06 72594222; email: francesco.cecconi@uniroma2.it
(2) Department of Molecular Medicine, University of Padova, Padova, Italy
(3) IRCCS Fondazione Santa Lucia, 00143 Rome, Italy
(4) Department of Biology, University of Padova, Padova, Italy
(5) Unit of Cell Stress and Survival, Danish Cancer Society Research Center, Copenhagen, Denmark

ABSTRACT N. 33

Telethon Research Projects - Neuromuscular Diseases		
Principal Investigator	**DI BLASI CLAUDIA**	
Telethon grant N.	**GEP12074**	
Total budget €	**50.000**	
Centres: **1**	*Duration (yrs):* **1**	*Starting year:* **2012**

ASSESSMENT OF THE PATHOGENIC ROLE OF A MISSENSE VARIANT IN A BENIGN AUTOSOMAL DOMINANT MYOPATHY WITH HYPERCKAEMIA

VALUTAZIONE DEL RUOLO PATOGENETICO DI UNA MUTAZIONE MISSENSO IN UNA MIOPATIA BENIGNA AUTOSOMICA DOMINANTE CON IPERCKEMIA

Mora Marina (1), Claudia Di Blasi (1), Serena Sansanelli (1), Alessandra Ruggieri (1), Manuela Moriggi (2), Michele Vasso (2,3), Adamo Pio D'Adamo (4), Flavia Blasevich (1), Simona Zanotti (1), Cecilia Paolini (5), Feliciano Protasi (5), Frediano Tezzon (6), Cecilia Gelfi (2,3), Lucia Morandi (1), Mauro Pessia (7)

(1) Neuromuscular Diseases and Neuroimmunology Unit, Foundation IRCCS Neurological Institute C. Besta, Milano, Italy - Muscle Cell Biology Lab, Neuromuscular Diseases and Neuroimmunology Unit Neurological Institute "C. Besta", Via Temolo 4, 20126 Milano, Italy Tel: +39-022394632; Fax: +39-022394619; Email: mmora@istituto-besta.it
(2) Department of Biomedical Sciences for Health, University of Milano, Milano, Italy
(3) CNR-Institute of Bioimaging and Molecular Physiology, Segrate, Milano, Italy
(4) Medical Genetics, University of Trieste, Trieste, Italy
(5) CeSI, Center for Research on Ageing & Department of Neuroscience, Imaging, and Clinical Sciences, University G. D'Annunzio of Chieti, Italy
(6) Neurology Unit, F. Tappeiner Hospital of Merano, Merano, Italy
(7) Faculty of Medicine, Section of Physiology and Biochemistry, Department of Experimental Medicine, University of Perugia, Perugia, Italy

ABSTRACT N. 34

Telethon Research Projects - Neuromuscular Diseases		
Principal Investigator	**PROTASI FELICIANO**	
Telethon grant N.	**GGP13213**	
Total budget €	**510.300**	
Centres: **3**	*Duration (yrs):* **3**	*Starting year:* **2013**
Partners	CARLO REGGIANI, VINCENZO SORRENTINO	

ALTERED CALCIUM HANDLING IN CENTRAL CORE DISEASE AND MALIGNANT HYPERTHERMIA: UNDERSTAND MOLECULAR

MECHANISMS AND GENETIC BACKGROUND TO DEVELOP INNOVATIVE THERAPEUTIC INTERVENTIONS

MIOPATIA CENTRAL CORE E IPERTERMIA MALIGNA: COMPRENDERNE I MECCANISMI MOLECOLARI E LE BASI GENETICHE PER SVILUPPARE TERAPIE FARMACOLOGICHE INNOVATIVE

Michelucci Antonio (1), De Marco Alessandro (1), Canato Marta (2), Rossi Daniela (3), Paolini Cecilia (1), Boncompagni Simona (1), Sorrentino Vincenzo (3), Reggiani Carlo (2), Protasi Feliciano (1)

(1) CeSI & DNICS, Università G. d'Annunzio di Chieti-Pescara - CeSI-Centro Scienze dell'invecchiamento; Via Luigi Polacchi 66100 Chieti (tel.: 0871 541423; e-mail: fprotasi@unich.it).
(2) Dept. of Neuroscience, Università di Padova
(3) Dept. of Molec. Med. & Dev, Università di Siena

ABSTRACT N. 35

Telethon Research Projects - Neuromuscular Diseases		
Principal Investigator	**ZORZATO FRANCESCO**	
Telethon grant N.	**GGP14003**	
Total budget €	**80.000**	
Centres: **1**	*Duration (yrs):* **2**	*Starting year:* **2014**

JP45 A FUNCTIONAL MODIFIER IN RYANODINOPATHIES

JP45 UN MODULATORE DEL FENOTIPO CAUSATO DA MUTAZIONI DEL GENE CODIFICANTE IL RECETTORE DELLA RIANODINA DEL MUSCOLO SCHELETRICO (RIANODINOPATIE)

Streye Monika (2), Vincze Janos (2), Csernoch Laszlo (2), Treves Susan (1,3), Zorzato Francesco (1,3)

(1) Dipartimento di Scienze della Vita e Biotecnologie, Sezione di Patologia Generale, Via Borsari 46 ,44100 Ferrara 0532455352 zor@unife.it
(2) Department of Physiology, University of Debrecen, Debrecen Hungary
(3) Deparment of Anesthesiology, University Hospital Basel, Basel, Switzerland

ABSTRACT N. 36

Telethon Research Projects - Neuromuscular Diseases		
Principal Investigator	**SZABADKAI GYORGY**	
Telethon grant N.	**GEP12066**	
Total budget €	**49.500**	
Centres: **1**	*Duration (yrs):* **1**	*Starting year:* **2012**

THE ROLE OF INOSITOL 1,4,5-TRISPHOSPHATE MEDIATED CA2+ SIGNALS IN CORE MYOPATHIES

IL RUOLO DEL SEGNALE DI CALCIO NUCLEARE INDOTTO DAL INOSITOLO 1,4,5-TRISFOSAFATO NELLO SVILUPPO DELLA MIOPATIA CONGENITA 'CENTRAL CORE'

Gyorgy Szabadkai, Matteo Suman

Department of Biomedical Sciences University of Padua - Via Ugo Bassi, 58/B 35131 Padua, Italy T.: +39 049 827 6359

ABSTRACT N. 37

Telethon Research Projects - Neuromuscular Diseases		
Principal Investigator	**CARRA SERENA**	
Telethon grant N.	**GEP12008**	
Total budget €	**50.000**	
Centres: **1**	*Duration (yrs):* **1**	*Starting year:* **2012**

CHARACTERIZATION OF THE R7S MUTATION OF HEAT SHOCK PROTEIN HSPB3 AND OF TWO NOVEL MUTATIONS FOUND IN PATIENTS SUFFERING OF CONGENITAL MYOPATHY: UNDERSTANDING THE MECHANISMS LEADING TO DISEASE

CARATTERIZZAZIONE DELLA MUTAZIONE R7S DELLA PROTEINA HSPB3 E DI ALTRE DUE NUOVE MUTAZIONI TROVATE IN PAZIENTI AFFETTI DA MIOPATIA CONGENITA: COMPRENSIONE DEI MECCANISMI GENETICI E MOLECOLARI

Morelli Federica F (1), Heldens Lonneke (1), Verbeek Dineke (2), Angelini Corrado (3), Cenacchi Giovanna (4), Tupler Rossella (5), Carra Serena (1)

(1) Department of Biomedical, Neuronal and Metabolic Sciences, University of Modena and Reggio Emilia, Modena, Italy - via Giuseppe Campi, 287, 41125 Modena, Italy; tel: 0592055265; fax: 0592055363; e-mail: serena.carra@unimore.it
(2) Department of Genetics, University Medical Center Groningen, University of Groningen, Groningen, The Netherlands
(3) IRCCS San Camillo, Lido di Venezia, Italy
(4) Department of Biomedical and Neuromotor Sciences, "Alma Mater Studiorum", University of Bologna, Bologna, Italy
(5) Department of Life Sciences, University of Modena and Reggio Emilia, Modena, Italy

ABSTRACT N. 38

Telethon Research Projects - Neuromuscular Diseases		
Principal Investigator	**VAZZA GIOVANNI**	
Telethon grant N.	**GEP12083**	
Total budget €	**43.000**	
Centres: **1**	*Duration (yrs):* **1**	*Starting year:* **2012**

IDENTIFICATION OF NEW GENES INVOLVED IN DISTAL MYOPATHY AND DHMN

IDENTIFICAZIONE DI GENI RESPONSABILI DI MIOPATIA DISTALE E DHMN

Gregianin E. (1), Salvoro C. (1), Crippa V. (2), Rusmini P. (2), Poletti A. (2), Pegoraro E. (3), Angelini C. (3), Petrucci A. (4), Cavallaro T. (5), Fabrizi GM. (5), Boaretto F. (1), Mostacciuolo ML. (1), Vazza G. (1)

(1) Department of Biology, University of Padova, Italy - via U. Bassi 58/b, 35131 Padova Tel: 049 8276226, Fax: 049 8276209, Email: giovanni.vazza@unipd.it
(2) Department of Pharmacological and Biomolecular Sciences, University of Milan
(3) Department of Neurosciences, University of Padova
(4) Neuromuscular and Rare Neurological Diseases Centre Neurology & Neurophysiopathology Unit, ASO San Camillo-Forlanini Hospital of Rome
(5) Section of Neuropathology, Neurological and Movement Sciences, University of Verona, Italy

ABSTRACT N. 39

DTI - Neuromuscular Diseases		
Principal Investigator	**ZITO ESTER**	
Telethon grant N.	**TCP12001**	
Total budget €	**517.000**	
Centres: **1**	*Duration (yrs):* **5**	*Starting year:* **2013**

DISSECTING THE MOLECULAR BASIS OF SEPN1-RELATED-MYOPATHIES

ANALISI DELLE BASI MOLECOLARI DELLE MIOPATIE CORRELATE A SEPN1

Zito Ester (1), Marino Marianna (1), Giorgi Carlotta (2), Bachi Angela (3), Cattaneo Angela (3), Auricchio Alberto (4), Pinton Paolo (2)

(1) Dulbecco Telethon Institute at IRCCS-Istituto di Ricerche Farmacologiche Mario Negri, Milan, Italy - Ester Zito, Dulbecco Telethon Assistant Scientist, IRCCS-Istituto di Ricerche Farmacologiche Mario Negri Via La Masa 19, 20156 Milano, Italy Tel: +39 0239014480 FAX:+39023546277 E-mail: ester.zito@marionegri.it
(2) Section of Pathology, Oncology and Experimental Biology, Laboratory for Technologies of Advanced Therapies (LTTA), Department of Morphology, Surgery and Experimental Medicine, University of Ferrara, Ferrara, Italy
(3) IFOM-FIRC Institute of Molecular Oncology, Milan, Italy
(4) Telethon Institute of Genetics and Medicine (TIGEM), Naples, and Medical Genetics, Department of Translational Medicine, Federico II University, Naples, Italy

ABSTRACT N. 40

Telethon Research Projects - Neuromuscular Diseases		
Principal Investigator	**CONTE CAMERINO DIANA**	
Telethon grant N.	**GGP14096**	
Total budget €	**176.500**	
Centres: **1**	*Duration (yrs):* **3**	*Starting year:* **2014**

PRECLINICAL EVALUATION OF PHARMACOGENETICS AND NEW THERAPEUTIC OPTIONS IN NON-DYSTROPHIC MYOTONIAS TOWARD PERSONALIZED MEDICINE

VALUTAZIONE PRECLINICA DI FARMACOGENETICA E NUOVE OPZIONI TERAPEUTICHE NELLE MIOTONIE NON-DISTROFICHE VERSO UNA MEDICINA PERSONALIZZATA

Desaphy Jean-François, Imbrici Paola, Roussel Julien, Altamura Concetta, Farinato Alessandro, Pierno Sabata, Liantonio Antonella, De Bellis Michela, Conte Elena, Fonzino Adriano, Camerino Giulia Maria, Conte Camerino Diana

Dipartimento di Farmacia - Scienze del Farmaco, Università degli Studi di bari - Aldo Moro - via Orabona 4 - campus, 70125 Bari, tel/fax: 080 544 2801, e-mail: diana.conte@uniba.it

ABSTRACT N. 41

Telethon Research Projects - Neuromuscular Diseases		
Principal Investigator	**MARTELLI FABIO**	
Telethon grant N.	**GGP14092**	
Total budget €	**345.300**	
Centres: **2**	*Duration (yrs):* **3**	*Starting year:* **2014**
Partners	GERMANA FALCONE	

SKELETAL MUSCLE AND CIRCULATING MICRORNAS IN MYOTONIC DYSTROPHY TYPE 1

RUOLO DEI MICRORNA NEL MUSCOLO SCHELETRICO E NEL SANGUE CIRCOLANTE DEI MALATI DI DISTROFIA MIOTONICA DI TIPO 1

Alessandra Perfetti (1), Simona Greco (1), Paola Fuschi (1), Giovanni Meola (1), Rosanna Cardani (1), Fabio Martelli (1), Beatrice Cardinali (2), Marisa Cappella (2), Claudia Provenzano (2), Germana Falcone (2), Jose Manuel Garcia-Manteiga (3), Davide Cittaro (3), Marco Bianchi (3)

(1) Gruppo Ospedaliero San Donato Foundation c/o Policlinico San Donato-IRCCS, via Morandi 30, San Donato Milanese, Milano; T: +390252774533; F: +390252774666; E: fabio.martelli@gruppo-sandonato.it
(2) Istituto di Biologia Cellulare e Neurobiologia, Consiglio Nazionale delle Ricerche, Monterotondo Scalo, Roma
(3) Center for Translational Genomics and Bioinformatics, San Raffaele Scientific Institute, Milan, Italy

ABSTRACT N. 42

Telethon Research Projects - Neuromuscular Diseases		
Principal Investigator	**TIZIANO FRANCESCO DANILO**	
Telethon grant N.	**GGP12116**	
Total budget €	**313.400**	
Centres: **2**	*Duration (yrs):* **2**	*Starting year:* **2012**
Partners	LUCIA DI MARCOTULLIO	

MUSCULAR MIRNOME AND TRANSCRIPTOME ANALYSIS AS A TOOL FOR THE IDENTIFICATION OF BIOMARKERS IN SPINAL MUSCULAR ATROPHY

ANALISI DI MIRNOMA E TRASCRITTOMA MUSCOLARI COME STRUMENTO PER L'IDENTIFICAZIONE DI BIOMARCATORI NELL'ATROFIA MUSCOLARE SPINALE

Di Marcotullio Lucia (2), Infante Paola (3), Abiusi Emanuela (1), Diano Federica (1), Di Pietro Lorena (1), D'Amico Davide (2), Fiori Stefania (1), Pasanisi Barbara (4), Bussolino Chiara (5), Mora Marina (4), Baranello Giovanni (5), Moroni Isabella (6), Tramontano Anna (7), Grassi Luigi (7), Lepera Loredana (3), D'Amico Adele (8), Morandi Lucia (4), Bertini Enrico (8), Tiziano Francesco Danilo (1)

(1) Istituto di Genetica Medica, Università Cattolica del Sacro Cuore, Roma - Largo Francesco Vito 1, Roma - Tel: 0630154606 - FAX: 0630157223 - fdtiziano@rm.unicatt.it
(2) Dipartimento di Medicina Molecolare, Università La Sapienza, Roma
(3) Istituto Italiano Tecnologie, Roma
(4) UO Patologia Muscolare-Neuroimmunologia, Fondazione Istituto Neurologico "Carlo Besta", Milano
(5) U.O. Neurologia dello sviluppo, Fondazione Istituto Neurologico "Carlo Besta, Milano
(6) U.O. Neuropsichiatria infantile, Fondazione Istituto Neurologico "Carlo Besta, Milano
(7) Dipartimento di Fisica, Università La Sapienza, Roma
(8) Unità di Medicina Molecolare, Ospedale Pediatrico Bambino Gesù, Roma

ABSTRACT N. 43

Telethon Research Projects - Neuromuscular Diseases		
Principal Investigator	**BATTAGLIA GIORGIO STEFANO**	
Telethon grant N.	**GGP13081**	
Total budget €	**451.300**	
Centres: **3**	*Duration (yrs):* **3**	*Starting year:* **2013**
Partners	ENRICO GARATTINI, FERDINANDO DI CUNTO	

RELEVANCE OF THE AXONAL SMN PROTEIN (A-SMN) FOR SPINAL MUSCULAR ATROPHY: NOVEL CELL MODELS, TRANSGENIC MICE AND THERAPEUTIC APPROACHES

IMPORTANZA DELLA PROTEINA A-SMN O SMN ASSONALE NELL'ATROFIA MUSCOLARE SPINALE: NUOVI MODELLI CELLULARI, TOPI TRANSGENICI E TERAPIE GENICHE

Locatelli Denise (1), Terao Mineko (2), Zanellati Maria Clara (1), Pletto Daniela (1), Berto Gaia (3), Chiotto Adelaide (3), Pallavicini Gianmarco (3), Di Cunto Ferdinando (3), Turco Emilia (3), Garattini Enrico (2), Battaglia Giorgio Stefano (1)

(1) Molecular Neuroanatomy and Pathogenesis Unit, IRCCS Foundation Neurological Institute "C. Besta" - via Temolo 4, 20126 Milano (Italy). Tel: +39-0223942606; Fax: +39-0223942619; email: giorgio.battaglia@istituto-besta.it
(2) Molecular Biology Lab, Mario Negri Institute of Pharmacological Research, Milan (Italy)
(3) Department of Molecular Biotechnology and Health Sciences, University of Turin, Turin (Italy)

ABSTRACT N. 44

Telethon Research Projects - Neuromuscular Diseases		
Principal Investigator	**RAFFA GRAZIA DANIELA**	
Telethon grant N.	**GGP13147**	
Total budget €	**117.100**	
Centres: **1**	*Duration (yrs):* **2**	*Starting year:* **2013**

A DROSOPHILA MODEL FOR SPINAL MUSCULAR ATROPHY (SMA): IDENTIFICATION AND CHARACTERIZATION OF SMN INTERACTORS AND PHENOTYPIC MODIFIERS

IL MOSCERINO DELLA FRUTTA COME MODELLO PER SMA: IDENTIFICAZIONE E CARATTERIZZAZIONE DI INTERATTORI E MODIFICATORI DELLA PROTEINA SMN

Maccallini Paolo (1), Di Giorgio Maria Laura (1), Di Schiavi Elia (2), Esposito Alessandro (2), Micheli Emanuela (1), Bavasso Francesca (1), Cacchione Stefano (1), Raffa Grazia Daniela (1)

(1) Dipartimento di Biologia e Biotecnologie Sapienza-Università di Roma Piazzale Aldo Moro, 5 00185 Rome, Italy Tel. +39 06 49912 843 mail: graziadaniela.raffa@uniroma1.it
(2) Institute of Biosciences and BioResources, IBBR Department of Biology, Agriculture and Food Science - CNR Via Pietro Castellino 111, 80131 Naples Italy

ABSTRACT N. 45

Telethon Research Projects - Neuromuscular Diseases		
Principal Investigator	**NIZZARDO MONICA**	
Telethon grant N.	**GGP14025**	
Total budget €	**291.500**	
Centres: **1**	*Duration (yrs):* **3**	*Starting year:* **2014**

PEPTIDE-CONJUGATED MORPHOLINO FOR TREATMENT OF SPINAL MUSCULAR ATROPHY

MORFOLINO CONIUGATO CON PEPTIDI PER IL TRATTAMENTO DELL'ATROFIA MUSCOLARE SPINALE

Simone Chiara, Rizzuti Mafalda, Ramirez Agnese, Rizzo Federica, Nizzardo Monica

Università degli Studi di Milano, Dipartimento di Fisiopatologia Medica-Chirurgica e dei Trapianti, sezione Neurologia - via Francesco Sforza 35, 20122 Milano, tel 02 55033833, fax: 02 55033800, email: monica.nizzardo1@gmail.com

ABSTRACT N. 46

Telethon Research Projects - Neuromuscular Diseases		
Principal Investigator	**SETTE CLAUDIO**	
Telethon grant N.	**GGP14095**	
Total budget €	**268.600**	
Centres: **1**	*Duration (yrs):* **3**	*Starting year:* **2014**

SAM68 IS A PHYSIOLOGICAL REGULATOR OF SMN2 SPLICING IN SPINAL MUSCULAR ATROPHY

SAM68 È UN REGOLATORE FISIOLOGICO DELLO SPLICING DEL GENE SMN2 NELL'ATROFIA MUSCOLARE SPINALE

Pagliarini Vittoria (1,2), Bustamante Maria Blaire (1,2), Pelosi Laura (3), Nobili Annalisa (2,4), Berardinelli Maria Grazia (3), D'Amelio Marcello (2,4), Musarò Antonio (3,5), Sette Claudio (1,2)

(1) Department of Biomedicine and Prevention, University of Rome "Tor Vergata" - Via Montpellier, 1 00133, Rome Italy Telephone: 3906 72596260 Fax: 3906 72596268 Email: claudio.sette@uniroma2.it
(2) Laboratories of Neuroembryology and Molecular Neuroscience, Fondazione Santa Lucia, Rome
(3) Institute Pasteur Cenci-Bolognetti, DAHFMO-Unit of Histology and Medical Embryology, IIM, University of Rome La Sapienza, Rome
(4) Medical School University Campus Bio-Medico, Rome
(5) Center for Life Nano Science@Sapienza, Istituto Italiano di Tecnologia, Rome

ABSTRACT N. 47

Telethon Research Projects - Neuromuscular Diseases		
Principal Investigator	**MERCURI EUGENIO**	
Telethon grant N.	**GSP13002**	
Total budget €	**226.000**	
Centres: **13**	*Duration (yrs):* **2**	*Starting year:* **2014**
Partners	ENRICO SILVIO BERTINI, ROBERTA BATTINI, ANGELA LUCIA BERARDINELLI, CLAUDIO BRUNO, MARIA GRAZIA D'ANGELO, KSENIJA GORNI, TIZIANA MONGINI, GIOVANNI BARANELLO, ELENA PEGORARO, LUISA POLITANO, SONIA MESSINA, ANTONELLA PINI	

DEVELOPMENT OF AN ITALIAN CLINICAL NETWORK FOR SPINAL MUSCULAR ATROPHY

CREAZIONE DI UNA RETE CLINICA ITALIANA PER L'ATROFIA MUSCOLARE SPINALE (SMA)

Mercuri Eugenio (1), Mazzone Elena (1), Messina Sonia (2), Berardinelli Angela (3), Bruno Claudio (4), D'Angelo Grazia (5), Gorni Ksenia (6), Mongini Tiziana (7), Morandi Lucia (8), Pegoraro Elena (9), Battini Roberta (10), Politano Luisa (11), Pini Antonella (12), Pane Marika (1), Bertini Enrico (13)

(1) Dipartimento di Scienze Pediatriche, Medico Chirurgiche e di Neuroscienze dello Sviluppo - Università Cattolica del Sacro Cuore, Roma; Tel. +39-06/30155386; Email: eumercuri@gmail.com
(2) Centro NEMO Sud, Università di Messina
(3) Istituto Gianna Gaslini, Genova
(4) IRCSS Fondazione Istituto Neurologico C. Mondino, Pavia
(5) IRCCS Eugenio Medea, Bosisio Parini
(6) Centro Clinico Nemo, Milano
(7) Università di Torino
(8) IRCCS Fondazione Istituto Neurologico C. Besta, Milano
(9) Università di Padova
(10) IRCCS Fondazione Stella Maris, Calambrone (Pisa)
(11) Seconda Università di Napoli
(12) Università di Bologna
(13) Ospedale Pediatrico Bambino Gesù, Roma

ABSTRACT N. 48

Telethon Research Projects - Neuromuscular Diseases		
Principal Investigator	**COMI GIACOMO PIETRO**	
Telethon grant N.	**GGP10062**	
Total budget €	**399.200**	
Centres: **1**	*Duration (yrs):* **3**	*Starting year:* **2010**

DEVELOPMENT OF A THERAPEUTIC APPROACH FOR SPINAL MUSCULAR ATROPHY WITH RESPIRATORY DISTRESS (SMARD1) USING HUMAN INDUCED PLURIPOTENT STEM CELL-DERIVED NEURAL STEM CELLS AND MOTOR NEURONS

SVILUPPO DI UN APPROCCIO TERAPEUTICO PER L'ATROFIA MUSCOLARE SPINALE CON DISTRESS RESPIRATORIO DI TIPO 1 (SMARD1) MEDIATO DA CELLULE STAMINALI NEURONALI E MOTONEURONI DIFFERENZIATI DA CELLULE STAMINALI PLURIPOTENTI INDOTTE

Simone Chiara, Nizzardo Monica, Rizzo Federica, Riboldi Giulietta, Salani Sabrina, Bucchia Monica, Bresolin Nereo, Corti Stefania, Comi Giacomo

Università degli Studi di Milano, IRCCS Fondazione Ca' Granda Ospedeale Maggiore Policlinico, +39 02 55033817, fax 02 55033800, giacomo.comi@unimi.it

ABSTRACT N. 49

Telethon Research Projects - Neuromuscular Diseases		
Principal Investigator	**POLETTI ANGELO**	
Telethon grant N.	**GGP14039**	
Total budget €	**244.700**	
Centres: **1**	*Duration (yrs):* **3**	*Starting year:* **2014**

MOTOR NEURON DEGENERATION IN SPINAL AND BULBAR MUSCULAR ATROPHY: MOLECULAR APPROACHES TO COUNTERACT MUTANT ANDROGEN RECEPTOR NEUROTOXICITY

DEGENERAZIONE DEI MOTONEURONI NELLA ATROFIA MUSCOLARE SPINALE E BULBARE. APPROCCI MOLECOLARI PER CONTRASTARE LA NEUROTOSSICITÀ DEL RECETTORE DEGLI ANDROGENI MUTATO

Rusmini Paola (1,2,3), Giorgetti Elisa (1,2,4), Crippa Valeria (1,2,3), Cristofani Riccardo (1,2,3), Cicardi Maria Elena (1,2,3), Meroni Marco (1,2,3), Galbiati Mariarita (1,2,3), Poletti Angelo (1,2,3)

(1) Dipartimento di Scienze Farmacologiche e Biomolecolari, Università degli Studi di Milano - via Balzaretti 9, 20133 Milano (Italy) ph +390250318215; fax +390250318204; e-mail: angelo.poletti@unimi.it
(2) Centro Interdipartimentale sulle Malattie Neurodegenerative dell'Università degli Studi di Milano
(3) Centro InterUniversitario sulle Malattie Neurodegenerative Università di Firenze, Roma "Tor Vergata", Genova e Milano
(4) Department of Pathology, University of Michigan, Ann Arbor, Michigan, USA

ABSTRACT N. 50

DTI - Neuromuscular Diseases		
Principal Investigator	**PENNUTO MARIA**	
Telethon grant N.	**TCP12013**	
Total budget €	**517.000**	
Centres: **1**	*Duration (yrs):* **5**	*Starting year:* **2013**

TARGETING AKT SIGNALING IN MUSCLE TO IDENTIFY NEW THERAPEUTIC STRATEGIES FOR SPINAL AND BULBAR MUSCULAR ATROPHY

RUOLO DEI SEGNALI INTRACELLULARI ATTIVATI NEL MUSCOLO DALLA CINASI AKT NELLA PATOGENESI DELLA ATROFIA MUSCOLARE SPINALE E BULBARE

Milioto Carmelo (1), Rocchi Anna (2), Chivet Mathilde (1), Parodi Sara (2,3), Armirotti Andrea (6), Urciolo Anna (4), Molon Sibilla (4), Bonaldo Paolo (4), Giorgetti Elisa (7), Lieberman Andrew (7), Vergani Lodovica (5), Soraru' Gianni (5), Pennuto Maria (1)

(1) Dulbecco Telethon Institute Lab of Neurodegenerative Diseases, CIBIO, University of Trento, Italy, - Via Sommarive 9, 38123 Trento, Italy Phone: Office +39 0461 285215 Phone: Lab +39 0461 283732 Fax: +39 0461 283937
(2) Department of Neuroscience and Brain Technologies, Istituto Italiano di Tecnologia, Genoa, Italy
(3) Neurogenetics Branch, NINDS, NIH, Bethesda MD, US
(4) Department of Histology, Microbiology & Medical Biotechnology, University of Padova, Padova, Italy
(5) Department of Neurosciences, University of Padova, Padova, Italy
(6) Drus Discovery Department, Istituto Italiano di Tecnologia, Genoa, Italy
(7) Michigan University Medical School, USA

ABSTRACT N. 51

Telethon Research Projects - Neuromuscular Diseases		
Principal Investigator	**RONCHI DARIO**	
Telethon grant N.	**GEP14049**	
Total budget €	**50.000**	
Centres: **1**	*Duration (yrs):* **1**	*Starting year:* **2015**

PATIENT-SPECIFIC CELLULAR MODELS FOR SPINAL MUSCULAR ATROPHY WITH PROGRESSIVE MYOCLONIC EPILEPSY: FROM PATHOGENESIS TO THERAPEUTICS DEVELOPMENT FOR A NEGLECTED NEUROLOGICAL DISORDER

MODELLI CELLULARI PAZIENTE-SPECIFICI PER L'AMIOTROFIA SPINALE CON MIOCLONO EPILESSIA PROGRESSIVA: DALLA PATOGENESI ALLO SVILUPPO DI TERAPIE PER UNA MALATTIA NEUROLOGICA NEGLETTA

Ronchi Dario

Centro Dino Ferrari per le malattie neuromuscolari e neurodegenerative, via Francesco Sforza 35, Milano

ABSTRACT N. 52

Telethon Research Projects - Neuromuscular Diseases		
Principal Investigator	**VITA GIUSEPPE**	
Telethon grant N.	**GUP10008**	
Total budget €	**170.300**	
Centres: **8**	*Duration (yrs):* **2**	*Starting year:* **2011**
Partners	LUCA PADUA, DAVIDE PAREYSON, ANGELO SCHENONE , GIAN MARIA FABRIZI, LUCIO SANTORO, FRANCO GEMIGNANI, ALDO QUATTRONE	

NOVEL OUTCOME MEASURES FOR CHARCOT-MARIE-TOOTH DISEASE

NUOVE MISURE DI OUTCOME NELLA MALATTIA DI CHARCOT-MARIE-TOOTH

Vita Giuseppe (1,2), Padua Luca (3), Pareyson Davide (4), Schenone Angelo (5), Fabrizi Gian Maria (6), Santoro Lucio (7), Gemignani Franco (8), Quattrone Aldo (9), Mazzeo Anna (1), Russo Massimo (2), Stancanelli Claudia (1), Gentile Luca (1), Pazzaglia Costanza (3), Iacovelli Chiara (3), Simbolotti Chiara (3), Piscosquito Giuseppe (4), Calabrese Daniela (4), Aiello Alessia (5), Bolla Simone (5), Cavallaro Tiziana (6), Manganelli Fiore (7), Pisciotta Chiara (7), Vitetta Francesca (8), Contini Mara (8), Valentino Paola (9)

(1) Dipartimento di Neuroscienze, Università di Messina, AOU Policlinico "G. Martino", Messina 98125 - Tel: 0902212793; Fax: 0902212789; vitag@unime.it
(2) Centro Clinico Nemo Sud, Messina
(3) Fondazione Don Carlo Gnocchi ONLUS, Centro Santa Maria della Pace, Roma
(4) Fondazione IRCCS Istituto Neurologico "Carlo Besta", Milano
(5) Dipartimento di Neuroscienze, Riabilitazione, Oftalmologia, Genetica e Scienze Materno Infantili, Università di Genova
(6) Dipartimento di Scienze Neurologiche e del Movimento, Università di Verona
(7) Dipartimento di Neuroscienze e Scienze Riproduttive ed Odontostomatologiche, Università "Federico II", Napoli
(8) Dipartimento di Neuroscienze, Università di Parma
(9) Dipartimento di Scienze Mediche e Chirurgiche, Università "Magna Graecia", Catanzaro

ABSTRACT N. 53

Telethon Research Projects - Neuromuscular Diseases		
Principal Investigator	**FERRARIN MAURIZIO**	
Telethon grant N.	**GUP10010**	
Total budget €	**283.775**	
Centres: **4**	*Duration (yrs):* **3**	*Starting year:* **2011**
Partners	DAVIDE PAREYSON, LUCA PADUA, MANUELA DIVERIO	

DEVELOPMENT OF AN INSTRUMENTED MOVEMENT ANALYSIS PROTOCOL FOR THE MULTI-TASKING ANALYSIS OF LOCOMOTOR FUNCTIONS IN ADULT AND YOUNG PATIENTS WITH CHARCOT-MARIE-TOOTH DISEASE: MULTICENTER STUDY TO CHARACTERISE RELIABILITY AND RESPONSIVENESS

SVILUPPO DI UN PROTOCOLLO STRUMENTALE DI ANALISI DEL MOVIMENTO PER L'ANALISI MULTI-TASKING DELLE FUNZIONI LOCOMOTORIE NELLA MALATTIA DI CHARCOT-MARIE-TOOTH NELL'ETÀ EVOLUTIVA E NELL'ADULTO: CARATTERIZZAZIONE DELLA RELIABILITY E RESPONSIVENESS TRAMITE STUDIO MULTICENTRICO

Lencioni Tiziana (1), Rabuffetti Marco (1), Piscosquito Giuseppe (2), Moroni Isabella (3), Pagliano Emanuela (3), Diverio Manuela (4), Aiello Alessia (5), Beghi Ettore (6), Di Sipio Enrica (7), Padua Luca (7), Pazzaglia Costanza (7), Schenone Angelo (5), Pareyson Davide (2), Ferrarin Maurizio (1)

(1) Biomedical Technology Department, IRCCS Foundation Don Gnocchi Onlus, Milan - Via Capecelatro 66, 20148 Milano, Tel. +39 02 40308305, Fax. +39 02 4048919, E-mail: mferrarin@dongnocchi.it
(2) Unit of Clinic of Central and Peripheral Degenerative Neuropathies, IRCCS Neurological Institute Besta, Milan
(3) Child Neurology Department, IRCCS Neurological Institute Besta, Milan
(4) Polo Riabilitativo del Levante Ligure, Foundation Don Gnocchi Onlus, Sarzana
(5) Department of Neuroscience, Rehabilitation, Ophthalmology, Genetics, and Maternal Infantile Sciences, University of Genoa, Genoa
(6) Lab. of Neurological Disorders, Institute for Pharmacological Research Mario Negri, Milan
(7) Centro S. Maria della Pace, Foundation Don Gnocchi Onlus, Rome

ABSTRACT N. 54

Telethon Research Projects - Neuromuscular Diseases		
Principal Investigator	**PAREYSON DAVIDE**	
Telethon grant N.	**GUP13006**	
Total budget €	**292.000**	
Centres: **10**	*Duration (yrs):* **2**	*Starting year:* **2014**
Partners	ANGELO SCHENONE , GIAN MARIA FABRIZI, STEFANO CARLO PREVITALI, ISABELLA ALLEGRI, LUCA PADUA, LUCIO SANTORO, ALDO QUATTRONE, GIUSEPPE VITA, ISABELLA MORONI	

CMT NATIONAL REGISTRY: TOWARDS DEFINITION OF STANDARDS OF CARE AND CLINICAL TRIALS

REGISTRO NAZIONALE CMT: VERSO LA DEFINIZIONE DEGLI STANDARD DI CURA E LE SPERIMENTAZIONI CLINICHE

Calabrese Daniela (1), Schenone Angelo (2), Fabrizi Gian Maria (3), Santoro Lucio (4), Vita Giuseppe (5), Quattrone Aldo (6), Padua Luca (7), Previtali Stefano (8), Allegri Isabella (9), Filippini Graziella (1), Pareyson Davide (1)

(1) IRCCS Besta Neurol. Inst. Milan - Via Giovanni Celoria 11 - 20133 Milano Tel 02.2394.3001 Fax 02.2394.2293 davide.pareyson@istituto-besta.it
(2) DINOGMI Genoa Univ; IRRCS San Martino IST, Genoa
(3) Neurology Unit, Verona Univ.
(4) Federico II Univ Neurological Sciences Dept, Naples
(5) Neurosciences Dept., Messina Univ. & Clinical Centre NEMO SUD, Messina
(6) Magna Graecia Univ., Neurology Clinic & National Research Council, Catanzaro
(7) Catholic University Neurosciences Dept. & Don Gnocchi Foundation, Rome
(8) INSPE Neurology Dept. IRCCS San Raffaele Milano
(9) Neurosciences Dept., Parma University

ABSTRACT N. 55

Telethon Research Projects - Neuromuscular Diseases		
Principal Investigator	**BORTOLOZZI MARIO**	
Telethon grant N.	**GGP12269**	
Total budget €	**341.400**	
Centres: **1**	*Duration (yrs):* **3**	*Starting year:* **2013**

STRUCTURAL AND FUNCTIONAL ANALYSIS OF SELECTED CONNEXIN32 MUTATIONS IMPLICATED IN THE PATHOGENESIS OF THE X-LINKED FORM OF CHARCOT-MARIE-TOOTH DISEASE

ANALISI STRUTTURALE E FUNZIONALE DI PARTICOLARI MUTAZIONI DELLA CONNESSINA 32 IMPLICATE NELLA PATOGENESI DELLA FORMA X-LINKED DELLA MALATTIA DI CHARCOT-MARIE-TOOTH

Carrer Andrea (2), Leparulo Alessandro (2,3), Crispino Giulia (2,3), Ciubotaru Catalin (2,4), Zonta Francesco (3), Bortolozzi Mario (1,3)

(1) Venetian Institute of Molecular Medicine (VIMM), Via G.Orus 2, 35129, Padova; tel/fax.: 049-7923237/250; mario.bortolozzi@unipd.it
(2) Venetian Institute of Molecular Medicine (VIMM), Via G.Orus 2, 35129, Padova
(3) Università di Padova, Dipartimento di Fisica e Astronomia, Via Marzolo 8, 35131, Padova
(4) CNR - Istituto di Neuroscienze, viale G. Colombo 3, 35121, Padova

ABSTRACT N. 56

Telethon Research Projects - Neuromuscular Diseases		
Principal Investigator	**BRUZZONE SANTINA**	
Telethon grant N.	**GGP12002**	
Total budget €	**115.500**	
Centres: **1**	*Duration (yrs):* **2**	*Starting year:* **2012**

ROLE OF PURINERGIC RECEPTORS IN MYELINATION: THERAPEUTIC IMPLICATIONS FOR TREATMENT OF THE PERIPHERAL NEUROPHATY CHARCOT-MARIE-TOOTH 1A

RUOLO DEI RECETTORI PURINERGICI NELLA MIELINIZZAZIONE: IMPLICAZIONI TERAPEUTICHE PER IL TRATTAMENTO DELLA NEUROPATIA PERIFERICA CHARCOT-MARIE-TOOTH 1A

Sociali Giovanna (1), Davide Visigalli (2), Prukop Thomas (3), Cervellini Ilaria (3), Zocchi Elena (1), Nobbio Lucilla (2), Schenone Angelo (2), Sereda Michael (3), Bruzzone Santina (1)

(1) Department of Experimental Medicine, Section of Biochemistry, and CEBR, University of Genova, Italy - Viale Benedetto XV, 1 16132 Genova tel 0103538161; fax 01035338162; email: santina.bruzzone@unige.it
(2) Department of Neurosciences, Rehabilitation, Ophthalmology,

Genetics and Mother and Child Sciences and CEBR, University of Genova, Italy
(3) Max Planck Institute of Experimental Medicine, Department of Neurogenetics, Göttingen, Germany

ABSTRACT N. 57

Telethon Research Projects - Neuromuscular Diseases		
Principal Investigator	**D'ANTONIO MAURIZIO**	
Telethon grant N.	**GGP14147**	
Total budget €	**314.300**	
Centres: **1**	*Duration (yrs):* **3**	*Starting year:* **2014**

PROTEIN MISFOLDING IN CHARCOT-MARIE-TOOTH DISEASE: TOWARDS THE DEVELOPMENT OF A THERAPEUTIC STRATEGY TARGETING THE UNFOLDED PROTEIN RESPONSE

SCORRETTO RIPIEGAMENTO DELLE PROTEINE NELLA MALATTIA DI CHARCOT-MARIE-TOOTH: VERSO LO SVILUPPO DI UNA STRATEGIA TERAPEUTICA CHE MODULI LA RISPOSTA ALLE PROTEINE NON NATIVE

Florio Francesca, Touvier Thierry, Ferri Cinzia, D'Antonio Maurizio

Division of Genetics and Cell Biology, San Raffaele Scientific Institute, Milan, Italy - Via Olgettina 58, 20132, Milan, Italy; phone: 02-26435307; fax: 02-26434767; email: dantonio.maurizio@hsr.it

ABSTRACT N. 58

Telethon Research Projects - Neuromuscular Diseases		
Principal Investigator	**BOLINO ALESSANDRA**	
Telethon grant N.	**GGP12017**	
Total budget €	**253.000**	
Centres: **1**	*Duration (yrs):* **3**	*Starting year:* **2012**

PHOSPHOLIPID METABOLISM AND MEMBRANE TRAFFICKING IN THE PATHOGENESIS OF CHARCOT-MARIE-TOOTH NEUROPATHIES

RUOLO DEI FOSFOLIPIDI E DEL TRAFFICO DI MEMBRANA NELLA PATOGENESI DELLE NEUROPATIE DI CHARCOT-MARIE-TOOTH

Guerrero Valero Marta (1), Alberizzi Valeria (1), Simons Michael (2), Bolino Alessandra (1)

(1) Human Inherited Neuropathies Unit, Division of Neuroscience, San Raffaele Scientific Institute, Via Olgettina 60, 20132 Milan-Italy - tel 0226434743; fax 0226435093; bolino.alessandra@hsr.it
(2) Max Planck Institute for Experimental Medicine, Goettingen, Germany

ABSTRACT N. 59

Telethon Research Projects - Neuromuscular Diseases		
Principal Investigator	**TAVEGGIA CARLA**	
Telethon grant N.	**GGP14040**	
Total budget €	**260.700**	
Centres: **1**	*Duration (yrs):* **3**	*Starting year:* **2014**

ROLE OF PROSTAGLANDIN D2 SYNTHASE IN PNS MYELINATION AND REMYELINATION

RUOLO DELLA PROSTAGLANDINA D2 SINTASI NELLA MIELINIZZAZIONE E NELLA RIMIELINIZZAZIONE

Trimarco Amelia (1), Forese Maria Grazia (1), Quattrini Angelo (2), Bolino Alessandra (3), Taveggia Carla (1)

(1) Axo-Glial Interaction Unit, Division of Neuroscience and INSPE, San Raffaele Scientific Institute - via Olgettina 60, 20132 Milano, ITALY tel: 02 26434439; fax: 02 26436164; e-mail: taveggia.carla@hsr.it
(2) Experimental Neuropathology Unit, Division of Neuroscience and INSPE, San Raffaele Scientific Institute
(3) Human inherited Neuropathies Unit, Division of Neuroscience and INSPE, San Raffaele Scientific Institute

ABSTRACT N. 60

Telethon Research Projects - Neuromuscular Diseases		
Principal Investigator	**CATTANEO ANTONINO**	
Telethon grant N.	**GGP11179**	
Total budget €	**189.200**	
Centres: **1**	*Duration (yrs):* **3**	*Starting year:* **2011**

TOWARDS AN NGF-BASED THERAPY FOR HEREDITARY SENSORY AND AUTONOMIC NEUROPATHIES IV AND V

SVILUPPO DI UNA TERAPIA A BASE DI NGF PER LE NEUROPATIE EREDITARIE SENSORIE ED AUTONOME DI TIPO IV E V

Testa Giovanna (1), Pancrazi Laura (1), Capsoni Simona (1), Costa Mario (2), Cattaneo Antonino (1)

(1) Laboratorio di Biologia, Scuola Normale Superiore, piazza dei Cavalieri 7, 56126 Pisa, 050509220, 0503153220, antonino.cattaneo@sns.it
(2) Istituto di Neuroscienze, CNR, Pisa

ABSTRACT N. 61

Telethon Research Projects - Neuromuscular Diseases		
Principal Investigator	**PALMIERI FERDINANDO**	
Telethon grant N.	**GGP11139**	
Total budget €	**395.900**	
Centres: **4**	*Duration (yrs):* **3**	*Starting year:* **2011**
Partners	COSTANZA LAMPERTI, PAOLO PINTON, BARBARA MONTI	

THE MITOCHONDRIAL ASPARTATE-GLUTAMATE CARRIER ISOFORM 1 (AGC1) DEFICIENCY: IDENTIFICATION OF NEW PATIENTS AND GENERATION OF BRAIN CELL MODELS THAT REVEAL PROLIFERATION, SURVIVAL AND POTENTIAL INTERCELLULAR CROSS-TALK DEFECTS

LA "DEFICIENZA DEL CARRIER MITOCONDRIALE DI ASPARTATO/GLUTAMMATO ISOFORMA 1 (AGC1)": IDENTIFICAZIONE DI NUOVI PAZIENTI E GENERAZIONE DI MODELLI CELLULARI DI CERVELLO CHE RIVELANO POTENZIALI DIFETTI DI VITALITÀ, PROLIFERAZIONE E COMUNICAZIONE INTERCELLULARE

Monti Barbara (6), Lasorsa Francesco Massimo (2), Pena-Altamira Luis Emiliano (6), Pinton Paolo (5), Giorgi Carlotta (5), Giannuzzi Giulia (1), Profilo Emanuela (1), Falk Marni J. (3), Li Dong (4), Hakonarson Hakon (3), Lamperti Costanza (7), Zeviani Massimo (7,8), Palmieri Ferdinando (1,2)

(1) Department of Biosciences, Biotechnology and Biopharmaceutics, University of the Studies of Bari Aldo Moro - Via E. Orabona, 4 70125 Bari - tel. 0805443323, FAX 0805442770, ferdinando.palmieri@uniba.it
(2) Institute of Biomembranes and Bioenergetics IBBE CNR Bari
(3) Division of Human Genetics, Department of Pediatrics, The Children's Hospital of Philadelphia and The Perelman School of Medicine, Philadelphia, PA USA
(4) The Center for Applied Genomics, Abramson Research Center, The Children's Hospital of Philadelphia, Philadelphia, PA USA
(5) Department of Morphology, Surgery and Experimental Medicine, Section of Pathology, Oncology and Experimental Biology and LTTA center, University of Ferrara, Italy
(6) Department of Pharmacy and Biotechnology, University of Bologna, Italy
(7) Unit of Molecular Neurogenetics, the Carlo Besta Institute of Neurology IRCCS, Milan, Italy
(8) MRC Mitochondrial Biology Unit, Cambridge, UK

ABSTRACT N. 62

Telethon Research Projects - Neuromuscular Diseases		
Principal Investigator	**SALVIATI LEONARDO**	
Telethon grant N.	**GGP13222**	
Total budget €	**202.400**	
Centres: **1**	*Duration (yrs):* **3**	*Starting year:* **2013**

PATHOGENESIS OF PRIMARY AND SECONDARY COENZYME Q DEFICIENCY

PATOGENESI DEL DEFICIT PRIMARIO E SECONDARIO DI COENZIMA Q

Doimo Mara, Desbats Maria Andrea, Cerqua Cristina, Vaquez Fonseca Luis, Acosta Manuel J., Pertegato Vanessa, Morbidoni Valeria, Zordan Roberta, Casarin Alberto, Trevisson Eva, Salviati Leonardo

Clinical Genetics Unit , Dept of Woman and Child Health, University of Padova. - IRP Città della Spèeranza, C.so Stati Uniti 4 35127 Padova,Tel 0498216164, fax 0498211174, e-mail leonardo.salviati@unipd.itmail

ABSTRACT N. 63

Telethon Research Projects - Neuromuscular Diseases		
Principal Investigator	**GHEZZI DANIELE**	
Telethon grant N.	**GGP11011**	
Total budget €	**388.600**	
Centres: **3**	*Duration (yrs):* **3**	*Starting year:* **2011**
Partners	RODOLFO COSTA, ILEANA FERRERO	

MITMED: A MULTICENTER CONSORTIUM FOR THE IDENTIFICATION AND CHARACTERIZATION OF NUCLEAR GENES RESPONSIBLE FOR HUMAN MITOCHONDRIAL DISORDERS

MITMED: UN CONSORZIO MULTICENTRICO PER L'IDENTIFICAZIONE E LA CARATTERIZZAZIONE DI GENI NUCLEARI RESPONSABILI DI MALATTIE MITOCONDRIALI UMANE

Melchionda Laura (1), Invernizzi Federica (1), Nasca Alessia (1), Diodato Daria (1), Lamantea Eleonora (1), Marchet Silvia (1), Lamperti Costanza (1), Baruffini Enrico (2), Dallabona Cristina (2), Donnini Claudia (2), Goffrini Paola (2), Lodi Tiziana (2), Ferrero Ileana (2), De Pittà Cristiano (3), Da Re Caterina (3), Corrà Samantha (3), Zordan Mauro (3), Costa Rodolfo (3), Zeviani Massimo (1,4), Ghezzi Daniele (1)

(1) Unit of Molecular Neurogenetics, Foundation IRCCS Istituto Neurologico "C. Besta" - via Temolo 4 Milano 20126 Italy; phone +39-0223942627; FAX +39-0223942619; dghezzi@istituto-besta.it (2) Department of Life Sciences, University of Parma, Parma, (3) Department of Biology, University of Padova, Padova, (4) Mitochondrial Biology Unit-Medical Research Council, Cambridge, UK

ABSTRACT N. 64

Telethon Research Projects - Neuromuscular Diseases		
Principal Investigator	**SCORRANO LUCA**	
Telethon grant N.	**GGP14187**	
Total budget €	**1.000.600**	
Centres: **4**	*Duration (yrs):* **3**	*Starting year:* **2014**
Partners	VALERIO CARELLI, PAOLO BERNARDI, LEONARDO SALVIATI	

MITCARE-2

MITCARE-2

Scorrano Luca (1,2), Carelli Valerio (3), Bernardi Paolo (4), Salviati Leonardo (5)

(1) Dipartimento di Biologia, Università di Padova
(2) Dulbecco-Telethon Institute, Venetian Institute of Molecular Medicine
(3) Unit of Neurology, Department of Biomedical and NeuroMotor Sciences (DIBINEM), University of Bologna
(4) Dipartimento di Scienze Biomediche, Università di Padova
(5) Department of Woman and Child Health, Università di Padova

ABSTRACT N. 65

Telethon Research Projects - Neuromuscular Diseases		
Principal Investigator	**VERGANI LODOVICA**	
Telethon grant N.	**GGP10145**	
Total budget €	**161.000**	
Centres: **1**	*Duration (yrs):* **3**	*Starting year:* **2010**

ROLE OF MITOCHONDRIAL DYNAMIC AND AUTOPHAGY IN THE SEGREGATION OF MUTANT MTDNA

RUOLO DELLA DINAMICA MITOCONDRIALE E DELLA AUTOFAGIA NELLA SEGREGAZIONE DEL DNA MITOCONDRIALE MUTATO

Vergani Lodovica (1), Malena Adriana (1), Pantic Boris (1), Borgia Doriana (1), Baracca Alessandra (2), Sgarbi Gialuca (2), Solaini Giancarlo (2), Sandri Marco (3,4,5), Spinazzola Antonella (6), Holt Ian (6)

(1) Dip Neuroscienze NPSRR-Università di Padova, Lab. Neuromuscolare c/o Centro Biomedico Pietro D'Abano, via Orus 2-35129 Padova-Italy - Phone: +39 049 8216162, fax: +39 049 8216143- email: lodovica.vergani@unipd.it
(2) Dip. di Scienze Biomediche e Neuromotorie Alma Mater-Università di Bologna, Bologna, Italy
(3) Dip di Scienze Biomediche, University of Padova-Padova-Italy
(4) Dulbecco Telethon Institute, Venetian Institute of Molecular Medicine, 35129 Padova, Italy
(5) Telethon Institute of Genetics and Medicine (TIGEM), 80131 Napoli, Italy
(6) MRC London, UK

ABSTRACT N. 66

Telethon Research Projects - Neuromuscular Diseases		
Principal Investigator	**BURATTI EMANUELE**	
Telethon grant N.	**GGP14192**	
Total budget €	**268.600**	
Centres: **2**	*Duration (yrs):* **3**	*Starting year:* **2014**
Partners	ANDREA ELENA DARDIS	

IDENTIFICATION OF NEW THERAPEUTIC AGENTS FOR THE TREATMENT OF GLYCOGENOSIS TYPE 2 DUE TO THE COMMON SPLICING MUTATION C.-32-13T>G

IDENTIFICAZIONE DI NUOVE TERAPIE PER IL TRATTAMENTO DELLA GLICOGENOSI DI TIPO 2 DOVUTA A UN DIFETTO DI SPLICING

Dardis Andrea (2), Goina Elisa (1), Zanin Irene (2), Stuani Cristiana (1), Zampieri Stefania (2), Romano Maurizio (3,1), Buratti Emanuele (1)

(1) ICGEB - Padriciano 99, Trieste, 34149, Italia. Tel: 040-3757316, Fax: 040-226555, e-mail: buratti@icgeb.org
(2) Ospedale Universitario "Santa Maria della Misericordia", Udine, Italia
(3) Università degli Studi di Trieste

ABSTRACT N. 67

Telethon Research Projects - Neuromuscular Diseases		
Principal Investigator	**TOSCANO ANTONIO**	
Telethon grant N.	**GUP13013**	
Total budget €	**203.250**	
Centres: **13**	*Duration (yrs):* **2**	*Starting year:* **2014**
Partners	TIZIANA MONGINI, CORRADO ANGELINI, CLAUDIO BRUNO, MAURIZIO MOGGIO, GABRIELE SICILIANO, PAOLA TONIN, LORENZO MAGGI, ANDREA MARTINUZZI, MASSIMILIANO FILOSTO, SERENELLA SERVIDEI, MARIA ALICE DONATI, BRUNO BEMBI	

BUILDING A NATION-WIDE ITALIAN COLLABORATIVE NETWORK FOR MUSCLE GLYCOGENOSES: REGISTRY AND NATURAL HISTORY

SVILUPPO DI UNA RETE COLLABORATIVA ITALIANA PER LA RACCOLTA DEI PAZIENTI CON GLICOGENOSI MUSCOLARI (MDG): CREAZIONE DI UN REGISTRO NAZIONALE E STUDIO DELLA STORIA NATURALE DELLE MGD

Mongini Tiziana (2), Angelini Corrado (3), Bruno Claudio (4), Moggio Maurizio (5), Siciliano Gabriele (6), Tonin Paola (7), Maggi Lorenzo (8), Martinuzzi Andrea (9), Filosto Massimiliano (10), Servidei Serena (11), Donati Alice (12), Bembi Bruno (13), Marrosu Gianni (14), Di Iorio Giuseppe (15), Fiumara Agata (16), Ravaglia Sabrina (17), Massa Roberto (18), Bertini Enrico (19), Di Muzio Antonio (20), Toscano Antonio (1)

(1) Dipartimento di Neuroscienze, Messina, UOC neurologia e Malattie Neuromuscolari AOU Policlinico di Messina via C. Valeria 1 98125 Messina 0902213500 atoscano@unime.it
(2) Università di Torino
(3) Università di Padova
(4) Istituto G. Gaslini, Genova
(5) Università di Milano
(6) Università di Pisa
(7) Università di Verona
(8) Istituto C. Besta, Milano
(9) IRCCS Medea, Conegliano (TV)
(10) Spedali Civili , Brescia
(11) Università Cattolica di Roma
(12) Ospedale Meyer, Firenze
(13) Università di Udine
(14) ASP Cagliari
(15) Università di Napoli
(16) Università di Catania
(17) Università di Pavia
(18) Università tor Vergata, Roma
(19) Ospedale Bambin Gesu', Roma
(20) Università di Chieti

ABSTRACT N. 68

Telethon Research Projects - Neuromuscular Diseases		
Principal Investigator	**ARCA MARCELLO**	
Telethon grant N.	**GGP14066**	
Total budget €	**358.500**	
Centres: **5**	*Duration (yrs):* **3**	*Starting year:* **2015**
Partners	ELENA MARIA PENNISI, DANIELA TAVIAN, ANTONIO MUSARÒ, CORRADO ANGELINI	

CLINICAL, MOLECULAR AND PATHOGENETIC STUDIES OF NEUTRAL LIPID STORAGE DISEASE (NLSD)

STUDIO DEGLI ASPETTI CLINICI E PATOGENETICI DELLE SINDROMI DA ACCUMULO DI LIPIDI NEUTRI (NLSD)

Arca Marcello (1), Pennisi Elena (2), Angelini Corrado (3), Tavian Daniela (4), Musarò Antonio (5)

(1) Dipartimento di Medicina Interna e Specialità Medica - Sapienza Università di Roma, Policlinico Umberto I, Viale del Policlinico 155, Roma, Italy; tel.+39 0654451354; fax +39 064463534; email: marcelloarca@libero.it
(2) Azienda Ospedaliera San Filippo Neri, Dipartimento di Neuroscienze, Divisione di Neurologia, Via Martinotti 20, 00135 Roma; 0633063054; elenapennisi@gmail.com
(3) IRCCS S.Camillo, via Alberoni 70, 35100 Lido Di Venezia; 049 /8216155; corrado.angelini@ospedalesancamillo.net
(4) CRIBENS, Università Cattolica del Sacro Cuore, Piazza Buonarroti, 30, 20145 Milano; +39 02 72348731; daniela.tavian@unicatt.it
(5) Dipartimento di Scienze Anatomiche, Istologiche, Medico Legali e dell'Apparato Locomotore, Sapienza Università di Roma, Via Antonio Scarpa 14, 00161 Roma; +39 0649766956; antonio.musaro@uniroma1.it

NEUROLOGICAL DISEASES

ABSTRACT N. 69

Telethon Research Projects - Neurological Diseases		
Principal Investigator	**D'AMATI GIULIA**	
Telethon grant N.	**GGP13097**	
Total budget €	**218.500**	
Centres: **1**	*Duration (yrs):* **2**	*Starting year:* **2013**

ISOLATED DOMAINS OF AMINOACYL TRNA SYNTETHASES AS A NOVEL THERAPEUTIC TOOL FOR MT TRNA MUTATION ASSOCIATED DISEASE

RUOLO TERAPEUTICO DELLE AMINOACIL TRNA SINTETASI NELLE MALATTIE DA MUTAZIONI DEI TRNA MITOCONDRIALI

Perli Elena (1), Di Micco Patrizio (2), Fiorillo Annarita (2), Montanari Arianna (3), Pisano Annalinda (1), Preziuso Carmela (1), Poser Elena (2), Genovese Ilaria (1), Francisci Silvia (3), Frontali Laura (3), Zeviani Massimo (4), Giordano Carla (1), Morea Veronica (2), Colotti Gianni (2), d'Amati Giulia (1)

(1) Dip Scienze Radiologiche, Oncologiche e Anatomo Patologiche, Sapienza, Università di Roma - Policlinico Umberto I, Viale Regina Elena 324, 00161, Tel 06 49973332, Fax 06 4461484, email: giulia.damati@uniroma1.it
(2) Istituto di Biologia e Patologia Molecolari, CNR, Dip. Scienze Biochimiche, Sapienza, Università di Roma
(3) Dip Biologia e Biotecnologie "Charles Darwin", Sapienza, Università di Roma
(4) Medical Research Council-Mitochondrial Biology Unit, Cambridge, UK

ABSTRACT N. 70

Telethon Research Projects - Neurological Diseases		
Principal Investigator	**CARELLI VALERIO**	
Telethon grant N.	**GGP11182**	
Total budget €	**289.000**	
Centres: **4**	*Duration (yrs):* **2**	*Starting year:* **2011**
Partners	PALMIRO CANTATORE, ADAMO PIO D'ADAMO, ANDREA DAGA	

SYSTEMATIC GENE HUNTING FOR NUCLEAR MODIFIERS IN LEBER'S HEREDITARY OPTIC NEUROPATHY AND THEIR VALIDATION IN MODEL SYSTEMS

IDENTIFICAZIONE DEI GENI MODIFICATORI NUCLEARI NELLA NEUROPATIA OTTICA EREDITARIA DI LEBER E LORO VALIDAZIONE IN LINEE CELLULARI E ORGANISMI MODELLO

Cantatore Palmiro (2), d'Adamo Pio (3), Daga Andrea (4), Carelli Valerio (1)

(1) Dipartimento di Scienze Biomediche e Neuromotorie, Università di Bologna, Italia, IRCCS Istituto delle Scienze Neurologiche di Bologna, Ospedale Bellaria, Padiglione G, Via Altura 3, 40139, Bologna, Italia, Tel. +39 051 4966747, email valerio.carelli@unibo.it
(2) Dipartimento di Bioscienze, Biotecnologie e Biofarmaceutica, Università degli Studi di Bari Aldo Moro, Bari, Italia
(3) Division of Medical Genetics, IRCCS Burlo Garofolo, University of Trieste, Trieste, Italy
(4) IRCCS "E. Medea", Conegliano Research Center, Conegliano TV, Italy

ABSTRACT N. 71

Telethon Research Projects - Neurological Diseases		
Principal Investigator	**SCORRANO LUCA**	
Telethon grant N.	**GGP12162**	
Total budget €	**371.500**	
Centres: **1**	*Duration (yrs):* **3**	*Starting year:* **2012**

EXTENDING THE OPTIC ATROPHY 1 DEPENDENT CRISTAE REMODELING: FROM MODELS TO A RATIONALE FOR THERAPY OF AUTOSOMAL DOMINANT OPTIC ATROPHY

IL RIMODELLAMENTO DELLE CRISTE CONTROLLATO DA OPA1: DAI MODELLI ALLE BASI PER LA TERAPIA DELL'ATROFIA OTTICA DOMINANTE

Scorrano Luca (1,2)

(1) Dipartimento di Biologia, Università di Padova - Via G. Colombo 3, 35121 Padova, Italy, tel+390498276320, fax+390497923257, email luca.scorrano@unipd.it
(2) Dulbecco-Telethon Institute, Istituto Veneto di Medicina Molecolare

ABSTRACT N. 72

DTI - Neurological Diseases		
Principal Investigator	**SCORRANO LUCA**	
Telethon grant N.	**TCR12001**	
Total budget €	**207.441**	
Centres: **1**	*Duration (yrs):* **5**	*Starting year:* **2013**

GENETIC DISEASES OF MITOCHONDRIAL SHAPE: INTEGRATED APPROACHES TO UNDERSTAND PATHOGENESIS AND ESTABLISH TREATMENTS

MALATTIE GENETICHE DELLA FORMA DEI MITOCONDRI: APPROCCI INTEGRATI PER COMPRENDERNE LA PATOGENESI E DEFINIRNE STRATEGIE TERAPEUTICHE

Scorrano Luca (1,2)

(1) Dulbecco-Telethon Institute, Venetian Institute of Molecular Medicine - Via G. Colombo 3, 35121 Padova, Italy, tel+390498276320, fax+390497923257, email luca.scorrano@unipd.it
(2) Dipartimento di Biologia, Università di Padova

ABSTRACT N. 73

Telethon Research Projects - Neurological Diseases		
Principal Investigator	**AMBROSINI ELENA**	
Telethon grant N.	**GEP14134**	
Total budget €	**50.000**	
Centres: **1**	*Duration (yrs):* **1**	*Starting year:* **2015**

MEGALENCEPHALIC LEUKOENCEPHALOPATHY WITH SUBCORTICAL CYSTS: STUDY OF MLC MOLECULAR PATHOGENESIS AND IDENTIFICATION OF POTENTIAL THERAPEUTIC TARGETS USING ASTROCYTES DERIVED FROM PATIENT INDUCIBLE PLURIPOTENT STEM CELLS

LEUCOENCEFALOPATIA MEGALENCEFALICA CON CISTI SUBCORTICALI: STUDIO DELLA PATOGENESI MOLECOLARE E IDENTIFICAZIONE DI BERSAGLI TERAPEUTICI MEDIANTE LA GENERAZIONE DI ASTROCITI DIFFERENZIATI DA CELLULE STAMINALI PLURIPOTENTI INDOTTE DERIVATE DA PAZIENTI

Lanciotti Angela (1), Brignone Maria Stefania (1), Catacuzzeno Luigi (2), Visentin Sergio (1), Mallozzi Cinzia (1), Macchia Gianfranco (1), Pessia Mauro (2), Bertini Enrico (3), Ambrosini Elena (1)

(1) Dept. Cell Biology and Neuroscience, Istituto Superiore di Sanità, Rome - Viale Regina Elena 299 Roma - Tel. 06/49902037 – E-mail: elena.ambrosini@iss.it
(2) Dept. of Chemistry, Biology and Biotechnology/University of Perugia
(3) Dept. of Neuromuscular Disorders/ Pediatric Hospital Bambino Gesù (OPBG), Rome

ABSTRACT N. 74

Telethon Research Projects - Neurological Diseases		
Principal Investigator	**FORNASARI DIEGO MARIA MICHELE**	
Telethon grant N.	**GGP13055**	
Total budget €	**185.900**	
Centres: **1**	*Duration (yrs):* **2**	*Starting year:* **2013**

NEW APPROACHES TO THE MOLECULAR PATHOGENESIS OF CCHS: IMPLICATIONS FOR THERAPEUTIC STRATEGIES

STUDIO DEI MECCANISMI MOLECOLARI NELLA PATOGENESI DELLA SINDROME DI ONDINE PER LO SVILUPPO DI NUOVE STRATEGIE TERAPEUTICHE

Di Lascio Simona (1), Belperio Debora (1), Moncini Silvia (1), Maroli Annalisa (1), Benfante Roberta (2), Fornasari Diego Maria Michele (1,2)

(1) Dipartimento di Biotecnologie Mediche e Medicina Traslazionale, Università degli Studi di Milano, Milano - via Vanvitelli 32, 20129 Milano tel: 02 503 16960; Fax: 02 503 17132; diego.fornasari@unimi.it
(2) CNR - Istituto di Neuroscienze, Milano

ABSTRACT N. 75

Telethon Research Projects - Neurological Diseases		
Principal Investigator	**GASPARINI LAURA**	
Telethon grant N.	**GGP10184**	
Total budget €	**420.700**	
Centres: **3**	*Duration (yrs):* **3**	*Starting year:* **2010**

CLINICAL, NEURORADIOLOGICAL AND MOLECULAR INVESTIGATION OF ADULT-ONSET AUTOSOMAL DOMINANT LEUKODYSTROPHY (ADLD): DISSECTION OF LAMIN B1-MEDIATED PATHOPHYSIOLOGICAL MECHANISMS IN CELLULAR AND MOUSE MODELS

LEUCODISTROFIA AUTOSOMICA DOMINANTE DELL'ETÀ ADULTA: STUDIO TRASLAZIONALE DEI MECCANISMI GENETICI, MOLECOLARI E CELLULARI DI MALATTIA MEDIATI DALLA PROTEINA LAMIN B1 E CORRELAZIONE CON PARAMETRI CLINICI E NEURORADIOLOGICI

Cortelli Pietro (2), Brusco Alfredo (3), Brussino Alessandro (3), Giorgio Elisa (3), Antonorakis Stylianos, E. (4), Pennacchio Lea (5),

Spielman Malte (6), Di Gregorio Eleonora (7), Capellari Sabina (2), Bartoletti-Stella Anna (2), Terlizzi Rosanna (2), Parchi Piero (2), Liguori Rocco (2), Zanigni Stefano (9), Tonon Caterina (9), Lodi Raffaele (9), Vaula Giovanna (8), Contestabile Andrea (1), Mahajani Sameehan (1), Giacomini Caterina (1), Gasparini Laura (1)

(1) Dept. Neuroscience and Brain Technologies, Istituto Italiano di Tecnologia, Via Morego 30, 16163 Genova, Italy, Tel +39 010 71781 519; Fax +39 010 71781 230; email; laua.gasparini@iit.it
(2) IRCCS Istituto delle Scienze Neurologiche di Bologna, UOC Clinica Neurologica, Ospedale Bellaria, Dept. of Biomedical and Neuromotor Sciences, University of Bologna, Bologna
(3) Dept of Medical Sciences, University of Torino, Torino, Italy
(4) University of Geneva Medical School, Department of Genetic Medicine and Development, 1211, Geneva, Switzerland
(5) Genomics Division, MS 84-171, Lawrence Berkeley National Laboratory, Berkeley, CA 9472, USA
(6) Institute for Medical Genetics and Human Genetics, Charité - Universitätsmedizin Berlin, Berlin, Germany
(7) A.O. Città della Salute e della Scienza, S.C.D.U. Medical Genetics, 10126, Torino, Italy
(8) AO Città della Salute e della Scienza, Dept of Neurology, Torino, Italy
(9) Functional MR Unit, Policlinico S. Orsola – Malpighi, Dept. Of Biomedical and Neuromotor Sciences, University of Bologna, Bologna

ABSTRACT N. 76

Telethon Research Projects - Neurological Diseases		
Principal Investigator	**BALESTRINO MAURIZIO**	
Telethon grant N.	**GEP13109**	
Total budget €	**49.980**	
Centres: **1**	*Duration (yrs):* **1**	*Starting year:* **2014**

A NOVEL STRATEGY TO DELIVER GLUCOSE TO THE BRAIN UNDER CONDITIONS OF GLUCOSE TRANSPORTER DEFICIENCY

INDIVIDUAZIONE DI UNA TERAPIA FARMACOLOGICA PER LA MALATTIA DI DE VIVO O SINDROME DA DEFICIT DEL TRASPORTATORE DEL GLUCOSIO

Garbati Patrizia (1), Millo Enrico (2), Salis Annalisa (2,3), Adriano Enrico (1), Damonte Gianluca (2), Balestrino Maurizio (1)

(1) Dipartimento di Neuroscienze, Riabilitazione, Oftalmologia, Genetica e Scienze Materno-Infantili (DiNOGMI), Università di Genova - Largo Paolo Daneo 3, 16132 Genova - Italia Tel. 0103537078 Fax 0103537080 Mail mbalestrino@neurologia.unige.it
(2) Center of Excellence for Biomedical Research, Università di Genova
(3) Dipartimento di Scienze della Terra dell'Ambiente e della Vita (DISTAV), Università di Genova

ABSTRACT N. 77

Telethon Research Projects - Neurological Diseases		
Principal Investigator	**CRESTANI MAURIZIO**	
Telethon grant N.	**GEP14129**	
Total budget €	**49.900**	
Centres: **1**	*Duration (yrs):* **1**	*Starting year:* **2015**

NOVEL PHARMACOLOGICAL APPROACHES TO INCREASE KETONE BODIES AVAILABILITY IN GLUT1 DEFICIENCY SYNDROME

NUOVI APPROCCI FARMACOLOGICI PER AUMENTARE I LIVELLI DI CORPI CHETONICI NEI PAZIENTI CON DEFICIT DEL TRASPORTATORE GLUT1

Longo Raffaella, Ferrari Alessandra, Fiorino Erika, Fidone Alessandra, Crestani Maurizio

"Giovanni Galli" Laboratory of Biochemistry and Molecular Biology of Metabolism-Mass Spectrometry - Dipartimento di Scienze Farmacologiche e Biomolecolari (DiSFeB), Università degli Studi di Milano, via Balzaretti 9, 20133, Milano -Phone: +39 02 50318393/1 - Fax: +39 02 50318391 - e-mail: Maurizio.Crestani@unimi.it

ABSTRACT N. 78

Telethon Research Projects - Neurological Diseases		
Principal Investigator	**ORLACCHIO ANTONIO**	
Telethon grant N.	**GGP10121**	
Total budget €	**345.000**	
Centres: **4**	*Duration (yrs):* **2**	*Starting year:* **2010**
Partners	FILIPPO M. SANTORELLI, MARCO SERI, ORSETTA ZUFFARDI	

IDENTIFICATION OF NEW DISEASE-CAUSING GENES IN HEREDITARY SPASTIC PARAPLEGIA

IDENTIFICAZIONE DI NUOVI GENI-MALATTIA NELLA PARAPLEGIA SPASTICA EREDITARIA

Lo Giudice Temistocle (1), Di Lullo Martina (1), Montecchiani Celeste (1), Mearini Marzia (1), Casella Antonella (1), Carosi Laura (1), Lombardi Federica (1), Babalini Carla (1), D'Aloia Maria Michela (1), Montieri Pasqua (1), Gaudiello Fabrizio (1), Miele Marialuisa (1), Tessa Alessandra (2), Nesti Claudia (2), Pippucci Tommaso (3), Magini Pamela (3), Baptista Julia Pereira (3), Ciccone Roberto (4), Vetro Annalisa (4), Nuzzo Angelo (4), Limongelli Ivan (4), Zuffardi Orsetta (4), Seri Marco (3), Santorelli Filippo Maria (2), Orlacchio Antonio (1)

(1) Laboratorio di Neurogenetica, CERC-IRCCS Santa Lucia, Roma; Dipartimento di Medicina dei Sistemi, Università di Roma "Tor Vergata", Roma - Laboratorio di Neurogenetica, CERC-IRCCS Santa Lucia, Via del Fosso di Fiorano 64, 00143 Roma. Tel: 06-501703308; fax: 06-501703312; email: a.orlacchio@hsantalucia.it
(2) Laboratorio di Medicina Molecolare e Neurogenetica delle Malattie Neurodegenerative, Dipartimento di Medicina Molecolare, IRCCS Stella Maris, Calambrone (Pisa)
(3) Dipartimento di Scienze Mediche e Chirurgiche, Università di Bologna, Bologna
(4) Dipartimento di Medicina molecolare, Università di Pavia, Pavia

ABSTRACT N. 79

Telethon Research Projects - Neurological Diseases		
Principal Investigator	**DAGA ANDREA**	
Telethon grant N.	**GGP11189**	
Total budget €	**306.000**	
Centres: **1**	*Duration (yrs):* **3**	*Starting year:* **2011**

MODELS OF ATLASTIN FUNCTION AND DYSFUNCTION

MODELLI DI FUNZIONE E DISFUNZIONE DI ATLASTINA

Misticoni Giulia (1), Pendin Diana (2), Daga Andrea (1)

(1) Istituto Scientifico E. Medea - Largo Meneghetti 2, Padova; tel 0498275778; fax: 0498275011; email: andrea.daga@gmail.com
(2) CNR, Neuroscience Institute University of Padova, Department of Biomedical Sciences Viale G. Colombo, 3 35131 Padova

ABSTRACT N. 80

Telethon Research Projects - Neurological Diseases		
Principal Investigator	**D'ADDA DI FAGAGNA FABRIZIO**	
Telethon grant N.	**GGP12059**	
Total budget €	**210.600**	
Centres: **1**	*Duration (yrs):* **3**	*Starting year:* **2012**

THE ROLE OF TRANSCRIPTION IN THE CONTROL OF ATM ACTIVATION

IL RUOLO DELLA TRASCRIZIONE NEL CONTROLLO DELL'ATTIVAZIONE DI ATM

Michelini Flavia (1), Sharma Sheetal (1), Vitelli Valerio (1), Gioia Ubaldo (1), D'Adda Di Fagagna Fabrizio (1,2)

(1) IFOM, the FIRC Institute of Molecular Oncology, Via Adamello 16 - 20139 Milan - ITALY, tel. 02574303227, fax 02574303088, E-mail: fabrizio.dadda@ifom.eu
(2) Istituto di Genetica Molecolare, National Research Council, Via Abbiategrasso 207 - 27100 Pavia - ITALY, tel. 03825461, fax 0382422286, E-mail: fabrizio.dadda@igm.cnr.it

ABSTRACT N. 81

Telethon Research Projects - Neurological Diseases		
Principal Investigator	**FOIANI MARCO**	
Telethon grant N.	**GGP12171**	
Total budget €	**331.500**	
Centres: **1**	*Duration (yrs):* **3**	*Starting year:* **2012**

MODEL SYSTEMS TO IDENTIFY GENES AND FACTORS IN THE SIGNAL TRANSDUCTION PATHWAY DEFECTIVE IN ATAXIA TELANGIECTASIA PATIENTS

SISTEMI MODELLO PER IDENTIFICARE GENI E FATTORI NEI PROCESSI MOLECOLARI DIFETTIVI NEI PAZIENTI DI ATAXIA TELANGIECTASIA

Ferrari Elisa, Kidiyoor Gururaj, Foiani Marco

IFOM, the FIRC Institute of Molecular Oncology, Via Adamello, 16 - 20139 Milan, Italy - Tel: +39 02 574303238 - Fax: +39 02 574303231 marco.foiani@ifom.eu

ABSTRACT N. 82

Telethon Research Projects - Neurological Diseases		
Principal Investigator	**COSTANZO VINCENZO**	
Telethon grant N.	**GGP13071**	
Total budget €	**283.800**	
Centres: **1**	*Duration (yrs):* **3**	*Starting year:* **2013**

UNDERSTANDING ATM DEPENDENT CONTROL OF CELLULAR METABOLISM

CAPIRE IL CONTROLLO DEL METABOLISMO CELLULARE DA PARTE DI ATM

Costanzo Vincenzo, Vinciguerra Maria

IFOM, istituto FIRC di Oncologia Molecolare, Via Adamello 16, Milano, 20139, 02 574303231

ABSTRACT N. 83

Telethon Research Projects - Neurological Diseases		
Principal Investigator	**DELIA DOMENICO**	
Telethon grant N.	**GGP14164**	
Total budget €	**445.690**	
Centres: **2**	*Duration (yrs):* **3**	*Starting year:* **2014**
Partners	LORENZO MAGRASSI	

DETERMINANTS OF NEURODEGENERATION IN ATAXIA TELANGIECTASIA

DETERMINANTI DELLA NEURODEGENERAZIONE NELL'ATASSIA TELANGIECTASIA

Carlessi Luigi, Corti Alessandro, Delia Domenico

Fondazione IRCCS Istituto Nazionale Tumori, DOSMM, Via Amadeo 42, 20133 Milano, Italy - (02-23902641-Domenico.Delia@istituto-tumori.mi.it)

ABSTRACT N. 84

Telethon Research Projects - Neurological Diseases		
Principal Investigator	**TESTI ROBERTO**	
Telethon grant N.	**GGP11102**	
Total budget €	**373.000**	
Centres: **1**	*Duration (yrs):* **3**	*Starting year:* **2011**

INVESTIGATING NEW THERAPEUTIC APPROACHES TO FRIEDREICH'S ATAXIA

STUDIO DI NUOVE STRATEGIE TERAPEUTICHE PER L'ATASSIA DI FRIEDREICH

Rufini Alessandra (1,2), Fortuni Silvia (1,2), Benini Monica (1), Cavallo Francesca (1), Condò Ivano (1), De Martino Gabriella (1), Incani Ottaviano (1), Massaro Damiano Sergio (1), Alfedi Giulia (1), Alaimo Giorgia (1), Di Venere Almerinda (3), Malisan Florence (1), Serio Dario (1), Testi Roberto (1,2)

(1) Laboratory of Signal Transduction, Department of Biomedicine and Prevention, University of Rome "Tor Vergata", Italy - Laboratory of Signal Transduction, Department of Biomedicine and Prevention, University of Rome "Tor Vergata", Via Montpellier 1, 00133, Rome, Italy, tel 06 72596503, roberto.testi@uniroma2.it
(2) Fratagene Therapeutics Ltd, 22 Northumberland Rd, Dublin, Ireland
(3) Department of Experimental Medicine and Surgery, University of Rome "Tor Vergata", Italy

ABSTRACT N. 85

Telethon Research Projects - Neurological Diseases		
Principal Investigator	**BRUSCO ALFREDO**	
Telethon grant N.	**GGP12217**	
Total budget €	**322.800**	
Centres: **2**	*Duration (yrs):* **3**	*Starting year:* **2012**
Partners	FILIPPO TEMPIA	

SPINOCEREBELLAR ATAXIA TYPE 28: CELLULAR AND ANIMAL MODELS TO UNRAVEL THE PATHOGENESIS AND TO IDENTIFY POTENTIAL THERAPEUTIC TARGETS

ATASSIA SPINOCEREBELLARE TIPO 28 (SCA28): MODELLI CELLULARI E ANIMALI PER IDENTIFICARE I MECCANISMI PATOGENETICI ED I POTENZIALI BERSAGLI TERAPEUTICI

Mancini Cecilia (1), Hoxha Eriola (2), Turco Emilia (3), Altruda Fiorella (3), Tempia Filippo (2), Brusco Alfredo (1,4)

(1) Department of Medical Sciences, University of Torino, Torino, Italy; - Alfredo Brusco, via Santena 19, Department of Medical Sciences, 10126 Torino. Phone +390116334480; Fax: +390116706582; e-mail: alfredo.brusco@unito.it
(2) Neuroscience Institute Cavalieri Ottolenghi (NICO), University of Torino, Italy
(3) Dipartimento di Biotecnologie Molecolari e Scienze per la Salute
(4) Medical Genetics Unit, "Città della Salute e della Scienza" Hospital, Torino, Italy

ABSTRACT N. 86

Telethon Research Projects - Neurological Diseases		
Principal Investigator	**CASARI GIORGIO**	
Telethon grant N.	**GGP12235**	
Total budget €	**423.900**	
Centres: **1**	*Duration (yrs):* **3**	*Starting year:* **2012**

GENETIC AND PHARMACOLOGICAL RESCUES OF SPINOCEREBELLAR ATAXIA IN THE SCA28 MODEL OPEN TO HUMAN THERAPY

LA RIDUZIONE DELLA STIMOLAZIONE GLUTAMMATERGICA NEL MODELLO MURINO SCA28 RISOLVE LA SINTOMATOLOGIA ATASSICA DEGENERATIVA

Maltecca Francesca (1), Baseggio Elisa (1), Consolato Francesco (1), Mazza Davide (2), Podini Paola (3), Young Jr. Samuel M. (4), Drago Ilaria (5,6), Bahr Ben A. (7), Puliti Alda Maria (8), Codazzi Franca (9), Quattrini Angelo (3), Casari Giorgio (1)

(1) Università Vita-Salute San Raffaele and Div. Genetics and Cell Biology, San Raffaele Scientific Institute, Milan, Italy - Via Olgettina 58, 20132 Milano, tel 0226433502 casari.giorgio@hsr.it
(2) Università Vita-Salute San Raffaele and Experimental Imaging Center, San Raffaele Scientific Institute, Milan, Italy
(3) Institute of Experimental Neurology, Division of Neuroscience, San Raffaele Scientific Institute, Milan, Italy
(4) Research Group Molecular Mechanisms of Synaptic Function, Max Planck Florida Institute for Neuroscience, Jupiter, FL USA
(5) Department of Biomedical Sciences, University of Padua, Italy
(6) Department of Neuroscience, The Scripps Research Institute Florida, Jupiter, FL, USA
(7) Biotechnology Research and Training Center, University of North Carolina-Pembroke, USA
(8) Medical Genetics Unit, Istituto Giannina Gaslini, Genoa, Italy and Department of Neurosciences, Rehabilitation, Ophthalmology, Genetics, Maternal and Child Health (DiNOGMI), University of Genoa, Genoa, Italy
(9) Cellular Neurophysiology Unit, Division of Neuroscience, San Raffaele Scientific Institute, Milan, Italy

ABSTRACT N. 87

Telethon Research Projects - Neurological Diseases		
Principal Investigator	**BORRONI BARBARA**	
Telethon grant N.	**GGP14225**	
Total budget €	**483.800**	
Centres: **5**	*Duration (yrs):* **3**	*Starting year:* **2014**
Partners	ALFREDO BRUSCO, DONATELLA CARUSO, LOREDANA BOCCONE, FILIPPO TEMPIA	

TRANSLATING MOLECULAR PATHOLOGY INTO A THERAPEUTIC STRATEGY IN SCA38, A NEWLY IDENTIFIED FORM OF SPINOCEREBELLAR ATAXIA

DAI MECCANISMI PATOGENETICI ALLA TERAPIA DELLA SCA38, UNA NUOVA FORMA DI ATASSIA AUTOSOMICA DOMINANTE

Eleonora Di Gregorio (2), Marta Ferrero (3), Neftj Ragusa (3), Federica Gottardi (1), Paolo Costa (1), Adele Zoppo (4), Eriola Hoxa (5), Filippo Tempia (5), Loredana Boccone (6), Nico Mitro (7), Donatella Caruso (7), Alfredo Brusco (2,3), Barbara Borroni (1)

(1) Dipartimento di Scienze Cliniche e Sperimentali, Clinica Neurologica, Università degli Studi di Brescia, Brescia, Italia - Piazzale Spedali Civili n. 1; 25123; Brescia, 030 3995632, 0303384086, bborroni@inwind.it
(2) Unità di Genetica Medica, Ospedale "Città della Salute e della Scienza", Torino, Italia
(3) Dipartimento di Scienze Mediche, Università di Torino, Torino, Italia
(4) Dipartimento di Endocrinologia, Ospedale S. Carlo, Paderno Dugnano, Milano, Italia
(5) Neuroscience Institute Cavalieri Ottolenghi (NICO), Università di Torino, Torino, Italia
(6) Ospedale Regionale Microcitemie, ASL 8, 09121, Cagliari, Italia
(7) Dipartimento di Scienze Farmacologiche e Biomolecolari, Università di Milano, Milano, Italia

ABSTRACT N. 88

Telethon Research Projects - Neurological Diseases		
Principal Investigator	**GALIETTA LUIS JUAN VICENTE**	
Telethon grant N.	**GEP14096**	
Total budget €	**50.000**	
Centres: **1**	*Duration (yrs):* **1**	*Starting year:*

ROLE OF ANO10 IN SPINOCEREBELLAR ATAXIA

RUOLO DELLA PROTEINA ANO10 NELL'ATASSIA SPINOCEREBELLARE DI TIPO 10

Scudieri Paolo (1), Valente Pierluigi (2), Sondo Elvira (1), Pedemonte Nicoletta (1), Benfenati Fabio (2), Galietta Luis J.V. (1)

(1) U.O.C. Genetica Medica, Istituto Giannina Gaslini, Genova - Via Gerolamo Gaslini 5, Genova - Tel. 010/5636711 - E-mail: galietta@unige.it
(2) DIMES, Università di Genova

ABSTRACT N. 89

Telethon Research Projects - Neurological Diseases		
Principal Investigator	**BERTINI ENRICO SILVIO**	
Telethon grant N.	**GGP13146**	
Total budget €	**519.400**	
Centres: **3**	*Duration (yrs):* **3**	*Starting year:* **2013**
Partners	ENZA MARIA VALENTE, GIANGIACOMO CONSALEZ	

CLINICAL, GENETIC AND FUNCTIONAL STUDIES ON JOUBERT SYNDROME AND RELATED DISORDERS: A MODEL TO UNDERSTAND THE COMPLEXITY OF CILIOPATHIES

STUDI CLINICI, GENETICI E FUNZIONALI NELLA SINDROME DI JOUBERT E DISORDINI CORRELATI: UN MODELLO PER COMPRENDERE LA COMPLESSITÀ DELLE CILIOPATIE

Romani Marta (2), Croci Laura (3), Travaglini Lorena (1), Micalizzi Alessia (2,4), Zanni Ginevra (1), Badaloni Aurora (3), Mazza Tommaso (2), Barresi Sabina (1), Bosone Camilla (5), Miccinilli Elide (2), Consalez Giacomo (3,5), Valente Enza Maria (2,6), Bertini Enrico Silvio (1)

(1) Laboratory of Molecular Medicine, Unit of Neuromuscular Disorders, Dept of Neurosciences, IRCCS Bambino Gesù Children's Hospital, Rome, Italy - Viale di San Paolo 15 00146 Rome - ITALY, Tel. +39.06.6859.2104/2105 Fax. +39.06.6859.2463; enricosilvio.bertini@opbg.net
(2) IRCCS Casa Sollievo della Sofferenza, Mendel Institute, Rome, Italy
(3) San Raffaele Scientific Institute, Milan, Italy
(4) Dept. of Biological and Environmental Science, University of Messina, Messina, Italy
(5) Università Vita-Salute San Raffaele, Milan, Italy
(6) Section of Neurosciences, Dept. of Medicine and Surgery, University of Salerno, Salerno, Italy

ABSTRACT N. 90

Telethon Research Projects - Neurological Diseases		
Principal Investigator	**SALLESE MICHELE**	
Telethon grant N.	**GGP12220**	
Total budget €	**357.700**	
Centres: **2**	*Duration (yrs):* **3**	*Starting year:* **2013**
Partners	ROBERTO CHIESA	

PURKINJE CELL DEGENERATION IN MARINESCO-SJOGREN SYNDROME: ROLE OF CELL STRESS, ALTERATIONS OF PROTEOSTASIS AND CALCIUM HOMEOSTASIS

RUOLO DELLO STRESS CELLULARE, PROTEOSTASI E OMEOSTASI DEL CALCIO NELLA DEGENERAZIONE DELLE CELLULE DEL PURKINJE NELLA SINDROME DI MARINESCO-SJOGREN

Capone Vanessa (1), Fragassi Giorgia (1), Ornaghi Francesca (2), Chiesa Roberto (2), Sallese Michele (1)

(1) Unit of Genomic Approaches to Membrane Traffic, Fondazione Mario Negri Sud - Via Nazionale 8/A, Santa Maria Imbaro (Chieti), Italy. Tel: +39-0872-570421, Fax: +39-0872-570412, E-mail: sallese@negrisud.it
(2) Department of Neuroscience, IRCCS - Mario Negri Institute for Pharmacological Research, Milan, Italy

ABSTRACT N. 91

Telethon Research Projects - Neurological Diseases		
Principal Investigator	**CHIABRANDO DEBORAH**	
Telethon grant N.	**GEP13065**	
Total budget €	**40.100**	
Centres: **1**	*Duration (yrs):* **1**	*Starting year:* **2014**

INVESTIGATION OF THE MOLECULAR BASIS OF POSTERIOR COLUM ATAXIA AND RETINITIS PIGMENTOSA

ANALISI DELLE BASI MOLECOLARI DELL'ATASSIA DEL CORDONE POSTERIORE-RETINITE PIGMENTOSA

Petrillo Sara, Mercurio Sonia, Altruda Fiorella, Silengo Lorenzo, Tolosano Emanuela, Chiabrando Deborah

Department of Molecular Biotechnology and Health Sciences. Molecular Biotechnology Centre (MBC). University of Torino, Torino - Department of Molecular Biotechnology and Health Sciences - Via Nizza 52, 10126, Torino - Tel: 011 670 6423 - Fax: 011 670 6432 - email: deborah.chiabrando@unito.it

ABSTRACT N. 92

Telethon Research Projects - Neurological Diseases		
Principal Investigator	**BENFENATI FABIO**	
Telethon grant N.	**GGP13033**	
Total budget €	**527.500**	
Centres: **3**	*Duration (yrs):* **3**	*Starting year:* **2013**
Partners	FEDERICO ZARA, FLAVIA VALTORTA	

ROLE OF THE NOVEL PRESYNAPTIC PROTEIN PRRT2 IN NEURONAL PHYSIOLOGY AND IN THE PATHOGENESIS OF PAROXYSMAL NEUROLOGICAL DISORDERS

RUOLO DELLA PROTEINA PRESINAPTICA PRRT2 NELLA FISIOLOGIA NEURONALE E NELLA PATOGENESI DEI DISORDINI PAROSSISTICI DEL SISTEMA NERVOSO

Benfenati Fabio (1), Valtorta Flavia (3), Zara Federico (2), Rossi Pia (1), Valente Pierluigi (1), Corradi Anna (1), Fadda Manuela (1), Giovedì Silvia (1), Fassio Anna (1), Fruscione Floriana (2), Baldassari Simona (2), Guarnieri Fabrizia (3), Mura Elisa (3)

(1) Department of Experimental Medicine, University of Genova, Viale Benedetto XV, 3 16132 Genova; tel: 010/3538183, fax: 010/3538194, email: Fabio.Benfenati@unige.it
(2) Department of Neuroscience, G. Gaslini Institute Genova
(3) Department of Neuroscience, San Raffaele Scientific Institute, Milano

ABSTRACT N. 93

Telethon Research Projects - Neurological Diseases		
Principal Investigator	**CANCEDDA LAURA**	
Telethon grant N.	**GGP10135**	
Total budget €	**132.000**	
Centres: **1**	*Duration (yrs):* **3**	*Starting year:* **2010**

ROLE OF GABA A-RECEPTOR MUTATIONS IN IDIOPATHIC GENERALIZED EPILEPSY: A DEVELOPMENTAL STUDY

RUOLO DELLE MUTAZIONI DEL RECETTORE GABA A NELLA EPILESSIA IDIOPATICA GENERALIZZATA: UNO STUDIO SULLO SVILUPPO

Szczurkowska Joanna (1), Cwetsch Andrzej (1), Deidda Gabriele (1), Allegra Manuela (2,3), Perlini Laura (1), Succol Francesca (1), Cossette Patrick (4), Barberis Andrea (1), Caleo Matteo (3), Cancedda Laura (1)

(1) Neuroscience and Brain Technologies Department, Istituto Italiano di Tecnologia, via Morego, 30, 16163 Genova
(2) Scuola Normale Superiore, Pisa, Italy
(3) CNR Neuroscience Institute, Pisa, Italy
(4) Université de Montréal, Canada

ABSTRACT N. 94

Telethon Research Projects - Neurological Diseases		
Principal Investigator	**NOBILE CARLO**	
Telethon grant N.	**GGP12078**	
Total budget €	**194.600**	
Centres: **1**	*Duration (yrs):* **2**	*Starting year:* **2012**

IDENTIFICATION OF NOVEL GENES FOR AUTOSOMAL DOMINANT LATERAL TEMPORAL EPILEPSY IN FAMILIES WITHOUT LGI1 MUTATIONS

IDENTIFICAZIONE DI NUOVI GENI PER L'EPILESSIA TEMPORALE LATERALE AUTOSOMICA DOMINANTE IN FAMIGLIE SENZA MUTAZIONI IN LGI1

Dazzo Emanuela (1), Fanciulli Manuela (2), Serioli Elena (1), Minervini Giovanni (3), Pulitano Patrizia (4), Binelli Simona (5), Di Bonaventura Carlo (4), La Neve Angela (6), Pasini Elena (7), Striano Salvatore (8), Striano Pasquale (9), Coppola Giangennaro (10), Chiavegato Angela (1), Radovic Slobodanka (11), Spadotto Alessandro (11), Uzzau Sergio (2), Giallonardo Anna Teresa (4), Mecarelli Oriano (4), Tosatto Silvio (3), Ottman Ruth (12), Michelucci Roberto (7), Nobile Carlo (1)

(1) CNR-Istituto di Neuroscienze, Sede di Padova - Viale G. Colombo 3, 35121 Padova; Tel. 0498276072; Fax: 0498276040; Email: nobile@bio.unipd.it
(2) Porto Conte Ricerche, Alghero, Sassari
(3) Dipartimento di Scienze Biomediche, Università di Padova, Padova
(4) Dipartimento di Scienze Neurologiche, Università La Sapienza, Roma
(5) Istituto Neurologico C. Besta, Milano
(6) Clinica Neurologica, Università di Bari

(7) IRCCS-Scienze Neurologiche, Ospedale Bellaria, Bologna
(8) Dipartimento di Scienze Neurologiche, Università Federico II, Napoli
(9) Dipartimento di Neuroscienze, Università di Genova, Istituto G. Gaslini, Genova
(10) Neuropsichiatria Infantile, Università di Salerno, Salerno
(11) IGA Technology, Udine
(12) Department of Epidemiology and Neurology, Columbia University, New York, USA

ABSTRACT N. 95

Telethon Research Projects - Neurological Diseases		
Principal Investigator	**BECCHETTI ANDREA**	
Telethon grant N.	**GGP12147**	
Total budget €	**179.700**	
Centres: **1**	*Duration (yrs):* **3**	*Starting year:* **2012**

THE ROLE OF NEURONAL NICOTINIC RECEPTORS IN THE PATHOGENESIS OF AUTOSOMAL DOMINANT NOCTURNAL FRONTAL LOBE EPILEPSY (ADNFLE): A STUDY ON WILD-TYPE AND CONDITIONAL TRANSGENIC MICE EXPRESSING THE BETA2-V287L SUBUNIT

RUOLO DEI RECETTORI NICOTINICI NELLA PATOGENESI DELL'EPILESSIA NOTTURNA AUTOSOMICA DOMINANTE DEL LOBO FRONTALE (ADNFLE): STUDIO SU UN MODELLO MURINO CONDIZIONALE

Meneghini Simone (1), Aracri Patrizia (1), Brusco Simone (1), Carraresi Laura (2), Arcangeli Annarosa (2), Amadeo Alida (3), Becchetti Andrea (1)

(1) Dipartimento di Biotecnologie e Bioscienze, Università di Milano-Bicocca, piazza della Scienza 2, 20126 Milano. - Tel. 02-64483301; fax 02-64483565; andrea.becchetti@unimib.it
(2) Dipartimento di Medicina Sperimentale e Clinica, Università di Firenze, Firenze
(3) Dipartimento di Scienze della Vita, Università di Milano, Milano

ABSTRACT N. 96

Telethon Research Projects - Neurological Diseases		
Principal Investigator	**TINUPER PAOLO**	
Telethon grant N.	**GGP13200**	
Total budget €	**442.850**	
Centres: **2**	*Duration (yrs):* **3**	*Starting year:* **2013**
Partners	TOMMASO PIPPUCCI	

IN-DEPTH CLINICAL AND GENETIC STUDY OF FAMILIAL AND SPORADIC PATIENTS WITH NOCTURNAL FRONTAL LOBE EPILEPSY (NFLE): IDENTIFICATION OF NEW GENES BY WES IN 192 CASES NEGATIVE FOR MUTATIONS IN THE NEURONAL NICOTINIC ACETYLCHOLINE RECEPTOR SUBUNITS GENES

STUDIO CLINICO E GENETICO DI CASI SPORADICI E FAMILIARI DI EPILESSIA FRONTALE NOTTURNA (EFN)

Pippucci Tommaso (1), Licchetta Laura (2,3), Baldassari Sara (1), Palombo Flavia (1), Magini Pamela (1), Provini Federica (2,3), Mostacci Barbara (2,3), Naldi Ilaria (2,3), Seri Marco (1), Bisulli Francesca (2,3), Tinuper Paolo (2,3)

(1) U.O. Medical Genetics, Polyclinic Sant'Orsola-Malpighi; Department of Medical and Surgical Sciences, University of Bologna, 40138 Bologna, Italy - paolo.tinuper@unibo.it; Ospedale Bellaria, via Altura 3 - 40139 Bologna; +390514966967; fax: +390514966993
(2) IRCCS Istituto delle Scienze Neurologiche of Bologna
(3) Department of Biomedical and Neuromotor Science, University of Bologna

ABSTRACT N. 97

Telethon Research Projects - Neurological Diseases		
Principal Investigator	**MALLAMACI ANTONELLO**	
Telethon grant N.	**GGP13034**	
Total budget €	**283.150**	
Centres: **3**	*Duration (yrs):* **3**	*Starting year:* **2013**
Partners	PASQUALE STRIANO, YURI BOZZI	

MODELLING ETIOPATHOGENESIS OF THE FOXG1-LINKED VARIANT OF WEST SYNDROME

CARATTERIZZAZIONE EZIOPATOGENETICA DELLA VARIANTE DELLA SINDROME DI WEST ASSOCIATA A DUPLICAZIONE DI FOXG1

Do Duc Minh (1), Pinzan Moira (1), Grudina Clara (1), Falcone Carmen (1), Provenzano Giovanni (2), Zara Federico (3), Vari Maria Stella (3), Goina Elisa (1), Calligaris Raffaella (1), Bozzi Yuri (2), Striano Pasquale (3), Mallamaci Antonello (1)

(1) Cerebral Cortex Development Lab/Neuroscience Area, SISSA, Trieste - via Bonomea 265 - 34136 Trieste - Italy, phone: +39 040 3787 717, e-mail: amallama@sissa.it
(2) Laboratory of Molecular Neuropathology/CIBIO, University of Trento, Trento
(3) Pediatric Neurology and Muscular Diseases Unit/DINOGMI-Department of Neurosciences, Rehabilitation, Ophtalmology, Genetics, Maternal and Child Health, University of Genua - "G. Gaslini" Institute, Genua

ABSTRACT N. 98

Telethon Research Projects - Neurological Diseases		
Principal Investigator	**MIANO MARIA GIUSEPPINA**	
Telethon grant N.	**GGP14198**	
Total budget €	**187.500**	
Centres: **1**	*Duration (yrs):* **3**	*Starting year:* **2014**

DISSECTING THE ARISTALESS-RELATED HOMEOBOX EPILEPSY PATH TO FIND DRUGGABLE TARGET MOLECULES

ANALISI DELLA FUNZIONE DEL GENE ARISTALESS-RELATED HOMEOBOX NELL'EPILESSIA PEDIATRICA MALIGNA E IDENTIFICAZIONE DI BERSAGLI MOLECOLARI A SCOPO TERAPEUTICO

Poeta Loredana (1), Padula Agnese (1), Zucchelli Silvia (2,3), Ranieri Augusto (1), D'Adamo Patrizia (4), Ursini Matilde Valeria (1), Tongiorgi Enrico (5), Filosa Stefania (6), Acampora Dario (1), Altucci Lucia (1), Gustincich Stefano (3), Miano Maria Giuseppina (1)

(1) Institute of Genetics and Biophysics "Adriano Buzzati Traverso", CNR, Naples, Italy, phone 0816132261, fax 0816132706, email mariag.miano@igb.cnr.it
(2) Department of Health Sciences, University of Eastern Piedmont "A. Avogadro", Novara, Italy
(3) SISSA, Area of Neuroscience, Trieste, Italy
(4) Vita-Salute San Raffaele University, Milan, Italy
(5) Department of Life Sciences, University of Trieste, Trieste, Italy
(6) Institute of Biosciences and BioResources, CNR, Naples, Italy

ABSTRACT N. 99

Telethon Research Projects - Neurological Diseases		
Principal Investigator	**FELLIN TOMMASO**	
Telethon grant N.	**GGP10138**	
Total budget €	**479.700**	
Centres: **5**	*Duration (yrs):* **3**	*Starting year:* **2010**
Partners	SILVANA FRANCESCHETTI, GIORGIO CARMIGNOTO, DANIELE AROSIO, ALBERTO BACCI	

CELLULAR AND SYNAPTIC ALTERATIONS UNDERLYING EPILEPTOGENESIS IN AN EXPERIMENTAL MODEL OF DRAVET SYNDROME

MODIFICAZIONI CELLULARI E SINAPTICHE DURANTE IL PERIODO EPILETTOGENICO IN UN MODELLO SPERIMENTALE DI SINDROME DI DRAVET

Scalmani Paolo (2), De Stasi Angela Michela (1), Farisello Pasqualina (1,3), Marcon Iacopo (4), Losi Gabriele (4), Ratto Gian Michele (5), Arosio Daniele (6), Franceschetti Silvana (2), De Curtis Marco (2), Carmignoto Giorgio (4), Bacci Alberto (3,7), Mantegazza Massimo (8), Fellin Tommaso (1)

(1) Istituto Italiano di Tecnologia, Genova, Italy - Via Morego 30, 16163 Genova; tel: 010 71781549; fax:010 71781230; email: tommaso.fellin@iit.it
(2) Fondazione IRCCS Istituto Neurologico C.Besta, Milano, Italia
(3) Fondazione EBRI "Rita Levi-Montalcini", Roma, Italia
(4) CNR-Istituto di Neuroscienze ed Università di Padova, Padova, Italia
(5) CNR- Istituto di Neuroscienze, Pisa, Italia
(6) Istituto di Biofisica, CNR, Trento, Italia
(7) ICM - Institut du Cerveau et de la Moelle épinière, Paris, France
(8) IPMC, CNRS UMR7275 and University of Nice-Sophia Antipolis, Valbonne, France

ABSTRACT N. 100

Telethon Research Projects - Neurological Diseases		
Principal Investigator	**CARMIGNOTO GIORGIO**	
Telethon grant N.	**GGP12265**	
Total budget €	**446.800**	
Centres: **3**	*Duration (yrs):* **3**	*Starting year:* **2012**
Partners	MARCO DE CURTIS, GIAN MICHELE RATTO	

ROLE OF DYSREGULATED ASTROCYTE-GABAERGIC INTERNEURON INTERACTIONS IN THE CONTROL OF SEIZURES IN MONOGENIC MODELS OF EPILEPSY

RUOLO DELLE INTERAZIONI TRA ASTROCITI ED INTERNEURONI GABAERGICI NEL CONTROLLO DELLE CRISI EPILETTICHE IN MODELLI DI MALATTIE NEUROLOGICHE MONOGENICHE ASSOCIATE AD EPILESSIA

Marcon Iacopo (1), Losi Gabriele (1), Sessolo Michele (1), Brondi Marco (2), Noè Francesco (3), Ratto Gian Michele (2), Marco DeCurtis (3), Giorgio Carmignoto (1)

(1) Neuroscience Institute, Consiglio Nazionale delle Ricerche (CNR) and University of Padova, Padova, Italy. - Viale G. Colombo 3, tel: 049 8276075, fax 049 8276049, giorgio.carmignoto@bio.unipd.it
(2) NEST, Istituto Nanoscienze-CNR, Pisa, Italy
(3) Foundation IRCCS Neurological Institute C. Besta, Milano, Italy

ABSTRACT N. 101

Telethon Research Projects - Neurological Diseases		
Principal Investigator	**PIETROBON DANIELA**	
Telethon grant N.	**GGP14234**	
Total budget €	**315.100**	
Centres: **1**	*Duration (yrs):* **3**	*Starting year:* **2014**

FAMILIAL HEMIPLEGIC MIGRAINE MECHANISMS

MECCANISMI DELL'EMICRANIA EMIPLEGICA FAMILIARE

Capuani Clizia (1), Tottene Angelita (1), Melone Marcello (2), Brigina Luca (2), Casari Giorgio (3), Brennan KC (4), Conti Fiorenzo (2), Pietrobon Daniela (1)

(1) Dipartimento di Scienze Biomediche, Universita' di Padova, Padova - Dipartimento di Scienze Biomediche, Università di Padova, Via U Bassi 58, 35121 Padova; Tel: 049 8276052; Fax: 049 8276049; daniela.pietrobon@unipd.it
(2) Dipartimento di Medicina Sperimentale e Clinica, Università Politecnica delle Marche, Ancona
(3) Centro di Genomica, Bioinformatica e Biostatistica, Università Vita Salute San Raffele, Milano
(4) Department of Neurology, University of Utah School of Medicine, Salt Lake City, USA

ABSTRACT N. 102

Telethon Research Projects - Neurological Diseases		
Principal Investigator	**BOLOGNESI MARTINO**	
Telethon grant N.	**GGP11057**	
Total budget €	**419.100**	
Centres: **3**	*Duration (yrs):* **3**	*Starting year:* **2011**
Partners	MAURO MANNO, MARIA ELENA MIRANDA BANOS	

THE ROLE OF NEUROSERPIN IN FAMILIAL ENCEPHALOPATHY WITH NEUROSERPIN INCLUSION BODIES

IL RUOLO DELLA NEUROSERPINA NELLA ENCEFALOPATIA FAMIGLIARE DA CORPI D'INCLUSIONE DI NEUROSERPINA

Saga Giorgia (1), Sessa Fabio (1), Ricagno Stefano (1), Manno Mauro (2), Martorana Vincenzo (2), Noto Rosina (2), Raccosta Samuele (2), Randazzo Loredana (2), Santagelo Maria Grazia (2), Moriconi Claudia (3), Guadagno Noemi A (3), Lupo Giuseppe (3), Carucci Nicoletta (3), Timpano Valentina (3), Miranda Elena (3), Bolognesi Martino (1)

(1) Dipartimento di Bioscienze, Università di Milano, Via Celoria 26, Milano, Italia
(2) Istituto di Biofisica, CNR, Palermo, Italia
(3) Dipartimento di Biologia e biotecnologie Charles Darwin, Università la Sapienza, Roma, Italia

ABSTRACT N. 103

Telethon Research Projects - Neurological Diseases		
Principal Investigator	**DI FEDE GIUSEPPE**	
Telethon grant N.	**GGP10120**	
Total budget €	**502.400**	
Centres: **2**	*Duration (yrs):* **3**	*Starting year:* **2010**
Partners	GIUSEPPE DI FEDE, LUISA DIOMEDE	

BAD GENE, GOOD GENE: A RECESSIVE APP MUTATION CAN BE BOTH. NEW THERAPEUTIC PERSPECTIVE FOR ALZHEIMER'S DISEASE BASED ON AN ABETA VARIANT WITH DOMINANT-NEGATIVE EFFECT ON AMYLOIDOGENESIS

NUOVE PROSPETTIVE TERAPEUTICHE PER LA MALATTIA DI ALZHEIMER BASATE SU UNA VARIANTE DI ABETA CHE INIBISCE L'AMILOIDOGENESI

Diomede Luisa (2), Catania Marcella (1), Romeo Margherita (2), Morbin Michela (1), Gobbi Marco (2), Maderna Emanuela (1), Moda Fabio (1), Palamara Luisa (1), Colombo Laura (2), Rossi Alessandro (2), Cagnotto Alfredo (2), Salmona Mario (2), Tagliavini Fabrizio (1), Di Fede Giuseppe (1)

(1) U.O. Neurology V - Neuropathology, Fondazione IRCCS "Carlo Besta" Neurological Institute, Milan, Italy - Dr. Giuseppe di Fede, Laboratorio di Genetica e Biochimica delle Demenze, Fondazione IRCCS "Carlo Besta" Neurological Institute, Via Amadeo 42, 20133 Milano, Tel. +390223944583, fax +390223944711, email: gdifede@istituto-besta.it
(2) Department of Molecular Biochemistry and Pharmacology, Istituto di Ricerche Farmacologiche IRCCS "Mario Negri", Milan, Italy

ABSTRACT N. 104

Telethon Research Projects - Neurological Diseases		
Principal Investigator	**BARTESAGHI RENATA**	
Telethon grant N.	**GGP12149**	
Total budget €	**296.400**	
Centres: **3**	*Duration (yrs):* **3**	*Starting year:* **2012**
Partners	LAURA CALZÀ, JACOPO MAGISTRETTI	

PREVENTIVE THERAPY OF MENTAL RETARDATION IN DOWN SYNDROME BY A NOVEL GAMMA-SECRETASE INHIBITOR: FOCUS ON APP-DEPENDENT MECHANISMS IN NEURODEVELOPMENT

TERAPIA PREVENTIVA DEL RITARDO MENTALE NELLA SINDROME DI DOWN CON UN NUOVO INIBITORE DELLA GAMMA-SECRETASI: MECCANISMI APP-DIPENDENTI NELLO SVILUPPO DEL SISTEMA NERVOSO

Giacomini Andrea (1), Raspanti Alessandra (2), Magistretti Jacopo (2), Calzà Laura (3), Bartesaghi Renata (1)

(1) Department of Biomedical and Neuromotor Sciences, University of Bologna, Italy. - Piazza di Porta San Donato 2, 40126 Bologna, Tel. 0512091727 - Fax 0512091737 - e-mail renata.bartesaghi@unibo.it
(2) Department of Biology and Biotechnology "L. Spallanzani", University of Pavia, Italy
(3) Health Sciences and Technologies - Interdepartmental Center for Industrial Research (HST-ICIR), University of Bologna, Italy

ABSTRACT N. 105

Telethon Research Projects - Neurological Diseases		
Principal Investigator	**CATTANEO ELENA**	
Telethon grant N.	**GGP12122**	
Total budget €	**192.000**	
Centres: **1**	*Duration (yrs):* **2**	*Starting year:* **2012**

IMPACT OF REDUCED GLIAL-DERIVED CHOLESTEROL IN HUNTINGTON'S DISEASE

IMPATTO DELLA MINOR PRODUZIONE DI COLESTEROLO DI ORIGINE GLIALE NELLA MALATTIA DI HUNTINGTON

Valenza Marta (1), Di Paolo Eleonora (1), Marullo Manuela (1), Cesana Elisabetta (2), Zuccato Chiara (1), Biella Gerardo (2), Cattaneo Elena (1)

(1) Department of Biosciences and Centre for Stem Cell Research, Università degli Studi di Milano (Italy) - via Viotti 3/5, 20133 Milano; tel: 02-50325840; elena.cattaneo@unimi.it
(2) Department of Biology and Biotechnology "Lazzaro Spallanzani", University of Pavia, Pavia

ABSTRACT N. 106

Telethon Research Projects - Neurological Diseases		
Principal Investigator	**ZUCCATO CHIARA**	
Telethon grant N.	**GGP13053**	
Total budget €	**180.900**	
Centres: **1**	*Duration (yrs):* **2**	*Starting year:* **2013**

ROLE OF ADAM10 IN HUNTINGTON'S DISEASE

IL RUOLO DI ADAM10 NELLA MALATTIA DI HUNTINGTON

Battaglia Elisa (1), Conforti Paola (1), Talpo Francesca (2), Saftig Paul (3), Biella Gerardo (2), Cattaneo Elena (1), Zuccato Chiara (1)

(1) Department of Biosciences and Center for Stem Cell Research, Università degli Studi di Milano - Via Viotti 3/5 20133 Milano (Italy) phone: +39 02 50325839 fax: +39 02 50325843 email: chiara.zuccato@unimi.it
(2) Department of Biology and Biotechnologies, University of Pavia
(3) Institute of Biochemistry, Christian Albrechts University of Kiel

ABSTRACT N. 107

DTI - Neurological Diseases		
Principal Investigator	**MARTELLO GRAZIANO**	
Telethon grant N.	**TCP13013**	
Total budget €	**517.000**	
Centres: **1**	*Duration (yrs):* **5**	*Starting year:* **2014**

DISSECTING THE MOLECULAR FUNCTION OF MUTANT HUNTINGTIN WITH STEM CELLS

CARATTERIZZARE A LIVELLO MOLECOLARE L'ATTIVITÀ DI HUNTINGTIN MUTATA UTILIZZANDO CELLULE STAMINALI

Ferlazzo Giorgia, Carbognin Elena, Martello Graziano

Dipartimento di Medicina Molecolare, Università di Padova - Complesso Vallisneri - Viale Colombo 3, 35131 Padova. - tel: +39 049 8276088 fax: +39 049 8276079 email: graziano.martello@unipd.it GMartello@Dti.Telethon.it

ABSTRACT N. 108

Telethon Research Projects - Neurological Diseases		
Principal Investigator	**AROSIO PAOLO**	
Telethon grant N.	**GGP10099**	
Total budget €	**585.800**	
Centres: **5**	*Duration (yrs):* **3**	*Starting year:* **2010**
Partners	OTTAVIO CREMONA, MARCO GIORGIO, FABIO GROHOVAZ, SONIA LEVI	

ANIMAL MODELS OF NEUROFERRITINOPATHIES FOR THE STUDY OF THE ROLE OF IRON IN NEURODEGENERATION

MODELLI ANIMALI DI NEUROFERRITINOPATIE PER LO STUDIO DEL RUOLO DEL FERRO NELLA NEURODEGENERAZIONE

Cremona Ottavio (2), Giorgio Marco (3), Grohovaz Fabio (2), Levi Sonia (2), Arosio Paolo (1)

(1) Laboratorio di Biologia Molecolare, Dipartimento di Medicina Molecolare e Traslazionale, Univesrità di Brescia - Viale Europa 11, 25123 Brescia, tel. 030-3717303/1, fax: 030-3717305, email: paolo.arosio@unibs.it
(2) Università Vita-Salute & San Raffaele Scientific Institute, Via Olgettina 58, 20132 Milano
(3) Dipartimento di Oncologia Sperimentale, Istituto Europeo di Oncologia, Via Adamello, 16 -20139 Milano

ABSTRACT N. 109

Telethon Research Projects - Neurological Diseases		
Principal Investigator	**LEVI SONIA**	
Telethon grant N.	**GGP11088**	
Total budget €	**502.400**	
Centres: **3**	*Duration (yrs):* **3**	*Starting year:* **2011**
Partners	SONIA LEVI, DARIO FINAZZI, VALERIA TIRANTI	

THE ROLE OF IRON AND MITOCHONDRIA IN THE PATHOGENESIS OF PANTOTHENATE KINASE ASSOCIATED NEURODEGENERATION (PKAN): DEVELOPMENT OF NEW NEURONAL CELLULAR SYSTEMS AND ANALYSIS OF A MOUSE MODEL

RUOLO DEL FERRO E DEI MITOCONDRI NELLA PATOGENESI DELLA NEURODEGENERAZIONE ASSOCIATA A PANTOTENATO CHINASI (PKAN): SVILUPPO DI NUOVI MODELLI CELLULARI NEURONALI E ANALISI DI UN MODELLO DI MALATTIA MURINO

Santambrogio Paolo (2), Guaraldo Michela (3), Dusi Sabrina (4), Colombelli Cristina (4), Aoun Manar (4), Finazzi Dario (5), Tiranti Valeria (4), Levi Sonia (1)

(1) University Vita-Salute San Raffaele, Milano and Division of Neuroscience, San Raffaele Scientific Institute, Milano - Via Olgettina 58, Milano, Italy, 0226434755, 0226434844, levi.sonia@hsr.it
(2) Division of Neuroscience, San Raffaele Scientific Institute, Milano
(3) University Vita-Salute San Raffaele, Milano
(4) IRCCS Foundation Neurological Institute Carlo Besta, Milano
(5) Università di Brescia, Brescia; Italy

ABSTRACT N. 110

Telethon Research Projects - Neurological Diseases		
Principal Investigator	**DE FRANCESCHI LUCIA**	
Telethon grant N.	**GGP13005**	
Total budget €	**185.800**	
Centres: **1**	*Duration (yrs):* **3**	*Starting year:* **2013**

ANALYSIS OF LYN CORE SIGNALING MACHINERY IN NEUROACANTHOCYTOSIS

ANALISI FUNZIONALE DEL SISTEMA LYN RELATO NELLA NEUROACANTOCITOSI

lupo Francesca (1), Tibaldi Elena (2), Matte' Alessandro (1), Brunati AnnaMaria (2), Siciliano Angela (1), Danek Adrian (3), Walker Ruth H (4), Hermann Andreas (5), De Franceschi Lucia (1)

(1) Department of Medicine, University of Verona; Policlinico GB Rossi, P.Le L. Scuro, 10; 37134 Verona +390458124918; +390458027473; lucia.defranceschi@univr.it
(2) Department of Department of Biochemstry, University of Padova, Padova; Padova, Italy
(3) Department of Neurology, Ludwig-Maximilians-Universität, Munich, Germany
(4) Department of Neurology, James J Peters VAMC, Bronx, New York, and Mount Sinai School of Medicine, New York, USA
(5) Department of Neurology, Dresden University of Technology and German Centre for Neurodegenerative Disease (DZNE), Dresden,Germany

ABSTRACT N. 111

Telethon Research Projects - Neurological Diseases		
Principal Investigator	**CHIEREGATTI EVELINA**	
Telethon grant N.	**GGP10109**	
Total budget €	**187.500**	
Centres: **1**	*Duration (yrs):* **3**	*Starting year:* **2010**

PATHOGENETIC MECHANISMS OF FAMILIAL PARKINSON'S DISEASE: WT AND A30P ALPHA-SYNUCLEINS AFFECT THE STRUCTURE OF MICROFILAMENTS AND INTERMEDIATE FILAMENTS. PATHWAYS AND EFFECTS ON CYTOSKELETAL DYNAMICS

MECCANISMI PATOGENETICI DEL MORBO DI PARKINSON FAMILIARE: ALPHA-SYNUCLEINA E LA SUA FORMA MUTATA A30P AGISCONO SULLA STRUTTURA DEI MICROFILAMENTI E DEI FILAMENTI INTERMEDI. VIE DI SEGNALAZIONE ED EFFETTI SULLE DINAMICHE CITOSCHELETRICHE

Emanuele Marco (1), Ronzitti Giuseppe (1,2), Bellani Serena (3), Bucci Giovanna (4,5), Mescola Andrea (6), Leo Damiana (1), Tilve Sharada (1), Canale Claudio (6), Difato Francesco (1), Gainetdinov Raul (1,7), Mochida Sumiko (8), Stephens Gary (4), Valtorta Flavia (3), Chieregatti Evelina (1)

(1) Department of Neuroscience and Brain Technologies, Istituto Italiano di Tecnologia, via Morego 30, 16163, Genova, Italy - +39-010-71781740, fax: +39-010-71781230, email: evelina.chieregatti@iit.it
(2) Genethon, Evry, 91000 France
(3) Division of Neuroscience, San Raffaele Scientific Institute and Vita-Salute University, 20132 Milano, Italy
(4) School of Pharmacy, University of Reading, Whiteknights, Reading, RG6 6AJ, UK
(5) Department of Physics, University of Oxford, Oxford, OX1 3PU, UK
(6) Department of Nanophysics, Istituto Italiano di Tecnologia, 16163, Genova, Italy
(7) Skolkovo Institute of Science and Technology Skolkovo, Moscow region, 143025 and Faculty of Biology and Soil Science, St. Petersburg State University, St. Petersburg, 199034, Russia
(8) Department of Physiology, Tokyo Medical University, 6-1-1 Shinjuku, Tokyo 160-8402, Japan

ABSTRACT N. 112

Telethon Research Projects - Neurological Diseases		
Principal Investigator	**VALENTE ENZA MARIA**	
Telethon grant N.	**GGP10140**	
Total budget €	**180.000**	
Centres: **1**	*Duration (yrs):* **2**	*Starting year:* **2010**

PINK1, MUTATED IN AUTOSOMAL RECESSIVE PARKINSON'S DISEASE, INTERACTS WITH THE PROAUTOPHAGIC PROTEIN BECLIN1 AND ITS ANTIAPOPTOTIC PARTNER BCL-XL: UNRAVELING THE SIGNIFICANCE OF THESE INTERACTIONS AT THE CROSSROAD OF MULTIPLE NEUROPROTECTIVE PATHWAYS

PINK1, UNA PROTEINA MUTATA NELLA MALATTIA DI PARKINSON A TRASMISSIONE AUTOSOMICA RECESSIVA, INTERAGISCE CON LA PROTEINA PROAUTOFAGICA BECLIN1 E CON IL SUO PARTNER ANTIAPOPTOTICO BCL-XL: CARATTERIZZAZIONE DEL RUOLO NEUROPROTETTIVO DI QUESTE INTERAZIONI ATTRAVERSO LA REGOLAZIONE DI MULTIPLE VIE CELLULARI

Gelmetti Vania (1), Torosantucci Liliana (1), De Rosa Priscilla (1), Arena Giuseppe (1), Romagnoli Alessandra (3), Di Rienzo Martina (3,5), Piacentini Mauro (3,5), Fimia Gianmaria (3,4), Valente Enza Maria (1,2)

(1) IRCCS Casa Sollievo della Sofferenza, CSS-Mendel Institute - viale Regina Margherita 261 - 00198 Rome, Italy. Tel +39 06 4416 0537, fax +39 06 4416 0548, email: e.valente@css-mendel.it
(2) Section of Neurosciences, Dept. of Medicine and Surgery, University of Salerno, Salerno, Italy
(3) National Institute for Infectious Diseases "Lazzaro Spallanzani" IRCCS
(4) Dept. of Biological and Environmental Sciences and Technologies (DiSTeBA), University of Salento, Lecce, Italy
(5) Dept. of Biology, University of Rome 'Tor Vergata', Rome, Italy

ABSTRACT N. 113

Telethon Research Projects - Neurological Diseases		
Principal Investigator	**GOLDWURM STEFANO**	
Telethon grant N.	**GGP11164**	
Total budget €	**94.500**	
Centres: **3**	*Duration (yrs):* **2**	*Starting year:* **2011**
Partners	JOHN LANDERS, STEFANO DUGA	

IDENTIFICATION OF RECESSIVE GENES CAUSATIVE FOR PARKINSON'S DISEASE USING EXOME SEQUENCING

IDENTIFICAZIONE DI GENI RECESSIVI CHE PROVOCANO LA MALATTIA DI PARKINSON TRAMITE SEQUENZA COMPLETA DELL'ESOMA

Duga Stefano (3), Landers John (2), Tesei Silvana (1), Cilia Roberto

(1), Siri Chiara (1), Asselta Rosanna (3), Soldà Giulia (3), Rimoldi Valeria (3), Pezzoli Gianni (1), Goldwurm Stefano (1)

(1) Parkinson Institute, Istituti Clinici di Perfezionamento, Milan, Italy. - Via Bignami 1, 20126, Mlano, tel +39 02 57993319; Fax +39 02 5799 3468; email: goldwurm@parkinson.it
(2) University of Massachusetts Medical School, Worcester, Massachusetts, USA
(3) Humanitas University, Rozzano (Milan), Italy

ABSTRACT N. 114

Telethon Research Projects - Neurological Diseases		
Principal Investigator	**GREGGIO ELISA**	
Telethon grant N.	**GGP12237**	
Total budget €	**350.000**	
Centres: **4**	*Duration (yrs):* **3**	*Starting year:* **2012**
Partners	GIOVANNI PICCOLI, FRANCO ONOFRI, MICHELE MORARI	

FUNCTION AND DYSFUNCTION OF THE PARKINSON'S DISEASE KINASE LRRK2 AT THE PRE-SYNAPTIC SITE

FUNZIONE E DISFUNZIONE A LIVELLO PRESINAPTICO DI LRRK2, UNA PROTEINA CHINASI ASSOCIATA ALLA MALATTIA DI PARKINSON

Civiero Laura (1), Cirnaru Maria-Daniela (2), Marte Antonella (3), Longo Francesco (4), Russo Isabella (1), Belluzzi Elisa (1), Gonnelli Adriano (1), Bubacco Luigi (1), Morari Michele (4), Onofri Franco (3), Piccoli Giovanni (2), Greggio Elisa (1)

(1) Department of Biology, University of Padova, Via Ugo Bassi 58/B, 35131, Padova, Italy. Tel: +39 0498276244, Fax: +39 0498276300, email: elisa.greggio@unipd.it
(2) Division of Neuroscience, San Raffaele Scientific Institute and Vita-Salute University, Milan, Italy
(3) Department of Experimental Medicine, University of Genova, Genova, Italy
(4) Department of Medical Sciences, Section of Pharmacology, University of Ferrara, Ferrara, Italy

ABSTRACT N. 115

Telethon Research Projects - Neurological Diseases		
Principal Investigator	**FORLONI GIANLUIGI**	
Telethon grant N.	**GGP10208**	
Total budget €	**237.200**	
Centres: **3**	*Duration (yrs):* **3**	*Starting year:* **2011**
Partners	BENEDETTO IGNAZIO ROITER, FABRIZIO TAGLIAVINI	

FATAL FAMILIAL INSOMNIA: PREVENTIVE TREATMENT WITH DOXYCYCLINE OF AT RISK INDIVIDUALS

INSONNIA FATALE FAMILIARE: TRATTAMENTO PREVENTIVO CON DOXICICLINA IN SOGGETTI A RISCHIO GENETICO DI MALATTIA

Forloni Gianluigi (1), Tettamanti Mauro (1), Lucca Ugo (1), Albanese Yasmin (1), Chiesa Roberto (1), Erbetta Alessandra (2), Villani Flavio (2), Redaelli Veronica (2), Tagliavini Fabrizio (2), Artuso Vladimiro (3), Roiter Ignazio (3)

(1) Dipartimento di Neuroscienze, Istituto di Ricerche Farmacologiche "Mario Negri", Via La Masa 19, 20156 Milano, Italy Tel.02.39014462; Fax 0239001916
(2) Fondazione IRCCS - Istituto Neurologico "Carlo Besta", Milano, Italy
(3) Dipartmento di Medicina Interna, Unità Operativa di Medicina, Oderzo - ASL 9 Treviso ASL 9 Treviso, Italy

ABSTRACT N. 116

Telethon Research Projects - Neurological Diseases		
Principal Investigator	**MATTEOLI MICHELA**	
Telethon grant N.	**GGP12115**	
Total budget €	**374.800**	
Centres: **2**	*Duration (yrs):* **3**	*Starting year:* **2012**
Partners	ROBERTO CHIESA	

MUTANT PRION PROTEIN IMPAIRS DELIVERY OF VOLTAGE GATED CALCIUM CHANNELS TO THE PRESYNAPTIC MEMBRANE: MECHANISMS OF NEUROTOXICITY AND POTENTIAL THERAPEUTIC STRATEGIES

LA PROTEINA PRIONICA MUTATA IMPEDISCE L'INSERZIONE DI CANALI PER IL CALCIO VOLTAGGIO-DIPENDENTI NELLA MEMBRANA PRESINAPTICA: ANALISI DEI MECCANISMI DI NEUROTOSSICITÀ E POTENZIALI STRATEGIE TERAPEUTICHE

Morini Raffaella (1), Restelli Elena (2), Ghirardini Elsa (1), Senatore Assunta (2), Pozzoli Manuela (2), Bertani Ilaria (2), Chiesa Roberto (2), Matteoli Michela (1)

(1) IRCCS Humanitas Research Institute, 20089 Rozzano (MI), Italy. - Laboratory of Pharmacology and Brain Pathology,IRCCS Humanitas Research Institute, via Manzoni 56, 20089 Rozzano (MI), Italy. Tel: 0282245202, michela.matteoli@humanitasresearch.it
(2) IRCCS - Mario Negri Institute for Pharmacological Research, 20156 Milano, Italy

ABSTRACT N. 117

DTI - Neurological Diseases		
Principal Investigator	**CHIESA ROBERTO**	
Telethon grant N.	**TCR08005**	
Total budget €	**800.000**	
Centres: **1**	*Duration (yrs):* **5**	*Starting year:* **2009**

CELLULAR MECHANISMS OF SYNAPTIC DYSFUNCTION IN INHERITED PRION DISEASES

MECCANISMI CELLULARI DI DISFUNZIONE SINAPTICA NELLE MALATTIE DA PRIONI FAMILIARI

Bouybayoune Ihssane (1), Mantovani Susanna (1), Del Gallo Federico (2), Bertani Ilaria (1), Restelli Elena (1), Comerio Liliana (1), Tapella Laura (1), Baracchi Francesca (2), Fernández-Borges Natalia (3), Mangieri Michela (4), Bisighini Cinzia (1), Beznoussenko Galina v (5), Paladini Alessandra (1), Balducci Claudia (1), Micotti Edoardo (1), Forloni Gianluigi (1), Castilla Joaquìn (3), Fiordaliso Fabio (1), Tagliavini Fabrizio (4), Imeri Luca (2), Chiesa Roberto (1)

(1) IRCCS - Istituto di Ricerche Farmacologiche Mario Negri - Via G. La Masa 19 20156 Milano, Italy Tel. 0239014428, e-mail: roberto.chiesa@marionegri.it
(2) Università degli Studi di Milano, Italy
(3) CIC bioGUNE, Parque Tecnológico de Bizkaia, Spain
(4) IRCCS - Istituto Neurologico Carlo Besta, Milano, Italy
(5) IFOM - FIRC Istituto di Oncologia Molecolare, Milano, Italy

ABSTRACT N. 118

Telethon Research Projects - Neurological Diseases		
Principal Investigator	**LANDSBERGER NICOLETTA**	
Telethon grant N.	**GGP10032**	
Total budget €	**398.600**	
Centres: **2**	*Duration (yrs):* **3**	*Starting year:* **2010**
Partners	SILVIA SODDU	

MECP2 PHOSPHORYLATION AND RELATED KINASES IN RETT SYNDROME AND EARLY INFANTILE EPILEPTIC ENCEPHALOPATHY 2

RUOLO DELLA FOSFORILAZIONE DI MECP2 E DELLE CHINASI IMPLICATE NELLA SINDROME DI RETT E NELL'ENCEFALOPATIA EPILETTICA INFANTILE PRECOCE-2

La Montanara Paolo (2), Rusconi Laura (2), Barbiero Isabella (2), Tramarin Marco (2), Kilstrup-Nielsen Charlotte (2), Landsberger Nicoletta (1,2)

(1) Division of Neuroscience, San Raffaele Scientific Institute, Via Olgettina 58, 20132 Milano - Tel.: 02-26436278; email: landsberger.nicoletta@hsr.it
(2) Department of Theoretical and Applied Sciences, Section of Biomedical Research, University of Insubria, Busto Arsizio

ABSTRACT N. 119

Telethon Research Projects - Neurological Diseases		
Principal Investigator	**BROCCOLI VANIA**	
Telethon grant N.	**GGP11110**	
Total budget €	**349.700**	
Centres: **2**	*Duration (yrs):* **3**	*Starting year:* **2011**
Partners	ALESSANDRA RENIERI	

MOLECULAR BASES AND IN VITRO MODELING OF CDKL5 DEPENDENT INFANTILE NEUROLOGICAL DISORDERS

BASI MOLECOLARI E MODELLI CELLULARI DELLE MALATTIE NEUROLOGICHE INFANTILI CAUSATE DA MUTAZIONI DEL GENE CDKL5

Ricciardi Sara (1,2), Stefanelli Gilda (1,3), Scarongella Helena (1), Biffo Stefano (2), Broccoli Vania (1)

(1) San Raffaele Scientific Institute, Via Olgettina 58, 20132 Milan
(2) INGM, Fondazione Istituto Nazionale Genetica Molecolare, Milan
(3) Department of Theoretical and Applied Sciences, Division of Biomedical Research, University of Insubria, Busto Arsizio

ABSTRACT N. 120

Telethon Research Projects - Neurological Diseases		
Principal Investigator	**CIANI ELISABETTA**	
Telethon grant N.	**GGP11147**	
Total budget €	**458.700**	
Centres: **4**	*Duration (yrs):* **3**	*Starting year:* **2011**
Partners	LAURA CALZÀ, TOMMASO PIZZORUSSO, GIOVANNI PERINI	

MECHANISMS UNDERLYING NEURODEVELOPMENTAL ALTERATIONS IN A MOUSE MODEL OF CDKL5 DISORDER

MECCANISMI ALLA BASE DELLE ALTERAZIONI CEREBRALI NEL DISORDINE CDKL5

De Franceschi Marianna (1), Pizzorusso Tommaso (2,3), Della Sala Grazia (3), Giustetto Maurizio (4), Pizzo Riccardo (4), Calzà Laura (6), Sivilia Sandra (6), Perini Giovanni (5), Ciani Elisabetta (1)

(1) Dipartimento di Scienze Biomediche e Neuromotorie, Università di Bologna - DIBINEM, Piazza di Porta San Donato 2, 40126 Bologna, tel 051 2091773, fax 051 2091737, email: elisabetta.ciani@unibo.it
(2) Instituto di Neuroscience CNR, Pisa
(3) Dipartimento NEUROFARBA, Università di Firenze
(4) Dipartimento di Neuroscience, Università di Torino
(5) Dipartimento di Farmacia e Biotecnologie, Università di Bologna
(6) Dipartimento di Scienze Mediche e Chirurgiche, Università di Bologna

ABSTRACT N. 121

Telethon Research Projects - Neurological Diseases		
Principal Investigator	**SALA CARLO**	
Telethon grant N.	**GGP11095**	
Total budget €	**429.000**	
Centres: **4**	*Duration (yrs):* **3**	*Starting year:* **2012**
Partners	ALEXANDER DITYATEV, VANIA BROCCOLI, MAURIZIO GIUSTETTO	

IDENTIFICATION OF NEURONAL ALTERATIONS UNDERLYING SHANK3 MUTATIONS AND THEIR RESCUE BY GENETIC/PHARMACOLOGICAL THERAPIES IN ANIMAL MODELS AND PATIENTS' DERIVED IPS CELLS

CARATTERIZZAZIONE DELLE ALTERAZIONI NEURONALI INDOTTE DALLE MUTAZIONI DI SHANK3 E SVILUPPO DI APPROCCI TERAPEUTICI FARMACOLOGICI E GENETICI UTILIZZANDO MODELLI ANIMALI E CELLULE IPS DEI PAZIENTI

Verpelli Chiara (1), Vicidomini Cinzia (1), Mossa Adele (1), Schmeisser Michael J. (2), Scalmani Paolo (3), Ponzoni Luisa (4), Sala Mariaelvina (4,1), Mantegazza Massimo (3), Böckers Tobias M. (2), Broccoli Vania (5), Giustetto Maurizio (6), Morello Noemi (6), Dityatev Alexander (7), Sala Carlo (1)

(1) CNR Istituto di Neuroscienze - Via Vanvitelli 32 - 20129 Milano - Tel. 0250317096 - Fax 0250317132 - Email c.sala@in.cnr.it
(2) Institute for Anatomy and Cell Biology, Ulm University, Ulm, Germany
(3) Dipartimento di Neurofisiopatogia, Fondazione Instituto Neurologico Carlo Besta, Milano
(4) BIOMETRA Università degli Studi di Milano, Milano
(5) Division of Neuroscience, Fondazione Centro San Raffaele del Monte Tabor, Milano
(6) Dipartimento di Anatomia, Farmacologia e Medicina Legale, Università di Torino e Istituto Nazionale di Neuroscienze, Torino
(7) Dipartimento di Neuroscienze Fondazione Istituto Italiano di Tecnologia, Genova

ABSTRACT N. 122

Telethon Research Projects - Neurological Diseases		
Principal Investigator	**CHERUBINI ENRICO**	
Telethon grant N.	**GGP11043**	
Total budget €	**385.600**	
Centres: **2**	*Duration (yrs):* **3**	*Starting year:* **2011**
Partners	ANDREA BARBERIS	

MECHANISMS UNDERLYING ALTERED GABAERGIC SIGNALING IN THE HIPPOCAMPUS OF TRANSGENIC MICE CARRYING THE HUMAN R451C MUTATION OF THE NLG3 GENE: AN ANIMAL MODEL OF AUTISM

MECCANISMI ALLA BASE DI UNA ALTERATA TRASMISSIONE GABAERGICA NELL'IPPOCAMPO DI TOPI TRANSGENICI PORTATORI DELLA MUTAZIONE UMANA R451C NEL GENE CODIFICANTE LA NLG3: UN MODELLO ANIMALE DI AUTISMO

Barberis Andrea (1), Bialowas Andrzej Jan (2), Cellot Giada (2), Cherubini Enrico (2,3), Petrini Enrica Maria (1), Ruggeri Federica (2), Stancheva Stefka (1), Wiera Grzegorz (2), Zacchi Paola (2)

(1) Department of Neuroscience and Brain Technologies, The Italian Institute of Technology, Via Morego 30, 16163 Genova, Italy. - Enrico Cherubini, SISSA, Via Bonomea 265, 34136 Trieste, Italy. Tel: 040-3787704; Fax: 040-3787702; E-mail: cher@sissa.it
(2) Department of Neuroscience, International School for Advanced Studies (SISSA), Via Bonomea 265, 34136 Trieste, Italy
(3) European Brain Research Institute (EBRI), Via del Fosso di Fiorano 64, 00143 Roma, Italy

ABSTRACT N. 123

Telethon Research Projects - Neurological Diseases		
Principal Investigator	**SICCA FEDERICO**	
Telethon grant N.	**GGP11188**	
Total budget €	**384.300**	
Centres: **3**	*Duration (yrs):* **3**	*Starting year:* **2011**
Partners	MAURO PESSIA, ELENA AMBROSINI	

ROLE OF ASTROCYTIC INWARDLY-RECTIFYING K CHANNELS IN THE PATHOGENESIS OF AUTISM SPECTRUM DISORDERS WITH SUSCEPTIBILITY TO SEIZURES (AUTISM-EPILEPSY PHENOTYPE)

RUOLO DEI CANALI RETTIFICATORI DI INGRESSO DEL K+ ASTROCITARI NELLA PATOGENESI DEI DISTURBI DELLO SPETTRO AUTISTICO CON SUSCETTIBILITÀ ALLE CRISI EPILETTICHE (FENOTIPO AUTISMO-EPILESSIA)

Ambrosini Elena (2), Pessia Mauro (3), Apicella Fabio (1), Baldini Sara (1), Brignone Maria Stefania (2), Conti Valerio (4), D'Adamo Maria Cristina (3), Grottesi Alessandro (5), Guerrini Renzo (4,1), Lanciotti Angela (2), Marchese Maria (1), Moro Francesca (1), Napolitano Carlo (6), Priori Silvia (6), Santorelli Filippo Maria (1), Tancredi Raffaella (1), Valvo Giulia (1), Sicca Federico (1)

(1) Clinical Neurophysiology and Neurogenetics Laboratories, Department of Developmental Neuroscience, IRCCS Stella Maris Foundation - Via dei Giacinti 2, Calambrone, 56128, Pisa - Italy - Phone number: +39050886234, Fax number: +39050886247, email: federico.sicca@fsm.unipi.it
(2) Department of Cell Biology and Neuroscience, Istituto Superiore di Sanità, Rome
(3) Department of Internal Medicine - Section of Human Physiology, University of Perugia, Perugia
(4) Pediatric Neurology Unit, University Hospital A. Meyer, Florence
(5) Computational Medicine and Biology Group, CASPUR, Rome
(6) Molecular Cardiology, IRCCS Salvatore Maugeri Foundation, Pavia

ABSTRACT N. 124

Telethon Research Projects - Neurological Diseases		
Principal Investigator	**MORONI ANNA**	
Telethon grant N.	**GEP14137**	
Total budget €	**49.500**	
Centres: **1**	*Duration (yrs):* **1**	*Starting year:* **2015**

STRUCTURAL AND FUNCTIONAL STUDIES OF HCN1 CHANNEL MUTATIONS CAUSING EARLY INFANTILE EPILEPTIC ENCEPHALOPATHY

STUDIO STRUTTURALE E FUNZIONALE DELLE MUTAZIONI DEI CANALI HCN1 RESPONSABILI DI ENCEFALOPATIA EPILETTICA INFANTILE PRECOCE

Alfieri Andrea, Porro Alessandro, Moroni Anna

Dipartimento di Bioscienze - Università degli Studi di Milano, via Celoria 23, 20133 Milano - Tel. +39-0250314826 - Email: anna.moroni@unimi.it

ABSTRACT N. 125

Telethon Research Projects - Neurological Diseases		
Principal Investigator	**CIRANNA LUCIA**	
Telethon grant N.	**GGP13145**	
Total budget €	**329.921**	
Centres: **3**	*Duration (yrs):* **3**	*Starting year:* **2013**
Partners	MARIA VINCENZA CATANIA, MARCELLO LEOPOLDO	

ACTIVATION OF SEROTONIN TYPE 7 (5-HT7) RECEPTORS AS A NOVEL THERAPEUTIC STRATEGY IN FRAGILE X SYNDROME

UNA NUOVA STRATEGIA TERAPEUTICA PER LA SINDROME DEL CROMOSOMA X FRAGILE BASATA SU NUOVE MOLECOLE ATTIVANTI I RECETTORI 5-HT7 PER LA SEROTONINA

Costa Lara (2), Sardone Lara Maria (1), Spatuzza Michela (3), Bonaccorso Carmela Maria (4), D'Antoni Simona (3), Puzzo Daniela (1), Gulisano Walter (1), Leopoldo Marcello (5), Lacivita Enza (5), Catania Maria Vincenza (3,4), Ciranna Lucia (1)

(1) Dipartimento di Scienze Biomediche e Biotecnologiche, Università di Catania, Italia - Viale A. Doria 6, Edificio 2, Città Universitaria, 95125 Catania; tel 095 7384111, fax 095 7384217; ciranna@unict.it
(2) Dipartimento di Medicina Clinica e Sperimentale, Università di Messina, Italia
(3) Istituto di Scienze Neurologiche, CNR, Catania, Italia
(4) Laboratorio di Neurobiologia, IRCCS Oasi Maria Santissima, Troina (EN), Italia
(5) Dipartimento di Farmacia, Università di Bari, Italia

ABSTRACT N. 126

Telethon Research Projects - Neurological Diseases		
Principal Investigator	**BOZZETTI MARIA GIUSEPPINA**	
Telethon grant N.	**GGP14181**	
Total budget €	**203.400**	
Centres: **1**	*Duration (yrs):* **3**	*Starting year:* **2014**

DROSOPHILA MELANOGASTER AS A MODEL TO STUDY THE ROLE OF THE FRAGILE X MENTAL RETARDATION PROTEIN IN THE GENOME STABILITY PATHWAY MEDIATED BY PIRNAS

DROSOPHILA MELANOGASTER COME MODELLO PER STUDIARE IL RUOLO DELLA PROTEINA FMRP, COINVOLTA NELLA SINDROME DELL'X FRAGILE NELLA STABILITÀ GENOMICA MEDIATA DAI PIRNA

Specchia Valeria (1), Laneve Pietro (2), Massari Serafina (1), Diebold Celine (3), Friscini Antonella (1), Giangrande Angela (3), Bozzetti Maria Giuseppina (1)

(1) Dipartimento di Scienze e Tecnologie Biologiche ed Ambientali (DiSTeBA) -Università del Salento - Via Monteroni 165, 73100 Lecce Telefono: 0832 298681, Fax: 0832 298626, email: maria.bozzetti@unisalento.it
(2) Center for Life NanoScience, Istituto Italiano di Tecnologia, Rome
(3) Institut de Génétique et de Biologie Moléculaire et Cellulaire, Illkirch-Strasbourg

ABSTRACT N. 127

Telethon Research Projects - Neurological Diseases		
Principal Investigator	**CALEO MATTEO**	
Telethon grant N.	**GGP11116**	
Total budget €	**348.600**	
Centres: **3**	*Duration (yrs):* **3**	*Starting year:* **2011**
Partners	CLAUDIA LODOVICHI, MARIA PASSAFARO	

ROLE OF OLIGOPHRENIN-1 IN CIRCUIT FORMATION AND FUNCTION IN A MOUSE MODEL OF X-LINKED MENTAL RETARDATION

SVILUPPO E FUNZIONE DEI CIRCUITI NEURONALI IN UN MODELLO DI RITARDO MENTALE ASSOCIATO AL CROMOSOMA X

Allegra Manuela (1), Spalletti Cristina (1,2), Azzimondi Stefano (1), Redolfi Nelly (3,4), Galla Luisa (3,4), Murru Luca (5), Vignoli Beatrice (6), Canossa Marco (6), Passafaro Maria (5), Lodovichi Claudia (3,4), Caleo Matteo (1)

(1) CNR Neuroscience Institute - via G. Moruzzi 1, 56124 Pisa (Italy); Tel.: 050-3153195, Fax: 050-3153220, e-mail: caleo@in.cnr.it
(2) Istituto di Scienze della Vita, Scuola Superiore Sant'Anna, Pisa, Italy
(3) CNR Neuroscience Institute, Padua, Italy
(4) VIMM, Padua, Italy
(5) CNR Neuroscience Institute, Milan, Italy
(6) Dipartimento di Farmacia e Biotecnologie, Bologna, Italy

ABSTRACT N. 128

Telethon Research Projects - Neurological Diseases		
Principal Investigator	**PASSAFARO MARIA**	
Telethon grant N.	**GGP12097**	
Total budget €	**255.400**	
Centres: **2**	*Duration (yrs):* **3**	*Starting year:* **2012**
Partners	PATRIZIA D'ADAMO	

ANALYSIS OF NEURONAL ALTERATIONS ASSOCIATED TO TM4SF2 MUTATIONS AND THEIR RESCUE BY GENETIC AND PHARMACOLOGICAL THERAPIES

ANALISI DELLE ALTERAZIONI NEURONALI ASSOCIATE A MUTAZIONI NEL GENE TM4SF2 E RIPRISTINO MEDIANTE TERAPIE GENETICHE E FARMACOLOGICHE

Folci Alessandra (1), Murru Luca (1), Vezzoli Elena (2), Bianchi Veronica (3), Bassani Silvia (1), D'Adamo Patrizia (3), Francolini Maura (1,2), Passafaro Maria (1)

(1) CNR Neuroscience Institute, Via Vanvitelli , N°32, 20129 Milan, Italy, - tel. 0250316973, fax 0250317132, m.passafar@in.cnr.it
(2) Università degli Studi di Milano – Dept. of Medical Biotechnology and Translational Medicine, Milano Italy
(3) DTI at San Raffaele Scientific Institute, Milan, Italy

ABSTRACT N. 129

Telethon Research Projects - Neurological Diseases		
Principal Investigator	**DE CURTIS IVAN**	
Telethon grant N.	**GGP12126**	
Total budget €	**284.800**	
Centres: **1**	*Duration (yrs):* **3**	*Starting year:* **2012**

ROLE OF RHO FAMILY GTPASES DURING NEURONAL DEVELOPMENT

RUOLO DELLE GTPASI DELLA FAMIGLIA RHO DURANTE LO SVILUPPO NEURONALE

Macco Romina (1), Pennucci Roberta (1), Talpo Francesca (2), Tonoli Diletta (1), Castoldi Valerio (1), Cursi Marco (1), Leocani Letizia (1), Biella Gerardo (2), D'Adamo Patrizia (1), de Curtis Ivan (1)

(1) Neuroscience Division, San Raffaele Scientific Institute and San Raffaele University - Cell Adhesion Unit, Neuroscience Division, San Raffaele Scientific Institute and San Raffaele University, via Olgettina, 58 - 20132 Milano; tel:0226434828; Fax: 02 2643 4844; e-mail: decurtis.ivan@hsr.it
(2) Dept of Biology and Biotechnology, Università di Pavia, Pavia

ABSTRACT N. 130

DTI - Neurological Diseases		
Principal Investigator	**D'ADAMO PATRIZIA**	
Telethon grant N.	**TCR11002**	
Total budget €	**235.567**	
Centres: **1**	*Duration (yrs):* **3**	*Starting year:* **2012**

ANALYSIS OF RAB39B ROLE IN X-LINKED INTELLECTUAL DISABILITY

ANALISI DEL RUOLO DI RAB39B NEL RITARDO MENTALE ASSOCIATO AL CROMOSOMA X

Mignogna Maria Lidia (1,2), Fjeldskaar Fatiha (2), Fanelli Francesca (3), Raimondi Francesco (3), Mapelli Lisa (4), Bassani Silvia (4), Passafaro Maria (4), Esteban José A. (5), Huganir Richard (6), Gatti Silvia (2), D'Adamo Patrizia (1)

(1) Dulbecco Telethon Institute at San Raffaele Scientific Institute, Division of Neuroscience, Milan - via Olgettina 58, 20132 Milan, Italy-tel 0226434604; fax 0226434844; p.dadamo@hsr.it
(2) F. Hoffmann-La Roche AG, pRED Pharma Research & Early Development, DTA Neuroscience, CH4070 Basel, Switzerland
(3) Dulbecco Telethon Institute at Department of Life Sciences, University of Modena and Reggio Emilia, 41125 Modena, Italy
(4) Dulbecco Telethon Institute at CNR Institute of Neuroscience, Department of BIOMETRIA, University of Milan, 20129 Milan, Italy
(5) Centro de Biología Molecular Severo Ochoa, Consejo Superior de Investigaciones Científicas/Universidad Autónoma de Madrid, Madrid 28049, Spain
(6) Department of Neuroscience and Howard Hughes Medical Institute, Johns Hopkins University School of Medicine, Baltimore, Maryland 21205, USA

ABSTRACT N. 131

Telethon Research Projects - Neurological Diseases		
Principal Investigator	**TESTA GIUSEPPE**	
Telethon grant N.	**GEP14115**	
Total budget €	**49.900**	
Centres: **1**	*Duration (yrs):* **1**	*Starting year:* **2015**

THE CHROMATIN BASIS OF NEUROLOGIC DYSFUNCTION IN THE SWI/SNF-RELATED AUTISM SYNDROME

LE BASI EPIGENETICHE DELLE ALTERAZIONI NEUROLOGICHE NELLA SINDROME AUTISTICA CORRELATA AL COMPLESSO SWI/SNF

Gabriele Michele, Testa Giuseppe

Istituto Europeo di Oncologia, Dipartimento di Oncologia sperimentale - Via Giuseppe Ripamonti, 435, 20141 Milano - Tel. 02/94375105 - Email: giuseppe.testa@ieo.eu

ABSTRACT N. 132

Telethon Research Projects - Neurological Diseases		
Principal Investigator	**CHINI BICE**	
Telethon grant N.	**GGP12207**	
Total budget €	**136.300**	
Centres: **1**	*Duration (yrs):* **2**	*Starting year:* **2012**

OXYTOCIN ANALOGS IN PRADER-WILLI SYNDROME: NEW TOOLS TO INVESTIGATE AND TREAT SOCIAL AND COGNITIVE AUTISTIC-LIKE SYMPTOMS

ANALOGHI DELL'OSSITOCINA NELLA SINDROME DI PRADER-WILLI: NUOVI STRUMENTI PER LO STUDIO ED IL TRATTAMENTO DEI DISTURBI COMPORTAMENTALI DI TIPO AUTISTICO

Busnelli Marta (1,2), Leonzino Marianna (1,2), Kleinau Gunnar (3), Muttenthaler Markus (5), Flavia Antonucci (1,2), Claudia Verderio (1), Daniela Braida (2,1), Mariaelvina Sala (2,1), Manning Maurice (4), Bellini Tommaso (2), Chini Bice (1)

(1) CNR Istituto di Neuroscienze, via Vanvitelli 32, Milano, 02 50316958, 02 50317132, b.chini@in.cnr.it
(2) UNIMI, Dipartimento di Biotecnologie Mediche e Medicina Traslazionale, Milano, Italy

(3) Institute of Experimental Pediatric Endocrinology, Charité-Universitätsmedizin Berlin, Berlin, Germany
(4) Dept. of Biochemistry and Cancer Biology, University of Toledo, Toledo, OH, USA
(5) Institute for Research in Biomedicine Barcelona, 08028 Barcelona, Spain

ABSTRACT N. 133

Telethon Research Projects - Neurological Diseases		
Principal Investigator	**RENIERI ALESSANDRA**	
Telethon grant N.	**GGP09117**	
Total budget €	**293.100**	
Centres: **2**	*Duration (yrs):* **3**	*Starting year:* **2009**
Partners	VANIA BROCCOLI	

CONGENITAL RETT SYNDROME: CELLULAR AND MOUSE MODELS FOR THE STUDY OF FOXG1 IMPACT ON FOREBRAIN NEUROGENESIS

SINDROME DI RETT CONGENITA: MODELLI CELLULARI E MURINI PER LO STUDIO DEL RUOLO DI FOXG1 NELLA NEUROGENESI

Meloni Ilaria (1), Amabile Sonia (1), Patriarchi Tommaso (1), Frullanti Elisa (2), Massimino Luca (3), Costa Mario (4), Broccoli Vania (3), Renieri Alessandra (1,2)

(1) Medical Genetics, University of Siena, Siena, Italy - Policlinico Le Scotte, Viale Bracci 2, 53100 Siena; Phone: 0577 233303; Fax: 0577 233325; e-mail: alessandra.renieri@unisi.it
(2) Genetica Medica, Azienda Ospedaliera Universitaria Senese, Siena, Italy
(3) San Raffaele Scientific Institute, Dibit, 20132 Milano, Italy
(4) Institute of Neuroscience, Italian National Research Council, Pisa, Italy

ABSTRACT N. 134

Telethon Research Projects - Neurological Diseases		
Principal Investigator	**RATTO GIAN MICHELE**	
Telethon grant N.	**GGP13187**	
Total budget €	**537.900**	
Centres: **4**	*Duration (yrs):* **3**	*Starting year:* **2013**
Partners	LAURA CANCEDDA, CLAUDIA LODOVICHI, CARLO SALA	

UNRAVELLING THE RETT SYNDROME: EFFECTS OF MECP2 MUTATIONS ON SYNAPTIC FUNCTION

CAPIRE I MECCANISMI CELLULARI ALLA BASE DELLA SINDROME DI RETT

Ratto Gian Michele (1,2), Cancedda Laura (3), Lodovichi Claudia (4,5), Sala Carlo (6), Verpelli Chiara (6)

(1) Istituto Nanoscienze CNR. - Piazza San Silvestro 12. Pisa. Tel: 050 509417 email gianmichele.ratto@sns.it
(2) Scuola Normale Superiore, Pisa
(3) Istituto Italiano di Tecnologia, Genova
(4) Istituto di Neuroscienze CNR, Padova
(5) Venetian Institute of Molecular Medicine, Padova
(6) Istituto di Neuroscienze CNR, Milano

ABSTRACT N. 135

Telethon Research Projects - Neurological Diseases		
Principal Investigator	**BATTAGLIOLI ELENA**	
Telethon grant N.	**GGP14074**	
Total budget €	**132.500**	
Centres: **1**	*Duration (yrs):* **3**	*Starting year:* **2014**

NEW STRATEGIES TO TARGET HYPER-EXCITABILITY IN RETT SYNDROME

NUOVE STRATEGIE PER CONTRASTARE L'IPER-ECCITABILITÀ PROPRIA DELLA SINDROME DI RETT

Rusconi Francesco, Grillo Barbara, Toffolo Emanuela, Ponzoni Luisa, Sala Mariaelvina

Dept. Medical Biotechnology and Translational Medicine, University of MIlan, Via Viotti 5, 20133 Milano. Phone:39-0250315859, fax: 39-0250315864; email: elena.battaglioli@unimi.it

ABSTRACT N. 136

Telethon Research Projects - Neurological Diseases		
Principal Investigator	**ZUFFARDI ORSETTA**	
Telethon grant N.	**GGP13060**	
Total budget €	**91.600**	
Centres: **1**	*Duration (yrs):* **2**	*Starting year:* **2013**

RING 14 SYNDROME: TOWARD A DETAILED GENOTYPE-PHENOTYPE CORRELATION

RING 14 SYNDROME: TOWARD A DETAILED GENOTYPE-PHENOTYPE CORRELATION

Novara Francesca (1), Rinaldi Berardo (1), Baldo Chiara (2), Giorda Roberto (3), Zuffardi Orsetta (1)

(1) Università degli Studi di Pavia, Dipartimento di Medicina Molecolare - Via Forlanini 14, 27100 Pavia, tel. 0382-987733, email: zuffardi@unipv.it
(2) E.O. Ospedale Galliera, Laboratorio di Genetica, Via A.Volta 6, Genova
(3) Laboratorio di Biologia Molecolare, Istituto Scientifico "E. Medea", IRCCS, V. Don L. Monza 20, Bosisio Parini (LC)

ABSTRACT N. 137

Telethon Research Projects - Neurological Diseases		
Principal Investigator	**TINTI LAURA**	
Telethon grant N.	**GEP13108**	
Total budget €	**31.400**	
Centres: **1**	*Duration (yrs):* **1**	*Starting year:* **2014**

IN VITRO FEASIBILITY STUDY OF A PROTEIN REPLACEMENT THERAPY FOR METHYLMALONIC ACIDEMIA WITH HOMOCYSTINURIA CBLC TYPE: DELIVERY OF RECOMBINANT HUMAN MMACHC PROTEINS INTO PRIMARY FIBROBLASTS FROM CBLC PATIENTS

STUDIO DI FATTIBILITÀ PER LO SVILUPPO IN VITRO DI UNA TERAPIA ENZIMATICA SOSTITUTIVA MIRATA AL TRATTAMENTO DELL'ACIDURIA METILMALONICA CON OMOCISTINURIA DI TIPO CBLC

Salvini Laura (1), Tinti Cristina (1), Giordano Cinzia (1), Chiariello Mario (2), La Marca Giancarlo (3), Donati Alice (3), Tinti Laura (1)

(1) Fondazione Toscana Life Sciences - via Fiorentina 1 53100 Siena tel:+39 0577 231219, fax:+39 0577 43444, l.tinti@toscanalifesciences.org
(2) Istituto di Fisiologia Clinica, Sezione di Siena, Consiglio Nazionale delle Ricerche (CNR)
(3) Ospedale Pediatrico Meyer, Firenze

ABSTRACT N. 138

Telethon Research Projects - Neurological Diseases		
Principal Investigator	**BIFULCO MAURIZIO**	
Telethon grant N.	**GEP14111**	
Total budget €	**50.000**	
Centres: **1**	*Duration (yrs):* **1**	*Starting year:* **2015**

ROLE OF UNPRENYLATED 2',3'-CYCLIC-NUCLEOTIDE 3'-PHOSPHODIESTERASE IN THE MOLECULAR MECHANISMS RESPONSIBLE FOR NEUROINFLAMMATION AND NEUROLOGICAL IMPAIRMENTS IN MEVALONATE KINASE DEFICIENCY

RUOLO DELLA DIFETTIVA PRENILAZIONE DELLA 2',3'- NUCLEOTIDE CICLICO 3'-FOSFODIESTERASI NEI MECCANISMI MOLECOLARI RESPONSABILI DELLA NEUROINFIAMMAZIONE E DELLE ALTERAZIONI NEUROLOGICHE NELLA SINDROME DA DEFICIT DI MEVALONATO CHINASI

Bifulco Maurizio

Dipartimento di Medicina e Chirurgia, Università degli Studi di Salerno, Via Salvatore Allende, Baronissi (SA)

ABSTRACT N. 139

Telethon Research Projects - Neurological Diseases		
Principal Investigator	**ALESSIO MASSIMO**	
Telethon grant N.	**GEP14102**	
Total budget €	**49.940**	
Centres: **1**	*Duration (yrs):* **1**	*Starting year:* **2015**

THERAPEUTIC POTENTIAL OF CERULOPLASMIN ADMINISTRATION IN ACERULOPLASMINEMIA

POTENZIALE TERAPEUTICO DELLA SOMMINISTRAZIONE DI CERULOPLASMINA IN ACERULOPLASMINEMIA

Zanardi Alan (1), Brambilla Riccardo (2), Politi Letterio Salvatore (3), David Samuel (4), Alessio Massimo (1)

(1) Proteome Biochemistry Unit, San Raffele Scientific Institute, Milano, Italy - Email: alessio.massimo@hsr.it
(2) Molecular Genetics Behaviour Unit, San Raffele Scientific Institute, Milano, Italy
(3) Mouse Clinic, San Raffele Scientific Institute, Milano, Italy
(4) Centre for Research in Neuroscience, The Research Institute of the McGill University Health Centre, Montreal (Quebec) Canada

ABSTRACT N. 140

Telethon Research Projects - Neurological Diseases		
Principal Investigator	**GIANSANTI MARIA GRAZIA**	
Telethon grant N.	**GEP14076**	
Total budget €	**50.000**	
Centres: **1**	*Duration (yrs):* **1**	*Starting year:* **2015**

A DROSOPHILA MODEL FOR STUDYING NEUROLOGICAL DEFECTS ASSOCIATED WITH CONGENITAL DISORDER OF GLYCOSYLATION TYPE IIE

LA DROSOPHILA COME SISTEMA MODELLO PER LO STUDIO DEI DIFETTI NEUROLOGICI ASSOCIATI ALLA MALATTIA COG7-CDG

Giansanti Maria Grazia

Dipartimento Di Biologia E Biotecnologia - Università La Sapienza, Roma; Piazzale A. Moro 5, Email: mariagrazia.giansanti@uniroma1.it

ABSTRACT N. 141

Telethon Research Projects - Neurological Diseases		
Principal Investigator	**PALMIERI LUIGI**	
Telethon grant N.	**GEP14141**	
Total budget €	**43.000**	
Centres: **1**	*Duration (yrs):* **1**	*Starting year:* **2015**

MOLECULAR AND CELLULAR UNDERPINNINGS OF THE NEUROLOGICAL PHENOTYPES ASSOCIATED TO MITOCHONDRIAL CITRATE CARRIER (SLC25A1) DEFICIENCY

BASI MOLECOLARI E CELLULARI DEL FENOTIPO NEUROLOGICO ASSOCIATO AL DIFETTO DEL CARRIER MITOCONDRIALE DEL CITRATO (SLC15A1)

Scarcia Pasquale (1), Lasorsa Massimo (2), Porcelli Vito (1), Di Schiavi Elia (3), Palmieri Luigi (1,2)

(1) Dipartimento di Bioscienze, Biotecnologie e Biofarmaceutica - Università degli Studi di Bari Aldo Moro, via E. Orabona n.4, 70125 Bari - Tel. 080/5442771 - Email: luigi.palmieri@uniba.it
(2) Istituto di Biomembrane e Bioenergetica (IBBE), CNR
(3) Istituto di Bioscienze e Biorisorse (IBBR), CNR

ABSTRACT N. 142

Telethon Research Projects - Neurological Diseases		
Principal Investigator	**PERCUDANI RICCARDO**	
Telethon grant N.	**GGP13149**	
Total budget €	**328.600**	
Centres: **2**	*Duration (yrs):* **3**	*Starting year:* **2013**
Partners	MARIA PIA RASTALDI	

DEVELOPMENT OF AN URICOLYTIC TREATMENT FOR HPRT-DEFICIENCY IN ANIMAL MODELS

SVILUPPO DI UNA TERAPIA ENZIMATICA PER LA SINDROME DI LESCH-NYHAN

Corsini Romina (1), Zennaro Cristina (4), Ronda Luca (3), Paredi Gianluca (6), Corbelli Alessandro (2), Zarattini Paola (5), Zanotti Giuseppe (7), Bettati Stefano (3), Rastaldi Maria Pia (2), Percudani Riccardo (1)

(1) Dipartimento di Bioscienze, Università di Parma, Parco area delle Scienze 23/a, Parma, Italia, Tel: 0521 905140; Fax: 0521 905151; email: riccardo.percudani@unipr.it
(2) Renal Research Laboratory, Fondazione IRCCS Ospedale Maggiore Policlinico, Milano
(3) Dipartimento di Neuroscienze, Università di Parma
(4) Dipartimento di Scienze Mediche Chirurgiche e della Salute, Università of Trieste
(5) Dipartimento di Scienze della Vita, Università of Trieste
(6) Dipartimento di Farmacia e Centro Interdipartimentale SITEIA.Parma, Università di Parma
(7) Dipartimento di Scienze Biomediche, Università di Padova

ABSTRACT N. 143

OSR TIGET - Neurological Diseases		
Principal Investigator	**BIFFI ALESSANDRA**	
Telethon grant N.	**TGTGSK07**	
Total budget €	**1.106.592**	
Centres: **1**	*Duration (yrs):* **5**	*Starting year:* **2011**

HEMATOPOIETIC STEM CELL GENE THERAPY FOR THE TREATEMENT OF GLOBOID CELL LEUKODYSTROPHY

SVILUPPO PRECLINICO DELLA TERAPIA GENICA CON CELLULE STAMINALI EMATOPOIETICHE PER LA CURA DELLA LEUCODISTROFIA A CELLULE GLOBOIDI

Ungari Silvia (1), Montepeloso Annita (2), Morena Francesco (3), Cocchiarella Fabienne (4), Gentner Bernhard (1), Recchia Alessandra (4), Martino Sabata (3), Naldini Luigi (1), Biffi Alessandra (1)

(1) San Raffaele Telethon Institute for Gene Therapy, San Raffaele Scientific Institute - Contact: Alessandra Biffi, San Raffaele Telethon Institute for Gene Therapy, San Raffaele Scientific Institute, Via Olgettina 58, 20132, Milano, Italy; a.biffi@hsr.it
(2) Vita-Salute San Raffaele University, Milano

(3) Department of Experimental Medicine and Biochemical Science, Perugia University, Perugia
(4) Center for Regenerative Medicine "Stefano Ferrari," Department of Life Sciences, University of Modena and Reggio Emilia, Modena

ABSTRACT N. 144

OSR-TIGET - Neurological Diseases		
Principal Investigator	**BIFFI ALESSANDRA**	
Telethon grant N.	**TGT11B01**	
Total budget €	**596.600**	
Centres: **1**	*Duration (yrs):* **5**	*Starting year:* **2011**

HSC GENE THERAPY FOR LSDS: UNDERSTANDING THE MODALITIES OF CELL REPLACEMENT IN THE LSD BRAIN FOR IMPROVING THERAPEUTIC EFFICACY

TERAPIA GENICA CON CELLULE STAMINALI EMATOPOIETICHE PER LE MALATTIE LISOSOMIALI: LA COMPRENSIONE DEI MECCANISMI DI TURNOVER CELLULARE NEL CERVELLO AFFETTO PER OTTIMIZZARE L'EFFICACIA TERAPEUTICA

Capotondo Alessia (1), Cecere Francesca (1), Peviani Marco (1), Milazzo Rita (1), Zonari Erika (1), Colombo Claudio (2), Gentner Bernhard (1), Moscatelli Davide (2), Politi Letterio Salvatore (3), Biffi Alessandra (1)

(1) San Raffaele Telethon Institute for Gene Therapy, San Raffaele Scientific Institute, Milano - Alessandra Biffi, San Raffaele Telethon Institute for Gene Therapy, San Raffaele Scientific Institute, Via Olgettina 58, 20132, Milano; tel: 0226434678; fax: 0226434668; e-mail: a.biffi@hsr.it
(2) Department of Chemistry, Materials and Chemical Engineering "G. Natta" , Politecnico of Milano, Milano
(3) Neuroradiology Department, Ospedale San Raffaele, Milano

ABSTRACT N. 145

OSR TIGET - Neurological Diseases		
Principal Investigator	**GRITTI ANGELA**	
Telethon grant N.	**TGT11B02**	
Total budget €	**1.094.900**	
Centres: **1**	*Duration (yrs):* **5**	*Starting year:* **2011**

CNS-DIRECTED GENE/CELL THERAPY OF LSDS

TERAPIA CELLULARE/GENICA DELLE LECODISTROFIE DIRETTA AL SISTEMA NERVOSO CENTRALE

Ricca Alessandra (1), Rufo Nicole (1), Meneghini Vasco (1), De Cicco Silvia (1), Frati Giacomo (1), Tiradani Luigi (1), Bravo Gabriele (1), Luciani Marco (1), Martino Sabata (2), Morena Francesco (2), Ungari Silvia (1), Biffi Alessandra (1), Gritti Angela (1)

(1) San Raffaele Scientific Institute, San Raffaele Telethon Institute for Gene Therapy (TIGET), Via Olgettina 58, Milano; tel +39 0226434623; fax +39 0226434668; gritti.angela@hsr.it
(2) University of Perugia, Italy

ABSTRACT N. 146

OSR TIGET - Neurological Diseases		
Principal Investigator	**BIFFI ALESSANDRA**	
Telethon grant N.	**TGTGSK02**	
Total budget €	**4.605.276**	
Centres: **1**	*Duration (yrs):* **4**	*Starting year:* **2011**

PHASE I/II CLINICAL TRIAL OF HEMATOPOIETIC STEM CELL GENE THERAPY FOR THE TREATMENT OF METACHROMATIC LEUKODYSTROPHY

PROTOCOLLO CLINICO DI FASE I/II DI TERAPIA GENICA CON CELLULE STAMINALI EMATOPOIETICHE PER IL TRATTAMENTO DELLA LEUCODISTROFIA METACROMATICA

Lorioli Laura (1,2,3,4)*, Fumagalli Francesca (2,5)*, Acquati Serena (1), Redaelli Daniela (1), Canale Sabrina (2), Martino Sabata (6), Baldoli Cristina (7), Morena Francesco (6), Calabria Andrea (1), Benedicenti Fabrizio (1), Antonioli Gigliola (2,3), Cicalese Maria Pia (2,3), Ciotti Francesca (2), Vallanti Giuliana (8), Natali Sora Maria Grazia (5), Fiori Rossana (9), Silvani Paolo (9), del Carro Ubaldo (5), Bordignon Claudio (8), di Serio Clelia (4), Rovelli Attilio (10), Ciceri Fabio (3), Roncarolo Maria Grazia (1,2,4), Montini Eugenio (1), Aiuti Alessandro (1,2,3,4), Sessa Maria (1), Naldini Luigi (1,4), Biffi Alessandra (1,2,3)

(1) San Raffaele Telethon Institute for Gene Therapy, San Raffaele Scientific Institute, Via Olgettina 58, 20132, Milano; tel: 0226434678; e-mail: a.biffi@hsr.it
(2) Pediatric Immunohematology Unit, San Raffaele Scientific Institute, Milano
(3) Stem Cell Program, San Raffaele Scientific Institute, Milano
(4) Vita Salute San Raffaele University, Milan
(5) Neurology Department, San Raffaele Scientific Institute, Milano
(6) Department of Experimental Medicine and Biochemical Science, Perugia University, Perugia
(7) Neuroradiology Department, San Raffaele Scientific Institute, Milano
(8) Molmed S.p.A., Milano
(9) Anesthesiology Department, San Raffaele Scientific Institute, Milano
(10) Bone Marrow Transplant Unit, MBBM Foundation, Pediatric Department, Milano-Bicocca University at San Gerardo Hospital, Monza
*equal contribution

ABSTRACT N. 147

OSR-TIGET - Neurological Diseases		
Principal Investigator	**GRITTI ANGELA**	
Telethon grant N.	**TGTGSK08**	
Total budget €	**119.864**	
Centres: **2**	*Duration (yrs):* **3**	*Starting year:* **2013**
Partners	LUIGI NALDINI	

INTRACEREBRAL ADMINISTRATION OF LENTIVIRAL VECTORS IN JUVENILE NON-HUMAN PRIMATES: A BIODISTRIBUTION STUDY

SOMMINISTRAZIONE INTRACEREBRALE DI VETTORI LENTIVIRALI IN GIOVANI PRIMATI NON-UMANI: STUDIO DI BIODISTRIBUZIONE

Meneghini Vasco (1), Lattanzi Annalisa (1), Tiradani Luigi (1), Bravo Gabriele (1), Morena Francesco (2), Martino Sabata (2), Bringas John (3), Bankiewicz Krystof (3), Gritti Angela (1)

(1) San Raffaele Scientific Institute, San Raffaele Telethon Institute for Gene Therapy (TIGET), Via Olgettina 58, Milano; tel +39 0226434623; fax +39 0226434668; gritti.angela@hsr.it
(2) University of Perugia, Italy
(3) University of California - Department of Neurological Surgery - San Francisco (CA)

ABSTRACT N. 148

OSR TIGET - Neurological Diseases		
Principal Investigator	**BIFFI ALESSANDRA**	
Telethon grant N.	**TGTGSK05**	
Total budget €	**3.456.744**	
Centres: **1**	*Duration (yrs):* **5**	*Starting year:* **2011**

HSC GENE THERAPY FOR THE TREATMENT OF MUCOPOLYSACCHARIDOSIS TYPE I: TOWARDS CLINICAL TESTING

TERAPIA GENICA CON CELLULE STAMINALI EMATOPOIETICHE DELLA MUCOPOLISACCARIDOSI DI TIPO I: VERSO LA SPERIMENTAZIONE CLINICA

Delai Stefania (1)*, Visigalli Ilaria (1)*, Ferro Francesca (1), Cecere Francesca (1), Sanvito Francesca (2), Vezzoli Michela (1), Cristofori Patrizia (1,3), Naldini Luigi (1), Biffi Alessandra (1)

(1) San Raffaele Telethon Institute for Gene Therapy, San Raffaele Scientific Institute, Via Olgettina 58, 20132, Milano; tel: 0226434678; e-mail: a.biffi@hsr.it
(2) Department of Pathology, San Raffaele Scientific Institute, Milano, Italy
(3) GlaxoSmithKline, R&D, Ware, SG12 0DP, UK
*equal contribution

ABSTRACT N. 149

Telethon Research Projects - Neurological Diseases		
Principal Investigator	**FIORENZA MARIA TERESA**	
Telethon grant N.	**GGP13183**	
Total budget €	**111.600**	
Centres: **1**	*Duration (yrs):* **2**	*Starting year:* **2013**

ENLIGHTENING MOLECULAR MECHANISMS OF ABNORMAL CEREBELLUM DEVELOPMENT IN MOUSE MODELS OF HUMAN NIEMANN-PICK C 1 DISEASE: THE EFFICACY OF HYDROXYPROPYL-BETACYCLODEXTRIN IN CORRECTING THE PHENOTYPE

STUDIO DEI MECCANISMI MOLECOLARI ALLA BASE DELLE ANOMALIE DELLO SVILUPPO DEL CERVELLETTO NELLA MALATTIA DI NIEMANN PICK C1: EFFICACIA DELLA CICLODESTRINA NEL CONTRASTARE I SINTOMI DELLA MALATTIA

Canterini Sonia (1), Nusca Stefania (1), Palladino Giampiero (1), Dragotto Jessica (1), Caporali Paola (1), Georgia Abate (1), Bruno Francesco (1), Erickson Robert (2), Fiorenza Maria Teresa (1)

(1) Dipartimento di Psicologia, Sezione Neuroscienze e Centro di ricerca in Neurobiologia “D. Bovet”, Sapienza Università di Roma - Via dei Sardi 70, 00185 Roma. Tel.: +39 0649917869; Fax: +39 0649917711; email: mariateresa.fiorenza@uniroma1.it
(2) Department of Pediatrics, University of Arizona, Tucson, AZ, USA

ABSTRACT N. 150

TIGEM - Neurological Diseases		
Principal Investigator	**BALLABIO ANDREA**	
Telethon grant N.	**TGM11CB6**	
Total budget €	**560.000**	
Centres: **1**	*Duration (yrs):* **5**	*Starting year:* **2011**

MODULATION OF CELLULAR CLEARANCE IN LYSOSOMAL STORAGE DISORDERS

IL LISOSOMA: UN CENTRO DI TRASMISSIONE CHE CONTROLLA CLEARANCE CELLULARE E METABOLISMO ENERGETICO

Ballabio Andrea (1, 2), Bouché Valentina (1, 2), Carmine Spampanato (1)

(1) Telethon Institute of Genetics and Medicine (TIGEM), Via Campi Flegrei 34, 80078 Pozzuoli (NA) - Tel. 081 6132207 - fax 081 19230650 - email: ballabio@tigem.it
(2) Department of Molecular and Human Genetics, Baylor College of Medicine, Houston, TX, USA

ABSTRACT N. 151

TIGEM - Neurological Diseases		
Principal Investigator	**PARENTI GIANCARLO**	
Telethon grant N.	**TGM11MT4**	
Total budget €	**350.000**	
Centres: **1**	*Duration (yrs):* **5**	*Starting year:* **2011**

SMALL MOLECULE-BASED THERAPIES WITH FOR LYSOSOMAL STORAGE DISEASES

TERAPIE CON PICCOLE MOLECOLE PER LE MALATTIE LISOSOMIALI

Parenti Giancarlo (1,2), Porto Caterina (1,2), Gatto Francesca (1), Tarallo Antonietta (1), Rossi Barbara (1), Coletta Marcella (1), Carrella Alessandra (1), Fecarotta Simona (2), Andria Generoso (2)

(1) Telethon Institute of Genetics and Medicine - Telethon Institute of Genetics and Medicine (TIGEM), via Campi Flegrei 34, Pozzuoli, tel 0816132227, email parenti@tigem.it
(2) Department of Translational Medical Sciences, Federico II University

ABSTRACT N. 152

TIGEM - Neurological Diseases		
Principal Investigator	**FRALDI ALESSANDRO**	
Telethon grant N.	**TGM11MT5**	
Total budget €	**190.000**	
Centres: **2**	*Duration (yrs):* **5**	*Starting year:* **2011**
Partners	ANDREA BALLABIO	

MODIFYING LYSOSOMAL ENZYMES TO IMPROVE SECRETION AND BRAIN DELIVERY [SURE]

MODIFICA DI ENZIMI LISOSOMIALI ALLO SCOPO DI MIGLIORARANE LA SECREZIONE ED IL TRASFERIMENTO AL SISTEMA NERVOSO CENTRALE

Sorrentino Nicolina Cristina, Cacace Vincenzo, Maffia Veronica, Strollo Sandra, Sambri Irene, Fraldi Alessandro

Telethon Institute of Genetics and Medicine - Telethon Institute of Genetics and Medicine (TIGEM), via Campi Flegrei 34, 80078 Pozzuoli, Naples, Italy, Tel. +39-081-19230632, Fax:+39-081-5609877, email: fraldi@tigem.it

ABSTRACT N. 153

Telethon Research Projects – Other Genetic Diseases		
Principal Investigator	**INVERNIZZI ROSANGELA**	
Telethon grant N.	**GGP13036**	
Total budget €	**66.900**	
Centres: **2**	*Duration (yrs):* **2**	*Starting year:* **2013**
Partners	PAOLO COLOMBO	

THALIDOMIDE FOR THE TREATMENT OF SEVERE RECURRENT EPISTAXIS IN HEREDITARY HEMORRHAGIC TELANGECTASIA: CLINICAL TRIAL ON THE EFFICACY OF ORAL ADMINISTRATION AND “IN VITRO” STUDY OF A NEW DRUG FORMULATION FOR A TOPICAL EFFECT

TRATTAMENTO CON TALIDOMIDE DEL SANGUINAMENTO NASALE SEVERO RICORRENTE NELLA TELEANGECTASIA EMORRAGICA EREDITARIA: STUDIO CLINICO SULL’EFFICACIA DELLA SOMMINISTRAZIONE ORALE E STUDIO “IN VITRO” DI UNA NUOVA FORMULAZIONE DEL FARMACO PER USO LOCALE

Quaglia Federica (1), Rossi Alessandra (2), Pagella Fabio (1), Colombo Gaia (3), Chiapponi Veronica (2), Chu Francesco (1), Matti Elina (1), Spinozzi Giuseppe (1), Ornati Federica (4), Canzonieri Cecilia (4), Olivieri Carla (4), Bastia Raffaella (1), Danesino Cesare (4),

Benazzo Marco (1), Balduini Carlo Luigi (1), Colombo Paolo (2), Invernizzi Rosangela (1)

(1) Fondazione IRCCS Policlinico San Matteo, Università di Pavia - Viale Golgi, 19 - 27100 Pavia, tel. 0382 502160, fax 0382 526223, e-mail: r.invernizzi@smatteo.pv.it
(2) Dipartimento di Farmacia, Università di Parma, Parma
(3) Dipartimento di Scienze della Vita e Biotecnologie, Università di Ferrara, Ferrara
(4) Dipartimento di Medicina Molecolare, Università di Pavia, Pavia

ABSTRACT N. 154

Telethon Research Projects - Neurological Diseases		
Principal Investigator	**DEJANA ELISABETTA**	
Telethon grant N.	**GGP14149**	
Total budget €	**349.800**	
Centres: **1**	*Duration (yrs):* **3**	*Starting year:* **2014**

NOVEL THERAPEUTIC INTERVENTIONS FOR CEREBRAL CAVERNOUS MALFORMATIONS

NUOVE TERAPIE FARMACOLOGICHE PER LA CURA DELLE MALFORMAZIONI CAVERNOSE CEREBRALI

Dejana Elisabetta, Bravi Luca

IFOM, the FIRC Institute of Molecular Oncology - via Adamello 16, 20139 Milano - Tel: 02574303372 - Mail elisabetta.dejana@ifom.eu

ABSTRACT N. 155

Telethon Research Projects - Neurological Diseases		
Principal Investigator	**ZARA FEDERICO**	
Telethon grant N.	**GEP14118**	
Total budget €	**50.000**	
Centres: **1**	*Duration (yrs):* **1**	*Starting year:* **2015**

IDENTIFICATION OF GENES FOR AICARDI SYNDROME BY EXOME SEQUENCING

IDENTIFICAZIONE DEL GENE PER LA SINDROME DI AICARDI ATTRAVERSO IL SEQUENZIAMENTO DELL'ESOMA

Zara Federico

Dipartimento di Neuroscienze, riabilitazione, oftalmologia, genetica e scienze materno-infantili - Università di Genova - Istituto Gianna Gaslini; Tel. (+39)010-56363816; Email: federicozara@ospedale-gaslini.ge.it

ABSTRACT N. 156

Telethon Research Projects - Neurological Diseases		
Principal Investigator	**DI CUNTO FERDINANDO**	
Telethon grant N.	**GGP12095**	
Total budget €	**270.000**	
Centres: **1**	*Duration (yrs):* **3**	*Starting year:* **2012**

IDENTIFICATION OF THERAPEUTIC TARGETS IN PRIMARY MICROCEPHALY THROUGH THE ANALYSIS OF THE CIT-K/ASPM PATHWAY

IDENTIFICAZIONE DI TARGET TERAPEUTICI NELLA MICROCEFALIA PRIMARIA ATTRAVERSO L'ANALISI DELLA VIA CIT-K/ASPM

Gai Marta (1), Bianchi Federico (1), Sgrò Francesco (1), Berto Gaia (1), Falcone Mattia (1), Chiotto Adelaide (1), Pallavicini Gianmarco (1), Turco Emilia (1), Chang YoonJeung (2), Priano Lorenzo (3), Mauro Alessandro (3), El Assawy Nadia (3), Huttner Wieland (2), Di Cunto Ferdinando (1)

(1) Università degli Studi di Torino, Centro di Biotecnologie Molecolari, Via Nizza 52, 10126, Torino, Phone: 0116706409, Fax: 0116706432, E-mail: ferdinando.dicunto@unito.it
(2) Max Planck Institute of Molecular Cell Biology and Genetics, Dresden, Germany
(3) Division of Neurology and Neurorehabilitation, San Giuseppe Hospital, IRCCS Istituto Auxologico Italiano, Piancavallo (VB), Italy

ABSTRACT N. 157

Telethon Research Projects - Neurological Diseases		
Principal Investigator	**GARAVELLI LIVIA**	
Telethon grant N.	**GEP14131**	
Total budget €	**50.000**	
Centres: **1**	*Duration (yrs):* **1**	*Starting year:* **2015**

GENOTYPE-PHENOTYPE CORRELATION FOR MAGNETIC RESONANCE IMAGING FEATURES OF MOWAT-WILSON SYNDROME WITH ZEB2 MUTATION/DELETION; FUTURE DISCOVERIES OF THE ROLE OF THE GENE ZEB2 IN THE DEVELOPMENT OF THE HUMAN BRAIN

CORRELAZIONE GENOTIPO-FENOTIPO E RISONANZA MAGNETICA CEREBRALE NELLA SINDROME DI MOWAT-WILSON DA MUTAZIONE DI ZEB2: CREAZIONE DI UN DATABASE INTERNAZIONALE E APERTURA A STUDI FUTURI SUL RUOLO DEL GENE ZEB2 NELLO SVILUPPO DELL'ENCEFALO UMANO

Garavelli Livia (1), Rosato Simonetta (1), Gelmini Chiara (1), Ivanovski Ivan (1), Maggi Massimo (2)

(1) Clinical Genetics Unit, IRCCS Arcispedale Santa Maria Nuova, Reggio Emilia (Italy) - Viale Risorgimento 80, Reggio Emilia - Tel. 0522/296244 -E-mail: garavelli.livia@asmn.re.it
(2) Neuroradiology Unit, IRCCS Arcispedale Santa Maria Nuova, Reggio Emilia (Italy)
(3) IT Technology Service, IRCCS Arcispedale Santa Maria Nuova, Reggio Emilia (Italy)
(4) Department of Research and Statistics Infrastructure, IRCCS Arcispedale Santa Maria Nuova, Reggio Emilia (Italy)
(5) Institute of Medical Genetics, Catholic University, Rome (Italy)

ABSTRACT N. 158

Telethon Research Projects - Neurological Diseases		
Principal Investigator	**ZOLLINO MARCELLA**	
Telethon grant N.	**GEP14089**	
Total budget €	**50.000**	
Centres: **1**	*Duration (yrs):* **1**	*Starting year:* **2015**

NGS TECHNIQUES TO EXPLORE UNUSUAL TCF4 MUTATIONS AND GENETIC HETEROGENEITY IN PATIENTS WITH PITT-HOPKINS SYNDROME PHENOTYPE

APPLICAZIONE DELLE TECNICHE DI SEQUENZIAMENTO DI NUOVA GENERAZIONE PER LA RICERCA DI MUTAZIONI ATIPICHE DEL GENE TCF4 E DI ETEROGENEITÀ GENETICA NELLA SINDROME DI PITT-HOPKINS

Zollino Marcella

Università Cattolica del Sacro Cuore, Policlinico A. Gemelli, Istituto di Genetica Medica, Largo F. Vito, 1, Roma

ABSTRACT N. 159

Telethon Research Projects - Other Genetic Diseases		
Principal Investigator	**GALLI ROSSELLA**	
Telethon grant N.	**GGP13022**	
Total budget €	**391.800**	
Centres: **2**	*Duration (yrs):* **3**	*Starting year:* **2013**
Partners	ROSSELLA GALLI, PIETRO LUIGI POLIANI	

EXPLOITING NEURAL STEM CELL-TARGETED MOUSE MODELS FOR IMPROVING THE UNDERSTANDING OF THE PATHOGENETIC MECHANISMS UNDERLYING TUBEROUS SCLEROSIS COMPLEX AND DEVELOPING NOVEL THERAPEUTIC APPROACHES

NUOVI MODELLI ANIMALI DI SCLEROSI TUBEROSA PER MIGLIORARE LA COMPRENSIONE DEI MECCANISMI PATOGENETICI E SVILUPPARE NUOVI APPROCCI TERAPEUTICI

Zordan Paola (1), Cominelli Manuela (2), Poliani Pietro Luigi (2), Galli Rossella (1)

(1) Neural Stem Cell Biology Unit, Division of Regenerative Medicine, Stem Cells and Gene Therapy, San Raffaele Scientific Institute - Via Olgettina 58 20132 Milan Italy Phone: +39 02 2643 4626 (office) Phone: +39 02 2643 4630 (lab) Fax: +39 02 2643 4621 Email: galli.rossella@hsr.it
(2) Pathology Unit, Department of Molecular and Translational Medicine, University of Brescia, Brescia, Italy

ABSTRACT N. 160

Telethon Research Projects - Neurological Diseases		
Principal Investigator	**PROIETTI DE SANTIS LUCA**	
Telethon grant N.	**GGP11176**	
Total budget €	**94.000**	
Centres: **1**	*Duration (yrs):* **2**	*Starting year:* **2014**

DISSECTING THE MOLECULAR BASIS OF NEURODEGENERATION IN COCKAYNE SYNDROME

STUDIO DELLE BASI MOLECOLARI DELLA NEURODEGENERAZIONE NELLA SINDROME DI COCKAYNE

Ciaffardini Flavia, Nicolai Serena, Caputo Manuela, Paccosi Elena, Costantino Michele, Proietti De Santis Luca

Dipartimento di Scienze Ecologiche e Biologiche, Università della Tuscia, 01100 Viterbo

ABSTRACT N. 161

Telethon Research Projects – Other Genetic Diseases		
Principal Investigator	**BRANZEI DANA**	
Telethon grant N.	**GGP12160**	
Total budget €	**223.500**	
Centres: **1**	*Duration (yrs):* **3**	*Starting year:* **2012**

CHARACTERIZATION OF THE DNA REPAIR DEFECTS AND CHROMOSOME STRUCTURAL ANOMALIES IN WARSAW BREAKAGE SYNDROME COHESINOPATHY CELLS

CARATTERIZZAZIONE DEI DIFETTI NELLA RIPARAZIONE DEL DNA E DELLE ANOMALIE CROMOSOMICHE NELLE CELLULE DI PAZIENTI AFFETTI DALLA SINDROME DELLA ROTTURA DI VARSAVIA

Takuya Abe, Fumasoni Marco, Branzei Dana

IFOM, the FIRC Institute of Molecular Oncology, Via Adamello 16 - 20139 Milan - Italy, tel. 02574303259, fax 02574303088, E-mail: dana.branzei@ifom.eu

ABSTRACT N. 162

Telethon Research Projects - Neurological Diseases		
Principal Investigator	**PLEVANI PAOLO**	
Telethon grant N.	**GGP11003**	
Total budget €	**295.300**	
Centres: **1**	*Duration (yrs):* **3**	*Starting year:* **2011**

THE ROLE OF RNASEH2 IN THE PATHOGENESIS OF AICARDI-GOUTIÉRES SYNDROME

IL RUOLO DELL'RNASIH2 NELLA PATOGENESI DELLA SINDROME DI AICARDI-GOUTIÈRES

Pizzi Sara (1), Sertic Sarah (1), Orcesi Simona (2), Cereda Cristina (3), Bianchi Marika (3), Jackson Andrew P. (4), Lazzaro Federico (1), Plevani Paolo (1), Muzi-Falconi Marco (1)

(1) Dipartimento di Bioscienze, Università degli Studi di Milano, 20133 Milano, Italy - Via Celoria 26, 02 50315032, 02 50315034, 02 50315044, email: paolo.plevani@unimi.it; marco.muzifalconi@unimi.it
(2) Child Neurology and Psychiatry Unit, C. Mondino National Neurological Institute, Pavia, Italy
(3) Laboratory of Experimental Neurobiology, C. Mondino National Neurological Institute, Pavia, Italy
(4) Medical Research Council Human Genetics Unit, MRC Institute of Genetics and Molecular Medicine, University of Edinburgh, Edinburgh EH4 2XU, UK

ABSTRACT N. 163

Telethon Research Projects - Neurological Diseases		
Principal Investigator	**BIANCHI VERA**	
Telethon grant N.	**GGP14005**	
Total budget €	**176.000**	
Centres: **1**	*Duration (yrs):* **2**	*Starting year:* **2014**

SAMHD1, A NEW REGULATOR OF DNA REPLICATION INVOLVED IN AICARDI-GOUTIÈRES SYNDROME

SAMHD1, UN NUOVO REGOLATORE DELLA SINTESI DEL DNA IMPLICATO NELLA SINDROME DI AICARDI-GOUTIÈRES

Ferraro Paola, Franzolin Elisa, Miazzi Cristina, Pontarin Giovanna, Rampazzo Chiara, Reichard Peter, Bianchi Vera

Department of Biology, University of Padova, Dipartimento di Biologia, Via Ugo Bassi 58B, 35131 Padova tel 049 8276282 fax: 049 8276280; vbianchi@bio.unipd.it

ABSTRACT N. 164

Telethon Research Projects - Neurological Diseases		
Principal Investigator	**PUSCH MICHAEL**	
Telethon grant N.	**GEP13101**	
Total budget €	**50.000**	
Centres: **1**	*Duration (yrs):* **1**	*Starting year:* **2014**

ROLE OF THE PUTATIVE ENDOSOMAL NA /H ANTIPORTER NHE6 IN ANGELMAN-LIKE SYNDROME AND OTHER FORMS OF X-LINKED MENTAL RETARDATION SYNDROMES

IL RUOLO DELLA PROTEINA NHE6 NELLA SINDROME DI ANGELMAN E IN ALTRE FORME DI RITARDO MENTALE DOVUTE A DIFETTI NEL CROMOSOMA X

Corso Lucia, Zifarelli Giovanni

Istituto di Biofisica, CNR, Genova- Via De Marini 6 - 16149 010 6475870 - zifarelli@ge.ibf.cnr.it

ABSTRACT N. 165

Telethon Research Projects - Neurological Diseases		
Principal Investigator	**CATTANEO LUIGI**	
Telethon grant N.	**GEP13004**	
Total budget €	**41.500**	
Centres: **1**	*Duration (yrs):* **1**	*Starting year:* **2014**

BRAIN MAPPING OF THE CORTICAL REPRESENTATION OF FACIAL MOVEMENTS IN PATIENTS WITH CONGENITAL FACIAL PALSY UN-

DERGOING SURGICAL PROCEDURES OF FACIAL ANIMATION

STUDIO DEI MECCANISMI DI CONTROLLO CEREBRALE DEI MOVIMENTI FACCIALI IN PAZIENTI CON SINDROME DI MOEBIUS DOPO L'INTERVENTO DI ANIMAZIONE FACCIALE

Cattaneo Luigi (1), Barchiesi Guido (1), Ferri Andrea (2), Bianchi Bernardo (2)

(1) Centro Interdipartimentale Mente/Cervello, Università di Trento - Via delle Regole 101, 38123 Trento. 0461802743. luigi.cattaneo@unitn.it
(2) Unità Operativa di Chirurgia Maxillo-Facciale, Azienda Ospedaliero-Universitaria di Parma

OTHER GENETIC DISEASES

ABSTRACT N. 166

Telethon Research Projects - Other Genetic Diseases		
Principal Investigator	**NICOLIS SILVIA KIRSTEN**	
Telethon grant N.	**GGP12152**	
Total budget €	**210.000**	
Centres: **1**	*Duration (yrs):* **3**	*Starting year:* **2012**

IDENTIFICATION AND FUNCTIONAL CHARACTERIZATION OF MOLECULAR TARGETS OF THE SOX2 TRANSCRIPTION FACTOR IN HUMAN INHERITED BRAIN DISEASE: AN APPROACH THROUGH SOX2 CONDITIONAL KNOCKOUT IN MOUSE

IDENTIFICAZIONE E CARATTERIZZAZIONE FUNZIONALE DEI BERSAGLI MOLECOLARI DEL FATTORE TRASCRIZIONALE SOX2 NELLA MALATTIA GENETICA DEL CERVELLO: UN APPROCCIO MEDIANTE ABLAZIONE CONDIZIONALE DI SOX2 NEL TOPO

Mercurio Sara (1), Favaro Rebecca (1), Bertolini Jessica (1), Barone Cristiana (1), Bottes Sara (1), Gesuita Lorenzo (1), Del Vecchio Simona (1), Alberti Chiara (1), Fucà Elisa (2), Leto Ketty (2), Zhang Yubo (3), Wei Chia-Lin (3), Martynoga Ben (4), Guillemot Francois (4), Vermunt Marit (5), Creyghton Menno (5), Bovolenta Paola (6), Nicolis Silvia (1)

(1) Dipartimento di Biotecnologie e Bioscienze, Università di Milano-Bicocca, piazza della Scienza 2, 20126 Milano, Italia - phone: +39 02 64483339 email: silvia.nicolis@unimib.it
(2) Dipartimento di Neuroscienze e Istituto di Neuroscienze della Fondazione Cavalieri-Ottolenghi, Università di Torino, Torino, Italia
(3) DOE Joint Genome Institute, Walnut Creek, California, USA
(4) National Institute for Medical Research, London, UK
(5) Hubrecht Institute, Utrecht, the Netherlands
(6) Centro de Biologia Molecular "Severo Ochoa", Universidad Autonoma de Madrid

ABSTRACT N. 167

TIGEM - Other Genetic Diseases		
Principal Investigator	**AURICCHIO ALBERTO**	
Telethon grant N.	**TGM11MT1**	
Total budget €	**450.000**	
Centres: **1**	*Duration (yrs):* **5**	*Starting year:* **2011**

OVERCOMING THE CHALLENGE OF LARGE GENE TRANSFER FOR THE THERAPY OF INHERITED PHOTORECEPTOR DISEASES

SUPERARE I LIMITI DEL TRASFERIMENTO ALLA RETINA DI GENI DI GRANDI DIMENSIONI PER LA CURA DI MALATTIE EREDITARIE A CARICO DEI FOTORECETTORI

Ivana Trapani (1), Pasqualina Colella (1), Agostina Puppo (1), Andrea Sommella (1), Carolina Iodice (1), Giulia Cesi (1), Sonia de Simone (1), Alberto Auricchio (1,2)

(1) Telethon Institute of Genetics and Medicine (TIGEM), Via Campi Flegrei 34, 80078 Pozzuoli (Naples), +39-081-6132228, +39.081.5790919, auricchio@tigem.it
(2) Medical Genetics, Dept. of Translational Medicine, "Federico II" University, Naples, Italy

ABSTRACT N. 168

TIGEM - Other Genetic Diseases		
Principal Investigator	**BANFI SANDRO**	
Telethon grant N.	**TGM11SB2**	
Total budget €	**500.000**	
Centres: **1**	*Duration (yrs):* **5**	*Starting year:* **2011**

MICRORNA-REGULATED GENE NETWORKS IN THE RETINA

IDENTIFICAZIONE DI NETWORK GENICI REGOLATI DA MICRORNA NELLA RETINA UMANA

Karali Marianthi (1), Carrella Sabrina (1), Pizzo Mariateresa (1), Barbato Sara (1), Bhat Rajeshwari S (1), Ambrosio Concetta (1), Persico Maria (1), Marrocco Elena (1), Ferrari Stefano (2), Ponzin Diego (2), Surace Enrico Maria (1), Black Graeme CM (3), di Bernardo Diego (1), Conte Ivan (1), Banfi Sandro (1,4)

(1) Telethon Institute of Genetics and Medicine (TIGEM) - Via Campi Flegrei, 34 - 80078 Pozzuoli (NA), tel. 0816132200, email banfi@tigem.it
(2) International Center for Ocular Physiopathology, The Veneto Eye Bank Foundation, Venice
(3) Institute of Human Development, Faculty of Medical and Human Sciences, University of Manchester, Manchester, UK
(4) Dipartimento di Biochimica, Biofisica e Patologia Generale, Seconda Università degli Studi di Napoli (SUN), Naples

ABSTRACT N. 169

Telethon Research Projects - Other Genetic Diseases		
Principal Investigator	**SANTORO MASSIMO**	
Telethon grant N.	**GGP10195**	
Total budget €	**338.800**	
Centres: **1**	*Duration (yrs):* **3**	*Starting year:* **2010**

METABOLIC AND REDOX STATE FUNCTIONS OF UBIAD1, A GENE PRODUCT ASSOCIATED TO SCHNYDER CRYSTALLINE CORNEAL DYSTROPHY

CARATTERIZZAZIONE MOLECOLARE E METABOLICA DI UBIAD1, UN NUOVO PRODOTTO GENICO COINVOLTO NELLA DISTROFIA DEL CRISTALLINO DI SCHNYDER

Panieri Emiliano, Santoro Massimo M.

Molecular Biotechnology Center, University of Torino, Torino

ABSTRACT N. 170

Telethon Research Projects - Other Genetic Diseases		
Principal Investigator	**SIMONELLI FRANCESCA**	
Telethon grant N.	**GGP10199**	
Total budget €	**221.800**	
Centres: **1**	*Duration (yrs):* **3**	*Starting year:* **2011**

A SAFETY AND EFFICACY STUDY IN SUBJECTS WITH LEBER CONGENITAL AMAUROSIS (LCA) USING ADENO-ASSOCIATED VIRAL

VECTOR TO DELIVER THE GENE FOR HUMAN RPE65 TO THE RETINAL PIGMENT EPITHELIUM (RPE) [AAV2-HRPE65V2-301]: TREATMENT AND FOLLOW UP OF 3 ITALIAN PATIENTS

STUDIO DI SICUREZZA ED EFFICACIA IN SOGGETTI CON AMAUROSI CONGENITA DI LEBER (ACL) TRAMITE VETTORE ADENO-ASSOCIATO PER TRASFERIRE IL GENE RPE65 UMANO NELL'EPITELIO PIGMENTATO DELLA RETINA (EPR): TRATTAMENTO E FOLLOW-UP DI 3 PAZIENTI ITALIANI

Testa Francesco (1), Rossi Settimio (1), Auricchio Alberto (2), Melillo Paolo (1), Simonelli Francesca (1)

(1) Clinica Oculistica, Dipartimento Multidisciplinare di Specialità Medico-Chirurgiche ed Odontoiatriche, Via Pansini,5 Ed. 15, Napoli, 081 7704501, 081 7704501, franctes@tin.it
(2) Telethon Institute of Genetics and Medicine (TIGEM), Pozzuoli (NA), Italy

ABSTRACT N. 171

TIGEM - Other Genetic Diseases		
Principal Investigator	**FRANCO BRUNELLA**	
Telethon grant N.	**TGM11CB3**	
Total budget €	**500.000**	
Centres: **1**	*Duration (yrs):* **5**	*Starting year:* **2011**

CILIA AND HUMAN DISEASES: INSIGHTS FROM THE OFD TYPE I SYNDROME

MALATTIE GENETICHE ASSOCIATE A DISFUNZIONE CILIARE: TUTTO QUELLO CHE POSSIAMO IMPARARE DALLA SINDROME OROFACIO-DIGITALE DI TIPO I

Iaconis Daniela (1), Morleo Manuela (1), Filomena Massa (1), Maria Monti (2), Tammaro Roberta (1), van Koppen Arianne (3), Venditti Rossella (1), Indrieri Alessia (1), Chiaravalli Marco (4), Cozzolino Flora (2), Pucci Piero (2), De Matteis Maria Antonietta (1), Boletta Alessandra (4), Belcastro Vincenzo (1), di Bernardo Diego (1), Giles Rachel (3), Pende Mario (5), Amato Roberto (1), Franco Brunella (1,6)

(1) Telethon Institute of Genetics and Medicine (TIGEM), Italy - Via Campi Flegrei 34, 80078, Pozzuoli (NA), Italy, franco@tigem.it; tel 0816132215, fax 0815609877
(2) Dipartimento di Scienze Chimiche and CEINGE Biotecnologie Avanzate, Università di Napoli, Federico II
(3) Department of Nephrology and Hypertension, University Medical Center Utrecht, the Netherlands
(4) Division of Genetics and Cell Biology, Dibit, San Raffaele Scientific Institute, Milan, Italy
(5) Institut National de la Santé et de la Recherche Médicale (INSERM) U1151, Institut Necker Enfants Malades, Université Paris Descartes, Sorbonne Paris Cité, Paris, France
(6) Department of Medical Translational Sciences, University of Naples "Federico II", Naples, Italy

ABSTRACT N. 172

Telethon Research Projects - Other Genetic Diseases		
Principal Investigator	**FANELLI FRANCESCA**	
Telethon grant N.	**GGP11210**	
Total budget €	**361.900**	
Centres: **2**	*Duration (yrs):* **3**	*Starting year:* **2011**
Partners	VALERIA MARIGO	

INTEGRATED IN SILICO, IN VITRO, AND IN VIVO STUDIES TOWARDS THE DESIGN OF MOLECULES WITH THERAPEUTIC POTENTIAL FOR RETINITIS PIGMENTOSA

STUDI INTEGRATI IN SILICO, IN VITRO ED IN VIVO VERSO LA PROGETTAZIONE DI POTENZIALI AGENTI TERAPEUTICI PER LA RETINITE PIGMENTOSA

Fanelli Francesca (1), Felline Angelo (2), Behnen Petra (2), Marigo Valeria (2)

(1) Dipartimento di Scienze della Vita, Università di Modena e Reggio Emilia, via Campi 183, 41125 Modena; Tel:059 2055114; Fax: 059 373543; e-mail: fanelli@unimo.it
(2) Dipartimento di Scienze della Vita, Università di Modena e Reggio Emilia

ABSTRACT N. 173

Telethon Research Projects - Other Genetic Diseases		
Principal Investigator	**LANZANI GUGLIELMO**	
Telethon grant N.	**GGP12033**	
Total budget €	**296.200**	
Centres: **3**	*Duration (yrs):* **2**	*Starting year:* **2012**
Partners	FABIO BENFENATI, SILVIA BISTI	

DEVELOPMENT AND APPLICATION OF OPTO-NEURAL PROSTHETIC DEVICES AS A THERAPEUTIC APPROACH FOR RETINITIS PIGMENTOSA

SVILUPPO DI INTERFACCE BIO-ORGANICHE FOTOVOLTAICHE E LORO APPLICAZIONE COME PROTESI RETINICHE PER LA CURA DELLA RETINITE PIGMENTOSA

Lanzani Guglielmo (1), Benfenati Fabio (2), Bisti Silvia (3), Ghezzi Diego (2), Antognazza Maria Rosa (1), Maccarone Rita (3), Mete Maurizio (4), Pertile Grazia (4)

(1) Department of Physics, Politecnico di Milano and Center for NanoScience and Technology@PoliMi, Istituto Italiano di Tecnologia, Milano, Italy - Piazza Leonardo da Vinci 32; 20133 Milano (tel. 02-23999877; fax 02-23996126; email guglielmo.lanzani@iit.it)
(2) Department of Neuroscience and Brain Technologies, Istituto Italiano di Tecnologia, Genova, Italy
(3) Dipartimento di Tecnologie Biomediche, Università dell'Aquila, L'Aquila, Italy
(4) UO Oculistica, Ospedale S. Cuore-Don Calabria, Negrar, Italy

ABSTRACT N. 174

Telethon Research Projects - Other Genetic Diseases		
Principal Investigator	**PERTILE GRAZIA**	
Telethon grant N.	**GGP14022**	
Total budget €	**322.300**	
Centres: **2**	*Duration (yrs):* **2**	*Starting year:* **2014**
Partners	FABIO BENFENATI	

DEVELOPMENT AND IMPLANT OF THE PHOTOVOLTAIC ARTIFICIAL RETINA IN THE PIG WITH PHOTORECEPTOR DEGENERATION: TOWARDS THE HUMAN PHASE-1 EXPERIMENTATION

SVILUPPO ED IMPIANTO DELLA RETINA ARTIFICIALE FOTOVOLTAICA NEL MAIALE CON DEGENERAZIONE DEI FOTORECETTORI: ULTIMO STADIO VERSO LA SPERIMENTAZIONE NELL'UOMO

Pertile Grazia (1), Benfenati Fabio (2), Lanzani Guglielmo (3), Bacci Maria Laura (4), Ghezzi Diego (2), Bisti Silvia (5), Antognazza Maria Rosa (3), Mete Maurizio (1)

(1) Department of Ophthalmology, Ospedale S. Cuore-Don Calabria, Negrar, via Don Sempreboni 5, 37024 Negrar (VR) (tel 045 6014629; fax 045 6013026; email grazia.pertile@sacrocuore.it)
(2) Department of Neuroscience and Brain Technologies, Istituto Italiano di Tecnologia, Genova, Italy

(3) Center for NanoScience and Technology@PoliMi, Istituto Italiano di Tecnologia, Milano, Italy
(4) Unità ASA, Department of Veterinary Medical Sciences, University of Bologna, Bologna, Italy
(5) Department of Biomedical Technologies, L'Aquila University, L'Aquila, Italy

ABSTRACT N. 175

Telethon Research Projects - Other Genetic Diseases		
Principal Investigator	**MARIGO VALERIA**	
Telethon grant N.	**GGP14180**	
Total budget €	**216.700**	
Centres: **1**	*Duration (yrs):* **3**	*Starting year:* **2014**

EXPLORING PEDF AS THERAPEUTIC AGENT FOR RETINITIS PIGMENTOSA

IL PEDF COME AGENTE TERAPEUTICO PER LA RETINITE PIGMENTOSA

Comitato Antonella (1), Subramanian Preeti (2), Kenealey Jason (2), Becerra Patricia S. (2), Marigo Valeria (1)

(1) Department of Life Sciences, University of Modena and Reggio Emilia
(2) National Institute of Health, National Eye Institute, USA

ABSTRACT N. 176

TIGEM - Other Genetic Diseases		
Principal Investigator	**SURACE ENRICO**	
Telethon grant N.	**TGM11MT2**	
Total budget €	**290.000**	
Centres: **1**	*Duration (yrs):* **5**	*Starting year:* **2011**

EFFICACY AND SAFETY OF TRANSCRIPTIONAL REPRESSORS AS BIOTHERAPEUTICS FOR THE TREATMENT OF AUTOSMAL DOMINANT RETINITIS PIGMENTOSA (ADRP)

EFFICACIA E SICUREZZA DI REPRESSORI TRASCRIZIONALI UTILIZZATI COME AGENTI TERAPEUTICI PER IL TRATTAMENTO DELLA RETINITE PIGMENTOSA AUTOSOMICA DOMINANTE (ADRP)

Botta Salvatore (1), Marrocco Elena (1), de Prisco Nicola (1), Curion Fabiola (1), Sofia Martina (1), Renda Mario (1), Bacci Maria Laura (2), Rossi Settimio (3), Simonelli Francesca (3), Surace Enrico Maria (1,4)

(1) Enrico Maria Surace - Telethon Institute of Genetics and Medicine (TIGEM), Via Campi Flegrei 34 - 800780 - Pozzuoli (NA), Italy
(2) Dipartimento di Scienze Mediche Veterinarie Università di Bologna, Ozzano dell'Emilia (BO)
(3) Seconda Università di Napoli (SUN)
(4) Dept. of Translational Medicine, "Federico II" University, Napoli

ABSTRACT N. 177

Telethon Research Projects - Other Genetic Diseases		
Principal Investigator	**DUGA STEFANO**	
Telethon grant N.	**GGP11177**	
Total budget €	**345.000**	
Centres: **3**	*Duration (yrs):* **3**	*Starting year:* **2011**
Partners	UMBERTO AMBROSETTI, LUCA DEL GIACCO	

MICS-BASED APPROACHES FOR THE IDENTIFICATION OF NOVEL INHERITED NON-SYNDROMIC SENSORINEURAL HEARING LOSS-RELATED GENES

APPROCCI "OMICI" PER L'IDENTIFICAZIONE DI NUOVI GENI RESPONSABILI DI SORDITÀ NON SINDROMICA NEUROSENSORIALE EREDITARIA

Soldà Giulia (1,2), Robusto Michela (1,2), Asselta Rosanna (1,2), Ghilardi Anna (3), Castorina Pierangela (4), Chiereghin Chiara (1,2), Primignani Paola (5), Ambrosetti Umberto (4), Del Giacco Luca (3), Duga Stefano (1,2)

(1) Humanitas University, Via Manzoni 113, 20089 Rozzano (Milan) - Italy; email: stefano.duga@hunimed.eu; tel.: 02.82245204
(2) Dipartimento di Biotecnologie Mediche e Medicina Traslazionale, Università degli Studi di Milano, Milano
(3) Dipartimento di Bioscienze, Università degli Studi di Milano, Milano
(4) Laboratorio di Genetica Medica e UO Audiologia, Fondazione IRCCS, Ospedale Maggiore Policlinico, Mangiagalli e Regina Elena, Milan
(5) S.O. Genetica Medica, Ospedale Niguarda Cà Granda, Milan

ABSTRACT N. 178

Telethon Research Projects - Other Genetic Diseases		
Principal Investigator	**MAMMANO FABIO**	
Telethon grant N.	**GGP13114**	
Total budget €	**399.300**	
Centres: **1**	*Duration (yrs):* **3**	*Starting year:* **2013**

INNER EAR CONNEXINS: ROLE IN HEARING ACQUISITION AND DFNB1 PATHOPHYSIOLOGY

PATOGENESI DELLA SORDITÀ EREDITARIA: RUOLO DELLE CONNESSINE NELL'ORECCHIO INTERNO PER LO SVILUPPO DEL SENSO DELL'UDITO

Zorzi Veronica (1,2), Crispino Giulia (1,2), Ceriani Federico (1,2), Carrer Andrea (1,2), Scimemi Pietro (3), Mammano Fabio (1,2,4)

(1) Istituto Veneto di Medicina Molecolare, Padova, Via G. Orus 2, 35129 Padova; Tel. 049 7923 231; Fax 049 7923266; email: fabio.mammano@unipd.it
(2) Università di Padova, Dipartimento di Fisica a Astronomia, Padova
(3) Università di Padova, Dipartimento Di Neuroscienze Scienze Npsrr, Padova
(4) CNR, Istituto di Biologia Cellulare e Neurobiologia, Monterotondo

ABSTRACT N. 179

Telethon Research Projects - Other Genetic Diseases		
Principal Investigator	**SERI MARCO**	
Telethon grant N.	**GGP13082**	
Total budget €	**299.000**	
Centres: **3**	*Duration (yrs):* **2**	*Starting year:* **2013**
Partners	PATRIZIA NORIS, ANNA SAVOIA	

COMBINING NEXT GENERATION SEQUENCING WITH CLINICAL STUDIES TO UNRAVEL NOVEL INHERITED THROMBOCYTOPENIAS AFFECTING HALF OF THE PATIENTS

IDENTIFICAZIONE DI NUOVI GENI COINVOLTI NELL'INSORGENZA DI PIASTRINOPENIE EREDITARIE TRAMITE TECNOLOGIE DI SEQUENZIAMENTO DI ULTIMA GENERAZIONE

Seri Marco (1), Bottega Roberta (2), Marconi Caterina (1), Faleschini Michela (3), Baj Gabriele (4), Cagioni Claudia (5), Pecci Alessandro (5), Pippucci Tommaso (1), Palombo Flavia (1), Balduini Carlo L. (5), Savoia Anna (2,3), Noris Patrizia (5)

(1) Department of Medical and Surgical Sciences, Medical Genetics Unit, S. Orsola-Malpighi Hospital, University of Bologna, Bologna, Italy, Via Massarenti 9 – Pav. 11, 40138, Bologna Phone 0039 051-2088421, Fax 0039 051-2088416, e-mail marco.seri@unibo.it

(2) Institute for Maternal and Child Health – IRCCS Burlo Garofolo, Trieste, Italy
(3) Department of Medical Sciences, University of Trieste, Trieste, Italy
(4) Department of Life Sciences, University of Trieste, Trieste, Italy
(5) Department of Internal Medicine, University of Pavia-IRCCS Policlinico San Matteo Foundation, Pavia, Italy

ABSTRACT N. 180

Telethon Research Projects - Other Genetic Diseases		
Principal Investigator	**DIANZANI IRMA**	
Telethon grant N.	**GGP13177**	
Total budget €	**473.100**	
Centres: **2**	*Duration (yrs):* **3**	*Starting year:* **2013**
Partners	FABRIZIO LORENI	

RESCUE OF DIAMOND-BLACKFAN ANEMIA HAPLOINSUFFICIENCY BY KNOCK-UP OF THE DEFICIENT PROTEIN

CORREZIONE DELLA CARENZA DI GLOBULI ROSSI NELL'ANEMIA DI DIAMOND-BLACKFAN TRAMITE UNA NUOVA TECNICA MOLECOLARE A BASE DI RNA

Aspesi Anna (1), Pavesi Elisa (1), Parrella Sara (1), Macrì Serena (1), Merlin Simone (1), Salmi Silvia (1), Chiesa Andrea (1), Cotella Diego (1), D'Amico Silvia (2), Sagar Vinay (2), Aria Valentina (2), Juli Giada (2), Garelli Emanuela (3), Ramenghi Ugo (3), Santoro Claudio (1), Follenzi Antonia (1), Loreni Fabrizio (2), Dianzani Irma (1)

(1) Dipartimento di Scienze della Salute, Univ. Piemonte Orientale, Via Solaroli 17, 28100 Novara, tel: 0321-660544, fax: 0321-6620421, irma.dianzani@med.unipmn.it
(2) Dipartimento di Biologia, Univ. Tor Vergata, Roma
(3) Dipartimento di Scienze della Sanità Pubblica e Pediatriche, Univ. Torino

ABSTRACT N. 181

Telethon Research Projects - Other Genetic Diseases		
Principal Investigator	**LA VOLPE ADRIANA**	
Telethon grant N.	**GGP11076**	
Total budget €	**457.800**	
Centres: **3**	*Duration (yrs):* **3**	*Starting year:* **2012**
Partners	MARIA CIARAMELLA, ANNA SAVOIA	

NEW PHARMACOLOGICAL TARGETS IN FANCONI ANEMIA

NUOVI BERSAGLI FARMACOLOGICI NELL'ANEMIA DI FANCONI

Adamo Adele (2), Santonicola Pamela (2), Germoglio Marcello (2), La Volpe Adriana (1), Vettone Antonella (2), Valenti Anna (2), Perugino Giuseppe (2), Ciaramella Maria (2), Faleschini Michela (3), Bottega Roberta (3), De Rocco Daniela (3), Savoia Anna (3)

(1) Institute of Biosciences and BioResources - Via Pietro Castellino 111, 80131, Naples, Italy tel. +390816132366 fax +390816132646 adriana.lavolpe@ibbr.cnr.it
(2) Institute of Biosciences and BioResources, Naples, Italy
(3) IRCCS Burlo Garofolo, Università degli Studi di Trieste

ABSTRACT N. 182

Telethon Research Projects - Other Genetic Diseases		
Principal Investigator	**FOLLENZI ANTONIA**	
Telethon grant N.	**GGP09280**	
Total budget €	**125.400**	
Centres: **1**	*Duration (yrs):* **3**	*Starting year:* **2009**

THERAPEUTIC ROLES OF HEALTHY DONOR HUMAN LIVER SINUSOIDAL ENDOTHELIAL CELLS (LSEC), BONE MARROW OR CORD BLOOD-DERIVED CELLS IN HEMOPHILIA A

POTENZIALI EFFETTI TERAPEUTICI DELLE CELLULE UMANE ENDOTELIALI DEI SINUSOIDI EPATICI, DEL MIDOLLO OSSEO E DEL SANGUE DI CORDONE NELL'EMOFILIA A

Zanolini Diego (1), Merlin Simone (1), Feola Maria (1), Ranaldo Gabriella (1), Zaffaroni Mauro (2), Ferrero Alessandro (3), Valente Guido (4), Prat Maria (1), Follenzi Antonia (1)

(1) Università del Piemonte Orientale, Dip di Scienze della Salute, Novara, Italia - Via Solaroli 17, 28100 Novara. Tel 0321-660674, antonia.follenzi@med.unipmn.it
(2) Azienda Ospedaliera Universitaria Maggiore della Carità, SCDU Pediatria, Novara, Italia
(3) Azienda Ospedaliera Mauriziano, SC Chirurgia Generale ed Oncologica, Torino, Italia
(4) Università del Piemonte Orientale, dip di Medicina Translazionale, Novara, Italia

ABSTRACT N. 183

Telethon Research Projects - Other Genetic Diseases		
Principal Investigator	**FALLARINO FRANCESCA**	
Telethon grant N.	**GGP14042**	
Total budget €	**132.600**	
Centres: **1**	*Duration (yrs):* **2**	*Starting year:* **2014**

INSTALLING FVIII-SPECIFIC TOLERANCE IN HEMOPHILIA A VIA TRYPTOPHAN CATABOLITES AND ARYL HYDROCARBON RECEPTOR (AHR) ACTIVATION

INDUZIONE DELLA TOLLERANZA VERSO LA PROTEINA RFVIII NELL'EMOFILIA A ATTRAVERSO METABOLITI DEL TRIPTOFANO E L'ATTIVAZIONE DEL RECETTORE AHR

Matino Davide (2), Gargaro Marco (2), Iorio Alfonso (3), Puccetti Paolo (2), Fallarino Francesca (1)

(1) Francesca Fallarino, Dept. of Experimental Medicine - Piazzale Gambuli, Polo Sant'Andrea delle Fratte, Perugia, Tel 075-5858242, email: fllfnc@tin.it.
(2) Dept. of Experimental Medicine,Piazzale Gambuli, Polo Sant'Andrea delle Fratte, Perugia
(3) Clinical Epidemiology and Biostatistics McMaster University Hamilton, ON Canada 905 525 9140

ABSTRACT N. 184

Telethon Research Projects - Other Genetic Diseases		
Principal Investigator	**PINOTTI MIRKO**	
Telethon grant N.	**GGP14190**	
Total budget €	**270.300**	
Centres: **2**	*Duration (yrs):* **2**	*Starting year:* **2014**
Partners	FRANCO PAGANI	

DEVELOPMENT OF A RNA-BASED THERAPEUTIC APPROACH FOR HEMOPHILIA B CAUSED BY EXON-SKIPPING MUTATIONS

SVILUPPO DI UN NUOVO APPROCCIO TERAPEUTICO PER L'EMOFILIA B CAUSATA DA MUTAZIONI CHE CAUSANO SALTO DELL'ESONE MEDIANTE MODULAZIONE DEL PROCESSAMENTO DI RNA MESSAGGERO

Balestra Dario (1), Dal Mas Andrea (2), Rogalska Malgorzata Ewa (2), Barbon Elena (1), Scalet Daniela (1), Donadon Irving (1), Ferrarese Mattia (1), Bussani Erica (2), Pianigiani Giulia (2), Ferraresi Paolo (1), Branchini Alessio (1), Bovolenta Matteo (1), Baroni Mar-

cello (1), Mattioli Chiara (2), Pagani Franco (2), Pinotti Mirko (1)

(1) Dipartimento di Scienze della Vita e Biotecnologie, Università di Ferrara, Via Fossato di Mortara 74, 44121 Ferrara, Italy. Tel. 0532-424424; Fax. 0532 424484; e-mail: pnm@unife.it
(2) International Centre for Genetic Engineering and Biotechnology, Trieste, Italy

ABSTRACT N. 185

OSR-TIGET - Other Genetic Diseases		
Principal Investigator	**NALDINI LUIGI**	
Telethon grant N.	**TGT11D03**	
Total budget €	**1.100.300**	
Centres: **1**	*Duration (yrs):* **5**	*Starting year:* **2011**

LIVER-DIRECTED LENTIVIRAL GENE THERAPY PROVIDES STABLE BENEFIT IN HEMOPHILIA B DOGS WITHOUT EVIDENCE OF GENOTOXICITY IN SENSITIZED MOUSE MODELS

LA TERAPIA GENICA DIRETTA AL FEGATO CON VETTORI LENTIVIRALI PRODUCE BENEFICI CLINICI IN CANI AFFETTI DA EMOFILIA B SENZA EVIDENZE DI GENOTOSSICITÀ IN MODELLI MURINI SENSIBILIZZATI

Cantore Alessio (1,2), Ranzani Marco (1,2), Bartholomae Cynthia (3), Volpin Monica (1,2), Della Valle Patrizia (4), Sanvito Francesca (5), Sergi Sergi Lucia (1), Gallina Pierangela (1), Benedicenti Fabrizio (1), Bellinger Dwight (6), D'Angelo Armando (4), VandenDriessche Thierry (7), Schmidt Manfred (3), Nichols Timothy (6), Montini Eugenio (1), Naldini Luigi (1,2)

(1) San Raffaele Telethon Institute for Gene Therapy, Via Olgettina 58, Milan, Italy, Phone: +39 02 2643 4681. Fax: +39 02 2643 4621. E-mail: naldini.luigi@hsr.it
(2) Vita Salute San Raffaele University, Milan, Italy
(3) German Cancer Research Center, Heidelberg, Germany
(4) Coagulation Service, San Raffaele Scientific Institute, Milan, Italy
(5) Pathology Unit, San Raffaele Scientific Institute, Milan, Italy
(6) University of North Carolina, Chapel Hill, USA
(7) Free University of Brussels, Brussels, Belgium

ABSTRACT N. 186

Telethon Research Projects - Other Genetic Diseases		
Principal Investigator	**BIFFO STEFANO**	
Telethon grant N.	**GGP10012**	
Total budget €	**263.400**	
Centres: **1**	*Duration (yrs):* **3**	*Starting year:* **2011**

MODULATION OF EUKARYOTIC INITIATION FACTOR 6 ACTIVITY AS A THERAPEUTIC TOOL IN RIBOSOME-BASED DISEASE

MODULAZIONE DELLA ATTIVITÀ DEL FATTORE DI INIZIO 6 (EIF6) E SUO UTILIZZO TERAPEUTICO NELLE MALATTIE A CAUSA RIBOSOMALE

Pesce Elisa (1), Miluzio Annarita (1), Peluso Ivana (2), Russo Arianna (1,4), Medina Diego (2), Rommens Johanna (3), Biffo Stefano (1,4), Calamita Piera (1)

(1) INGM, Istituto Nazionale Genetica Molecolare "Romeo ed Enrica Invernizzi", Milan, Italy - Via Francesco Sforza 35, 20122, Milano, Italy, Fax 39 02 00660216, Tel 39 02 00660304, biffo@ingm.org
(2) TIGEM, Napoli, Italy
(3) Sick Children Hospital, Toronto, Canada
(4) Dip. Bioscienze, University of Milan, Italy

ABSTRACT N. 187

Telethon Research Projects - Other Genetic Diseases		
Principal Investigator	**TOLOSANO EMANUELA**	
Telethon grant N.	**GGP12082**	
Total budget €	**180.800**	
Centres: **1**	*Duration (yrs):* **2**	*Starting year:* **2012**

THERAPEUTIC STRATEGIES TO AMELIORATE HEME-DRIVEN TISSUE OXIDATIVE INJURY IN SICKLE CELL ANEMIA

STRATEGIE TERAPEUTICHE VOLTE AD ALLEVIARE IL DANNO OSSIDATIVO INDOTTO DALL'EME NELL'ANEMIA FALCIFORME

Vinchi Francesca, Ingoglia Giada, Chiabrando Deborah, Fiorito Veronica, Mercurio Sonia, Petrillo Sara, Tolosano Emanuela

Dept. Molecular Biotechnology and Health Sciences, via Nizza 52, 10126 Torino, Italy

ABSTRACT N. 188

OSR-TIGET - Other Genetic Diseases		
Principal Investigator	**FERRARI GIULIANA**	
Telethon grant N.	**TGT11C02**	
Total budget €	**736.200**	
Centres: **1**	*Duration (yrs):* **5**	*Starting year:* **2011**

HUMAN HEMATOPOIETIC CD34 CELLS ISOLATED FROM DIFFERENT SOURCES: EXPLORING THEIR BIOLOGY FOR FUTURE CLINICAL PERSPECTIVES

CELLULE EMATOPOIETICHE UMANE CD34 ISOLATE DA DIVERSE FONTI: CARATTERIZZAZIONE BIOLOGICA E MOLECOLARE PER UNA FUTURA PROSPETTIVA CLINICA

Lidonnici Maria Rosa (1), Aprile Annamaria (1), Frittoli Marta (2), Mandelli Giacomo (1), Gentner Bernhard (1,2), Bellio Laura (3), Cassinerio Elena (4), Zanaboni Laura (4), Rossini Silvano (3), Cappellini Maria Domenica (4), Ciceri Fabio (2), Marktel Sarah (2), Ferrari Giuliana (1,5)

(1) San Raffaele-Telethon Institute for Gene Therapy (HSR-TIGET), Milan, Italy - Via Olgettina, 58, 20132 Milano; phone 02-26434705, fax 02-26434668; g.ferrari@hsr.it
(2) Hematology and Bone Marrow Transplantation Unit, San Raffaele Scientific Institute, Milan, Italy
(3) Immunohematology and Transfusion Medicine Unit, San Raffaele Scientific Institute, Milan, Italy
(4) Università di Milano, Ca Granda Foundation IRCCS, Milan, Italy
(5) Università Vita-Salute San Raffaele, Milan, Italy

ABSTRACT N. 189

Telethon Research Projects - Other Genetic Diseases		
Principal Investigator	**UDA MANUELA**	
Telethon grant N.	**GGP13246**	
Total budget €	**262.800**	
Centres: **3**	*Duration (yrs):* **2**	*Starting year:* **2013**
Partners	PAOLO MOI, ANDREA ANGIUS	

POST GWAS FUNCTIONAL CHARACTERIZATION OF BCL11A LOCUS TOWARD THE DEVELOPMENT OF A TREATMENT FOR ß-THALASSEMIA

CARATTERIZZAZIONE FUNZIONALE DEL GENE BCL11A, VOLTA ALLO SVILUPPO DI UNA NUOVA TERAPIA PER LA CURA DELLA BETA-TALASSEMIA

Caria Cristian Antonio (1), Anedda Francesca (1), Danjou Fabrice Fabrice (1), Loi Alessia (1), Asunis Isadora (1), Fancello Tatiana (1), Per-

seu Luciana (1), Manunza Laura (2), Usala Gianluca (1), Desogus Alessia (1), Vacca Claudia (1), Sanna Sonia (1), Marini Maria Giuseppina (1), Sidore Carlo (1), Pala Mauro (1), Berutti Riccardo (3), Ristaldi Maria Serafina (1), Busonero Fabio (1), Cusano Roberto (3), Satta, Stefania (2), Demartis, Francarosa (2), Maccioni Liliana (2), Goncalo Abecasis (4), Schlessinger David (5), Angius Andrea (1,3), Moi Paolo (2), Cucca Francesco (1,6), Sanna Serena (1), Uda Manuela (1)

(1) Istituto di Ricerca Genetica e Biomedica (IRGB), Consiglio Nazionale delle Ricerche (CNR), Cittadella Universitaria di Monserrato, SS 554 bivio per Sestu, Monserrato, Cagliari, Italy; Tel 0706754620; Fax 07064652; email manuela.uda@irgb.cnr.it;
(2) Clinica Pediatrica 2a, Dipartimento di Scienze Biomediche e Biotecnologie, Università di Cagliari, Ospedale Regionale Microcitemie ASL8, Cagliari, Italy
(3) Centro di Ricerca, Sviluppo e Studi Superiori in Sardegna(CRS4) Parco Scientifico e tecnologico della Sardegna, Pula, Italy
(4) Center for Statistical Genetics, Department of Biostatistics, University of Michigan, Ann Arbor, Michigan, USA
(5) Laboratory of Genetics, National Institute on Aging, Baltimore, Maryland, USA
(6) Dipartimento di Scienze Biomediche, Università di Sassari, Sassari, Italy

ABSTRACT N. 190

Telethon Research Projects - Other Genetic Diseases		
Principal Investigator	**RISTALDI MARIA SERAFINA**	
Telethon grant N.	**GGP14065**	
Total budget €	**166.000**	
Centres: **1**	*Duration (yrs):* **3**	*Starting year:* **2014**

VALIDATION OF THE HUMAN DELTA GLOBIN GENE AS A THERAPEUTIC TARGET FOR BETA THALASSEMIA AND SICKLE CELL DISEASE

VALIDAZIONE DEL GENE DELTA GLOBINICO UMANO QUALE TARGET TERAPEUTICO PER LA BETA TALASSEMIA E L'ANEMIA FALCIFORME

Porcu Susanna, Simbula Michela, Marongiu Maria Franca, Manchinu Francesca, Perseu Luciana, Daniela Poddie, Maria Serafina Ristaldi

IRGB/CNR, Cittadella Universitaria Di Monserrato SS554 Km 4,5, 09042 Monserrato (CA), tel. 0706754592, fax 0706754652, mail ristaldi@irgb.cnr.it

ABSTRACT N. 191

OSR-TIGET - Other Genetic Diseases		
Principal Investigator	**FERRARI GIULIANA**	
Telethon grant N.	**TGTGSK04**	
Total budget €	**3.674.545**	
Centres: **1**	*Duration (yrs):* **5**	*Starting year:* **2011**

PRECLINICAL SAFETY EVALUATION OF GENE THERAPY MEDICINAL PRODUCTS: TUMORIGENICITY, TOXICOLOGY AND BIODISTRIBUTION GLP STUDIES TO SUPPORT A BETA-THALASSEMIA GENE THERAPY CLINICAL TRIAL

VALUTAZIONE PRECLINICA DELLA BIOSICUREZZA DI PRODOTTI MEDICINALI BASATI SU TERAPIA GENICA: STUDI GLP DI TUMORIGENICITÀ, TOSSICITÀ E BIODISTRIBUZIONE PER L'APPLICAZIONE CLINICA DELLA TERAPIA GENICA DELLA BETA-TALASSEMIA

Paleari Ylenia (1,3), Lidonnici Maria Rosa (1), Tiboni Francesca (1), Rossi Claudia (1), Mandelli Giacomo (1), Vezzoli Michela (1), Aprile Annamaria (1), Montini Eugenio (1), Calabria Andrea (1), Sanvito Francesca (2), Cristofori Patrizia (4), Ferrari Giuliana (1,3)

(1) San Raffaele-Telethon Institute for Gene Therapy (HSR-TIGET), Milan, Italy - Via Olgettina, 58, 20132 Milano; phone 02-26434705, fax 02-26434668; g.ferrari@hsr.it
(2) San Raffaele Scientific Institute, Milan, Italy
(3) Vita-Salute San Raffaele University, Milan, Italy
(4) Glaxo Smith Kline, R&D, Ware, UK

ABSTRACT N. 192

Telethon Research Projects - Other Genetic Diseases		
Principal Investigator	**BALDUINI CARLO LUIGI**	
Telethon grant N.	**GGP10089**	
Total budget €	**371.000**	
Centres: **4**	*Duration (yrs):* **3**	*Starting year:* **2011**
Partners	SILVERIO PERROTTA, MARCO SERI, ANNA SAVOIA	

A NEW GENE FOR INHERITED THROMBOCYTOPENIAS: CLINICAL, PATHOGENETIC AND PHARMACOLOGICAL STUDIES

UN NUOVO GENE RESPONSABILE DI PIASTRINOPENIA EREDITARIA: STUDI CLINICI, PATOGENETICI E FARMACOLOGICI

Seri Marco (2), Savoia Anna (3), Perrotta Silverio (4), Pecci Alessandro (1), Marconi Caterina (2), Gnan Chiara (3), Noris Patrizia (1), Magini Pamela (2), Faleschini Michela (3), Pippucci Tommaso (2), Balduini Alessandra (5), Balduini Carlo L. (1)

(1) Dipartimento di Medicina Interna, Fondazione IRCCS Policlinico San Matteo-Università di Pavia, piazzale Golgi, 27100 Pavia, tel 0382 502183, fax 0382 526223, email c.balduini@smatteo.pv.it
(2) Dipartimento di Scienze Mediche Chirurgiche, Policlinico Sant'Orsola-Malpighi, Università di Bologna, Bologna
(3) Dipartimento di Scienze Mediche, Università di Trieste, Trieste
(4) Dipartimento di Pediatria, Seconda Università di Napoli, Napoli
(5) Dipartimento di Medicina Molecolare, Università di Pavia, Pavia

ABSTRACT N. 193

Telethon Research Projects - Other Genetic Diseases		
Principal Investigator	**DI VIRGILIO FRANCESCO**	
Telethon grant N.	**GGP11014**	
Total budget €	**190.000**	
Centres: **1**	*Duration (yrs):* **3**	*Starting year:* **2011**

THE P2X7 RECEPTOR/ADENOSINE-GENERATION SYSTEM: A NOVEL TARGET FOR THE THERAPY OF AUTOINFLAMMATORY DISEASES

IL SISTEMA RECETTORE P2X7/ADENOSINA: UN NUOVO BERSAGLIO PER LA TERAPIA DELLE MALATTIE AUTOINFIAMMATORIE

Sarti Alba Clara (1), Franceschini Alessia (1), Salaro Erica (1), Falzoni Simonetta (1), Ciancio Giovanni (2), Govoni Marcello (2), Di Virgilio Francesco (1)

(1) Dipartimento di Morfologia, Chirurgia e Medicina Sperimentale, Sezione di Patologia, Oncologia e Biologia Sperimentale, Università degli Studi di Ferrara, Via Borsari 46, 44121, Ferrara (Italy). Tel: 0532 455353; Fax: 0532 455351; email: fdv@unife.it
(2) Dipartimento di Scienze Mediche, Sezione di Reumatologia, Arcispedale S. Anna, Cona, Ferrara, Italy

ABSTRACT N. 194

Telethon Research Projects - Other Genetic Diseases		
Principal Investigator	**GATTORNO MARCO**	
Telethon grant N.	**GGP14144**	
Total budget €	**377.200**	
Centres: **2**	*Duration (yrs):* **3**	*Starting year:* **2014**
Partners	ANNA RUBARTELLI	

CRYOPYRIN ASSOCIATED PERIODIC SYNDROMES (CAPS): INVESTIGATIONS ON PATIENTS BLOOD CELLS AND IN A KNOCK-IN MOUSE MODEL TO EXPLOIT NOVEL APPROACHES FOR THE MODULATION OF THE NLRP3 INFLAMMASOME

SINDROME PERIODICA ASSOCIATA A DEFICIT DI CRIOPIRINA (CAPS): STUDI SU CELLULE PRIMARIE DI PAZIENTI E SU UN MODELLO ANIMALI PER IDENTIFICARE NUOVI APPROCCI TERAPAUTICI PER LA MODULAZIONE DEL NLRP3 INFLAMMASOMA

Gattorno Marco (1), Sonia Carta (2), Arinna Bertoni (1), Federica Penco (1), Rosa Lavieri (2), Sabrina Chiesa (1), Anna Rubartelli (2)

(1) UO Pediatria 2, Istituto Gaslini - Via Gaslini 5, 16145, Genova. 010 56362793, fax: 010 4211018, email: marcogattorno@ospedale-gaslini.ge.it
(2) Cell Biology Unit, IRCCS San Martino IST, Genova

ABSTRACT N. 195

Telethon Research Projects - Other Genetic Diseases		
Principal Investigator	**GIACCA MAURO**	
Telethon grant N.	**GGP11068**	
Total budget €	**204.600**	
Centres: **1**	*Duration (yrs):* **3**	*Starting year:* **2011**

TOWARDS BETTER AAV GENE THERAPY: WHOLE GENOME SIRNA AND MICRORNA HIGH THROUGHPUT SCREENING FOR THE IDENTIFICATION OF THE MOLECULAR DETERMINANTS GOVERNING AAV VECTOR TRANSDUCTION, VECTOR PRODUCTION AND VECTOR-INDUCED GENE CORRECTION

VERSO UNA PIÙ EFFICACE TERAPIA GENICA CON VETTORI AAV; SCREENING AD ALTA PROCESSIVITÀ PER L'IDENTIFICAZIONE DEI DETERMINANTI MOLECOLARI CHE REGOLANO LA TRASDUZIONE DA AAV, LA PRODUZIONE DEI VETTORI E LA CORREZIONE GENICA

Zentilin Lorena, Mano Miguel, Ippodrino Rudy, Zacchigna Serena, Backovic Ana, Giacca Mauro

International Center for Genetic Engineering and Biotechnology (ICGEB), Padriciano 99, 34149 Trieste, Italia, Tel. +39.040.3757324, Fax +39.040.226555, E-mail: giacca@icgeb.org

ABSTRACT N. 196

OSR-TIGET - Other Genetic Diseases		
Principal Investigator	**MONTINI EUGENIO**	
Telethon grant N.	**TGT11D01**	
Total budget €	**731.850**	
Centres: **1**	*Duration (yrs):* **5**	*Starting year:* **2011**

IMPROVING SAFETY OF LENTIVIRAL GENE TRANSFER

MIGLIORANDO LA BIOSICUREZZA DEL TRASFERIMENTO GENICO CON VETTORI LENTIVIRALI

Cesana Daniela, Volpin Monica, Calabria Andrea, Spinozzi Giulio, Brasca Stefano, Benedicenti Fabrizio, Rudilosso Laura, Gallina Pierangela, Tenderini Erika, Montini Eugenio

San Raffaele Telethon Institute for Gene Therapy, Via Olgettina 58, 201132 Milano, Tel: +39 02 26434069; montini.eugenio@hsr.it

ABSTRACT N. 197

OSR-TIGET - Other Genetic Diseases		
Principal Investigator	**RONCAROLO MARIA GRAZIA**	
Telethon grant N.	**TGT11E01**	
Total budget €	**954.350**	
Centres: **1**	*Duration (yrs):* **5**	*Starting year:* **2011**

LIVER-DIRECTED GENE TRANSFER FOR THE INDUCTION OF ANTIGEN-SPECIFIC TOLERANCE: MECHANISM AND THERAPEUTIC APPLICATIONS

INDUZIONE DI TOLLERANZA AG-SPECIFICA ATTRAVERSO IL TRASFERIMENTO GENICO NEL PARENCHIMA EPATICO: MECCANISMO E APPLICAZIONI

Annoni Andrea (1), Cantore Alessio (1), Akbarpour Mahzad (1), Naldini Luigi (1,2), Roncarolo Maria Grazia (1,2)

(1) San Raffaele Telethon Institute for Gene Therapy (HSR-TIGET), Division of Regenerative Medicine, Stem Cells and Gene Therapy, IRCCS San Raffaele Scientific Institute, Via Olgettina 58, 20132 Milan, Italy
(2) Vita e Salute University, Milan, Italy

ABSTRACT N. 198

OSR-TIGET - Other Genetic Diseases		
Principal Investigator	**GREGORI SILVIA**	
Telethon grant N.	**TGT11E02**	
Total budget €	**710.650**	
Centres: **1**	*Duration (yrs):* **5**	*Starting year:* **2011**

CELL THERAPY STRATEGIES FOR TOLERANCE INDUCTION IN HUMANS

STRATEGIE DI TERAPIA CELLULARE PER INDUZIONE DELLA TOLLERANZA IMMUNOLOGICA

Amodio Giada (1), Andolfi Grazia (1), Locafaro Grazia (1), Comi Michela (1), Russo Fabio (1), Roncarolo Maria Grazia (1,2), Gregori Silvia (1)

(1) San Raffaele Telethon Institute for Gene therapy (HSR-TIGET), Division of Regenerative Medicine, Stem Cells and Gene Therapy, IRCCS San Raffaele Scientific Institute - Via Olgettina 58, 20132 Milan, Italy
(2) Vita-Salute San Raffaele University, Milan, Italy

ABSTRACT N. 199

OSR-TIGET - Other Genetic Diseases		
Principal Investigator	**AIUTI ALESSANDRO**	
Telethon grant N.	**TGT11C01**	
Total budget €	**739.000**	
Centres: **1**	*Duration (yrs):* **5**	*Starting year:* **2011**

TRACKING AND MODELING OF HSC CLONAL DYNAMICS BY VECTOR MARKING

STUDIO DELLE DINAMICHE CLONALI DELLE CELLULE STAMINALI EMATOPOIETICHE E MODELLI DI EMATOPOIESI UMANA MEDIANTE L'ANALISI DELLE INTEGRAZIONI VIRALI

Luca Biasco (1), Francesca Dionisio (1,2), Danilo Pellin (3), Samantha Scaramuzza (1), Serena Scala (1), Luca BassoRicci (1), Cristina Baricordi (1), Stefania Giannelli (1), Victor Neduva (4), David J Dow (4), Andrea Calabria (1), Paola Vicard (5), Ernst Wit (6), Eugenio Montini (1), Clelia Di Serio (2,3), Luigi Naldini (1,2), Alessandro Aiuti (1,7)

(1) San Raffaele Telethon Institute for Gene Therapy (TIGET), Division of Regenerative Medicine Stem cells and Gene Therapy, IRCCS San Raffaele Scientific Institute, Via Olgettina, 58 20132, Milan, Italy email: aiuti.alessandro@hsr.it tel:02-2643-4875 fax:02-2643-4668
(2) Univ. Vita-Salute San Raffaele, Milano, Italy
(3) CUSSB, Università Vita-Salute, Milan Italy
(4) Molecular and Cellular Technologies, GlaxoSmithKline, Stevenage Herts, UK
(5) Univ. degli studi Roma Tre, Rome Italy

(6) Univ. of Groningen Faculty of Mathematics and Natural Sciences, Groningen, NE
(7) Univ. of Rome "Tor Vergata", Rome, Italy

ABSTRACT N. 200

OSR-TIGET - Other Genetic Diseases		
Principal Investigator	**KAJASTE RUDNITSKI ANNA**	
Telethon grant N.	**TGT11D04**	
Total budget €	**286.357**	
Centres: **1**	*Duration (yrs):* **2**	*Starting year:* **2014**

DISSECTING LENTIVIRUS-HOST INTERACTIONS TO IMPROVE GENE TRANSFER

STUDIO DELLE INTERAZIONI LENTIVIRUS-OSPITE PER MIGLIORARE IL TRASFERIMENTO GENICO

Petrillo Carolina (1), Piras Francesco (1), Riba Michela (2), Matafora Vittoria (3), Lazarevic Dejan (2), Cittaro Davide (2), Stupka Elia (4), Bachi Angela (3), Naldini Luigi (1), Kajaste-Rudnitski Anna (1)

(1) San Raffaele Telethon Institute for Gene Therapy, Milan, Italy - Via Olgettina 58, Milan, Italy, Tel. +390226436546, fax +390226434668/4621, e-mail: kajaste.anna@hsr.it
(2) San Raffaele Center for Translational Genomics and Bioinformatics, Milan, Italy
(3) FIRC Institute of Molecular Oncology Foundation (IFOM), Milan, Italy
(4) Neu-Ulm, Bayern, Germany

ABSTRACT N. 201

OSR-TIGET - Other Genetic Diseases		
Principal Investigator	**LOMBARDO ANGELO**	
Telethon grant N.	**TGT11D05**	
Total budget €	**316.142**	
Centres: **1**	*Duration (yrs):* **2**	*Starting year:* **2014**

PERMANENT EPIGENETIC SILENCING OF HUMAN GENES BY TRANSIENT DELIVERY OF ARTIFICIAL TRANSCRIPTIONAL REPRESSORS

SILENZIAMENTO PERMANENTE EPIGENETICO DI GENI UMANI TRAMITE ESPRESSIONE TRANSIENTE DI REPRESSORI TRASCRIZIONALI ARTIFICIALI

Amabile Angelo (2,3), Migliara Alessandro (2,3), Di Tomaso Tiziano (2), Biffi Mauro (2), Firrito Claudia (2), Naldini Luigi (2,3), Lombardo Angelo (1)

(1) San Raffaele Telethon Institute for Gene Therapy and Vita Salute San Raffaele University, Milan, Italy - Via Olgettina 58, 20132 Milan, ITALY; Tel: 0226435680 Fax: 0226434621; lombardo.angelo@hsr.it
(2) San Raffaele Telethon Institute for Gene Therapy, Milan, Italy
(3) Vita Salute San Raffaele University, Milan, Italy

ABSTRACT N. 202

OSR-TIGET - Other Genetic Diseases		
Principal Investigator	**AIUTI ALESSANDRO**	
Telethon grant N.	**TGTGSK06**	
Total budget €	**1.183.468**	
Centres: **1**	*Duration (yrs):* **5**	*Starting year:* **2011**

DUAL-REGULATED LENTIVIRAL VECTOR FOR GENE THERAPY OF X-LINKED CHRONIC GRANULOMATOUS DISEASE

SVILUPPO DI UN VETTORE BI-REGOLATO PER LA TERAPIA GENICA DELLA MALATTIA GRANULOMATOSA CRONICA LEGATA AL CROMOSOMA X

Farinelli Giada (1), Chiriaco Maria (2), Capo Valentina (1), Zonari Erika (1), Migliavacca Maddalena (1), Jofra-Hernandez Raisa (1), Scaramuzza Samantha (1), Di Matteo Gigliola (2), Sergi Sergi Lucia (1), Kajaste-Rudnitski Anna (1), Trono Didier (3), Grez Manuel (4), Rossi Paolo (2), Bragonzi Alessandra (5), Rossi Alice (5), Ranucci Serena (5), Finocchi Andrea (2), Naldini Luigi (1,6), Gentner Bernhard (1), Aiuti Alessandro (1,6)

(1) San Raffaele Telethon Institute for Gene Therapy (TIGET), IRCCS San Raffaele Scientific Institute, Milan, Italy - Via Olgettina 58 20132 Milano Italy Tel. +390226436324; Fax +390226434668; e mail: alessandro.aiuti@hsr.it
(2) Department of Pediatrics, Children's hospital Bambino Gesù and University of Rome Tor Vergata School of medicine, Rome, Italy
(3) Ecole Polytechnique Federale de Lausanne, Lausanne, Switzerland
(4) Georg-Speyer Haus, Frankfurt, Germany
(5) Infection and Cystic Fibrosis Unit, IRCCS San Raffaele Scientific Institute, Milan, Italy
(6) "Vita-Salute" S. Raffaele University, Milan, Italy

ABSTRACT N. 203

Telethon Research Projects - Other Genetic Diseases		
Principal Investigator	**BALDARI COSIMA T.**	
Telethon grant N.	**GGP11021**	
Total budget €	**264.000**	
Centres: **1**	*Duration (yrs):* **3**	*Starting year:* **2011**

THE INTRAFLAGELLAR TRANSPORT SYSTEM, A NOVEL REGULATOR OF IMMUNE SYNAPSE ASSEMBLY: FUNCTIONAL DISSECTION IN T-CELL ANTIGEN RECEPTOR TRAFFICKING AND ASSESSMENT AS DISEASE TARGET IN COMMON VARIABLE IMMUNODEFICIENCY

IL SISTEMA DI TRASPORTO INTRAFLAGELLARE NEI LINFOCITI T: DISSEZIONE FUNZIONALE NEL TRASPORTO DEL RECETTORE DELL'ANTIGENE E VALUTAZIONE QUALE BERSAGLIO DELLA MALATTIA NELL'IMMUNODEFICIENZA COMUNE VARIABILE

Finetti Francesca (1), Patrussi Laura (1), Onnis Anna (1), Galgano Donatella (1), Gottardo Marco (1), Cassioli Chiara (1), Spanò Stefania (2), Galan Jorge (2), Pazour Gregory J (3), Baldari Cosima T (1)

(1) Department of Life Sciences, University of Siena, Siena, Italy - via A. Moro, 2 53100 Siena tel +39 0577 234400 fax +39 0577234476 baldari@unisi.it
(2) Section of Microbial Pathogenesis, Yale University School of Medicine, New Haven, USA
(3) Program in Molecular Medicine, University of Massachusetts Medical School, Worcester, USA

ABSTRACT N. 204

Telethon Research Projects - Other Genetic Diseases		
Principal Investigator	**FERRARI SIMONA**	
Telethon grant N.	**GGP12052**	
Total budget €	**50.0000**	
Centres: **1**	*Duration (yrs):* **1**	*Starting year:* **2012**

IDENTIFICATION OF THE GENE RESPONSIBLE FOR AN AUTOSOMAL DOMINANT FORM OF COMMON VARIABLE IMMUNODEFICIENCY

IDENTIFICAZIONE DEL GENE RESPONSABILE DI UNA FORMA AUTOSOMICA DOMINANTE DI IMMUNODEFICIENZA COMUNE VARIABILE (CVID)

Di Pierro Valentina (1), Bacchelli Elena (2), Zuntini Roberta (1),

Schaffer Alejandro (3), Grimbacher Bodo (4), Quinti Isabella (5), and Ferrari Simona (1)

(1) Medical Genetic Unit, S.Orsola-Malpighi University Hospital - Via Massarenti 9 - 40138 Bologna. phone: *39 051 2088410; Fax: +39 051 2088416; email: simo.ferrari@unibo.it
(2) FaBiT, University of Bologna
(3) National Center for Biotechnology Information, NIH, Bethesda, USA
(4) Centre of Chronic Immunodeficiency, University Medical Centre Freiburg, Germany
(5) Department of Immunology, University of Roma "La Sapienza"

ABSTRACT N. 205

Telethon Research Projects - Other Genetic Diseases		
Principal Investigator	**SCHMITZ KERSTIN MAIKE**	
Telethon grant N.	**GGP13155**	
Total budget €	**121.000**	
Centres: **1**	*Duration (yrs):* **2**	*Starting year:* **2013**

A NOVEL AID STRUCTURE PROVIDING NEW INSIGHT INTO HIGM2

NUOVA LUCE SUL GENE AID E LA SINDROME DI IMMUNODEFICIENZA CON IPER-IGM DI TIPO II

Schmitz Kerstin-Maike (1), Mastrovito Marialaura (1), La Mastra Federica (1), Arakawa Hiroshi (1), Conticello Silvo (2), Casola Stefano (1), Mondino Anna (3), Petersen-Mahrt Svend (1)

(1) IFOM The FIRC Institute of Molecular Oncology, Via Adamello 16, 20139 Milan, Italy, Phone (39)-02574303323, Fax (39)-02574303231 kerstin.schmitz@ifom.eu
(2) Molecular Mechanisms of Oncogenesis, ITT Core Research Laboratory (CRL), Villa delle Rose, via Cosimo il Vecchio 2, 50139 Firenze, Italy
(3) San Raffaele Scientific Institute, DIBIT, Via Olgettina 58, 20132 Milan, Italy, Phone (39)-0226434801, fax (39)-0226434844anna.mondino@hsr.it

ABSTRACT N. 206

OSR-TIGET - Other Genetic Diseases		
Principal Investigator	**BACCHETTA ROSA**	
Telethon grant N.	**TGT11A04**	
Total budget €	**946.050**	
Centres: **1**	*Duration (yrs):* **5**	*Starting year:* **2011**

CELL/GENE TRANSFER BASED THERAPIES FOR IPEX SYNDROME AND FOXP3-GENE INDEPENDENT DISEASES WITH IMMUNE DYSREGULATION

TERAPIA GENICA E CELLULARE PER LA SINDROME IPEX E SIMILI PATOLOGIE (IPEX-LIKE) CON IMMUNODISREGOLAZIONE FOXP3-INDIPENDENTI

Passerini Laura (1), Santoni de Sio Francesca Romana (1), Barzaghi Federica (1,2), Rossi Mel Eva (1,3), Sartirana Claudia (1), Restelli Silvia (1), Maccari Maria Elena (2), Naldini Luigi (1,2), Roncarolo Maria Grazia (2,4), Bacchetta Rosa (1,4)

(1) San Raffaele Telethon Institute for Gene Therapy, Division of Regenerative Medicine, Stem Cells and Gene Therapy, IRCCS San Raffaele Scientific Institute, Milan, Italy - San Raffaele Telethon Institute for Gene Therapy, Division of Regenerative Medicine, Stem Cells and Gene Therapy, IRCCS San Raffaele Scientific Institute, Via Olgettina 58, 20132, Milan, Italy; Tel: +390226434669; Fax: +390226434668; email: bacchetta.rosa@hsr.it; rosab@stanford.edu
(2) Vita-Salute San Raffaele University, Milan, Italy
(3) University of Rome Tor Vergata, Rome, Italy
(4) Department of Pediatrics, Stanford Medical Center, Stanford, CA, USA

ABSTRACT N. 207

OSR-TIGET - Other Genetic Diseases		
Principal Investigator	**NALDINI LUIGI**	
Telethon grant N.	**TGT11D02**	
Total budget €	**918.400**	
Centres: **1**	*Duration (yrs):* **5**	*Starting year:* **2011**

SITE-SPECIFIC INTEGRATION AND GENE CORRECTION IN HUMAN PRIMARY CELLS BY ARTIFICIAL-NUCLEASES

INTEGRAZIONE SITO SPECIFICA E CORREZIONE GENICA MIRATA IN CELLULE STAMINALI UMANE MEDIANTE NUCLEASI ARTIFICIALI

Genovese Pietro (2), Schiroli Giulia (2,3), Firrito Claudia (2), Di Tomaso Tiziano (2), Gregory Philip D (4), Gabriel Richard (5), von Kalle Christof (5), Rio Paula (6), Bueren Juan (6), Bonini Chiara (7), Gentner Bernhard (2), Montini Eugenio (2), Lombardo Angelo (2,3), Naldini Luigi (1)

(1) San Raffaele Telethon Institute for Gene Therapy and Vita Salute San Raffaele University - Via Olgettina, 58, 20132 Milan, Italy; Tel: 02-2643-4681 Fax: 02-2643-4621; naldini.luigi@hsr.it
(2) San Raffaele Telethon Institute for Gene Therapy, Milan, Italy
(3) Vita Salute San Raffaele University, Milan, Italy
(4) Sangamo BioSciences, Richmond, CA, United States
(5) NCT-DKFZ, Heidelberg, Germany
(6) CIEMAT/CIBERER, Madrid, Spain
(7) San Raffaele Scientific Institute, Milan, Italy

ABSTRACT N. 208

Telethon Research Projects - Other Genetic Diseases		
Principal Investigator	**GRAZIANI ANDREA**	
Telethon grant N.	**GGP13254**	
Total budget €	**375.000**	
Centres: **2**	*Duration (yrs):* **3**	*Starting year:* **2013**
Partners	GIAN CESARE TRON	

INHIBITION OF DIACYLGLYCEROL KINASE ALPHA RESCUES TCR-INDUCED DIACYLGLYCEROL SIGNALLING AND RESTIMULATION INDUCED CELL DEATH IN XLP T LYMPHOCYTES

UNA NUOVA STRATEGIA FARMACOLOGICA PER LA SINDROME LINFOPROLIFERATIVA LEGATA AL CROMOSOMA X BASATA SULL'INIBIZIONE DELL'ENZIMA DIACILGLICEROLO CHINASI ALFA

Das Rupali (2), Malacarne Valeria (1), Ruffo Elisa (1), Patrussi Laura (4), Baldari Tatiana Cosima (4), Rubio Ignacio (5), Nichols Kim E. (2), Snow Andrew (3), Larens Sasha (3), Baldanzi Gianluca (1), Graziani Andrea (1)

(1) Dept. Traslational Medicine and Institute for Research and Cure of Autoimmune Diseases, University of Piemonte Orientale, via Solaroli 17, 28100 Novara, Italy, email: andrea.graziani@med.unipmn.it
(2) Division of Oncology; Children's Hospital of Philadelphia; Philadelphia, PA USA
(3) Dept. of Pharmacology, Uniformed Services University of the Health Sciences, Bethesda, MD 20814, USA
(4) Department of Life Sciences, University of Siena, Siena, Italy.
(5) Jena University Hospital, Jena, Germany

ABSTRACT N. 209

OSR-TIGET - Other Genetic Diseases		
Principal Investigator	**VILLA ANNA**	
Telethon grant N.	**TGT11A03**	
Total budget €	**512.600**	
Centres: **1**	*Duration (yrs):* **5**	*Starting year:* **2011**

MOLECULAR AND CELLULAR BASES OF INTESTINAL MUCOSAL PATHOLOGY IN OMENN SYNDROME

ANALISI DEI MECCANISMI CELLULARI E MOLECOLARI ALLA BASE DELL'AUTOIMMUNITÀ NELLA SINDROME DI OMENN

Cassani Barbara (2,8), Rigoni Rosita (2), Maina Virginia (1,8), Marrella Veronica (2,8), Pesole Graziano (5), Vezzoni Paolo (2,8), Grassi Fabio (4,6), Mora J.Rodrigo (7), Villa Anna (1,8)

(1) San Raffaele Telethon Institute for Gene Therapy (TIGET), San Raffaele Scientific Institute - Via Olgettina 58, Milan, Italy; phone +390226435273; fax: +390226434668; villa.anna@hsr.it
(2) Humanitas Clinical and Research Center, Rozzano, Italy
(3) San Raffaele Telethon Institute for Gene Therapy (TIGET), San Raffaele Scientific Institute, Milan, Italy
(4) Dipartimento di Biologia e Genetica per le Scienze Mediche, Università degli Studi, Milan, Italy
(5) Dipartimento di Bioscienze, Biotecnologie e Biofarmaceutica, Università di Bari, Italy
(6) Institute for Research in Biomedicine (IRB), Bellinzona (Switzerland)
(7) Gastrointestinal Unit, Massachusetts General Hospital, Harvard Medical School, Boston, Massachusetts, USA
(8) UOS/IRGB, Milan Unit, CNR, Milan, Italy

ABSTRACT N. 210

OSR-TIGET - Other Genetic Diseases		
Principal Investigator	**AIUTI ALESSANDRO**	
Telethon grant N.	**TGT11A01**	
Total budget €	**576.850**	
Centres: **1**	*Duration (yrs):* **5**	*Starting year:* **2011**

LOSS OF CENTRAL AND PERIPHERAL TOLERANCE MECHANISMS LEADING TO AUTOIMMUNE MANIFESTATIONS IN ADA-DEFICIENT SCID

STUDIO DEI MECCANSIMI DI PERDITA DELLA TOLLERANZA CENTRALE E PERIFERICA CHE COMPORTANO MANIFESTAZIONI AUTOIMMUNI NELL'ADA-SCID

Sauer AV (1), Brigida I (1), Carriglio N (1), Di Lorenzo B (1), Jofra Hernandez R (1), Giannelli S (1), Carlucci F (3), Poliani L (4), Di Marzo V (5), Hollander GA (6,7), Van der Burg M (8), Aiuti A (1,2)

(1) Telethon Institute for Gene Therapy (HSR-TIGET), Milan, Italy - Via Olgettina 58, 20132 Milano, +390226434671 +390226434668, a.aiuti@hsr.it
(2) Pediatric Immunohematology and Bone Marrow Transplantation Unit, San Raffaele Scientific Institute, Milano, Italy
(3) Department of Internal Medicine, Endocrine-Metabolic Sciences and Biochemistry, University of Siena, Italy
(4) Department of Pathology, University of Brescia, Brescia, Italy
(5) Endocannabinoid Research Group, Institute of Biomolecular Chemistry - CNR, Pozzuoli, Italy
(6) Laboratory of Pediatric Immunology, Department of Biomedicine, University of Basel and Basel University Children's Hospital, Basel, Switzerland
(7) Developmental Immunology, Department of Paediatrics, University of Oxford, Oxford, United Kingdom
(8) Department of Immunology, Erasmus MC, University Medical Center, Rotterdam, The Netherlands

ABSTRACT N. 211

OSR-TIGET - Other Genetic Diseases		
Principal Investigator	**NALDINI LUIGI**	
Telethon grant N.	**TGTGSK01**	
Total budget €	**134.763**	
Centres: **1**	*Duration (yrs):* **2**	*Starting year:* **2011**

GENETIC ENGINEERING AND TRANSPLANTATION OF HIGHLY PURIFIED HEMATOPOIETIC STEM CELLS (HSC) FOR IMPROVED EX VIVO GENE THERAPY

OTTIMIZZAZIONE DELLA TERAPIA GENICA EX VIVO MEDIANTE INGEGNERIZZAZIONE GENETICA E TRAPIANTO DI CELLULE STAMINALI EMATOPOIETICHE (CSE) ARRICCHITE

Zonari Erika (1), Boccalatte Francesco (1), Plati Tiziana (1), Ranghetti Anna (1), Escobar Giulia (1), Montini Eugenio (1), Biffi Alessandra (1), Ferrari Giuliana (1), Aiuti Alessandro (1), Gentner Bernhard (1,3)*, Naldini Luigi (1,2)*

(1) San Raffaele Telethon Institute for Gene Therapy (TIGET), Via Olgettina 58, 20132, Milan, Italy - Phone: +39 02 26434681; Fax: +39 02 26434621; E-mail: naldini.luigi@hsr.it
(2) Vita-Salute San Raffaele University, Milan, Italy
(3) Department of Hematology - San Raffaele Hospital, Milan, Italy
* Shared Senior Authorship

ABSTRACT N. 212

Telethon Research Projects - Other Genetic Diseases		
Principal Investigator	**VIOLA ANTONELLA**	
Telethon grant N.	**GGP10170**	
Total budget €	**284.800**	
Centres: **2**	*Duration (yrs):* **3**	*Starting year:* **2011**
Partners	RAFFAELE BADOLATO	

UNDERSTANDING THE WHIM SYNDROME: MOLECULAR ANALYSIS OF CXCR4 FUNCTIONS IN LEUKOCYTE TRAFFICKING AND ACTIVATION

LA PATOGENESI DELLA SINDROME WHIM: ANALISI DELLE FUNZIONI DEL CXCR4 NELLA ATTIVAZIONE E NEL TRAFFICO DEI LINFOCITI

Roselli Giuliana (1), Kallikourdis Marinos (1), Viola Antonella (2,3)

(1) Fondazione Humanitas per la Ricerca
(2) Dipartimento di Scienze Biomediche Sperimentali, Università di Padova
(3) Istituto Veneto di Medicina Molecolare, Padova

ABSTRACT N. 213

Telethon Research Projects - Other Genetic Diseases		
Principal Investigator	**BENVENUTI FEDERICA**	
Telethon grant N.	**GGP14281**	
Total budget €	**201.800**	
Centres: **1**	*Duration (yrs):* **3**	*Starting year:* **2015**

PLASMACYTOID DENDRITC CELLS FUNCTIONS AND AUTOIMMUNITY IN WISKOTT-ALDRICH SYNDROME

CELLULE DENDRITICHE PLASMACITOIDI E INTERFERONE DI TIPO PRIMO NELLO SVILUPPO DEI FENOMENI AUTOIMMUNI NELLA SINDROME DI WISKOTT-ALDRICH

Naseem Asma (1), Cervantes Luevano Karla (1), Piperno Maria Giulia (1), Bosticardo Marita (2), Villa Anna (2,3), Benvenuti Federica (1)

(1) Cellular Immunology, ICGEB, Area di Ricerca, Padriciano 99, Trieste, Italy. Tel: +39 040 3757389. Fax: +39 040 266555, benvenut@icgeb.org

(2) San Raffaele Scientific Institute, Telethon Institute for Gene Therapy, 20132 Milan, Italy
(3) Milan Unit, Istituto di Ricerca Genetica e Biomedica, Consiglio Nazionale delle Ricerche, Milan, Italy

ABSTRACT N. 214

OSR-TIGET - Other Genetic Diseases		
Principal Investigator	**VILLA ANNA**	
Telethon grant N.	**TGT11A02**	
Total budget €	**533.700**	
Centres: **1**	*Duration (yrs):* **5**	*Starting year:* **2011**

B CELL RECONSTITUTION IN WISKOTT-ALDRICH SYNDROME PATIENTS TREATED WITH GENE THERAPY

MECCANISMI CELLULARI E MOLECOLARI ALLA BASE DELL'AUTOIMMUNITÀ NELLA SINDROME DI WISKOTT ALDRICH

Castiello Maria Carmina (1), Bosticardo Marita (1), Scaramuzza Samantha (1), Pala Francesca (1), Brigida Immacolata (1), Sereni Lucia (1), van der Burg Mirjam (2), Ottaviano Giorgio (3), Ferrua Francesca (1,3), Naldini Luigi (1,4), Aiuti Alessandro (1,4), Meffre Eric (5), Villa Anna (1,6)

(1) San Raffaele Telethon Institute for Gene Therapy (TIGET), San Raffaele Scientific Institute, via Olgettina 58, Milan, Italy, phone +39 02 26435273 fax: +39 02 26434668; villa.anna@hsr.it
(2) Department of Immunology, Erasmus MC, University Medical Center Rotterdam, GE Rotterdam, The Netherlands
(3) Pediatric Immunohematology and Bone Marrow Transplantation Unit, San Raffaele Scientific Institute, Milan, Italy
(4) Vita-Salute San Raffaele University, Milan, Italy
(5) Department of Immunobiology, Yale University School of Medicine, New Haven, Conneticut, USA
(6) IRGB CNR, Milan Unit, Milan, Italy

ABSTRACT N. 215

OSR-TIGET - Other Genetic Diseases		
Principal Investigator	**AIUTI ALESSANDRO**	
Telethon grant N.	**TGTGSK03**	
Total budget €	**3.510.976**	
Centres: **1**	*Duration (yrs):* **4**	*Starting year:* **2011**

CLINICAL TRIAL OF HEMATOPOIETIC STEM CELL GENE THERAPY FOR WISKOTT-ALDRICH-SYNDROME

TERAPIA GENICA MEDIATA DA VETTORI LENTIVIRALI PER IL TRATTAMENTO DELLA SINDROME DI WISKOTT-ALDRICH

Scaramuzza Samantha (1), Ferrua Francesca (2), Cicalese Maria Pia (2), Pajno Roberta (2), Giannelli Stefania (1), Biasco Luca (1), Castiello Maria Carmina (1), Assanelli Andrea (2), Casiraghi Miriam (2), Bosticardo Marita (1), Rizzardi Paolo (3), Finocchi Andrea (4), Metin Ayse (5), Albert Micheal (6), Biffi Alessandra (1,2), Villa Anna (1,7), Ciceri Fabio (8), Roncarolo Maria Grazia (1,2,9), Naldini Luigi (1,9), Aiuti Alessandro (1,2,9)

(1) San Raffaele Telethon Institute for Gene Therapy (HSR-TIGET), Milan, Italy - Via Olgettina 58, 20132 Milan, Italy, tel: +390226436324, fax: +390226434668, email: a.aiuti@hsr.it
(2) Pediatric Immuno-hematology and Bone Marrow Transplant, Scientific Institute HS Raffaele, Milan, Italy
(3) MolMed SpA, Milan, Italy
(4) Univ. of Rome "Tor Vergata", Rome, Italy
(5) Ankara Dispaki Children's Hospital, Ankara, Turkey
(6) Ludwig-Maximilians-Universität München, Germany
(7) IRGB-CNR, Milan, Italy
(8) Hematology and Bone Marrow Transplant, Scientific Institute HS Raffaele, Milan, Italy
(9) Università Vita-Salute San Raffaele, Milan, Italy

ABSTRACT N. 216

Telethon Research Projects - Other Genetic Diseases		
Principal Investigator	**SANTUCCI ANNALISA**	
Telethon grant N.	**GGP10058**	
Total budget €	**233.200**	
Centres: **1**	*Duration (yrs):* **3**	*Starting year:* **2010**

SET UP OF EXPERIMENTAL MODELS OF ALKAPTONURIA AND PRECLINICAL TESTING OF THERAPEUTIC AGENTS FOR THE TREATMENT OF OCHRONOTIC ARTHROPATHY

ALLESTIMENTO DI MODELLI SPERIMENTALI DI ALCAPTONURIA E VALUTAZIONE PRECLINICA DI AGENTI TERAPEUTICI PER IL TRATTAMENTO DELL'ARTROPATIA OCRONOTICA

Braconi Daniela (1), Millucci Lia (1), Bernardini Giulia (1), Gambassi Silvia (1), Laschi Marcella (1), Geminiani Michela (1), Ghezzi Lorenzo (1), Lupetti Pietro (2), Bernini Andrea (1), Spiga Ottavia (1), Niccolai Neri (1), Santucci Annalisa (1)

(1) Dipartimento di Biotecnologie, Chimica e Farmacia, Università degli Studi di Siena, Siena - Prof. Annalisa Santucci, via Aldo Moro 2, 53100 Siena (SI); tel: +39 0577234958; fax: +39 0577234254; email: annalisa.santucci@unisi.it
(2) Dipartimento di Scienze della Vita, Università degli Studi di Siena, Siena

ABSTRACT N. 217

Telethon Research Projects - Other Genetic Diseases		
Principal Investigator	**BELLOTTI VITTORIO**	
Telethon grant N.	**GGP14127**	
Total budget €	**136.400**	
Centres: **1**	*Duration (yrs):* **2**	*Starting year:* **2014**

FAMILIAL ß2-MICROGLOBULIN AMYLOIDOSIS: FROM THE ELUCIDATION OF THE PATHOGENIC MECHANISM TO THE DISCOVERY OF NOVEL EFFECTIVE DRUGS

AMILOIDOSI FAMILIARE DA ß2-MICROGLOBULINA: DALLA DELUCIDAZIONE DEL MECCANISMO PATOGENICO ALLA SCOPERTA DI NUOVI FARMACI ATTIVI

Mangione Patrizia (1,2), Giorgetti Sofia (1), Soria Cristina (1), Raimondi Sara (1), Marchese Loredana (1), Porcari Riccardo (2), Verona Guglielmo (1,2), Zorzoli Irene (3), Gallanti Angelo (1), Stoppini Monica (1), Bellotti Vittorio (1,2)

(1) Dipartimento di Medicina Molecolare, Istituto di Biochimica, Università degli Studi di Pavia, Via Taramelli 3b, 27100 Pavia, Tel 0382 987932; fax: 0382 423108; email: vbellot@unipv.it
(2) Centre for Amyloidosis and Acute Phase Proteins, University College London, London, UK
(3) Dipartimento di Medicina Interna e Terapia Medica, Università di Pavia

ABSTRACT N. 218

Telethon Research Projects - Other Genetic Diseases		
Principal Investigator	**HEPPENSTALL PAUL**	
Telethon grant N.	**GGP14012**	
Total budget €	**227.900**	
Centres: **1**	*Duration (yrs):* **3**	*Starting year:* **2014**

GENERATION OF ANIMAL MODELS TO DEFINE MOLECULAR MECHANISMS AND NOVEL THERAPEUTIC STRATEGIES FOR FAMILIAL PRIMARY LOCALIZED CUTANEOUS AMYLOIDOSIS

GENERAZIONE DI MODELLI ANIMALI ALLO SCOPO DI DEFINIRE I MECCANISMI MOLECOLARI ALLA BASE DELL'AMILOIDOSI CUTANEA LOCALIZZATA FAMILIARE PER LA RICERCA DI NUOVE STRATEGIE TERAPEUTICHE

Nocchi Linda, de Castro Reis Fernanda, Asaro Antonino, Portulano Carla, Heppenstall Paul

European Molecular Biology Laboratory, Via Ramarini Monterotondo 00015 (RM), paul.heppenstall@embl.de

ABSTRACT N. 219

Telethon Research Projects - Other Genetic Diseases		
Principal Investigator	**MURO ANDRES FERNANDO**	
Telethon grant N.	**GGP10051**	
Total budget €	**435.900**	
Centres: **5**	*Duration (yrs):* **3**	*Starting year:* **2010**
Partners	CLAUDIO TIRIBELLI, GIANLUCA TELL, STEFANO GUSTINCICH, LORENA ZENTILIN	

NEW DIAGNOSTIC AND THERAPEUTIC APPROACHES FOR THE CRIGLER-NAJJAR SYNDROME TYPE I

ELABORAZIONE DI NUOVI APPROCCI DIAGNOSTICI E TERAPEUTICI ALLA SINDROME DI CRIGLER-NAJJAR DI TIPO I

Bortolussi Giulia (1), Zentillin Lorena (1), Vodret Simone (1), Bockor Luka (1), Codarin Erica (2), Baj Gabriele (3), Bellarosa Cristina (6), Vanikova Jana (4), Calligaris Raffaella (5), Antoniali Giulia (2), Vascotto Carlo (2), Arena Simona (7), Giraudi Pablo (6), Viviani Giulia (1), Vlachouli Christina (5), Chiaruttini Giulia (1), Bittolo Tamara (3), Cesaratto Laura (2), Vatta Paolo (5), Mancarella Antonio (6), Scaloni Andrea (7), Schreuder Andrea (8), Vianello Eleonora (6), Benvenutti Federica (1), Verkade Henkjan (8), Vitek Libor (4), Giacca Mauro (1), Gustincich Stefano (5), Tell Gianluca (2), Tiribelli Claudio (6), Muro Andrés F. (1)

(1) International Centre for Genetic Engineering and Biotechnology (ICGEB), Padriciano, 99, 34149 - Trieste, Italia, Tel: 040-3757369; Fax: 040-226555; email: muro@icgeb.org
(2) Dipartimento di Scienze Mediche e Biologiche, Università di Udine, Udine, Italy
(3) Basic Research and Integrative Neuroscience (BRAIN) Centre for Neuroscience, Department of Life Sciences, University of Trieste, Italy
(4) Institute of Medical Biochemistry and Laboratory Medicine, First Faculty of Medicine, Charles University, Prague, Czech Republic
(5) Sector of Neurobiology, International School for Advanced Studies (SISSA), Trieste, Italy
(6) Centro Studi Fegato, Fondazione Italiana Fegato, Campus Basovizza Trieste, Italy
(7) Proteomics & Mass Spectrometry Laboratory, ISPAAM, National Research Council, Naples, Italy
(8) Pediatric Gastroenterology and Hepatology, University of Groningen, Groningen, the Netherlands

ABSTRACT N. 220

Telethon Research Projects - Other Genetic Diseases		
Principal Investigator	**ALTRUDA FIORELLA**	
Telethon grant N.	**GGP14028**	
Total budget €	**312.400**	
Centres: **2**	*Duration (yrs):* **3**	*Starting year:* **2014**
Partners	GIOVANNI CAMUSSI	

CELL THERAPY FOR CRIGLER-NAJJAR TYPE I SYNDROME USING HUMAN ADULT LIVER STEM CELLS

TERAPIA CELLULARE PER LA SINDROME DI CRIGLER-NAJJAR DI TIPO I CON CELLULE STAMINALI EPATICHE UMANE

Camussi Giovanni (2), Fagoonee Sharmila (1), Famulari Elvira (1), Muro Andres (3), Bruno Stefania (2), Altruda Fiorella (1)

(1) Dept. Molecular Biotechnology and Health Sciences, University of Torino, via Nizza 52,10126 Torino, 011 6706414, 011 6706432, fiorella.altruda@unito.it
(2) Dept. Medical Sciences, University of Torino
(3) ICGEB, Trieste

ABSTRACT N. 221

Telethon Research Projects - Other Genetic Diseases		
Principal Investigator	**GALIETTA LUIS JUAN VICENTE**	
Telethon grant N.	**GGP10026**	
Total budget €	**391.000**	
Centres: **1**	*Duration (yrs):* **3**	*Starting year:* **2010**

IDENTIFICATION OF NOVEL STRATEGIES TO CORRECT THE CHLORIDE TRANSPORT DEFECT IN CYSTIC FIBROSIS

IDENTIFICAZIONE DI NUOVE STRATEGIE PER LA CORREZIONE DEL DIFETTO DI BASE NELLA FIBROSI CISTICA

Pedemonte Nicoletta, Caci Emanuela, Sondo Elvira, Ferrera Loretta, Tomati Valeria, Scudieri Paolo, Zegarra-Moran Olga, Ravazzolo Roberto, Galietta Luis J.V.

U.O.C. Genetica Medica, Istituto Giannina Gaslini, via Gerolamo Gaslini 5, 16147 Genova; Tel.: 010-56362801; Fax: 010-3779797; E-mail: galietta@unige.it

ABSTRACT N. 222

Telethon Research Projects - Other Genetic Diseases		
Principal Investigator	**MAIURI LUIGI**	
Telethon grant N.	**GGP12128**	
Total budget €	**293.700**	
Centres: **3**	*Duration (yrs):* **2**	*Starting year:* **2012**
Partners	VALERIA RAIA, MARIA CHIARA MAIURI	

THE HEME-OXYGENASE 1 (HO-1) AS MODULATOR OF CYSTIC FIBROSIS LUNG DISEASE

L'EME-OSSIGENASI 1(HO-1) COME MODULATORE DELLA PATOLOGIA POLMONARE ASSOCIATA ALLA FIBROSI CISTICA

Raia Valeria (3), Maiuri Maria Chiara (4), Maiuri Luigi (1,2)

(1) European Institute for Research in Cystic Fibrosis (IERFC), Division of Genetics and Cell Biology, San Raffaele Scientific Institute, Milan - Via Olgettina,60, 20132, Milan. Tel. +39 0226434301; Fax +39 0226434328; maiuri.luigi@hsr.it
(2) Department of Health Sciences, University of Piemonte Orientale, Novara
(3) Department of Translational Medicine, University of Naples Federico II, Naples
(4) Department of Pharmacy, University of Naples Federico II, Naples

ABSTRACT N. 223

Telethon Research Projects - Other Genetic Diseases		
Principal Investigator	**STRAZZABOSCO MARIO**	
Telethon grant N.	**GGP12133**	
Total budget €	**152.200**	
Centres: **1**	*Duration (yrs):* **3**	*Starting year:* **2013**

CFTR-DEFICIENCY CAUSES A DYSREGULATION IN TOLL-LIKE RECEPTOR-MEDIATED INNATE IMMUNE RESPONSES: PATHOGENETIC AND THERAPEUTIC IMPLICATIONS FOR CYSTIC FIBROSIS-RELATED LIVER DISEASE

IL DIFETTO DI FUNZIONE DEL CFTR ALTERA LE RISPOSTE DI IMMUNITÀ INNATA MEDIATE DA RECETTORI TOLL-LIKE: IMPLICAZIONI PATOGENETICHE E TERAPEUTICHE PER LA MALATTIA EPATICA ASSOCIATA A FIBROSI CISTICA

Fiorotto Romina (2), Scirpo Roberto (1), Villani Ambra (2), Strazzabosco Mario (1,2)

(1) Dipartimento di Chirurgia e Medicina Traslazione, Università di Milano-Bicocca, Monza, Italia - Lab 1.04, Edificio U8, via Cadore 48, 20900 Monza (MB), Tel. 0264488052, Fax: 0264488250, email: mario.strazzabosco@unimib.it
(2) Department of Internal Medicine, Yale University, New Haven, USA

ABSTRACT N. 224

Telethon Research Projects - Other Genetic Diseases		
Principal Investigator	**MILANI SILVANO**	
Telethon grant N.	**GGP12258**	
Total budget €	**66.000**	
Centres: **1**	*Duration (yrs):* **2**	*Starting year:* **2012**

IMPROVING THE DIAGNOSTIC ACCURACY OF A NEONATAL SCREENING PROTOCOL FOR CYSTIC FIBROSIS: FACTORS AFFECTING THE DISTRIBUTION OF IMMUNOREACTIVE TRYPSINOGEN (IRT) BLOOD LEVELS

MIGLIORARE L'ACCURATEZZA DIAGNOSTICA DI UN PROTOCOLLO DI SCREENING NEONATALE PER LA FIBROSI CISTICA: FATTORI CHE HANNO EFFETTO SULLA DISTRIBUZIONE DEI LIVELLI EMATICI DEL TRIPSINOGENO IMMUNOREATTIVO (IRT)

Spada Elena, Milani Silvano

Department of Clinical Sciences and Community Health - Unit of Medical Statistics and Biometry - Università degli Studi di Milano - via Venezian 1 20133 Milano, tel 02 50320855, fax 02 50320866 email silvano.milani@unimi.it

ABSTRACT N. 225

Telethon Research Projects - Other Genetic Diseases		
Principal Investigator	**CAMASCHELLA CLARA**	
Telethon grant N.	**GGP12025**	
Total budget €	**349.800**	
Centres: **1**	*Duration (yrs):* **3**	*Starting year:* **2012**

HEMOCHROMATOSIS: FROM GENES TO CLINICS AND BACK

EMOCROMATOSI: DAI GENI ALLA CLINICA E RITORNO

Nai Antonella (1,2), Pagani Alessia (1,2), Lidonnici Maria Rosa (3), Rausa Marco (1,2), Mandelli Giacomo (3), Ferrari Giuliana (3), Silvestri Laura (1,2), Camaschella Clara (1,2)

(1) Vita Salute San Raffaele University - Via Olgettina, 58, 20132 Milano, Italy - phone +390226437782,Fax +39-0226432640 email camaschella.clara@hsr.it
(2) Division of Genetics and Cell Biology, IRCCS San Raffaele Scientific Institute, Milan, Italy
(3) San Raffaele Telethon Institute for Gene Therapy (HSR-TIGET), Division of Regenerative Medicine, Gene Therapy and Stem Cells, IRCCS San Raffaele Scientific Institute, Milan, Italy.

ABSTRACT N. 226

Telethon Research Projects - Other Genetic Diseases		
Principal Investigator	**PIETRANGELO ANTONELLO**	
Telethon grant N.	**GGP14285**	
Total budget €	**328.900**	
Centres: **1**	*Duration (yrs):* **3**	*Starting year:* **2014**

THE METABOLIC ABNORMALITY OF HEREDITARY HEMOCHROMATOSIS: MECHANISMS AND CONSEQUENCES OF HEPCIDIN DEFICIENCY ON GLUCOSE HOMEOSTASIS AND INSULIN SIGNALING

ANOMALIE E ADATTAMENTO METABOLICO NELL'EMOCROMATOSI EREDITARIA: MECCANISMI E CONSEGUENZE DELLA CARENZA DI EPCIDINA

Vecchi Chiara (2), Montosi Giuliana (2), Garuti Cinzia (2), Corradini Elena (2), Sabelli Manuela (2), Pietrangelo Antonello (1)

(1) Università di Modena e Reggio Emilia, Via del Pozzo 71, 41100 Modena, Italy; tel: +39-059-4222714; fax +39-059-4223041; email: antonello.pietrangelo@unimore.it
(2) Università di Modena e Reggio Emilia, Via del Pozzo 71, 41100 Modena, Italy

ABSTRACT N. 227

Telethon Research Projects - Other Genetic Diseases		
Principal Investigator	**NORATA GIUSEPPE DANILO**	
Telethon grant N.	**GGP13002**	
Total budget €	**225.100**	
Centres: **2**	*Duration (yrs):* **3**	*Starting year:* **2013**
Partners	MASSIMO LOCATI	

ENGINEERED T REGULATORY CELLS TO CONTROL THE IMMUNE-INFLAMMATORY RESPONSE AND THE ACCELERATED ONSET OF ATHEROSCLEROSIS IN FAMILIAL HYPERCHOLESTEROLEMIA

UTILIZZO DI LINFOCITI T INGEGNERIZZATI NELL'IPERCOLESTEROLEMIA FAMILIARE

Bonacina Fabrizia (1), Kallikourdis Marinos (2), Garetto Stefano (2), Baragetti Andrea (1), Catapano Alberico (1), Locati Massimo (2), Norata Giuseppe Danilo (1)

(1) Dipartimento di Scienze Farmacologiche e Biomolecolari, Università degli Studi di Milano, Via Balzaretti 9, 20133 Milano, Tel 02 50318313, Fax 02 50318386, Danilo.Norata@unimi.it
(2) Fondazione Humanitas per la Ricerca, Rozzano

ABSTRACT N. 228

Telethon Research Projects - Other Genetic Diseases		
Principal Investigator	**VOLTATTORNI CARLA**	
Telethon grant N.	**GGP10092**	
Total budget €	**213.200**	
Centres: **2**	*Duration (yrs):* **3**	*Starting year:* **2010**
Partners	ANTONIO AMOROSO	

DEVELOPMENT OF NEW STRATEGIES FOR THE TREATMENT OF PRIMARY HYPEROXALURIA TYPE I

SVILUPPO DI NUOVE STRATEGIE PER IL TRATTAMENTO DELL'IPEROSSALURIA PRIMARIA DI TIPO I

Cellini Barbara (2), Montioli Riccardo (2), Oppici Elisa (2), Dindo Mirco (2), Mandrile Giorgia (3), De Marchi Mario (3), Amoroso Antonio (4), Voltattorni Carla (1)

(1) Department of Life Sciences and Reproduction, Section of Biological Sciences, University of Verona, Strada Le Grazie 8, 37134 Verona, Italy, Tel. 0458027175, e-mail: carla.borrivoltattorni@univr.it

(2) Department of Life Sciences and Reproduction, Section of Biological Sciences, University of Verona, Verona, Italy
(3) Department of Clinical and Biological Sciences, University of Torino, Torino, Italy
(4) Department of Medical Sciences, University of Torino, Torino, Italy

ABSTRACT N. 229

TIGEM - Other Genetic Diseases		
Principal Investigator	**BRUNETTI PIERRI NICOLA**	
Telethon grant N.	**TGM11MT3**	
Total budget €	**410.000**	
Centres: **1**	*Duration (yrs):* **5**	*Starting year:* **2011**

METABOLIC DIVERSION TOWARDS NON-TOXIC METABOLITES FOR THERAPY OF PRIMARY HYPEROXALURIA TYPE 1

ATTIVAZIONE DI PATHWAYS ALTERNATIVI PER LA PRODUZIONE DI METABOLITI NON-TOSSICI PER LA TERAPIA DELL'IPEROSSALURIA PRIMARIA DI TIPO 1

Castello Raffaele (1), Borzone Roberta (1), Annunziata Patrizia (1), Piccolo Pasquale (1), Brunetti-Pierri Nicola (1,2)

(1) Telethon Institute of Genetics and Medicine (TIGEM), Via Campi Flegrei 34, 80078 Pozzuoli (Napoli), ITALY. Phone +39 081 6132361; fax. +39 081 5609877; e-mail: brunetti@tigem.it
(2) Department of Translational Medicine, Federico II University, Naples, Italy

ABSTRACT N. 230

TIGEM - Other Genetic Diseases		
Principal Investigator	**DI BERNARDO DIEGO**	
Telethon grant N.	**TGM11SB1**	
Total budget €	**500.000**	
Centres: **1**	*Duration (yrs):* **5**	*Starting year:* **2011**

SYSTEMS BIOLOGY OF GENETIC DISEASES: ELUCIDATING GENE FUNCTION AND DRUG MODE OF ACTION

LA BIOLOGIA COMPUTAZIONALE PER LO STUDIO DELLE MALATTIE GENETICHE: IDENTIFICAZIONE DELLA FUNZIONE GENICA E DEL MECCANISMO DI AZIONE DEI FARMACI

Pagliarini Roberto (1), Napolitano Francesco (1), Sirci Francesco (1), Gambardella Gennaro (1,5), Lai Ching-Hung (1), Pinelli Michele (1), Mutarelli Margherita (1), Carissimo Annamaria (1), Carrella Diego (1), Cutillo Luisa (1), Moretti Nicoletta (1), Ponzin Diego (4), Ferrari Stefano (4), Banfi Sandro (1,3), Brunetti-Pierri Nicola (1,2), di Bernardo Diego (1,2)

(1) TIGEM, Via dei Campi Flegrei 34, Pozzuoli (Napoli), Tel: 081 6132319, Fax 081 6132 351, email: dibernardo@tigem.it
(2) Università degli Studi di Napoli, Federico II
(3) Seconda Universita di Napoli
(4) International Center for Ocular Physiopathology, The Veneto Eye Bank Foundation, Venice
(5) Present Address: King's College, London, UK

ABSTRACT N. 231

Telethon Research Projects - Other Genetic Diseases		
Principal Investigator	**CALABRESI LAURA**	
Telethon grant N.	**GGP14125**	
Total budget €	**158.900**	
Centres: **1**	*Duration (yrs):* **2**	*Starting year:* **2014**

RENAL DISEASE IN GENETIC LCAT DEFICIENCY: FROM PATHOGENESIS TO THERAPY

MALATTIA RENALE NEL DEFICIT DI LCAT: DALLA PATOGENESI ALLA TERAPIA

Ossoli Alice (1), Lucca Fabio (1), Viganò Valentina (1), Remaley Alan (2), Calabresi Laura (1)

(1) Dipartimento di Scienze Farmacologiche e Biomolecolari, via Balzaretti 9, 20133 Milano, email: laura.calabresi@unimi.it tel 02 50319906 fax 02 50319900
(2) Lipoprotein Metabolism Laboratory, NIH, Bethesda, United States

ABSTRACT N. 232

Telethon Research Projects - Other Genetic Diseases		
Principal Investigator	**SITIA ROBERTO**	
Telethon grant N.	**GGP11077**	
Total budget €	**343.200**	
Centres: **1**	*Duration (yrs):* **3**	*Starting year:* **2011**

PROTEOSTASIS IN THE EARLY SECRETORY COMPARTMENT AS A PATHOGENETIC MECHANISM AND THERAPEUTIC TARGET

PATOGENESI DELLE MALATTIE DA ALTERATO TRASPORTO E ACCUMULO PROTEICO NELLA VIA SECRETORIA

Anelli Tiziana (1), Cortini Margherita (1), Fagioli Claudio (1), Fra Annamaria (2), Lougaris Vassilis (3), Sannino Sara (1), Vavassori Stefano (1), Sitia Roberto (1)

(1) Università Vita-Salute San Raffaele Scientific Institute, Via Olgettina 58, 20132 Milano +39 02 2643 4722; r.sitia@hsr.it
(2) Università di Brescia
(3) Spedali Civili di Brescia

ABSTRACT N. 233

Telethon Research Projects - Other Genetic Diseases		
Principal Investigator	**BONATTI STEFANO**	
Telethon grant N.	**GGP14002**	
Total budget €	**309.100**	
Centres: **1**	*Duration (yrs):* **3**	*Starting year:* **2014**

DEVELOPING PEPTIDES PHARMACOLOGICALLY ACTIVE FOR THE MOST FREQUENT WILSON DISEASE-CAUSING ATP7B MUTANT

SVILUPPO DI PEPTIDI FARMACOLOGICAMENTE ATTIVI CONTRO LA PIU' FREQUENTE FORMA DELLA MALATTIA DI WILSON

Allocca Simona (1), Ciano Michela (1), Caporaso Gabriella (1), Stornaiuolo Mariano (2), Mallardo Massimo (1), Polishchuk Roman (3), Bonatti Stefano (1)

(1) Dipartimento Medicina Molecolare e Biotecnologie Mediche, Università di Napoli Federico II, via S. Pansini 5, 80131 Napoli, +39-081-7463200, fax +39081-7463205, bonatti@unina.it
(2) Dipartimento di Farmacia, Università di Napoli Federico II
(3) Tigem, Napoli

ABSTRACT N. 234

TIGEM - Other Genetic Diseases		
Principal Investigator	**POLISHCHUK ROMAN**	
Telethon grant N.	**TGM11CB4**	
Total budget €	**350.000**	
Centres: **1**	*Duration (yrs):* **5**	*Starting year:* **2011**

PATHOGENESIS OF WILSON DISEASE: MOLECULAR MECHANISMS

OF ATP7B TRAFFICKING IN THE MAINTENANCE OF COPPER HOMEOSTASIS

PATOGENESI DELLA MALATTIA DI WILSON: I MECCANISMI MOLECOLARI DELLA VEICOLAZIONE DI ATP7B NEL MANTENIMENTO DELL'OMEOSTASI DI RAME

Polishchuk Elena, Chesi Giancarlo, Concilli Mafalda, Iacobacci Simona, Polishchuk Roman

Telethon Institute of Genetics and Medicine (TIGEM), Via Campi Flegrei, 34, 80078, Pozzuoli (NA), Italy, phone: +39 081 6132336, fax: +39 081 5609877, e-mail polish@tigem.it

ABSTRACT N. 235

Telethon Research Projects - Other Genetic Diseases		
Principal Investigator	**HIRSCH EMILIO**	
Telethon grant N.	**GGP14106**	
Total budget €	**327.800**	
Centres: **1**	*Duration (yrs):* **3**	*Starting year:* **2014**

IMPROVING MC4R SIGNALING VIA ISOFORM SELECTIVE PI3K TARGETING TO FIGHT MELANOCORTIN OBESITY SYNDROME

COMBATTERE L'OBESITÀ GENETICA INDOTTA DA MUTAZIONI DI MC4R ATTRAVERSO LA MODULAZIONE DELLA SEGNALAZIONE RECETTORIALE

Alessia Perino (1,3), Martina Beretta (1,2), Alessandra Ghigo (1), Hirsch Emilio (1)

(1) Molecular Biotechnology Center, Department of Molecular Biotechnology and Health Sciences, University of Turin, Turin - Via Nizza 52, 10126 Torino, Italy, Tel +39 011-6706425, email: emilio.hirsch@unito.it
(2) Department of Anesthesiology and Intensive Care Medicine, Jena University Hospital, Jena, Germany
(3) Metabolic Signaling, Institute of Bioengineering, School of Life Sciences, Ecole Polytechnique Fédérale de Lausanne, Lausanne, Switzerland

ABSTRACT N. 236

Telethon Research Projects - Other Genetic Diseases		
Principal Investigator	**SVELTO MARIA**	
Telethon grant N.	**GGP12040**	
Total budget €	**114.400**	
Centres: **1**	*Duration (yrs):* **2**	*Starting year:* **2012**

STATINS AS POTENTIAL THERAPEUTIC AGENTS FOR HANDLING NEPHROGENIC DIABETES INSIPIDUS

STATINE COME POTENZIALI STRUMENTI TERAPEUTICI PER IL TRATTAMENTO DEL DIABETE INSIPIDO NEFROGENICO

Procino Giuseppe (1), Milano Serena (1), Carmosino Monica (1), Gullo Damiano (3), Portincasa Piero (2), Svelto Maria (1)

(1) Dipartimento di Bioscienze, Biotecnologie e Biofarmaceutica, Università degli Studi di Bari, Via Orabona 4, 70126, Bari, Tel 080-5443328, Fax: 080-5443388, e-mail: maria.svelto@uniba.it
(2) Dipartimento di Scienze Biomediche ed Oncologia Umana, Università degli studi di Bari
(3) Unità di Endocrinologia, Ospedale Garibaldi-Nesima, Policlinico di Catania

ABSTRACT N. 237

Telethon Research Projects - Other Genetic Diseases		
Principal Investigator	**TARONE GUIDO**	
Telethon grant N.	**GGP12047**	
Total budget €	**182.600**	
Centres: **1**	*Duration (yrs):* **2**	*Starting year:* **2012**

MELUSIN GENE THERAPY: A POSSIBLE APPROACH TO PREVENT CARDIOMYOPATHY

MELUSINA COME AGENTE DI TERAPIA GENICA: UN NUOVO APPROCCIO PER CONTRASTARE LE CARDIOMIOPATIE

Moiso Enrico, Rubinetto Cristina, Cimino James, Brancaccio Mara, Tarone Guido

Dipartimento di Biotecnologie Molecolari e Scienze per la Salute, Università di Torino, Via Nizza 52, 10126, Torino, Tel: +390116706433, Fax: +390116706432, Mail: guido.tarone@unito.it

ABSTRACT N. 238

Telethon Research Projects - Other Genetic Diseases		
Principal Investigator	**BANG MARIE-LOUISE**	
Telethon grant N.	**GGP12282**	
Total budget €	**428.600**	
Centres: **3**	*Duration (yrs):* **3**	*Starting year:* **2012**
Partners	VINCENZO NIGRO, MARCO LINARI	

MYOPALLADIN IN DILATED CARDIOMYOPATHY AND LIMB GIRDLE MUSCULAR DYSTROPHY

LA MIOPALLADINA NELLA CARDIOMOPATIA DILATATIVA E NELLA DISTROFIA MUSCOLARE DEI CINGOLI

Filomena Maria Carmela (1,2), Yamamoto Daniel Ludwig (3,5), Caremani Marco (7), Mastrototaro Giuseppina (1,4,6), Marmonti Enrica (1), Chen Ju (8), Nigro Vincenzo (9), Linari Marco (7), Marie-Louise Bang (1,4)

(1) Humanitas Research Hospital, Via Manzoni 113, 20089 Rozzano, Milan, Tel: 0282245210; Fax: 0282245290; email: marie-louise.bang@cnr.it
(2) University of Milan, Italy
(3) Institute of Biomedical Technologies (ITB) - Nazional Research Countil (ITB-CNR), Milan, Italy
(4) Institute of Genetic and Biomedical Research, Milan Unit - National Research Council (IRGB-CNR), Milan, Italy
(5) Multimedica Hospital, Scientific and Technology Pole, Milan, Italy
(6) University of Milano-Bicocca, Italy
(7) Department of Biology, University of Florence, Italy
(8) Department of Medicine, University of California San Diego, USA
(9) Telethon Institute of Genetics and Medicine (TIGEM), Napoli, Italy
(10) Department of General Pathology, Second University of Naples, Italy

ABSTRACT N. 239

Telethon Research Projects - Other Genetic Diseases		
Principal Investigator	**POGGESI CORRADO**	
Telethon grant N.	**GGP13162**	
Total budget €	**128.400**	
Centres: **2**	*Duration (yrs):* **2**	*Starting year:* **2013**
Partners	LEONARDO SACCONI	

HYPERTROPHIC CARDIOMYOPATHY CAUSED BY MUTATIONS IN THE THIN FILAMENT REGULATORY PROTEINS OF THE SARCOMERE

CARATTERISTICHE CLINICHE E BIOFISICHE DELLA CARDIOMIOPATIA IPERTROFICA ASSOCIATA A UN DIFETTO GENETICO DELLE PROTEINE REGOLATORIE DEL FILAMENTO SOTTILE

Ferrantini Cecilia (1), Coppini Raffaele (1), Crocini Claudia (2), Tosi Benedetta (1), Mazzoni Luca (1), Pioner J Manuel (1), Scardigli Marina (2), Scellini Beatrice (1), Piroddi Nicoletta (1), Tesi Chiara (1), Olivotto Iacopo (1), Cerbai Elisabetta (1), Sacconi Leonardo (2), Poggesi Corrado (1)

(1) Centro Interuniversitario di Medicina Molecolare e Biofisica Applicata (CIMMBA), Università di Firenze - Dipartimento di Medicina Sperimentale e Clinica e CIMMBA, Università di Firenze, Viale Morgagni 63, 50137 Firenze. Tel 0552758035 Cel 3404693508, Fax 0554379506, mail corrado.poggesi@unifi.it
(2) Istituto Nazionale di Ottica (INO) e Laboratorio Europeo di Spettroscopia Non Lineare (LENS), Firenze

ABSTRACT N. 240

Telethon Research Projects - Other Genetic Diseases		
Principal Investigator	**PIERONI MAURIZIO**	
Telethon grant N.	**GGP10186**	
Total budget €	**336.000**	
Centres: **2**	*Duration (yrs):* **3**	*Starting year:* **2010**
Partners	ANTONIO OLIVA	

IDENTIFICATION OF GENETIC, ELECTROANATOMICAL AND STRUCTURAL PREDICTORS OF MALIGNANT VENTRICULAR ARRHYTHMIAS IN PATIENTS WITH BRUGADA SYNDROME

IDENTIFICAZIONE DEI PREDITTORI GENETICI, ELETTROANATOMICI E STRUTTURALI DI ARITMIE VENTRICOLARI MALIGNE IN PAZIENTI CON SINDROME DI BRUGADA

Pieroni Maurizio (1), Oliva Antonio (2), Notarstefano Pasquale (1), Partemi Sara (2), Camporeale Antonia (3), Guida Raffaele (1), Rio Teresa (1,3), Grotti Simone (1), Brugada Ramon (4), Bolognese Leonardo (1)

(1) Dipartimento Cardiovascolare e Neurologico, Ospedale San Donato, Arezzo, Italia, - Via Pietro Nenni 22, 52100 Arezzo, 0575/255524, mauriziopieroni@yahoo.com
(2) Istituto di Medicina Legale, Università Cattolica del Sacro Cuore, Roma, Italia
(3) Dipartimento Cardiovascolare, Università Cattolica del Sacro Cuore, Roma, Italia
(4) Cardiovascular Genetics Centre, IDIBGI-University of Girona

ABSTRACT N. 241

Telethon Research Projects - Other Genetic Diseases		
Principal Investigator	**PRIORI SILVIA GIULIANA**	
Telethon grant N.	**GGP11141**	
Total budget €	**465.600**	
Centres: **3**	*Duration (yrs):* **3**	*Starting year:* **2011**
Partners	POMPEO VOLPE, FELICIANO PROTASI	

MUTATIONS OF CARDIAC CALSEQUESTRIN AND CARDIAC ARRHYTHMIAS: NOVEL INSIGHTS ON PATHOGENESIS AND THERAPY

MUTAZIONI DEL GENE CASQ2 (CALSEQUESTRINA) ED ARITMIE CARDIACHE: APPROCCIO SPERIMENTALE ALLA PATOGENESI E TERAPIA

Bongianino Rossana (1), Lodola Francesco (1), Denegri Marco (1), Boncompagni Simona (2), Valle Giorgia (3), Persampieri Simone (1), Avelino-Cruz Josè Everardo (8), Pietrangelo Laura (2), Liu Nian (1), Auricchio Alberto (4,5), Protasi Feliciano (2), Volpe Pompeo (3), Napolitano Carlo (1), Priori Silvia Giuliana (1,6,7)

(1) Molecular Cardiology, IRCCS Fondazione Salvatore Maugeri, Pavia, Italy, Via Salvatore Maugeri 10, 27100, Pavia, Italy; Tel: 0382592040; Fax: 0382592059; email: silvia.priori@fsm.it
(2) CeSi, Center for Research on Ageing & DNA, Department of Neuroscience and Imaging, G. d'Annunzio University, Chieti, Italy.
(3) Department of Biomedical Sciences, University of Padova, Padova, Italy
(4) Telethon Institute of Genetics and Medicine (TIGEM), Naples, Italy
(5) Department of Pediatrics, "Federico II" University, Naples, Italy
(6) Department of Molecular Medicine , University of Pavia, Pavia, Italy
(7) Cardiovascular Genetic Program, The Leon H. Charney Division of Cardiology, New York University School of Medicine, New York, USA
(8) Laboratorio de Cardiología Molecular, Insituto de Fisiología, Benemérita Universidad Autónoma de Puebla, Puebla, México

ABSTRACT N. 242

Telethon Research Projects - Other Genetic Diseases		
Principal Investigator	**MONGILLO MARCO**	
Telethon grant N.	**GGP11224**	
Total budget €	**215.600**	
Centres: **1**	*Duration (yrs):* **3**	*Starting year:* **2011**

NOVEL OPTOGENETIC APPROACH TO INVESTIGATE ARRHYTHMOGENESIS IN DOMINANT CATECHOLAMINERGIC POLYMORPHIC VENTRICULAR TACHYCARDIA (CPVT)

NUOVO METODO PER LO STUDIO DELLE ARITMIE CATECOLAMINERGICHE FAMILIARI BASATO SULL'USO DI PROTEINE FOTOATTIVATE

Zaglia Tania (1,2), Pianca Nicola (1,2), Borile Giulia (1,2), Da Broi Francesca (1,2), Luther Stefan (5,6), Campione Marina (2,4), Corrado Domenico (3), Miquerol Lucile (7), Mongillo Marco (1,2,4)

(1) Istituto Veneto di Medicina Molecolare - via Orus 2, Padova, 049 7923229, fax 049 7923250, marco.mongillo@unipd.it
(2) dipartimento di scienze biomediche, Universita di Padova, via U. Bassi, Padova
(3) dipartimento di scienze cardio-toraco-vascolari, Università di Padova, via Giustiniani 2, Padova
(4) CNR Institute of Neuroscience, Via Ugo Bassi 58, 35121 Padova, Italy.
(5) Biomedical Physics, Max Planck Institute for Dynamics and Self-Organization, Göttingen, Germany
(6) Heart Research Center Göttingen, Göttingen, Germany
(7) Developmental Biology Institute of Marseille, CNRS UMR 7288, Aix Marseille University, Marseille, France

ABSTRACT N. 243

Telethon Research Projects - Other Genetic Diseases		
Principal Investigator	**COLOMBI MARINA**	
Telethon grant N.	**GGP13167**	
Total budget €	**91.300**	
Centres: **1**	*Duration (yrs):* **2**	*Starting year:* **2013**

ARTERIAL TORTUOSITY SYNDROME: A VITAMIN C COMPARTMENTATION DISEASE?

SINDROME DELLE ARTERIE TORTUOSE: UN PROBLEMA NEL METABOLISMO DELLA VITAMINA C?

Zoppi Nicoletta, Chiarelli Nicola, Cinquina Valeria, Ritelli Marco, Colombi Marina

Division of Biology and Genetics, Department of Molecular and Translational Medicine, School of Medicine, University of Brescia, Viale Europa 11, 25123 Brescia, Italy, Phone/Fax 0303717265, marina.colombi@unibs.it

ABSTRACT N. 244

Telethon Research Projects - Other Genetic Diseases		
Principal Investigator	**CESARENI GIANNI**	
Telethon grant N.	**GGP09243**	
Total budget €	**254.600**	
Centres: **1**	*Duration (yrs):* **3**	*Starting year:* **2009**

A NETWORK ANALYSIS STRATEGY TO IDENTIFY GENES IMPLICATED IN NOONAN AND LEOPARD SYNDROMES

ANALISI DI RETI DI INTERAZIONE GENICA PER L'IDENTIFICAZIONE DI GENI IMPLICATI NELLE SINDROMI DI NOONAN E DI LEOPARD

Corallino Salvatore (1), Iwai Leo K. (2), Huang Paul H. (2), Sacco Francesca (1), Casatgnoli Luisa (1), Cesareni Gianni (1,3)

(1) Department of Biology, University of Rome Tor Vergata, Via della ricerca scientifica, 00133 Rome, Italy
(2) Protein Networks Team, Division of Cancer Biology, Institute of Cancer Research, London SW3 6JB, United Kingdom
(3) IRCCS Fondazione Santa Lucia, 00143, Rome, Italy

ABSTRACT N. 245

Telethon Research Projects - Other Genetic Diseases		
Principal Investigator	**TARTAGLIA MARCO**	
Telethon grant N.	**GGP13107**	
Total budget €	**225.400**	
Centres: **1**	*Duration (yrs):* **2**	*Starting year:* **2013**

MOLECULAR BASES OF NOONAN SYNDROME AND RELATED DISORDERS

BASI MOLECOLARI DELLA SINDROME DI NOONAN E DI MALATTIE GENETICHE CORRELATE

Flex Elisabetta (1), Martinelli Simone (1), Pantaleoni Francesca (1), Motta Maria Letizia (1), Stellacci Emilia (1), Cordeddu Viviana (1), Ciolfi Andrea (1), Bruselles Alessandro (1), Pannone Luca (1), Paolacci Stefano (1), Caputo Viviana (2), Dhandapany Perundurai (3), Zampino Giuseppe (4), Digilio Maria Cristina (5), Melis Daniela (6), Selicorni Angelo (7), Mazzanti Laura (8), Rossi Cesare (9), Lepri Francesca (5), De Luca Alessandro (10), Ferrero Giovanni B (11), Clementi Maurizio (12), Zenker Martin (13), Dallapiccola Bruno (5), Gelb Bruce D (3), Cave' Helene (14), Ahmadian M Reza (15), Tartaglia Marco (1)

(1) Istituto Superiore di Sanità, Rome - Dipartimento di Ematologia, Oncologia e Medicina Molecolare, Istituto Superiore di Sanità, Viale Regina Elena, 299 - 00161 Roma. Tel: 0649902569, Fax: 0649902850, Email: mtartaglia@iss.it
(2) Università "La Sapienza", Rome
(3) Mount Sinai School of Medicine, New York
(4) Università Cattolica del Sacro Cuore, Rome
(5) Ospedale Bambino Gesù, Rome
(6) Università "Federico II", Naples
(7) Università di Milano Bicocca, Monza
(8) Policlinico S. Orsola-Malpighi, Bologna
(9) Università di Bologna, Bologna
(10) Istituto Mendel-IRCCS ''Casa Sollievo della Sofferenza'', Rome
(11) Università di Torino, Turin
(12) Università di Padova, Padua
(13) Institute of Human Genetics, University Hospital, Magdeburg
(14) Hopital Robert Debre', Paris
(15) Heinrich-Heine University Medical Center, Düsseldorf

ABSTRACT N. 246

Telethon Research Projects - Other Genetic Diseases		
Principal Investigator	**POLO SIMONA LAURA ANNA**	
Telethon grant N.	**GEP13069**	
Total budget €	**49.950**	
Centres: **1**	*Duration (yrs):* **1**	*Starting year:* **2014**

EVALUATING THE ROLE OF UBIQUITIN AND ENDOCYTOSIS IN GITELMAN SYNDROME

STUDIO DEI MECCANISMI MOLECOLARI ALLA BASE DELLA SINDROME DI GITELMAN

Fajner Valentina (1), Maspero Elena (1), Polo Simona (1,2)

(1) IFOM, Fondazione Istituto FIRC di Oncologia Molecolare, Via Adamello 16, 20139, Milan, Italy tel +39 02 574303242, fax +39 02 574303231, simona.polo@ifom.eu
(2) Dipartimento di Medicina, Chirurgia ed Odontoiatria, Università degli Studi di Milano, Via di Rudinì 8, 20122 Milan, Italy

ABSTRACT N. 247

Telethon Research Projects - Other Genetic Diseases		
Principal Investigator	**COTECCHIA SUSANNA**	
Telethon grant N.	**GGP13227**	
Total budget €	**302.300**	
Centres: **2**	*Duration (yrs):* **3**	*Starting year:* **2013**
Partners	FRANCESCA FANELLI	

GAIN-OF-FUNCTION MUTATIONS OF THE V2 VASOPRESSIN RECEPTOR IN NEPHROGENIC SYNDROME OF INAPPROPRIATE ANTIDIURESIS (NSIAD): MOLECULAR CHARACTERIZATION AND IN SILICO IDENTIFICATION OF POTENTIAL THERAPEUTIC AGENTS

MUTAZIONI ATTIVANTI DEL RECETTORE V2 DELLA VASOPRESSINA NELLA SINDROME NEFROGENICA DA ANTIDIURESI NON APPROPRIATA (NSIAD): CARATTERIZZAZIONE MOLECOLARE ED IDENTIFICAZIONE IN SILICO DI POTENZIALI AGENTI TERAPEUTICI

Cotecchia Susanna (1), Fanelli Francesca (2), Costa Tommaso (3), Vezzi Vanessa (3), Valenti Giovanna (1), Tamma Grazia (1), Ranieri Marianna (1)

(1) Dipartimento di Bioscienze, Biotecnologie e Biofarmaceutica, Università degli Studi di Bari "Aldo Moro", Via Orabona 4, 70126 Bari, tel +39-080-5543333, susanna.cotecchia@uniba.it
(2) Dipartimento di Scienze della Vita, Università di Modena e Reggio Emilia, Modena
(3) Istituto Superiore di Sanità, Roma

ABSTRACT N. 248

Telethon Research Projects - Other Genetic Diseases		
Principal Investigator	**BRESIN ELENA**	
Telethon grant N.	**GGP11201**	
Total budget €	**271.500**	
Centres: **1**	*Duration (yrs):* **3**	*Starting year:* **2011**

GENETIC BASIS OF STEROID RESISTANT NEPHROTIC SYNDROME AND IMPLICATIONS FOR THERAPY

BASI GENETICHE DELLA SINDROME NEFROSICA STEROIDO-RESISTENTE E IMPLICAZIONI PER LA TERAPIA

Bresin Elena, Mele Caterina, Paraskevas Iatropoulos, Maranta Ra-

mona, Breno Matteo, Alberti Marta, Cassis Paola, Remuzzi Giuseppe, Noris Marina

IRCCS - Istituto di Ricerche Farmacologiche “Mario Negri”, Clinical Research Center for Rare Diseases “Aldo e Cele Daccò”, Via Camozzi 3, 24020, Ranica, Bergamo, Italy tel 0039 035 4535327 fax 0039 035 4535377, e-mail elena.bresin@marionegri.it

ABSTRACT N. 249

Telethon Research Projects - Other Genetic Diseases		
Principal Investigator	**PUSCH MICHAEL**	
Telethon grant N.	**GGP12008**	
Total budget €	**226.700**	
Centres: **1**	*Duration (yrs):* **3**	*Starting year:* **2012**

MOLECULAR MECHANISMS OF TRANSPORT, SMALL LIGAND MODULATION, AND SUBUNIT INTERACTION OF CHLORIDE TRANSPORTING CLC PROTEINS INVOLVED IN HUMAN GENETIC DISEASES

MECCANISMI MOLECOLARI RIGUARDANTI IL TRASPORTO, LA REGOLAZIONE DA PARTE DI PICCOLI LIGANDI E L’INTERAZIONE CON ALTRE SUBUNITÀ DELLE PROTEINE CLC COINVOLTE IN MALATTIE GENETICHE UMANE

Gradogna Antonella, Fiore Michele, Zanardi Ilaria, Zifarelli Giovanni, Pusch Michael

Istituto di Biofisica, CNR - Via De Marini 6, 16149 Genova, Tel. 0106475330, Fax 0106475500, pusch@ge.ibf.cnr.it

ABSTRACT N. 250

TIGEM - Other Genetic Diseases		
Principal Investigator	**DE MATTEIS MARIA ANTONIETTA**	
Telethon grant N.	**TGM11CB1**	
Total budget €	**300.000**	
Centres: **1**	*Duration (yrs):* **5**	*Starting year:* **2011**

DISSECTING THE MOLECULAR MECHANISMS UNDERLYING ENDOCYTIC DYSFUNCTIONS INDUCED BY MUTATIONS IN OCRL AND CLC5 TO IDENTIFY CORRECTORS FOR LOWE SYNDROME AND DENT DISEASE

DEFINIZIONE DEI MECCANISMI MOLECOLARI ALLA BASE DELLE DISFUNZIONI DELLA VIA ENDOCITICA INDOTTA DA MUTAZIONI DEI GENI OCRL E CLC5 ALLO SCOPO DI IDENTIFICARE CORRETTORI FARMACOLOGICI PER LA SINDROME DI LOWE E PER LA MALATTIA DI DENT

De Leo Maria Giovanna, Staiano Leopoldo, Polishchuk Elena, Di Tullio Giuseppe, Medina Diego, Ballabio Andrea, De Matteis Maria Antonietta

Telethon Institute of Genetics and Medicine (TIGEM), Via Campi Flegrei 34, 80078 Pozzuoli (Napoli), ITALY. Phone +39 081 6132361; fax. +39 081 5609877

ABSTRACT N. 251

Telethon Research Projects - Other Genetic Diseases		
Principal Investigator	**RAMPOLDI LUCA**	
Telethon grant N.	**GGP14263**	
Total budget €	**419.200**	
Centres: **1**	*Duration (yrs):* **3**	*Starting year:* **2014**

MOLECULAR MECHANISMS OF PATHOGENESIS AND PRECLINICAL TREATMENT IN RENAL DISORDERS ASSOCIATED WITH UROMODULIN MUTATIONS

MECCANISMI MOLECOLARI DI PATOGENESI E TERAPIA IN UN MODELLO PRECLINICO DI MALATTIE RENALI ASSOCIATE A MUTAZIONI DI UROMODULINA

Schaeffer Céline (1), Trudu Matteo (1), Brunati Martina (1), Creatore Anna (1), Pasqualetto Elena (1), Scolari Francesco (2), Rastaldi Maria Pia (3), Devuyst Olivier (4), Rampoldi Luca (1)

(1) Molecular Genetics of Renal Disorders Unit, Division of Genetics and Cell Biology, San Raffaele Scientific Institute - Via Olgettina 58, 20132 Milan, Italy. Phone: +39 0226434947; Fax: +39 9226434767; E-mail: rampoldi.luca@hsr.it
(2) Division of Nephrology, Hospital of Montichiari, Brescia
(3) Fondazione IRCCS Ospedale Maggiore Policlinico, Milan
(4) Institute of Physiology, University of Zurich, Zurich

ABSTRACT N. 252

Telethon Research Projects - Other Genetic Diseases		
Principal Investigator	**BOLETTA ALESSANDRA**	
Telethon grant N.	**GGP12183**	
Total budget €	**388.800**	
Centres: **1**	*Duration (yrs):* **3**	*Starting year:* **2012**

TOWARDS THE COMPREHENSION OF POLYCYSTIN-1 FUNCTION AND IDENTIFICATION OF SPECIFIC TARGETS FOR THERAPY IN ADPKD

VERSO LA COMPRENSIONE DELLA FUNZIONE DI POLICISTINA-1 E L’IDENTIFICAZIONE DI TARGET TERAPEUTICI SPECIFICI PER ADPKD

Castelli Maddalena (1), Distefano Gianfranco (1), Pesenti Gritti Angela (1), Matafora Vittoria (2), Chiaravalli Marco (1), Bachi Angela (2), Boletta Alessandra (1)

(1) San Raffaele Scientific Institute, Via Olgettina, 58 20132 Milano
(2) IFOM-IEO campus, Milano

ABSTRACT N. 253

Telethon Research Projects - Other Genetic Diseases		
Principal Investigator	**GENNARI LUIGI**	
Telethon grant N.	**GGP11119**	
Total budget €	**125.200**	
Centres: **2**	*Duration (yrs):* **2**	*Starting year:* **2011**
Partners	FERNANDO GIANFRANCESCO	

GENETICS AND PHARMACOGENETICS OF PAGET’S DISEASE OF BONE

GENETICA E FARMACOGENETICA DELLA MALATTIA OSSEA DI PAGET

Fernando Gianfrancesco (2), Giuseppina Divisato (2), Daniela Formicola (2), Teresa Esposito (2), Daniela Merlotti (1), Laura Pazzaglia (3), Maria Serena Benassi (3), Jaques Brown (5), Pasquale Strazzullo (4), Ranuccio Nuti (1), Domenico Rendina (4), Laetitia Michou (5), Luigi Gennari (1)

(1) Department of Medicine, Surgery and Neurosciences, University of Siena, Siena, Italy - Policlinico Santa Maria alle Scotte, Viale Bracci, 53100-Siena, Italy, Phone: +390577585364, Fax: +390577233446, Email: gennari@unisi.it
(2) Institute of Genetics and Biophysics “Adriano Buzzati-Traverso”, CNR, Naples, Italy
(3) Laboratory of Experimental Oncology, Rizzoli Orthopaedic Institute, Bologna, Italy
(4) Department of Clinical and Experimental Medicine, Federico II University of Naples, Naples, Italy

(5) Department of Medicine, Faculté de Médecine de l'Université Laval, Quebec City, Canada

ABSTRACT N. 254

Telethon Research Projects - Other Genetic Diseases		
Principal Investigator	**MATTEVI ANDREA**	
Telethon grant N.	**GGP12007**	
Total budget €	**221.000**	
Centres: **1**	*Duration (yrs):* **3**	*Starting year:* **2012**

PEROXISOMAL ENZYME DEFICIENCIES IN RHIZOMELIC CHONDRODYSPLASIA PUNCTATA: BIOCHEMISTRY AND THERAPEUTIC AVENUES

DIFETTI ENZIMATICI NELLA CONDRODISPLASIA PUNTATA RIZOMELICA: BIOCHIMICA E POSSIBILITÀ TERAPEUTICHE

Piano Valentina (1), Nenci Simone (1), Pandini Vittorio (2), Aliverti Alessandro (2), Mattevi Andrea (1)

(1) Dipartimento Biologia e Biotecnologie "L. Spallanzani", Università degli Studi di Pavia, Via Ferrata 1, 27100 Pavia - Via Ferrata 1, 27100 Pavia, 0382985534, andrea.mattevi@unipv.it
(2) Dipartimento di Bioscienze, Università degli Studi di Milano, Via Celoria 26, 20133 Milano

ABSTRACT N. 255

Telethon Research Projects - Other Genetic Diseases		
Principal Investigator	**ROSSI ANTONIO**	
Telethon grant N.	**GGP11079**	
Total budget €	**174.200**	
Centres: **1**	*Duration (yrs):* **3**	*Starting year:* **2011**

THE FUNCTIONAL ROLE OF THE CALCIUM ACTIVATED NUCLEOTIDASE 1 (CANT1) GENE IN THE SKELETON: AN IN VIVO STUDY WITH A MOUSE MODEL OF DESBUQUOIS DYSPLASIA

CARATTERIZZAZIONE FUNZIONALE DEL RUOLO DELLA NUCLEOTIDASI 1 ATTIVATA DAL CALCIO (CANT1) NELLO SCHELETRO: UNO STUDIO IN VIVO CON UN MODELLO ANIMALE DI DISPLASIA DI DESBUQUOIS

Monti Luca (1), Paganini Chiara (1), Lecci Silvia (1), Maruelli Silvia (1), Costantini Rossella (1), Cormier-Daire Valerie (2), Forlino Antonella (1), Rossi Antonio (1)

(1) Dipartimento di Medicina Molecolare - Università di Pavia - Via Taramelli 3/B, 27100 Pavia, Italy, e-mail: antrossi@unipv.it
(2) INSERM U781, Institut Imagine, Hopital Necker-Enfants Malades,Paris, France

ABSTRACT N. 256

DTI - Other Genetic Diseases		
Principal Investigator	**SETTEMBRE CARMINE**	
Telethon grant N.	**TCP12008**	
Total budget €	**342.000**	
Centres: **1**	*Duration (yrs):* **5**	*Starting year:* **2013**

IDENTIFICATION OF MOLECULAR TARGETS FOR THE TREATMENT OF THE SKELETAL PHENOTYPE IN LYSOSOMAL STORAGE DISORDERS

IDENTIFICAZIONE DI NUOVI APPROCCI TERAPEUTICI PER IL TRATTAMENTO DELLE MALFORMAZIONI OSSEE NELLE MALATTIE DA ACCUMULO LISOSOMIALE

Cinque Laura (1,2), Forrester Alison (1,2,3), Bartolomeo Rosa (1,2), Venditti Rossella (2), Montefusco Sandro (2), Polishchuk Elena (2), Rossi Antonio (4), Medina Diego (2), De Matteis Maria Antonietta (2), Settembre Carmine (1,2,3)

(1) Dulbecco Telethon Institute c/o Telethon Institute of Genetics and Medicine (TIGEM), Via Campi Flegrei, 34, 80078 Pozzuoli (NA), Italy , email: Settembre@Tigem.it
(2) Telethon Institute of Genetics and Medicine (TIGEM), Pozzuoli (NA), Italy
(3) Medical Genetics, Department of Medical and Translational Science Unit, Federico II University, Naples, Italy
(4) Department of Molecular Medicine, Biochemistry Unit, University of Pavia, Pavia, Italy

ABSTRACT N. 257

DTI - Other Genetic Diseases		
Principal Investigator	**SERAFINI MARTA**	
Telethon grant N.	**TCR14001**	
Total budget €	**136.000**	
Centres: **1**	*Duration (yrs):* **2**	*Starting year:* **2015**

NEONATAL BONE MARROW TRANSPLANTATION PREVENTS BONE PATHOLOGY IN A MOUSE MODEL OF MUCOPOLYSACCHARIDOSIS TYPE I

IL TRAPIANTO DI MIDOLLO OSSEO IN EPOCA NEONATALE PREVIENE IL DANNO SCHELETRICO NEL MODELLO MURINO DI MUCOPOLISACCARIDOSI DI TIPO I

Serafini Marta (1), Pievani Alice (1), Azario Isabella (1), Antolini Laura (2), Shimada Tsutomu (3), Patel Pravin (3), Remoli Cristina (4), Rambaldi Benedetta (1), Valsecchi Maria Grazia (2), Riminucci Mara (4), Biondi Andrea (5), Tomatsu Shunji (3), Serafini Marta (1)

(1) Dulbecco Telethon Institute at Centro Ricerca M.Tettamanti, Department of Paediatrics, University of Milano-Bicocca, San Gerardo Hospital, via Pergolesi, 33 20900 Monza (MB), Italy Phone: +39-039-2332232 Fax: +39-039-2332167 E-mail: serafinim72@gmail.com
(2) Centro di Biostatistica per l'epidemiologia clinica, Dep of Health Sciences, Milano–Bicocca University, Monza
(3) Dep of Biomedical Research, Alfred I. duPont Hospital for Children, Wilmington, DE, US
(4) Dep of Molecular Medicine, Sapienza University, Rome
(5) Dep of Pediatrics, Milano-Bicocca University, San Gerardo Hospital/Fondazione MBBM, Monza

ABSTRACT N. 258

Telethon Research Projects - Other Genetic Diseases		
Principal Investigator	**FORLINO ANTONELLA**	
Telethon grant N.	**GGP13098**	
Total budget €	**321.700**	
Centres: **2**	*Duration (yrs):* **3**	*Starting year:* **2013**
Partners	LAURA BIANCHI	

TARGETING ER STRESS TO TREAT OSTEOGENESIS IMPERFECTA

NUOVE STRATEGIE PER IL TRATTAMENTO DELL'OSTEOGENESI IMPERFETTA

Gioia Roberta (1), Gagliardi Assunta (2), Landi Claudia (2), Besio Roberta (1), Tonelli Francesca (1), Bini Luca (2), Leikin Sergey (3), Schinke Thorsten (4), Cotelli Franco (5), Tenni Ruggero (1), Bianchi Laura (2), Forlino Antonella (1)

(1) Department of Molecular Medicine, Biochemistry Unit, University of Pavia, Pavia, Italy - Via Taramelli 3/B, 27100 Pavia, phone: +39-0382-987235, fax: +39-0382-423108, aforlino@unipv.it
(2) Functional Proteomics Lab, Department of Life Sciences, University of Siena, Siena, Italy

(3) Section of Physical Biochemistry, NICHD/NIH, Bethesda, Maryland, USA
(4) University of Hamburg, Hamburg, Germany
(5) Department of Biosciences, University of Milano, Milan, Italy

ABSTRACT N. 259

Telethon Research Projects - Other Genetic Diseases		
Principal Investigator	**SOBACCHI CRISTINA**	
Telethon grant N.	**GGP12178**	
Total budget €	**123.000**	
Centres: **1**	*Duration (yrs):* **2**	*Starting year:* **2012**

MESENCHYMAL STEM CELL TRANSPLANTATION AS A THERAPEUTIC APPROACH TO RANKL-DEPENDENT OSTEOPETROSIS

TRAPIANTO DI CELLULE MESENCHIMALI STAMINALI COME APPROCCIO TERAPEUTICO PER L'OSTEOPETROSI RANKL- DIPENDENTE

Menale Ciro (1,2), Lo Iacono Nadia (1,2), Schena Francesca (3), Marrella Veronica (1,2), Caldana Elena (1,2), Mantero Stefano (1,2), Vezzoni Paolo (1,2), Villa Anna (1,2), Sobacchi Cristina (1,2)

(1) CNR-IRGB, Milan Unit, Milan, Italy - via Manzoni 113, Rozzano (MI); tel 0282245153; fax 0282245290; cristina.sobacchi@humanitasresearch.it
(2) Humanitas Clinical and Research Center, Rozzano, Italy
(3) Laboratory of Immunology and Rheumatic Disease, Istituto G. Gaslini IRCCS, Genova, Italy

ABSTRACT N. 260

Telethon Research Projects - Other Genetic Diseases		
Principal Investigator	**TETI ANNA MARIA**	
Telethon grant N.	**GGP14014**	
Total budget €	**202.300**	
Centres: **1**	*Duration (yrs):* **3**	*Starting year:* **2014**

IN-DEPTH PHENOTYPING AND EXPERIMENTAL THERAPY OF AUTOSOMAL DOMINANT OSTEOPETROSIS

STUDIO DEL FENOTIPO E TERAPIA SPERIMENTALE DELL'OSTEOPETROSI AUTOSOMICA DOMINANTE

Capulli Mattia, Maurizi Antonio, Rucci Nadia, Teti Anna

Dipartimento di Scienze Cliniche Applicate e Biotecnologie, Università dell'Aquila, Via Vetoio - Coppito 2, 67100 L'Aquila. Tel. 0862 433513. Fax 0862 433523. E-mail teti@univaq.it

ABSTRACT N. 261

TIGEM - Other Genetic Diseases		
Principal Investigator	**DE MATTEIS MARIA ANTONIETTA**	
Telethon grant N.	**TGM11CB2**	
Total budget €	**300.000**	
Centres: **1**	*Duration (yrs):* **5**	*Starting year:* **2011**

DEFINING THE CELLULAR AND MOLECULAR BASIS OF THE SPONDYLOEPIPHYSEAL DYSPLASIA TARDA AND IDENTIFICATION OF TARGETS FOR PHARMACOLOGICAL INTERVENTION

DEFINIZIONE DELLE BASI MOLECOLARI E CELLULARI DELLA DISPLASIA SPONDILOEPIFISIARIA TARDA E IDENTIFICAZIONE DI TARGET PER L'INTERVENTO FARMACOLOGICO

Zappa Francesca, Venditti Rossella, Santoro Michele, Wilson Cathal, De Matteis Maria Antonietta

Telethon Institute of Genetics and Medicine (TIGEM), Via Campi Flegrei 34, 80078 Pozzuoli (Napoli), Italy. Phone +39 081 6132361; fax +39 081 5609877

ABSTRACT N. 262

Telethon Research Projects - Other Genetic Diseases		
Principal Investigator	**ZIPPO ALESSIO**	
Telethon grant N.	**GEP13057**	
Total budget €	**40.100**	
Centres: **1**	*Duration (yrs):* **1**	*Starting year:* **2014**

DEVELOPMENT OF AN IN VITRO DISEASE MODEL SYSTEM FOR DISSECTING THE EPIGENETIC MECHANISMS UNDERLYING PATHOGENESIS OF KABUKI SYNDROME

SVILUPPO DI UN MODELLO SPERIMENTALE PER STUDIARE I MECCANISMI EPIGENETICI ALLA BASE DELLA SINDROME KABUKI

Zippo Alessio, Fasciani Alessandra

INGM Via Francesco Sforza 35, Milano tel. 02 0066321 e-mail: zippo@ingm.org

ABSTRACT N. 263

Telethon Research Projects - Other Genetic Diseases		
Principal Investigator	**MERLA GIUSEPPE**	
Telethon grant N.	**GGP13231**	
Total budget €	**413.200**	
Centres: **3**	*Duration (yrs):* **3**	*Starting year:* **2013**
Partners	STEFANO CASOLA, GIUSEPPE TESTA	

AN INTEGRATED STRATEGY TO FUNCTIONALLY DISSECT THE GENETIC AND EPIGENETIC MECHANISMS UNDERLYING KABUKI SYNDROME

UNA STRATEGIA INTEGRATA PER COMPRENDERE I MECCANISMI GENETICI ED EPIGENETICI ALLA BASE DELLA SINDROME KABUKI

Micale Lucia (1), Gabriele Michele (3), Valentina Petrocelli (2), De Nittis Pasquelena (1), Germain Pierre-Luc (3), Mainoldi Federica (2), Augello Bartolomeo (1), Fusco Carmela (1), Casola Stefano (2), Testa Giuseppe (3), Merla Giuseppe (1)

(1) IRCCS Casa Sollievo delle Sofferenza, Medical Genetics Unit - viale Cappuccini, 71013 San Giovanni Rotondo (FG), tel: 088 2416350, fax: 088 2411616, email: g.merla@operapadrepio.it
(2) IFOM Fondazione Istituto FIRC di Oncologia Molecolare, Milano
(3) Dipartimento di Oncologia Sperimentale, Istituto Europeo di Oncologia, Milano

ABSTRACT N. 264

Telethon Research Projects - Other Genetic Diseases		
Principal Investigator	**RICCIO ANDREA**	
Telethon grant N.	**GGP11122**	
Total budget €	**231.000**	
Centres: **1**	*Duration (yrs):* **3**	*Starting year:* **2011**

GROWTH DISORDERS AND GENOMIC IMPRINTING: GENETIC DEFECTS AND MOLECULAR MECHANISMS

IMPRINTING GENOMICO E DISORDINI DELLA CRESCITA: DIFETTI GENETICI E MECCANISMI MOLECOLARI

Riccio Andrea (1,2)

(1) Istituto di Genetica e Biofisica A. Buzzati-Traverso, CNR, via P. Castellino 111, Napoli
(2) Dipartimento DiSTABiF, Seconda Università di Napoli, Caserta

ABSTRACT N. 265

Telethon Research Projects - Other Genetic Diseases		
Principal Investigator	**SOBACCHI CRISTINA**	
Telethon grant N.	**GEP13060**	
Total budget €	**21.350**	
Centres: **1**	*Duration (yrs):* **1**	*Starting year:* **2014**

UNDERSTANDING THE GENETIC BASIS OF ACROFRONTOFACIONASAL DYSOSTOSIS 1

STUDIO DELLE BASI GENETICHE DELLA DISOSTOSI ACRO-FRONTO-FACIO-NASALE DI TIPO 1

Palagano Eleonora (1,2), Strina Dario (1,2), Menale Ciro (1,2), Mantero Stefano (1,2), Prontera Paolo (3), Guion-Almeida Maria Leine (4), Uva Paolo (5), Angius Andrea (5,6), Villa Anna (1,2), Sobacchi Cristina (1,2)

(1) CNR-IRGB, Milan Unit, Milan, Italy - via Manzoni 113, 20089 Rozzano (MI); tel 02 82245153; fax 02 82245290; cristina.sobacchi@humanitasresearch.it
(2) Humanitas Clinical and Research Center, Rozzano, Italy
(3) CRR Genetica Medica, Università ed Azienda Ospedaliera di Perugia, Perugia, Italy
(4) Department of Clinical Genetics, Hospital of Rehabilitation of Craniofacial Anomalies, University of Sao Paulo, Sao Paulo, Brasil
(5) CRS4, Science and Technology Park Polaris, Piscina Manna, Pula, Italy
(6) CNR-IRGB, Monserrato, Italy

ABSTRACT N. 266

Telethon Research Projects - Other Genetic Diseases		
Principal Investigator	**MISSERO CATERINA**	
Telethon grant N.	**GGP12239**	
Total budget €	**259.700**	
Centres: **1**	*Duration (yrs):* **3**	*Starting year:* **2012**

DISSECTING THE MOLECULAR MECHANISMS UNDERLYING EPIDERMAL DEFECTS IN AEC SYNDROME

STUDIO DEI MECCANISMI COINVOLTI NEI DIFETTI CUTANEI CARATTERIZZANTI LA SINDROME DI HAY-WELLS

Cirillo Luisa (1,3), Mollo Maria Rosaria (1,3), Sirico Anna (1), Polishchuk Elena (4), Polishchuk Roman (4), Antonini Dario (5), Missero Caterina (1,2)

(1) CEINGE Biotecnologie Avanzate, Napoli, Italy - Via G. Salvatore 486 - 80145 Napoli, Italy. Tel 081-3737853. e-mail: missero@ceinge.unina.it
(2) Department of Biology, University of Naples Federico II, Napoli, Italy
(3) Department of Molecular Medicine, University of Naples Federico II, Napoli, Italy
(4) Telethon Institute of Genetics and Medicine (TIGEM), Napoli, Italy
(5) IRCCS SDN, Napoli, Italy

ABSTRACT N. 267

Telethon Research Projects - Other Genetic Diseases		
Principal Investigator	**VANONI MARIA ANTONIETTA**	
Telethon grant N.	**GGP10090**	
Total budget €	**206.800**	
Centres: **1**	*Duration (yrs):* **3**	*Starting year:* **2010**

STRUCTURE-FUNCTION STUDIES ON 24-DEHYDROCHOLESTEROL REDUCTASE, THE AFFECTED ENZYME IN DESMOSTEROLOSIS, A SEVERE INHERITED DISORDER OF STEROL METABOLISM

STUDI STRUTTURA-FUNZIONE SULLA 24-DEIDROCOLESTEROLO RIDUTTASI, L'ENZIMA DIFETTOSO NELLA DESMOSTEROLOSI, UNA GRAVE MALATTIA EREDITARIA A CARICO DEL METABOLISMO DEGLI STEROLI

Zucchini Daniela (1), Sorrentino Luca (1), Zito Arianna (2), Caruso Donatella (2), Tedeschi Gabriella (3), Galbiati Rita (2), Aliverti Alessandro (1), Maggi Roberto (2), Camattari Andrea (4,5), Pichler Harald (4,5), Vanoni Maria Antonietta (1)

(1) Dipartimento di Bioscienze, Università degli Studi di Milano, Milano - Via Celoria 26, 20133 Milano, tel. 0250314901, fax 0250314895, maria.vanoni@unimi.it
(2) Dipartimento di Scienze Farmacologiche e Biomolecolari, Università degli Studi di Milano, Milano
(3) Dipartimento di Scienze Veterinarie e Sanità Pubblica, Università degli Studi di Milano, Milano
(4) Institute of Molecular Biotechnology, Graz University of Technology, Graz, Austria
(5) Austrian Centre of Industrial Biotechnology, Graz, Austria

ABSTRACT N. 268

Telethon Research Projects - Other Genetic Diseases		
Principal Investigator	**MERLO GIORGIO ROBERTO**	
Telethon grant N.	**GGP11097**	
Total budget €	**489.800**	
Centres: **3**	*Duration (yrs):* **3**	*Starting year:* **2011**
Partners	LUISA FRANCESCA GUERRINI, ANTONIO COSTANZO	

TRANSLATING CURRENT AND NEW KNOWLEDGE ON THE DEVELOPMENTAL FUNCTIONS OF P63 INTO RESTORING NORMAL DEVELOPMENT IN MODELS OF ORGAN CULTURE AND IN VIVO

TRASFERIRE LE CONOSCENZE SUL RUOLO DEL GENE MALATTIA P63 DURANTE LO SVILUPPO EMBRIONALE ALLA POSSIBILITÀ DI RIPRISTINARE LO SVILUPPO NORMALE IN MODELLI DI COLTURE DI TESSUTO E IN VIVO

Restelli Michela (2), Guerrini Luisa F. (2), Gnesutta Nerina (2), Costanzo Antonio (3), Marinari Barbara (3), Conte Daniele (1), Merlo Giorgio R. (1)

(1) Dept. Molecular Biotechnology, Universtity of Torino, Via Nizza 52 Torino. Tel 011-6706449 Fax 011-6706432 e-mail giorgioroberto.merlo@unito.it
(2) Dept. Biosciences, University of Milano
(3) Dept. Dermatology, University of Rome "Tor Vergata", Roma

ABSTRACT N. 269

Telethon Research Projects - Other Genetic Diseases		
Principal Investigator	**TALORA CLAUDIO**	
Telethon grant N.	**GGP12264**	
Total budget €	**252.600**	
Centres: **3**	*Duration (yrs):* **3**	*Starting year:* **2012**
Partners	GIANFRANCO BIOLCATI, CLAUDIO PALLESCHI	

CALCIUM DYSREGULATION AND OXIDATIVE STRESS: FROM MOLECULAR MECHANISMS TO THERAPEUTIC IMPLICATIONS IN HAILEY-HAILEY DISEASE

DEREGOLAZIONE DEL CALCIO E STRESS-OSSIDATIVO: DAI MECCANISMI MOLECOLARI ALLE IMPLICAZIONI TERAPEUTICHE NELLA PATOLOGIA DI HAILEY-HAILEY

Cialfi Samantha (1), Ficocello Graziella (2), Zanni Elena (2), De Blasio Carlo (1), Barbieri Luca (3), Aurizi Caterina (3), Biolcati Gianfranco (3), Palleschi Claudio (2), Screpanti Isabella (1), Uccelletti Daniela (2), Talora Claudio (1)

(1) Dipartimento di Medicina Molecolare Sapienza Universita" di Roma, Viale Regina Elena 291, 00161 Roma, Italy Tel. 06 49255674 claudio.talora@uniroma1.it
(2) Dipartimento di Biologia e Biotecnologie, Charles Darwin, Sapienza Università di Roma, Piazzale Aldo Moro 5, 00161 Roma, Italy
(3) Centro Porfirie, Istituto San Gallicano IRCCS, Via Elio Chianesi 53, 00144 Roma, Italia

ABSTRACT N. 270

Telethon Research Projects - Other Genetic Diseases		
Principal Investigator	**BOTTA ELENA**	
Telethon grant N.	**GEP13022**	
Total budget €	**44.000**	
Centres: **1**	*Duration (yrs):* **1**	*Starting year:* **2014**

DISEASE GENE IDENTIFICATION IN NON-PHOTOSENSITIVE TRICHOTHIODYSTROPHY BY EXOME SEQUENCING

IDENTIFICAZIONE DI GENI RESPONSABILI DELLA FORMA NON-FOTOSENSIBILE DELLA TRICOTIODISTROFIA

Caligiuri Giuseppina, Carriero Roberta, Ricotti Roberta, Lisa Antonella, Bione Silvia, Botta Elena

IGM CNR Pavia - IGM CNR Via Abbiategrasso, 207 Pavia -tel. 0382/546331-fax 0382/422286-email botta@igm.cnr.it

ABSTRACT N. 271

Telethon Research Projects - Other Genetic Diseases		
Principal Investigator	**BARCHI MARCO**	
Telethon grant N.	**GGP12189**	
Total budget €	**324.200**	
Centres: **2**	*Duration (yrs):* **3**	*Starting year:* **2012**
Partners	CLAUDIO SETTE	

UNDERSTANDING XY AND AUTOSOME CHROMOSOME SEGREGATION DEFECTS IN MAMMALS: NEW INSIGHTS FROM THE REGULATION OF EXPRESSION AND FUNCTION OF SPO11 SPLICE ISOFORMS

IDENTIFICAZIONE DEI MECCANISMI MOLECOLARI ALLA BASE DEI DIFETTI DI SEGREGAZIONE DEI CROMOSOMI SESSUALI E DEGLI AUTOSOMI NEI MAMMIFERI: IL RUOLO DEL GENE SPO11

Faieta Monica (1), Cesari Eleonora (1), Loiarro Maria (2), Di Cecca Stefano (1), Bielli Pamela (2), Sette Claudio (1,2), Barchi Marco (1)

(1) Department of Biomedicine and Prevention, University of Rome Tor Vergata, Rome, Italy - Via Montpellier n.1, 00133, Rome Italy, tel +390672596266, fax +390672596268, marco.barchi@uniroma2.it
(2) Laboratory of Neuroembryology, Fondazione Santa Lucia, Rome, Italy

ABSTRACT N. 272

Telethon Research Projects - Other Genetic Diseases		
Principal Investigator	**FIUMARA FERDINANDO**	
Telethon grant N.	**GGP11223**	
Total budget €	**147.300**	
Centres: **1**	*Duration (yrs):* **3**	*Starting year:* **2011**

POLYALANINE REPEATS AS PHYLOGENETIC BARCODES AND MEDIATORS OF PROTEIN FUNCTION AND GENETIC DISEASE-RELATED DYSFUNCTION THROUGH COILED COIL ASSEMBLY

TRATTI DI POLIALANINA COME BARCODE FILOGENETICI E MEDIATORI DELLA FUNZIONE E DISFUNZIONE PROTEICA ASSOCIATA A MALATTIE GENETICHE DA ESPANSIONE ATTRAVERSO LA FORMAZIONE DI STRUTTURE COILED COIL

Pelassa Ilaria, Corà Davide, Arnaboldi Sofia, Ferdinando Fiumara

Dipartimento di Neuroscienze, Università degli Studi di Torino, ferdinando.fiumara@unito.it

ABSTRACT N. 273

Telethon Research Projects - Other Genetic Diseases		
Principal Investigator	**VACCARI THOMAS**	
Telethon grant N.	**GGP13225**	
Total budget €	**250.800**	
Centres: **1**	*Duration (yrs):* **3**	*Starting year:* **2013**

MULTIPLE FUNCTIONS OF THE SNARE PROTEIN SNAP29 DURING EPITHELIAL FORMATION

IL RUOLO DELLA PROTEINA SNARE SNAP29 DURANTE LA FORMAZIONE DEGLI EPITELI

Morelli Elena (1), Mastrodonato Valeria (1), Ginefra Pierpaolo (2), Beznusenko Galina (1), Mironov Alexandre (1), Vaccari Thomas (1)

(1) IFOM, the FIRC Institute of Molecular Oncology, Via Adamello 16 - 20139 Milan - ITALY, tel. 02574303823, fax 02574303088, E-mail: thomas.vaccari@ifom.eu
(2) École Polytechnique Fédérale de Lausanne, Swiss Institute for Experimental Cancer Research, Lausanne, Switzerland

ABSTRACT N. 274

Telethon Research Projects - Other Genetic Diseases		
Principal Investigator	**CARETTI GIUSEPPINA**	
Telethon grant N.	**GEP13056**	
Total budget €	**47.800**	
Centres: **1**	*Duration (yrs):* **1**	*Starting year:* **2014**

FUNCTIONAL ROLE OF EZH2 MUTATIONS IN WEAVER SYNDROME

STUDIO DELLE BASI MOLECOLARI DELLA SINDROME DI WEAVER

Camilli Giulia, Claudio Fenizia, Caretti Giuseppina

Dipartimento di Bioscienze, Università degli Studi di Milano, Via Celoria 26, 20133 Milano, Tel:02-50315002, fax: 02-50315044, email: giuseppina.caretti@unimi.it

ABSTRACT N. 275

Telethon Research Projects - Other Genetic Diseases		
Principal Investigator	**PRIMO LUCA**	
Telethon grant N.	**GEP13015**	
Total budget €	**49.500**	
Centres: **1**	*Duration (yrs):* **1**	*Starting year:* **2014**

CELLULAR AND ANIMAL MODELS OF CLOVES SYNDROME

MODELLI CELLULARI E ANIMALI PER LO STUDIO DELLA SINDROME CLOVES

diBlasio Laura (1,2), Primo Luca (1,2)

(1) Dep. of Oncology, University of Torino - Str. Provinciale 142 Km 3.95 - Candiolo 10100 (TO) - +39 011 9933505 luca.primo@unito.it
(2) Candiolo Cancer Institute-FPO IRCCS

ABSTRACT N. 276

Telethon Research Projects - Other Genetic Diseases		
Principal Investigator	**TESTA GIUSEPPE**	
Telethon grant N.	**GEP13105**	
Total budget €	**49.940**	
Centres: **1**	*Duration (yrs):* **3**	*Starting year:* **2014**

NEURODEVELOPMENTAL ALTERATIONS IN WEAVER SYNDROME: A CELL REPROGRAMMING-BASED APPROACH TO THE ELUCIDATION OF EPIGENETIC DISEASE MECHANISMS

SINDROME DI WEAVER: NUOVE TECNOLOGIE DI RIPROGRAMMAZIONE CELLULARE PER LO STUDIO DEI MECCANISMI PATOGENETICI ALL'ORIGINE DI QUESTA MALATTIA

Buontempo Serena (1), Laise Pasquale (1), Testa Giuseppe (1,2)

(1) Istituto Europeo di Oncologia, Via Adamello 16, 20139 Milano giuseppe.testa@ieo.eu Tel. 0294375105
(2) Università di Milano

ABSTRACT N. 277

Telethon Research Projects - Other Genetic Diseases		
Principal Investigator	**BALDINI ANTONIO**	
Telethon grant N.	**GGP14211**	
Total budget €	**341.600**	
Centres: **1**	*Duration (yrs):* **3**	*Starting year:* **2015**

PHENOTYPIC RESCUE OF THE DIGEORGE SYNDROME PHENOTYPE IN MOUSE MODELS

SINDROME DI DIGEORGE: APPROCCI TERAPEUTICI NEL MODELLO MURINO

Fulcoli Filomena Gabriella (2), Caprio Cinzia (2), Baldini Antonio (1,2)

(1) Department of Molecular Medicine and Medical Biotechnology, University of Naples Federico II, 80131 Naples, Italy
(2) Consiglio Nazionale delle Ricerche Institute of Genetics and Biophysics Adriano Buzzati Traverso, 80131 Naples, Italy

ABSTRACT N. 278

Telethon Research Projects - Other Genetic Diseases		
Principal Investigator	**TESTA GIUSEPPE**	
Telethon grant N.	**GGP14265**	
Total budget €	**431.100**	
Centres: **2**	*Duration (yrs):* **3**	*Starting year:* **2014**
Partners	GIUSEPPE MERLA	

TRANSCRIPTIONAL AND EPIGENETIC DYSFUNCTION IN WILLIAMS BEUREN AND 7Q11.23 MICRODUPLICATION SYNDROMES

STUDIO DELLE DISFUNZIONI TRASCRIZIONALI ED EPIGENETICHE NELLA SINDROME DI WILLIAMS BEUREN E NELLA SINDROME DA MICRODUPLICAZIONE DEL 7Q11.23

Adamo Antonio (1), Atashpaz Sina (1), Germain Pierre-Luc (1), Zanella Matteo (1), D'Agostino Giuseppe (1), Albertin Veronica (1), Chenoweth Josh (3), Micale Lucia (4), Fusco Carmela (4), Unger Christian (5), Augello Bartolomeo (4), Palumbo Orazio (4), Hamilton Brad (6), Carella Massimo (4), Donti Emilio (7), Pruneri Giancarlo (1), Biamino Elisa (9), Selicorni Angelo (8), Prontera Paolo (7), McKay Ronald (3), Merla Giuseppe (4), Testa Giuseppe (1,2)

(1) Istituto Europeo di Oncologia, Via Adamello 16, 20139 Milano giuseppe.testa@ieo.eu Tel. 0294375105
(2) Dipartimento di Scienze della salute, Università di Milano, Milano
(3) Lieber Institute for Brain Development, Baltimore, USA
(4) Unità di Genetica Medica, Ospedale IRCCS Casa Sollievo della Sofferenza, San Giovanni Rotondo
(5) Department of Biomedical Sciences, University of Sheffield, Sheffield, United Kingdom
(6) Stemgent, Cambridge, USA
(7) Unità di Genetica Medica, Ospedale S. M. della Misericordia, University of Perugia, Perugia
(8) UOS Genetica Clinica Pediatrica, Fondazione MBBM, AO San Gerardo, Monza
(9) Dipartimento di Pediatria, Università di Torino, Torino

ABSTRACT N. 279

DTI - Other Genetic Diseases		
Principal Investigator	**CORONA DAVIDE**	
Telethon grant N.	**TCR09002**	
Total budget €	**800.000**	
Centres: **1**	*Duration (yrs):* **5**	*Starting year:* **2010**

TRANS-ACTIVATION: A NEW EPIGENETIC PHENOMENON UNDERLYING TRANSCRIPTIONAL MEMORY

TRANS-ACTTIVAZIONE: UN NUOVO FENOMENO EPIGENETICO CHE SPIEGA LA MEMORIA TRASCRIZIONALE

Onorati Maria Cristina, Arancio Walter, Vincenzo Cavalieri, Davide F.V. Corona

Istituto Telethon Dulbecco c/o Dipartimento STEBICEF, Università di Palermo, Viale delle Scienze - Edificio 16, 091 238 97345, davide.corona@unipa.it

ABSTRACT N. 280

Telethon Research Projects - Other Genetic Diseases		
Principal Investigator	**CARIBONI ANNA MARIA**	
Telethon grant N.	**GGP13142**	
Total budget €	**125.600**	
Centres: **1**	*Duration (yrs):* **3**	*Starting year:* **2013**

SEMAPHORIN 3E AND CHD7 SIGNALLING PATHWAYS IN THE CONTROL OF OLFACTION AND REPRODUCTION: LINK FOR KALLMANN AND CHARGE SYNDROMES?

RUOLO DELLA SEMAFORINA 3E E DI CHD7 NELL'OLFATTO E NELLA RIPRODUZIONE: UN POSSIBILE COLLEGAMENTO TRA LE SINDROMI CHARGE E DI KALLMANN?

Andrè Valentina (1), Chauvet Sophie (3), Davidson Kathryn (2), Cassatella Daniele (4), Fantin Alessandro (2), Bouloux Pierre (2), Mann Fanny (3), Ruhrberg Christiana (2), Cariboni Anna (1,2)

(1) University of Milan, Department of Pharmacological and Biomolecular Sciences - Via Balzaretti, 9, 20133, Milan, t. 02/50318216, f. 02/50318204, e. anna.cariboni@unimi.it
(2) University College London, London, UK
(3) Aix Marseille Université, Marseille, France
(4) Centre Hospitalier Universitaire Vaudois, Lausanne, Switzerland

ABSTRACT N. 281

Telethon Research Projects - Other Genetic Diseases		
Principal Investigator	**DIONISI-VICI CARLO**	
Telethon grant N.	**GEP13085**	
Total budget €	**49.500**	
Centres: **1**	*Duration (yrs):* **1**	*Starting year:* **2014**

DECHIPERING THE MECHANISM OF IMMUNE DYSFUNCTION IN VICI SYNDROME

DECIFRARE IL MECCANISMO RESPONSABILE DEL DEFICIT IMMUNITARIO NELLA SINDROME DI VICI

Carsetti Rita (2), Piano Mortari Eva (2), Farroni Chiara (2), Marcellini Valentina (2), Martinelli Diego (1), Dionisi-Vici Carlo (1)

(1) Unità Operativa Complessa Patologia Metabolica, Dipartimento Medicina Pediatrica, Ospedale Pediatrico Bambino Gesù IRCCS, Roma, Italia - Piazza S. Onofrio 4, 00165, Roma - tel. +39.06.68592275 - fax +39.06.68592791 - carlo.dionisivici@opbg.net
(2) Unità di Ricerca Immunologia, Ospedale Pediatrico Bambino Gesù IRCCS, Roma, Italia

ABSTRACT N. 282

Telethon Research Projects - Other Genetic Diseases		
Principal Investigator	**PICHIERRI PIETRO**	
Telethon grant N.	**GGP12144**	
Total budget €	**225.500**	
Centres: **1**	*Duration (yrs):* **3**	*Starting year:* **2012**

UNCOVERING THE MOLECULAR PATHOLOGY OF WERNER SYNDROME: ANALYSIS OF THE FUNCTIONAL RELATIONSHIP BETWEEN ATR-RELATED WRN FUNCTION, REPLICATION STRESS AND PREMATURE CELLULAR SENESCENCE

VERSO LA COMPRENSIONE DELLA PATOGENESI DELLA SINDROME DI WERNER: STUDIO DELLA CORRELAZIONE FUNZIONALE TRA LA PROTEINA ATR E LA PROTEINA WRN NELL'INSORGENZA DELLA SENESCENZA CELLULARE PREMATUTA DELLA SINDROME

Iannascoli Chiara (1), Palermo Valentina (1), Basile Giorgia (2), Franchitto Annapaola (2), Pichierri Pietro (1)

(1) Section of Experimental and Computational Carcinogenesis - Istituto Superiore di Sanità - Viale Regina Elena 299 - 00161 Roma; Tel. 0649902994 (Studio) Tel. 0649902954 (Lab) Fax. 0660513138; pietro.pichierri@iss.it
(2) Section of Molecular Epidemiology - Istituto Superiore di Sanità, Roma

ABSTRACT N. 283

Telethon Research Projects - Other Genetic Diseases		
Principal Investigator	**CRISPONI LAURA**	
Telethon grant N.	**GEP13093**	
Total budget €	**44.110**	
Centres: **1**	*Duration (yrs):* **1**	*Starting year:* **2014**

IDENTIFICATION OF NOVEL GENE(S) ASSOCIATED WITH CRISPONI/COLD INDUCED SWEATING SYNDROME-LIKE PHENOTYPES BY WHOLE-EXOME SEQUENCING

IDENTIFICAZIONE DI NUOVI GENI IMPLICATI NELLA SINDROME DI CRISPONI/SUDORAZIONE INDOTTA DAL FREDDO TIPO 1 ATTRAVERSO IL SEQUENZIAMENTO ESOMICO

Uva Paolo (1), Puddu Alessandro (2), Oppo Manuela (3), Onano Stefano (2), Marcia Loredana (2,3), Deiana Manila (2), Loi Angela (2), Marongiu Mara (2), Cuccuru Gianmauro (1), Fotia Giorgio (1), Rutsch Frank (4), Cucca Francesco (2,3), Crisponi Giangiorgio (5), Angius Andrea (1,2), Crisponi Laura (1,2)

(1) Centro di Ricerca, Sviluppo e Studi Superiori in Sardegna (CRS4), Pula, Italy - Loc. Piscina Manna, 09010 Pula (CA) - Tel. +39 070/6754591 -Email: laura.crisponi@irgb.cnr.it
(2) Istituto di Ricerca Genetica e Biomedica (IRGB), Consiglio Nazionale delle Ricerche (CNR), Monserrato (CA), Italy
(3) Dipartimento di Scienze Biomediche, Università di Sassari, Sassari, Italy
(4) Department of General Pediatrics, Munster University Children's Hospital, Munster, Germany
(5) Clinica Sant'Anna, Cagliari, Italy

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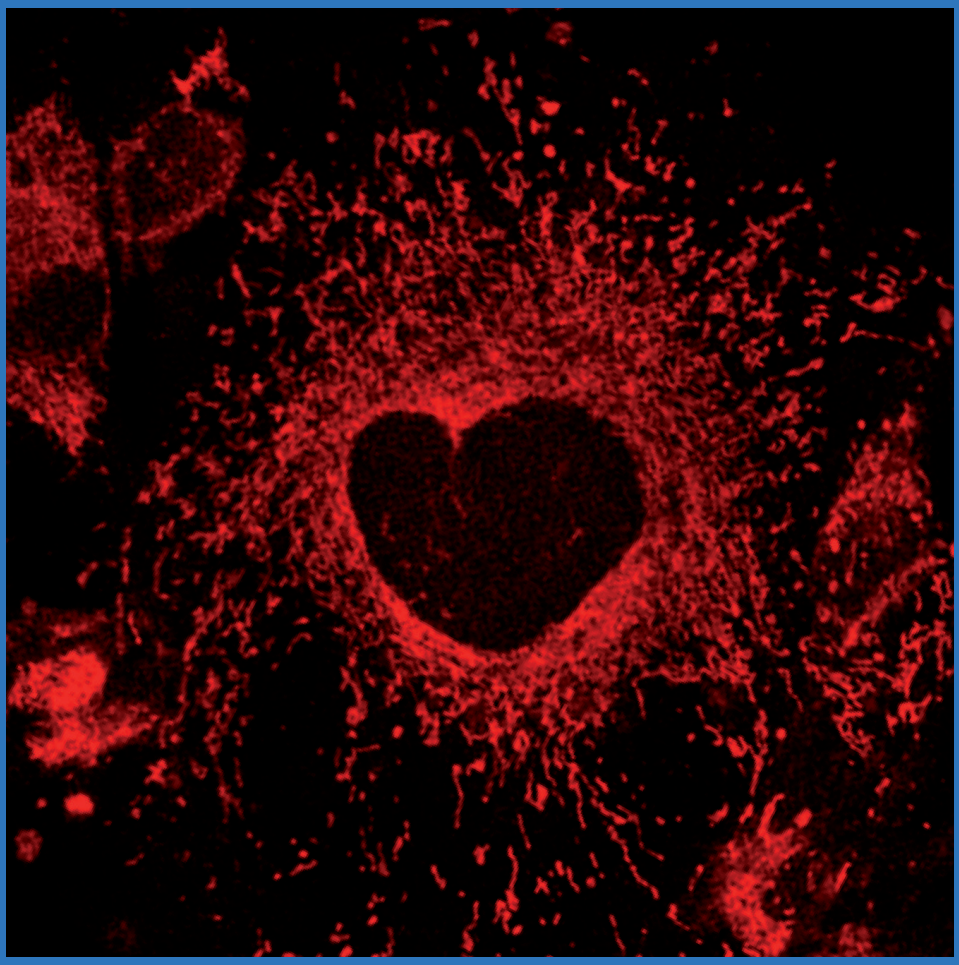

FONDAZIONE TELETHON

Rome headquarter
Via Varese, 16/B
00185 Rome, Italy
Tel. +39 06 440151
Fax +39 06 44015521

Milan headquarter
Piazza Cavour, 1
20121 Milan, Italy
Tel. +39 02 2022171
Fax +39 06 44015600

www.telethon.it e-mail: telethon@telethon.it