Fondazione Telethon

XX Scientific Convention

October 28-30, 2019

Riva del Garda (TN) - Palazzo dei Congressi

FONDAZIONE Telethon

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Telethon

XX SCIENTIFIC CONVENTION

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Riva del Garda (TN)

PROVINCIA
AUTONOMA
DI TRENTO

Front cover - Fondazione Telethon mission: from research to therapy.
SR-Tiget, 2011: child during gene therapy treatment / Telethon scientist at work.

Immagine di copertina - La missione di Fondazione Telethon: dalla ricerca alla cura.
SR- Tiget, 2011: un bambino durante il trattamento di terapia genica / Una ricercatrice Telethon al lavoro.

Back cover - Fondazione Telethon stakeholders: Researchers, Representatives of Patient Organizations and Volunteer at the XVII Scientific Convention, Riva del Garda, March 11-13, 2013

Retro di copertina - Le anime di Fondazione Telethon: Ricercatori, Rappresentanti delle Associazioni di Pazienti e Volontari alla XVII Convention scientifica, Riva del Garda, 11-13 marzo 2013.

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Fondazione Telethon would like to express its gratitude to those who, through their generosity, have made it possible to hold the XX Scientific Convention

RINGRAZIAMENTI

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

PROVINCIA AUTONOMA DI TRENTO

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Tebu-Bio SRL
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Thermo Fisher Scientific SRL
Twin Helix
Vinci-Biochem SRL
Voden

SCIENTIFIC PROGRAM

Monday, October 28th 2019

10.00 - 14.00 Registration and poster setting up

14.00 - 15.00 **Welcome and Opening by Fondazione Telethon**
Francesca Pasinelli, General Manager
Manuela Battaglia, Head of Research

15.00 - 17.00 **PLENARY SESSION 1 - Genetic MUSCLE DISEASES: FROM KNOWLEDGE ON PHYSIO-PATHOLOGY TO IMPLEMENTATION OF ADVANCED THERAPIES**
The complexity of the pathological mechanisms underlying Muscular dystrophies and myopathies will be discussed.
Chair: Enrico Bertini - Ospedale Pediatrico Bambino Gesù, Roma

15.00 - 15.15 **Introduction**

15.15 - 15.35 **Store-operated calcium entry (SOCE): role in skeletal muscle function and disease**
Feliciano Protasi - Università degli Studi G. d'Annunzio Chieti, Pescara **(Talk 1)**

15.40 - 15.50 ***Flash Talk - Neuromuscular biobanks: a support to research and patients' needs***
Giacomo Comi - Ospedale Maggiore Policlinico, Università degli Studi di Milano

15.50 - 16.10 **Autophagy as a key pathogenic mechanism in COL6-related diseases and its significance for prospective therapies**
Paolo Bonaldo - Università degli Studi di Padova **(Talk 2)**

16.10 - 16.30 **From reverse genetics to gene therapy: Duchenne Muscular Dystrophy**
Alessandra Ferlini - Università degli Studi di Ferrara **(Talk 3)**

16.30 - 16.50 **Cross-fertilization between motor neuron disorders and Muscular dystrophies: improving care and targeting treatments in Myotonic Dystrophy Type 1** **(Talk 4)**
Valeria Sansone - Centro Clinico Nemo and Università degli Studi di Milano

17.00 - 17.30 **Flash poster presentations**
The selected poster presenters will have the chance to give a Flash Talk presentation of 3-minutes to "advertise" their work prior to the poster session.

17.30 - 18.00 *Coffee break*

18.00 - 19.15 **POSTER SESSION 1**

19.15 - 20.15 **SCIENCE AND SOCIETY - WHOSE DATA ARE MY DATA? SHARING AND PROTECTING PERSONAL HEALTH DATA** **(Talk 5)**
An interactive session addressing patients' needs, concerns and expectations on personal health data and illustrating key principles relevant to health data management in research.
Moderator: Lucia Monaco - Research Impact and Strategic Analysis, Fondazione Telethon
Sandra Courbier - EURORDIS, Paris
Michela Maggi - Fondazione Telethon Data Protection Officer (DPO), Studio Legale Maggi, Milano

20.15 - 21.30 *Welcome buffet*

Tuesday, October 29th 2019

08.30 - 09.00 Registration and poster setting up

09.00 - 11.00 **PLENARY SESSION 2 - NEW THERAPEUTIC AVENUES FOR MITOCHONDRIAL DISEASES**
Different approaches to tackle mitochondrial diseases will be presented and discussed.
Chairs:
Giorgio Casari - TIGEM, Pozzuoli (Napoli) and Università Vita Salute San Raffaele, Milano
Gabriele Siciliano - Clinica Neurologica, Università degli Studi di Pisa

09.00 - 9.15 **Introduction**

9.20 - 9.40 **Omics approaches to improve diagnostics (and optimize treatment) for patients with mitochondrial disease**
Daniele Ghezzi - Fondazione IRCCS Istituto Neurologico "C. Besta", Milano **(TALK 6)**

9.40 - 10.00 **Mitochondrial disorders: from gene discovery to pathomechanisms and experimental therapy**
Massimo Zeviani - Università degli Studi di Padova **(TALK 7)**

10.00 - 10.20 **miR-181a and miR-181b downregulation ameliorates mitochondrial-associated neuro-degeneration by enhancing mitochondrial biogenesis and mitophagy**
Brunella Franco - TIGEM, Pozzuoli (Napoli) **(TALK 8)**

10.20 - 10.40 **Mitochondria as signalling hubs in neurodegeneration**
Rosario Rizzuto - Università degli Studi di Padova **(TALK 9)**

10.40 - 10.50 **Flash Talk - The 'Cell line and DNA Bank of Genetic Movement Disorders and Mitochondrial Diseases' of the TNGB as a key resource for research**
Barbara Garavaglia - Fondazione IRCCS Istituto Neurologico "C. Besta", Milano

11.00 - 11.30 *Coffee break*

11.30 - 13.00 **POSTER SESSION 2** - Meeting Patient Organizations
This poster session will be shared with Patient Organizations who were informed of the posters of interest. A useful slot for connecting Researchers and Patients.

13.00 - 14.00 *Buffet lunch*

14.00 - 14.15 **Fondazione Telethon partners with Patient Organizations in a Seed Grant Special Program**
Alessandra Camerini, Patient Associations Relationship - Fondazione Telethon
Manuela Battaglia, Head of Research - Fondazione Telethon

14.15 - 15.30 **FOCUS - INTERNATIONAL ACTIVITIES AT FONDAZIONE TELETHON**
International partnerships and European projects on research infrastructures, policy recommendations and strategic research agendas: why to engage and the potential impact for a Researcher.
Moderator: Graciana Diez Roux - TIGEM, Pozzuoli (Napoli)
RESTORE - Hans-Dieter Volk, BCRT and Charité - Universitätsmedizin Berlin, Germany
EJP rare diseases - Daria Julkowska, Inserm, Paris, France
IRDiRC - Lucia Monaco, Fondazione Telethon and IRDiRC
RARE 2030 - Stefano Benvenuti, Fondazione Telethon
ENMC - Anna Ambrosini, Fondazione Telethon

15.10 - 15.30 **Roundtable**

15.30 - 17.30 **PLENARY SESSION 3 - GOING TRANSLATIONAL**
The path towards therapy development: what it takes, the tools available and some good news.
Chair: Michael Caplan - Yale University (CT, USA)

15.30 - 15.40 **Introduction**

15.40 - 16.00 **What it takes to go translational**
Dieter Volk - BCRT and Charité - Universitätsmedizin Berlin, Germany

16.00 - 16.10 **The Sofinnova - Telethon fund: a new opportunity**
Annamaria Merico - Fondazione Telethon

16.10 - 16.30 **Orphan drug development guidebook: the IRDiRC toolkit to increase efficiency in the development of new treatments for rare disease**
Diego Ardigò - Chiesi Farmaceutici, Parma

16.30 - 16.50 **Artificial Intelligence in Biomedical Research: where we are and where we are going**
Robert Alexander - IBM Healthcare & Life Science, Roma

Chair: Robertson Parkman - Standford University (CA, USA)

16.50 - 17.00 **Update on clinical trials**
Alessandro Aiuti - SR-TIGET, Milano
Nicola Brunetti Pierri - TIGEM, Pozzuoli (Napoli)

17.00 - 17.30 **A focus on mucopolysaccharidoses (MPS) I**
Bernardo Maria Esther and Bernhard Gentner - SR-TIGET, Milano

17.30 - 18.30 ***Poster Session*** *& Coffee Break*

18.30 - 19.30 SCIENCE AND SOCIETY - ACCESSIBILITY AND SUSTAINABILITY OF ADVANCED THERAPIES
The roadblocks between EMA positive opinion for marketing authorization and patients: analysis of challenges, possible solutions and pioneers' experiences.
Moderator: Francesco Macchia - Osservatorio Terapie Avanzate
Diego Ardigò - Chiesi Farmaceutici, Parma
Christos Sotirelis - Patient Expert in the Committee for Advanced Therapies (CAT), European Medicines Agency (EMA)
Simona Paratore - Novartis Oncology

Roundtable

Wednesday, October 30th 2019

9.00 - 10.30 FOCUS - GOING BEYOND ANIMAL EXPERIMENTATION: WHEN AND WHERE
Cutting-edge alternatives to animal experimentation: are we there yet?
Moderator: Manuela Battaglia - Fondazione Telethon

9.00 - 9.20 **Let's hear from the Telethon-Institutes' Directors**
Andrea Ballabio - TIGEM, Pozzuoli (Napoli)
Luigi Naldini - SR-TIGET, Milano

9.20 - 9.40 **How can C. elegans worm your way into the study of Genetic diseases**
Elia Di Schiavi - CNR, Napoli **(Talk 10)**

9.40 - 10.00 **One by one: convergence, multiplexing and single-cell resolution in the study of neuro-developmental disorders through brain organoids**
Giuseppe Testa - Università degli Studi di Milano - Istituto Europeo di Oncologia, Milano **(Talk 11)**

10.00 - 10.20 **Organs-on-Chips: the promises and limits of microfluidics**
Diego Di Bernardo - TIGEM, Pozzuoli (Napoli) **(Talk 12)**

10.20 - 10.30 **Concluding remarks: MANIFESTO "Salviamo la ricerca Biomedica" and next steps**
Giuliano Grignaschi - Research4life, IRCCS Istituto M. Negri, Milano

10.30 - 11.00 Coffee break

11.00 - 13.00 PLENARY SESSION 4 - Fighting blindness: an eye on new therapeutic approaches and challenges
Novel and various approaches to tackle eye diseases will be presented and discussed.
Chair: Sandro Banfi - TIGEM, Pozzuoli (Napoli)

11.00 - 11.15 **Introduction**

11.20 - 11.40 **Expanding AAV transfer capacity in the retina**
Alberto Auricchio - TIGEM, Pozzuoli (Napoli) **(Talk 13)**

11.40 - 12.00 **Targeting common cell death pathways for the neuroprotection of degenerating photo-receptors**
Valeria Marigo, Università degli Studi di Modena e Reggio Emilia, Modena **(Talk 14)**

12.00 - 12.20 **Cone dystrophies and retinal degeneration from protein structures to biological networks**
Daniele Dell'Orco, Università degli Studi di Verona **(Talk 15)**

12.20 - 12.40 **Inhibition of autophagy curtails visual loss in a model of autosomal dominant optic atrophy**
Luca Scorrano, Università degli Studi di Padova **(Talk 16)**

12.40 - 12.50 **Flash Talk** - **Fondazione Telethon and the Unit of Advanced Therapies for Hereditary Ocular Diseases**
Stefano Zancan, Fondazione Telethon

13.00 - 13.30 LATE BREAKING NEWS
Authors of just-published important results will have the chance to present their data.

13.30 - 13.45 CLOSING REMARKS
Manuela Battaglia - Head of Research, Fondazione Telethon

"Fondazione Telethon e le Associazioni di pazienti: un percorso insieme"

28 OTTOBRE 2019

10.00 - 13.45 *Registrazione dei partecipanti e ritiro cuffie per traduzione simultanea*

14.00 - 15.00 **SESSIONE PLENARIA (Sala GARDA)**
Benvenuto e apertura dei lavori
Francesca Pasinelli, Direttore Generale di Fondazione Telethon
Manuela Battaglia, Direzione Scientifica di Fondazione Telethon

15.00 - 17.00 **WORKSHOP CON LE ASSOCIAZIONI (Sala DOLOMITI)**
Moderatore: *Alessandra Camerini*, Relazioni con Associazioni di pazienti di Fondazione Telethon

Fondazione Telethon e le Associazioni di pazienti: un percorso insieme
Francesca Pasinelli, Direttore Generale di Fondazione Telethon

Eccellenza, trasparenza e innovazione: le tre caratteristiche della ricerca da finanziare
Manuela Battaglia, Direzione Scientifica di Fondazione Telethon

Valorizzazione del percorso e dei risultati della ricerca
Annamaria Merico, Trasferimento Tecnologico di Fondazione Telethon
Simona Varani, Proprietà Intellettuale di Fondazione Telethon

Discussione

17.00 - 17.10 **UN SALUTO DALLA FONDAZIONE (Sala DOLOMITI)**
Omero Toso, Vice Presidente di Fondazione Telethon

17.10 - 17.20 **PRESENTAZIONE GAME "WORLD CAFE'– COSTRUIAMO UN PERCORSO INSIEME" (Sala DOLOMITI)**
Alessandra Camerini, Relazioni con Associazioni di pazienti di Fondazione Telethon

17.20 - 17.45 *Coffee break*

17.45 - 19.15 **"WORLD CAFE'– COSTRUIAMO UN PERCORSO INSIEME" (location varie)**

19.15 - 20.15 **SESSIONE PLENARIA (Sala GARDA)**
Di chi sono i miei dati? La condivisione e la protezione dei dati sanitari personali
Sandra Courbier, Eurordis, Paris
Michela Maggi, Data Protection Officer (DPO) di Fondazione Telethon
Lucia Monaco, Responsabile Centro Studi di Fondazione Telethon

20.15 - 21.15 *Welcome Cocktail*

29 OTTOBRE 2019

09.00 - 10.15 **WORKSHOP CON LE ASSOCIAZIONI (Sala DOLOMITI)**
Moderatore: *Annamaria Zaccheddu,* Content Management di Fondazione Telethon

Biobanche: avvertenze e modalità d'uso
Luca Sangiorgi, Coordinatore Reti Biobanche di Fondazione Telethon

Per uscire dal buio: il progetto *Malattie senza diagnosi*
Vincenzo Nigro, Partner fondatore del progetto "Malattie senza diagnosi"
Angelo Selicorni, Partner fondatore del progetto "Malattie senza diagnosi"

Discussione

10.15 - 11.15 **Restituzione risultati Game World Cafè e Costruiamo un percorso insieme: i prossimi passi (Sala DOLOMITI)**
Team dei facilitatori
Alessandra Camerini, Relazioni con Associazioni di pazienti di Fondazione Telethon
Manuela Battaglia, Direzione Scientifica di Fondazione Telethon

11.15 - 11.30 *Coffee break*

11.30 - 13.00 **POSTER SESSION: Incontro con i Ricercatori (Palameeting)**

13.00 - 14.00 *Pranzo*

Dalle 14.30 *Partenza navette*

Talk 1

Store-operated calcium entry (SOCE): role in skeletal muscle function and disease

Feliciano Protasi - Università degli Studi G. d'Annunzio Chieti, Pescara

Dysregulation of Ca2+ homeostasis is associated to several pathological conditions including neurodegenerative and skeletal muscle diseases. Store-operated Ca2+ entry (SOCE) is a ubiquitous Ca2+ entry mechanism, first described in non-excitable cells, that is triggered by depletion of intracellular Ca2+ stores (endoplasmic/sarcoplasmic reticulum, respectively ER and SR). SOCE is coordinated by the communication between: a) stromal interaction molecule-1 (STIM1), which acts as the Ca2+ sensor in the ER lumen, and b) Orai1, the Ca2+-permeable channel in the plasma membrane (PM). SOCE in skeletal muscle is similarly mediated by interactions between STIM1 and Orai1 channels and is proposed to limit muscle fatigue during repetitive stimulation. A reduction in SOCE activity was proposed to contribute to muscle impairment in aging and mutations in STIM1 and Orai1 have been linked to Tubular Aggregate Myopathy (TAM), an autosomal dominant muscle disease that is clinically characterized by myalgia, cramps and muscle stiffness, with or without proximal muscle weakness.
Despite the general agreement about the importance of SOCE in skeletal muscle function and disease, the precise subcellular sites of STIM1-Orai1 was not specifically investigated for about a decade. To give our contribute to this emerging field in muscle:

1. We discovered (using a combination of electron microscopy, immunofluorescence, immunogold, ex vivo muscle contractility, and Ca2+ imaging) that exercise promotes the formation of unique intracellular junctions that contain the molecular machinery required to activate SOCE (i.e. STIM1 and Orai1). We proposed that these not-previously-identified junctions function as Ca2+ entry units (CEUs), newly discovered organelles that i) promote recovery of extracellular Ca2+ during repetitive stimulation and ii) reduce/delay muscle fatigue optimizing muscle function.
2. As mutations in STIM1 and Orai1 have been linked to TAM (with the support of Telethon GGP19219), we are now investigating i) the mechanisms that lead to formation of Tubular Aggregates (TAs), and ii) the dysfunctional properties associated to their accrual in muscle fibers. With this goal in mind, we also generated new knock-in mice of the human disease (Orai1-G98S mice) and collected the first preliminary data.

Talk 2

Autophagy as a key pathogenic mechanism in COL6-related diseases and its significance for prospective therapies

Paolo Bonaldo, Dept. of Molecular Medicine, University of Padova

Collagen VI (COL6) is a distinctive extracellular matrix protein playing a remarkably major role in different cell processes[1]. At difference from other collagens, COL6 has a unique process of intracellular assembly involving multiple steps where different chains coded by separate genes give rise to large tetramers which once secreted form a branched network of beaded microfilaments in the matrix[1]. Mutations of COL6 genes are causative for a subclass of congenital Muscular dystrophies with a broad spectrum of clinical symptoms, collectively known as 'COL6-related myopathies', which include Bethlem Myopathy (BM) and Ullrich congenital Muscular Dystrophy (UCMD)[2].
Several years ago we generated a COL6 knockout (Col6a1–/–) mouse, whose phenotypical defects confirmed that COL6 has a critical role for muscle homeostasis and mutations. This mouse represents a valuable animal model of BM and UCMD, and its detailed characterization provided valuable information on the pathophysiological mechanisms underlying COL6-related myopathies[3-6]. Thanks to Telethon support, we demonstrated that lack of COL6 leads to organelle defects, with mitochondrial dysfunction and spontaneous apoptosis in Col6a1–/– muscle fibers. These studies allowed to understand that the mitochondrial defects are associated with an increased opening of the permeability transition pore, which can be desensitized by cyclosporin A treatment, leading also to an amelioration of muscle structure and function in mice[3]. Notably, work in patients' samples revealed similar defects[7], thus opening the way for targeted therapeutic approaches in COL6-related myopathies. A pilot clinical trial with cyclosporin A in BM and UCMD patients showed beneficial effects in muscle cells, leading to increased myofiber survival and muscle regeneration[8].
In further studies aimed at dissecting the pathomolecular mechanisms underlying COL6-related diseases, we showed that the autophagic machinery is impaired in muscles of Col6a1–/– mice and BM/UCMD patients, leading to the accumulation of dysfunctional mitochondria and organelles and to the subsequent myofiber defects[4]. Remarkably, reactivation of autophagy in Col6a1–/– mice by different approaches (e.g., prolonged starvation or low-protein diet) is beneficial for myofiber homeostasis, leading to improved muscle strength[4]. A recent pilot clinical trial, based on a 1-year low protein diet, confirmed that autophagy can be reactivated in BM and UCMD patients, with beneficial effects in counteracting the decline of functional parameters[9]. The great advantage of targeting autophagy relies on the fact that it is easily tunable by dietary means or by different nutraceuticals, such as resveratrol and spermidine (Spd). Along this line, we recently demonstrated that in vivo Spd administration to Col6a1–/– mice, by either i.p. injection or supplement to drinking water, leads to a significantly improved muscle homeostasis[10].
We now aim at understanding in detail the therapeutic efficacy of oral Spd administration, by testing different doses and regimens and monitoring their efficacy in ameliorating muscle structure and strength in Col6a1–/– mice, as well as by evaluating the effects of Spd treatment in patients' derived cells. These studies will prove the efficacy of Spd and autophagy-targeted nutraceutical approaches for COL6-related disorders, also in the perspective of their combinatorial use with mitochondria-targeted agents in the quest for the most effective and safe therapeutic strategies for these life-threatening diseases.

1 Cescon *et al.*, J. Cell Sci. 2015.
2 Bönnemann, Nature Rev. Neurol. 2011.
3 Irwin *et al.*, Nature Genet. 2003.
4 Grumati *et al.*, Nature Med. 2010.
5 Urciuolo *et al.*, Nature Comm. 2013.
6 Cescon *et al.*, Acta Neuropathol. 2018.
7 Angelin *et al.*, PNAS 2007.
8 Merlini *et al.*, PNAS 2008.
9 Castagnaro *et al.*, Autophagy 2016
10 Chrisam *et al.*, Autophagy 2015.

Talk 3

From reverse genetics to gene therapy: Duchenne Muscular Dystrophy

Alessandra Ferlini, MD, PHD
Medical Genetics Unit, Department of Medical Sciences,
University of Ferrara, Italy
Dubowitz Neuromuscular Unit, UCL, London, UK

Duchenne Muscular Dystrophy (DMD) is the most common childhood Muscular Dystrophy affecting ~ 1:5,000 live male births. Following the identification of the defective dystrophin gene in 1986 by reverse genetics, gene function, genotype/phenotype correlations and pathogenic mechanisms have been elucidated in skeletal, smooth and cardiac muscles as well as in the brain. Thanks to new high throughput methods, a DMD Genetic definition is now fully achievable and represents a requirement in order to have clinical diagnosis confirmation, family planning and preventive measures and clinical trials access.
Indeed, advances in the understanding of the molecular pathways affected in DMD have led to both the development of multiple therapeutic strategies tackling different aspects of disease pathogenesis and recently, the approval of first successful drugs for this condition. Antisense oligonucleotides, stop codon reversion and gene therapy are now a reality and have a crucial role in changing the natural history of the disease and ultimately, the whole lives of DMD-affected boys. An overview of the DMD gene story from discovery to new treatments will be presented, with a look into the future of oncoming therapeutic approaches and their wide repercussions in the neuromuscular disease field.

Talk 4

Cross-fertilization between motor neuron disorders and Muscular dystrophies: improving care and targeting treatments in Myotonic Dystrophy Type 1

Valeria A. Sansone - Centro clinico Nemo, Milano
The NEMO Clinical Center, Neurorehabilitation Unit,
University of Milan

Background: Motor neuron disorders and Muscular dystrophies are characterized by common features like muscle atrophy, weakness, fatigue and motor functional limitations. However, pathways of care and management may differ in many respects to the extent that a specific approach and decision process needs to be implemented to target better care and cure. The NEMO Center is a multidisciplinary tertiary center for the care and cure of different neuromuscular disorders, the most frequent being motor neuron disorders and the Muscular dystrophies. Myotonic dystrophies (DM1) represent 25% of the patients at the site. The diagnostic and management protocols are targeted for this patient population.
Aims: To discuss how cross-fertilization between motor neuron disorders and Muscular dystrophies may improve care and contribute to targeted treatments in DM1 while creating the basis for trial readiness and endpoint assessments.
Methods: Ventilatory support, nutrition protocols and motor function assessments used in ALS and the Muscular dystrophies will be described and the way these have been adapted to the care of DM1 patients will be discussed. Ongoing observational studies in both the adult and congenital and pediatric variants of DM1 will be presented as part of national and international networks. An update on targeted treatments and future therapeutic trials in this field will be discussed as well as the lessons learned from the experience with innovative therapies applied to the SMA field.
Clinical Relevance: DM1 is the most common form of adult Muscular Dystrophy and is perhaps the most variable amongst the different diseases in medicine, ranging from a congenital presentation, to a pediatric or adult onset for to a late-onset form. Multiple organs are involved, clinical presentation varies widely and death usually occurs between the 5th and 6th decade of life. There is still a significant diagnostic delay despite a blood draw is sufficient to identify a CTG repeat expansion > 50 which is associated with the disease.
Conclusions: There are upcoming drugs which may potentially target muscle tissue or the abnormal expansions. It is mandatory to identify appropriate outcome measures in preparation for clinical trials to improve care and target treatments in DM1.

Talk 5

Whose data are my data? Sharing and protecting personal health data

Sandra Courbier - Rare Barometer survey programme
Senior Manager, EURORDIS, Paris
Michela Maggi - Data Protection Officer,
Fondazione Telethon, Milan
Moderation by Lucia Monaco, Head, Research Impact
and strategic analysis, Fondazione Telethon

Personal health data are a core resource for biomedical research, clinical care and patient management. Sharing health data with and among researchers and healthcare professionals is key to shortening the time to diagnosis, advancing knowledge on the disease, and progressing towards the identification and development of care and therapies.
This is particularly true for rare Genetic diseases, which require matching and comparing data from affected people scattered across the globe, in order to take full advantage from the bioinformatics revolution.
Privacy protection, data stewardship, and compliance with legal regulations require engagement, competence and commitment by all parties involved.
This interactive session with the audience of patient representatives and researchers will address needs, concerns and expectations expressed by the patients' community, as well as the key principles of the European regulation on data privacy relevant to the management of health data in the research setting.

Talk 6

Omics approaches to improve diagnostics (and optimize treatment) for patients with mitochondrial disease

Daniele Ghezzi - Fondazione I.R.C.C.S Istituto Neurologico
"C. Besta"

Mitochondrial disorders (MD) are a genetically heterogeneous group of individually rare human diseases characterized by energy deficiency due to mitochondrial dysfunction. MD may result from pathogenic mutations of the mitochondrial or nuclear DNA, affecting components or key factors of the oxidative phosphorylation system responsible for ATP production. MD

typically are multi-organ diseases affecting high-energy demand tissues such as muscle, brain and liver. Their multi-system presentation, together with their complex Genetic bases, makes molecular diagnosis difficult.
The introduction of next generation sequencing has dramatically improved diagnostic yield for MD. Nevertheless, about half of MD patients still remain without molecular diagnosis despite whole exome sequencing. More recently, additional "omics" approaches (whole genome sequencing, transcriptomics, proteomics) have been considered to investigate unsolved cases and have been proven to increase the percentage of Genetic diagnoses, as well as to be useful for identifying new disease-genes.
Genetic confirmation of MD and the identification of the exact molecular defect are important for patients/families to remove uncertainty and end their diagnostic odyssey, to guide Genetic counseling and family planning, but they can also be fundamental for treatment. Although an effective therapeutic strategy is still missing for most of MD, a growing subgroup is amenable to treatment with cofactors (e.g. riboflavin in patients with ACAD9 deficiency); a rapid and precise diagnosis is thus crucial for these subjects.
Integration of multiple "omics" data will allow a more comprehensive view of human diseases. In addition to improve diagnosis, "omics" are expected to guide treatment and will likely become the starting point for personalized medicine.

Talk 7

Mitochondrial disorders: from gene discovery to pathomechanisms and experimental therapy

Massimo Zeviani - University of Padova, Department of Neurosciences, Padova, Italy

Mitochondria are the major source of ATP that is synthesized by the respiratory chain through the process of oxidative phosphorylation (OXPHOS), a complex biochemical process carried out through the dual control of physically separated, but functionally interrelated, genomes, nuclear and mitochondrial DNAs. The Genetic and biochemical intricacy of mitochondrial bioenergetics explains the extreme heterogeneity of mitochondrial disorders, a group of highly invalidating human conditions, for which no effective treatment is nowadays available. In addition to bioenergetic failure, other mechanisms are probably predominant in the pathogenesis of specific syndromes, such as alterations of cellular redox status, the production of reactive oxygen species, compromised Ca^{2+} homeostasis, mitochondrial protein and organelle quality control, and mitochondrial pathways of apoptosis. By investigating selected families and patients, we have identified several new disease genes, each responsible of distinct defects of the respiratory chain, mtDNA metabolism, or both, associated with paediatric or adult-onset clinical presentations. Structural analysis and the creation of ad hoc recombinant lines in yeast, flies, and mice have allowed us to dissect out the molecular consequences of the ablation or defects of some of these proteins, and their physical status in normal and disease conditions. These models have also been exploited to implement experimental therapeutic strategies, based on gene and cell replacement, or pharmacological control of mitochondrial biogenesis. For instance, coordinated increase of autophagy and lysosomal clearance based on inhibition of mTORC1 by rapamycin is effective to markedly prolong survival in OXPHOS impairment of brain or skeletal muscle. In addition, editing of mtDNA in a mutant mouse has been successfully achieved in our Unit through zinc-finger recombinant technology, opening the possibility to the controlled reduction of heteroplasmic load in vivo. Finally the use of new AAV vectors in vivo to convey therapeutic genes warrants promising developments for effectively crossing the BBB and targeting the CNS in mitochondrial encephalopathies.

Talk 8

miR-181a and miR-181b downregulation ameliorates mitochondrial-associated neurodegeneration by enhancing mitochondrial biogenesis and mitophagy

A. Indrieri[1,2], S Carrella[1], A. Spaziano[1], A Romano[1], E. Fernandez-Vizarra[3], S. Barbato[1], M. Zeviani[3], EM. Surace[2], E. De Leonibus[1], S. Banfi[1], B. Franco[1,2]

[1] Telethon Institute of Genetics and Medicine, Pozzuoli, Italy
[2] Department of Translational Medical Science, University of Naples "Federico II", Naples, Italy
[3] MRC Mitochondrial Biology Unit, University of Cambridge, Cambridge, United Kingdom

Mitochondrial dysfunction underlies the pathogenesis of a variety of human neurodegenerative diseases, either directly, in the case of rare mitochondrial diseases (MDs), or indirectly, as in more common neurodegenerative disorders, such as Parkinson's disease (PD). Despite the efforts, effective therapies are still not available for these devastating conditions. We demonstrated that microRNAs miR-181a and miR-181b (miR-181a/b) regulate key genes involved in mitochondrial biogenesis and function. We also showed that these miRNAs are involved in global regulation of mitochondrial turnover in the central nervous system through the coordinated activation of mitochondrial biogenesis and mitophagy. We thus tested whether the modulation of these miRNAs could be therapeutically exploited in neurodegenerative conditions associated with primary impairment of mitochondrial activity. We first showed that miR-181a/b downregulation effectively protects neurons from cell death and significantly ameliorates the disease phenotype in different animal models of MDs, such as two medakafish models of Microphthalmia with Linear Skin Lesions, and chemical and Genetic models of Leber Hereditary Optic Neuropathy[1]. In addition, our preliminary data also demonstrated amelioration of the disease phenotype in a mouse model of Leigh Syndrome, an often-fatal MD characterized by severe neurodegeneration. We then tested whether miR-181a/b downregulation could also be effective in chemical models of secondary mitochondrial dysfunction. To this aim we generated medakafish and murine models of PD using the neurotoxin 6-OHDA, which is widely used for this purpose. Our data demonstrate that inactivation of miR-181a/b reduces the extent of nigrostriatal dopaminergic neurons death in both models and results in improved motor performances in the mouse PD model. Altogether our results indicate that miR-181a/b act as hubs in mitochondrial homeostasis in the central nervous system. We propose these miRNAs may represent novel gene-independent therapeutic targets for a wide-range of neurodegenerative disorders caused by mitochondrial dysfunction.

1. Indrieri A., Carrella S., Romano A., Spaziano A., Marrocco E., Fernandez-Vizarra E., Barbato S., Pizzo M., Ezhova Y., Golia FM., Ciampi L., Tammaro R., Henao-Mejia J., Williams A., Flavell RA., De Leonibus E., Zeviani M., Surace EM., Banfi S., Franco B. EMBO Mol Med. 2019 May;11(5). pii: e8734. doi: 10.15252/emmm.201708734.

Talk 9

Mitochondria as signaling hubs in neurodegeneration

Beatrice D'Orsi[1], Luisa Galla[1,2], Elisa Greotti[1,2], Edward Beamer[3], Mariana Alves[3], Tobias Engel[3], Paola Pizzo[1,2], Diego De Stefani[1], Tullio Pozzan[1,2,4] and Rosario Rizzuto[1]

[1] Department of Biomedical Sciences, University of Padova, Padova, Italy.
[2] Neuroscience Institute - Italian National Research Council (CNR), Padova, Italy;
[3] Department of Physiology & Medical Physics, Royal College of Surgeons in Ireland, Dublin 2, Ireland.
[4] Venetian Institute of Molecular Medicine, Padova, Italy.

In Alzheimer 's disease (AD), the role of Genetic mutations in the pathogenesis is firmly established, despite their role in determining neuronal dysfunction and death is still unknown. Mitochondrial Ca^{2+} overload has been proposed as the no-return signal triggering neuronal death, and we have demonstrated that familial AD (FAD) due to PS2 mutations favors Ca^{2+} transfer from the endoplasmic reticulum (ER) to mitochondria. Recently, the molecular nature of the mitochondrial Ca^{2+} channel was unveiled, allowing the investigation of the role of mitochondrial Ca^{2+} dysregulation by new Genetic tools. Our final goal is to test the genuine contribution of mitochondrial Ca^{2+} overload to FAD pathogenesis.
To do this, we compared mRNA profiles of the neurotransmitters, cell death pathways and MCU complex (MCUC) components between wild Type (WT) and PS2-N141I/APPswe (PS2APP) mouse brains, during disease progression. We included PS2KO mice to test if the hypothesis of a loss-of-function phenotype associated to FAD-PS1 mutations Cn be extended also to FAD-PS2 mutations. PCR arrays reveal an early transcriptional impairment in PS2APP and PS2KO brains, with a non-overlapping profile between them. An early remodelling of the MCU complex was also evident, with a significant up-regulation of the MCUC enhancers. Given the transcriptional remodelling of excitatory neurotransmission, cytosolic Ca^{2+} dynamics of hippocampal slices have been studied. An altered NMDA-induced Ca^{2+} signalling is evident in 1.5-month-old PS2APP and PS2KO mice. Mitochondrial Ca^{2+} handling is currently under investigation.
As mentioned above, we found increased mRNA levels of genes involved in cell death, together with an up-regulated expression of MCU enhancers. We manipulated MCU protein levels to investigate how mitochondrial Ca^{2+} handling controls neuronal death. Since only *MCU*$^{+/-}$ mice are viable and fertile with no evident phenotype, we employed primary neuronal cultures from *MCU*$^{+/+}$ and *MCU*$^{+/-}$ mice. The latter display a decreased mitochondrial Ca^{2+} uptake and neuronal death in response to NMDA-induced excitotoxicity. We could not detect a decreased cell death when neurons were exposed to a milder and transient NMDA stimulus. In line with this, increasing mitochondrial Ca^{2+} levels by overexpressing of MCU is *per se* sufficient to cause neuronal death *in situ* and to trigger gliosis and neuronal loss *in vivo*. Accordingly, *MCU*$^{+/-}$ mice were more resistant to excitotoxicity *in vivo*, protecting neurons from kainite acid-induced injury (a model of epilepsy).
In summary, our results suggest that a substantial rearrangement of gene expression occurs early in PS2APP and PS2KO mice, especially of those involved in Ca^{2+} homeostasis and cell death regulation, with no evidence of a loss-of function phenotype associated to FAD-PS2 mutations. Furthermore, we provided important new insights into the role of MCU in neuronal excitotoxicity both *in situ* and *in vivo*.

Talk 10

How can C.elegans worm your way into the study of Genetic diseases

Elia Di Schiavi - Institute of Bioscience and BioResources, IBBR, CNR, Naples

Studying Genetic diseases in animal models has been crucial to understand human disease pathogenesis, the function played by mutated genes and to identify potential therapies. Among the most diffused animal models, invertebrates such as C.elegans, allowed rapid analyses of the molecular mechanisms leading to diseases and the identification of new potential therapeutic targets in several diseases (e.g. SMA, obesity, Huntington) (Ashrafi *et al*., Nature 2003; Grice *et al*., BioEssays 2011; Parker *et al*., Nature genetics 2005). Moreover C.elegans lead to the discovery of basic processes that unexpectedly became fundamental to set new strategies to cure human diseases (e.g. RNA-interference, miRNAs, apoptotic pathway). This has been acknowledged by the Nobel Prize awarding Institutions that awarded several times the prize to researchers working with C.elegans, including prizes for Medicine. The use of C.elegans as a model for human diseases provides: a) a powerful, easy and rapid system to directly assess the consequences of mutations at the organismal level, in vivo; b) the unique advantage of visualizing individual cells in living transparent animals; c) more than 70% of disease genes presenting an ortholog. Importantly, the use of C.elegans allows to strongly reducing the number of vertebrate animals used, fulfilling the 3Rs principles (Replacing the use of mammals; Reducing the number of mammals used to a minimum; Refining the way experiments are carried out). Moreover, the use of an invertebrate model has few ethical concerns for the public and for private foundations donors and is highly supported by EU (Resolution on the protection of animals used for scientific purposes, 5/05/2009) and Italian legislation (DL N°26, March 4th, 2014). These advantages together with the small dimensions (1 mm), the high rate of fertility and hermaphroditism (300 isogenic progeny per animal) and very cheap costs, have recently caused an expansion of its use also to high troughput screenings for toxicological studies (NIEHS National Toxicology Program), to improve diagnosis and care of patients with undiagnosed diseases (NIH Undiagnosed Diseases Network) and for drug discovery. The work from several C.elegans laboratories, including Telethon Grantees, will be presented to demonstrate the power of C.elegans to study Genetic diseases and to show how "As incredible as it seems, future research on flies and worms will quite often provide the shortest and most efficient path to curing human disease" (Alberts, Science 2010).

Talk 11

One by one: convergence, multiplexing and single-cell resolution in the study of neurodevelopmental disorders through brain organoids

Giuseppe Testa - Università degli Studi di Milano and Istituto Europeo di Oncologia (IEO), Milano

Talk 12

Organs-on-Chips: the promises and limits of microfluidics

Diego Di Bernardo - Telethon Institute of Genetics and Medicine (TIGEM), Pozzuoli (Napoli)

Animal models recapitulate human diseases and are still necessary to transform lab-based discovery into actual therapies for patients, starting from basic research into disease mechanisms, going into proof-of-principle studies and culminating in pre-clinical studies prior to clinical trials in human patients. In the course of the presentation I will illustrate efforts ongoing world-wide to reduce the need of animal models in biomedical research, including alternative models based on organ-on-chip and organoids and their advantages and current limitations.

Talk 13

Expanding AAV transfer capacity in the retina

Alberto Auricchio, MD - Telethon Institute of Genetics and Medicine (TIGEM) and Medical Genetics, Dept. of Advanced Biomedicine, University of Naples “Federico II”, Italy

Inherited retinal degenerations (IRDs) are a major cause of blindness worldwide. In vivo retinal gene therapy with adeno-associated viral (AAV) vectors has emerged as an effective and safe strategy to counteract retinal neurodegeneration associated with IRDs. Indeed the first approved gene therapy product for an ocular disease is based on AAV. However, the DNA cargo capacity of AAV vectors is limited to about 5 kb which precludes their application to IRDs due to mutations in genes with a coding sequence (CDS) larger than 5 kb, e.g. Stargardt disease or Usher syndrome Type IB. To overcome this limitation, we have developed two different strategies based on the co-delivery of two AAV vectors each containing one half of a large gene CDS. In one strategy, recombination occurs between the genomes of the two AAV vectors leading to the reconstitution of a single large expression cassette. In another strategy, the two AAV vectors separately encode for the two half polypeptides of large protein which undergo protein trans-splicing mediated by split inteins which results in reconstitution of the full length protein. Advantages and limitations of each system will be discussed as well as their application to gene therapy of IRDs.

Talk 14

Targeting common cell death pathways for the neuroprotection of degenerating photoreceptors

Marigo Valeria[1], Comitato Antonella[1], Subramanian Preeti[2], Becerra, S. Patricia[2]

[1] Department of Life Sciences, University of Modena and Reggio Emilia
[2] National Institute of Health, National Eye Institute, USA

Retinitis pigmentosa (RP) is a form of retinal degeneration (RD) and a major cause for legal blindness during working age. Over 70 different genes have been associated with RP. This Genetic heterogeneity has hampered the development of therapeutic interventions, but therapies based on neuroprotection, targeting common denominators activated in different forms of RD, could benefit a large cohort of patients. Pigment epithelium-derived factor (PEDF) is a natural protein in the eye with potent retinoprotective properties and high potential to be applied in retinal degeneration therapeutics.
We have characterized the molecular pathways targeted by PEDF in murine models of RP and found that PEDF acts on PMCA calcium pumps, present at the plasma membrane of rod photoreceptors, facilitating the decrease of intracellular calcium, a key player in photoreceptor cell death ([1]). Reduced levels of intracellular calcium limit the activation of calpains, calcium regulated proteases, that during degeneration activate Bax and the Apoptosis Inducing Factor (AIF) as triggers of photoreceptor cell death ([2]). We also identified a small protein domain (17 amino acids) in the PEDF molecule that mediates the neuroprotective activity of the factor ([3]).
Given that smaller molecules can be more permeable and facilitate delivery with limited side effects, likely caused by other regions of the entire molecule, we tested mutagenized small peptides, derived from the neurotrophic region of PEDF, that retain binding affinity for PEDF receptor but increase the neuroprotective activity. This study identified a small peptide of 17 amino acids, with a mutation in the histidine 105 into an alanine (17mer[H105A]), with enhanced neuroprotective activity compared to PEDF ([3]). We have delivered the 17mer[H105A] in the retina of murine models of RP via an AAV vector, a virus recently approved for gene therapy in the eye. The neuroprotective effects of intravitreal or subretinal injections of the therapeutic virus were analyzed histologically and electrophysiologically.

1. Comitato A., Subramanian P., Turchiano G., Montanari M., Becerra S.P., Marigo V. (2018) Pigment epithelium-derived factor (PEDF) hinders photoreceptor cell death by reducing intracellular calcium in the degenerating retina. Cell Death & Disease 9: 560.
2. Comitato A., Schiroli D., Montanari M., Marigo V. (2019) Calpain Activation Is the Major Cause of Cell Death in Photoreceptors Expressing a Rhodopsin Misfolding Mutation. Molecular Neurobiology, in press.
3. Kenealey J., Subramanian P., Comitato A., Bullock J, Keehan L., Polato F., Hoover D., Marigo V., Becerra S.P. (2015) Small Retinoprotective Peptides Reveal a Receptor Binding Region on Pigment Epithelium-derived Factor. Journal of Biological Chemistry 290:25241-25253.

Talk 15

Cone dystrophies and retinal degeneration from protein structures to biological networks

Daniele Dell’Orco - Department of Neurosciences, Biomedicine and Movement Sciences Section of Biological Chemistry University of Verona

Cone Dystrophy (COD) is a severe form of retinal disorder affecting photoreceptors, the cells where the visual signal originates. Common symptoms include decreased central and color vision and photophobia. In several patients, cone degeneration is followed by that of rods (CORD), which results in the progressive loss in peripheral vision. Currently, no cure exists for CORD, which affects 1 in 40,000 people. To date, up to 20 missense mutations in GUCA1A, the gene encoding the calcium sensor guanylate-

cyclase-activating protein (GCAP1) have been associated with autosomal dominant COD/CORD. The consequence of alterations in GCAP1 have been only partly explored and mechanisms leading to the onset of the disease remain largely unclear, although a connection with the dysregulation of intracellular cGMP and Ca^{2+} homeostasis has been established. Human GCAP1 variants associated with COD/CORD and their interaction with the target GC were thoroughly characterized by biochemical, biophysical and electrophysiological approaches, which have all been integrated by computer simulations. Under physiological conditions GCAP1 presents a dynamic monomer-dimer equilibrium that renders its crystallization process particularly tricky. SAXS studies corroborated by protein docking simulations allowed the building of a three-dimensional model of the GCAP1 dimer. A thorough structural and functional characterization was performed of the previously known COD-associated variants affecting the Ca^{2+} binding sites, namely p.E155A/G and p.D100G. All variants show a constitutive activation of the GC target at physiological concentrations of Ca^{2+} and altered affinity for Ca^{2+}. Finally, a novel GCAP1variant (p.E111V) associated with a severe form of CORD has been identified in an Italian family and the protein has been fully characterized.
Nano-sized liposomes with lipid composition mimicking that of photoreceptor outer segment were produced and their biodistribution was investigated in mouse retina both ex-vivo and following intra-vitreal injections. The liposomes fuse with retinal membranes and reach all layers including photoreceptor outer segments. When encapsulated with E111V-GCAP1 and delivered in vivo and ex vivo, liposomes perturbed the photoresponses of mouse photoreceptors in a way consistent with numerical simulations of the phototransduction cascade, thus opening the way to powerful tools for testing protein therapeutics hypotheses based on in vivo delivery of recombinant wild-Type protein.

Talk 16

Inhibition of autophagy curtails visual loss in a model of autosomal dominant optic atrophy

Luca Scorrano - University Padova

In Autosomal Dominant Optic Atrophy (ADOA) caused by mutations in the mitochondrial cristae biogenesis and fusion protein Optic Atrophy 1 (Opa1), retinal ganglion cell (RGC) dysfunction and visual loss occur by unknown mechanisms. Here we show an unexpected role for autophagosome accumulation at RGC axonal hillocks in ADOA pathogenesis. Expression of mutated Opa1 in RGCs causes heterogenous mitochondrial dysfunction and triggers AMPK- and tubulin acetylationdependent autophagosome accumulation at axonal hillocks, reducing axonal mitochondrial content. Pharmacological or Genetic inhibition of this pathway restores axonal mitochondrial content and curtails apoptosis caused by mutated Opa1. In *C. elegans*, deletion of AMPK or of key autophagy genes rescues axonal mitochondrial content reduced in neurons where mitochondrial dysfunction was induced. In conditional, RGC specific *Opa1*-deficient mice, depletion of the essential autophagy gene *Atg7* normalizes the excess autophagy and corrects the visual defects caused by *Opa1* ablation. Thus, axonal hillock accumulation of autophagosomes is a conserved mechanism crucial for ADOA pathogenesis.

VI CONVEGNO DELLE ASSOCIAZIONI AMICHE
PRESENTAZIONI

Fondazione Telethon e le Associazioni di pazienti: un percorso insieme

Francesca Pasinelli
Direttore Generale di Fondazione Telethon

Telethon nasce nel 1990 dalla volontà di una comunità di pazienti e familiari, assumendosi un ruolo di responsabilità sociale, dove la valorizzazione della ricerca coincide con la valorizzazione del paziente.
Il progetto collettivo che porta avanti da allora, si fonda su un ecosistema in cui cooperano diversi portatori di interesse, che condividono valori e intenti per un comune obiettivo: la cura delle malattie genetiche rare.
A guidare l'operato della Fondazione è la volontà di far sì che i pazienti si sentano garantiti da una ricerca di qualità, che i donatori sappiano come sono investiti i loro soldi e che i ricercatori siano valutati e sostenuti per competenza e impegno.
Per assicurare il mantenimento di un corretto equilibrio tra questi diversi attori, Telethon garantisce la trasparenza e l'autonomia di ciascun soggetto rispetto agli altri, in tre ambiti fondamentali: nel sistema di finanziamento (che assicura la giusta distanza tra chi chiede, chi decide e chi eroga), nelle strategie operative (dove nessuna pressione politica o commerciale deve condizionare le scelte e gli obiettivi), nel rispetto delle regole della scienza (che impongono qualità, rigore, pazienza e costante confronto internazionale, evitando le promesse di soluzioni miracolose e immediate).
Fondazione Telethon mira quindi alla creazione di alleanze e di partnership coi pazienti, con le aziende farmaceutiche e con le istituzioni regolatorie, cercando di ampliare sempre di più il tavolo di lavoro e creando un modello condiviso da parte di tutti gli attori, per ottimizzare il processo e definire modalità efficaci che permettano anche di gestire fattori limitanti (quali tempo e denaro). Vuole infatti trovare terapie e renderle sostenibili, per entrare in una dimensione fattuale dove, negoziando adozioni di un nuovo sviluppo regolatorio, si possa ridurre il percorso necessario per arrivare alla diagnosi, all'individuazione delle terapie e alla loro messa a disposizione dei pazienti, pur mantenendo inalterata la qualità.

Eccellenza, trasparenza e innovazione: le tre caratteristiche della ricerca da finanziare

Manuela Battaglia
Direzione Scientifica di Fondazione Telethon

Fondazione Telethon è un ente senza scopo di lucro – riconosciuto dal Ministero dell'Istruzione, dell'Università e della Ricerca – che si impegna ogni giorno per fare avanzare la ricerca biomedica verso la cura delle malattie genetiche rare che, proprio per la loro rarità, sono trascurate dai grandi investimenti pubblici e industriali.
Per essere efficace la Fondazione basa la propria strategia su un metodo rigoroso per selezionare le migliori idee, sostenere le attività di ricerca e tradurre i risultati raggiunti in vantaggi concreti per i pazienti. A questo processo è applicato un sistema certificato di gestione della qualità che rappresenta un modello unico tra gli Enti che finanziano ricerca in Italia.
Fondazione Telethon utilizza un metodo di finanziamento che si basa sulla valutazione dell'eccellenza scientifica affidata ad un gruppo di esperti di caratura internazionale: un gruppo di 30 ricercatori (i.e., la commissione medico scientifica) che si avvale a sua volta dell'aiuto specialistico di oltre 8000 scienziati. Fondazione Telethon, con l'impiego di una squadra dedicata in modo permanente, garantisce competenza e indipendenza nella gestione del processo di valutazione. L'elevata qualità scientifica della ricerca finanziata da Fondazione è provata dai risultati scientifici finora ottenuti che sono riconosciuti a livello internazionale e che hanno avuto un impatto decisivo sulla vita di pazienti provenienti da tutto il mondo. Questi risultati concreti confermano anche la bontà dei sistemi di selezione adottati.
Grazie al modello implementato, oggi Fondazione Telethon è riconosciuta come una delle realtà che contribuisce a livello internazionale all'avanzamento della ricerca biomedica sulle malattie genetiche rare. Non esistono scorciatoie in ricerca: la creazione di competenze uniche e processi trasparenti e di qualità sono fondamentale per il raggiungimento della cura.

Valorizzazione del percorso e dei risultati della ricerca

Annamaria Merico
Trasferimento Tecnologico di Fondazione Telethon

Simona Varani
Proprietà Intellettuale di Fondazione Telethon

Università, ospedali e centri di ricerca conducono ricerche che generano invenzioni rivoluzionarie, salvano vite e migliorano il modo in cui viviamo e lavoriamo tutti i giorni. Il trasferimento tecnologico ha un ruolo fondamentale nel condurre queste idee dal laboratorio al mercato: richiede competenze specifiche affinché le scoperte sviluppate nelle accademie vengano protette e tramite accordi con l'industria generino prodotti e servizi. Alcune invenzioni hanno successo, altre no; alcune vengono trasferite ad industrie esistenti, altre portano alla creazione di nuove aziende, che a loro volta portano alla creazione di nuovi posti di lavoro e ad un circolo virtuoso di innovazione. Vi sono vari passaggi chiave e sfide in questo processo: averne consapevolezza per chi lavora o ha interesse nell'ambito della ricerca facilita il processo e favorisce il successo.

Di chi sono i miei dati? La condivisione e la protezione dei dati sanitari personali

Sandra Courbier
Eurordis, Paris

Michela Maggi
Data Protection Officer (DPO) di Fondazione Telethon

Lucia Monaco
Responsabile Centro Studi di Fondazione Telethon

I dati sanitari personali sono una risorsa centrale per la ricerca biomedica, l'assistenza clinica e la gestione del paziente. La condivisione dei dati sanitari con e tra ricercatori e professionisti sanitari è indispensabile per abbreviare i tempi della diagnosi, per l'avanzamento della conoscenza sulla malattia e per i progressi nell'identificare e sviluppare trattamenti e terapie.
Questo è particolarmente vero per le malattie genetiche rare, che richiedono il collegamento e il confronto di dati da pazienti sparsi

in tutto il globo per poter beneficiare appieno della rivoluzione bioinformatica. La protezione della privacy, la gestione dei dati e l'applicazione delle norme di legge necessitano di coinvolgimento, competenza ed impegno da tutti gli attori interessati.
Questa sessione interattiva con il pubblico dei rappresentanti dei pazienti e dei ricercatori affronterà i bisogni, le preoccupazioni e le aspettative espressi dalla comunità dei pazienti, come pure i principi del regolamento europeo sulla privacy rilevanti per la gestione dei dati sanitari in contesto di ricerca.

Biobanche: avvertenze e modalità d'uso

Luca Sangiorgi
Coordinatore Reti Biobanche di Fondazione Telethon

La presentazione, mutuando l'impostazione di un bugiardino per farmaci, illustrerà la descrizione (principio attivo), destinazione d'uso, modalità d'uso, posologia, controindicazioni, interazioni e tutti gli effetti indesiderati sperimentalmente raccolti e segnalati dagli utilizzatori di biobanche e registri nel mondo delle malattie rare.

Per uscire dal buio: il progetto *Malattie senza diagnosi*

Vincenzo Nigro, Angelo Selicorni
Partner fondatore del progetto "Malattie senza diagnosi"

Per dare una risposta a pazienti senza diagnosi e iniziare a colmare questo bisogno Telethon ha ideato il programma "Malattie Senza Diagnosi".

Nonostante i numerosi sforzi della comunità medico-scientifica e i progressi dell'analisi del Dna, esistono ancora migliaia di malattie genetiche rarissime e con cause sconosciute che rimangono non diagnosticabili. Secondo Orphanet, a fronte di oltre 7500 malattie rare conosciute (l'80% delle quali di origine genetica), sono disponibili test diagnostici soltanto per circa 4200 di esse.
È proprio per colmare questo bisogno che Telethon ha avviato questo programma, che coinvolge una rete di centri clinici italiani di riferimento per la genetica medica e un centro di ricerca, l'Istituto Telethon di genetica e medicina di Pozzuoli, dalla consolidata esperienza nelle tecniche di sequenziamento di nuova generazione (Next Generation Sequencing). Ottenere una diagnosi è il punto di partenza per chiunque soffra di una malattia genetica: permette di dare un nome alla propria malattia, di individuare altri casi simili nel mondo da cui dedurre come evolverà, ma anche di avere più informazioni per gestire sia la quotidianità sia le situazioni di emergenza, e programmare controlli medici adeguati.
Essere coinvolti in questo programma rappresenta un'opportunità in più per chi oggi non ha una diagnosi: con quasi 700 casi ricevuti, ad oggi abbiamo identificati i geni causativi in circa il 40% dei pazienti coinvolti, con una resa diagnostica paragonabile a quella di altri programmi internazionali. È tuttavia probabile che le percentuali di successo aumentino progressivamente nel corso degli anni grazie agli avanzamenti della ricerca biomedica e attraverso la rianalisi periodica dei dati. Inoltre, le informazioni ottenute sono conservate nel database del progetto: è quindi possibile che una risposta possa arrivare successivamente, grazie al confronto con dati presenti in altri database internazionali.

POSTERS

FONDAZIONE

NEUROMUSCULAR DISORDERS

Muscular and neuromuscular disorders

Poster 01

A NATION-WIDE ITALIAN REGISTRY FOR PATIENTS WITH MUSCULAR DYSTROPHIES AND MYOPATHIES

Coordinator: **Adele D'Amico**
Partners: Claudio Bruno, Giacomo Comi, Rossella Tupler
Duration (N. Years): **2** *Starting year:* **2019**
Telethon Project (nr): GSP18002
Disease Name:
CMD, LGMD, FSHD, CM

Poster 02

UPDATE ON THE BON-DMD (GUP11011) STUDY: THE BIOCHEMICAL MARKERS

Aggiornamenti sul progetto Bone-DMD: marcatori biochimici

Coordinator: **Maria Luisa Bianchi**
Duration (N. Years): **3** *Starting year:* **2012**
Telethon Project (nr): GUP11011
Disease Name:
Duchenne Muscular Dystrophy

Poster 03

DETRIMENTAL ROLE OF COMPLEMENT C1/WNT AXIS IN DYSTROPHIC MUSCLE

Coordinator: **Stefano Biressi**
Duration (N. Years): **5** *Starting year:* **2014**
Telethon Project (nr): TCP 13007
Disease Name:
Duchenne Muscular Dystrophy

Poster 04

IDENTIFICATION OF A TWO NOVEL SUBPOPULATIONS OF SATELLITE CELLS WITH DIFFERENT KINETICS OF ACTIVATION

Coordinator: **Stefano Biressi**
Duration (N. Years): **5** *Starting year:* **2014**
Telethon Project (nr): TCP 13007
Disease Name:
Duchenne Muscular Dystrophy

Poster 05

A POSSIBLE STRATEGY TO INDUCE EXON 45 SKIPPING IN DMD-D44 PATIENTS THROUGH THE MODULATION OF CELF2A SPLICING FACTOR

Coordinator: **Irene Bozzoni**
Duration (N. Years): **3** *Starting year:* **2017**
Telethon Project (nr): GPP16213
Disease Name:
Duchenne Muscular Dystrophy

Poster 06

EXTRACELLULAR ATP AND T REGULATORY CELLS: NEW THERAPEUTICS TARGETS IN ALPHA-SARCOGLYCAN DEFICIENT MUSCULAR DYSTROPHY (LGMD2D)

Il ruolo dell'ATP e dei recettori purinergici nella distrofia muscolare dei cingoli da deficit di alfa-sarcoglicano (LGMD2D): nuove prospettive terapeutiche

Coordinator: **Claudio Bruno**
Duration (N. Years): **2** *Starting year:* **2017**
Telethon Project (nr): GGP17192
Disease Name:
Limb Girdle Muscular Dystrophy Type 2D (LGMD2D)

Poster 07

MODULATION OF THE CYCLIN INHIBITOR P27 TO AMELIORATE MEROSIN DEFICIENT CONGENITAL MUSCULAR DYSTROPHY (MDC1A)

Modulazione della molecula p27 per migliorare la distrofia muscolare congenita da deficit di merosina (MDC1A).

Coordinator: **Stefano Carlo Previtali**
Duration (N. Years): **3** Staring year: **2018**
Telethon Project (nr): GGP17009
Disease Name:
Merosin Deficient Congenital Muscular Dystrophy

Poster 08

USEFUL: USER- CENTRED ASSISTIVE SYSTEM FOR ARM FUNCTIONS IN NEUROMUSCULAR SUBJECTS

USEFUL: sistemi assistivi paziente-centrici per il supporto dell'arto superiore in pazienti neuromuscolari

Coordinator: **Alessandra Pedrocchi**
Partners: Grazia D'Angelo, Franco Molteni
Duration (N. Years): **2** *Starting year:* **2016**
Telethon Project (nr): GUP15021
Disease Name:
Muscular Dystrophy

Poster 09

GENE EDITING IN MYOTONIC DYSTROPHY TYPE 1: ASSESSMENT OF EFFICIENCY, SAFETY AND THERAPEUTIC EFFECT OF CTG-REPEAT DELETION IN A MOUSE MODEL OF DISEASE

Terapia genica nella Distrofia Miotonica di tipo 1: studio dell'efficienza, specificità ed effetto terapeutico della delezione delle espansioni di CTG in un modello murino della patologia

Coordinator: **Germana Falcone**
Partner: Fabio Martelli
Duration (N. Years): **3** *Starting year:* **2019**
Telethon Project (nr): GGP19035
Disease Name:
Myotonic Dystrophy Type 1

Poster 10

SMALL MOLECULES TO RESCUE FOLDING-DEFECTIVE SARCOGLYCANS: IN VIVO ASSESSMENT OF NOVEL THERAPEUTIC STRATEGIES

Piccole molecole per il recupero di sarcoglicani con difetti di ripiegamento: verifica in vivo di nuove strategie terapeutiche

Coordinator: **Dorianna Sandonà**

Duration (N. Years): **3** *Starting year:* **2015** *Project ending year:* **2019**

Telethon Project (nr): GGP15140

Disease Name:
Sarcoglycanopathies (LGMD2D-F)

Poster 11

SMN-PRIMED RIBOSOMES MODULATE THE TRANSLATION OF TRANSCRIPTS RELATED TO SPINAL MUSCULAR ATROPHY

Coordinator: **Gabriella Viero**
Partner: Marina Boido

Duration (N. Years): **3** *Starting year:* **2019**

Telethon Project (nr): GGP19115

Disease Name:
Spinal Muscular Atrophy

Poster 12

A MITOCHONDRIAL THERAPY FOR MUSCULAR DYSTROPHIES

Una terapia mitocondriale per le distrofie muscolari

Coordinator: **Paolo Bernardi**

Duration (N. Years): **3** Staring year: **2017**

Telethon Project (nr): GGP17092

Disease Name:
Ullrich Congenital MD; Duchenne MD

Poster 13

SPERMIDINE AS NEW CANDIDATE FOR THE TREATMENT OF COL6 MYOPATHIES (SPECTRE-COL6)

Dal laboratorio alla clinica: la spermidina come nuovo candidato per il trattamento delle miopatie da deficit di COL6

Coordinator: **Paolo Bonaldo**

Duration (N. Years): **2** *Starting year:* **2020**

Telethon Project (nr): GGP19229

Disease Name:
Ullrich Muscular Dystrophy; Bethlem Myopathy

Myopathies and cardiomyopathies

Poster 14

REMODELING OF MITOCHONDRIAL FUNCTION AND GENE EXPRESSION IN CORE MYOPATHY PATIENTS

Rimodellamento della espressione genica e funzione mitocondriale nelle malattie 'central core' del muscolo scheletrico

Coordinator: **Gyorgy Szabadkai**
Partner: Anna Raffaello

Duration (N. Years): **3** *Starting year:* **2017**

Telethon Project (nr): GGP16026

Disease Name:
Central Core Disease

Poster 15

A NOVEL IN VITRO DUCHENNE MUSCULAR DYSTROPHY CARDIOMYOPATHY MODEL: HUMAN IPSC-DERIVED CARDIOMYOCYTES FOR MECHANISTIC STUDIES

Un nuovo modello in vitro, basato su cardiomiociti derivati da cellule ips, per studiare i meccanismi della cardiomiopatia associata alla distrofia muscolare

Coordinator: **Cecilia Ferrantini**
Partner: Leonardo Sacconi

Duration (N. Years): **2** *Starting year:* **2016**

Telethon Project (nr): GGP16191

Disease Name:
Duchenne Muscular Dystrophy

Poster 16

DEVELOPING TOOLS FOR TRIAL READINESS IN PRIMARY MITOCHONDRIAL MYOPATHIES OF THE ADULTHOOD

Sviluppo dei necessari strumenti per "essere pronti" a trial clinici nelle miopatie mitocondriali dell'adulto.

Coordinator: **Michelangelo Mancuso**
Partners: Costanza Lamperti, Giacomo Comi, Tiziana Enrica Mongini, Paola Tonin, Valerio Carelli, Serenella Servidei, Olimpia Musumeci, Massimiliano Filosto, Elena Pegoraro

Duration (N. Years): **3** *Starting year:* **2017**

Telethon Project (nr): GSP16001

Disease Name:
Mitochondrial Myopathies

Poster 17

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

Coordinator: **Marcello Arca**
Partners: Daniela Tavian, Corrado Angelini, Elena Maria Pennisi, Antonio Musarò

Duration (N. Years): **3** *Starting year:* **2014** *Project ending year:* **2018**

Telethon Project (nr): GGP14066

Disease Name:
Neutral Lipid Storage Disease

Poster 18

STORE-OPERATED CALCIUM ENTRY (SOCE): ROLE IN SKELETAL MUSCLE FUNCTION AND DISEASE

Ingresso di Calcio operato dagli stores intracellulari (o SOCE): ruolo nella funzione e nella malattia del muscolo scheletrico

Coordinator: **Feliciano Protasi**
Partner: Vincenzo Sorrentino

Duration (N. Years): **3** Starting date: October **2019**

Telethon Project (nr): GGP19231

Disease Name:
Tubular Aggregate Myopathy

Myotonic disorders

Poster 19

SKELETAL MUSCLE AND CIRCULATING MICRORNAS IN MYOTONIC DYSTROPHY TYPE 1

MicroRNA del muscolo scheletrico e circolanti nella distrofia miotonica di tipo 1

Coordinator: **Fabio Martelli**
Partner: Germana Falcone

Duration (N. Years): **3** *Starting year:* **2014**

Project ending year: **2018**

Telethon Project (nr): GGP14092

Disease Name:
Myotonic Dystrophy Type 1

Neuromuscular diseases

Poster 20

MODULATING NEUREGULIN-1 SIGNALS TO TREAT HEREDITARY DEMYELINATING NEUROPATHIES

Modulazione della Neuregulina 1 come approccio terapeutico per il trattamento di neuropatie ereditarie.

Coordinator: **Carla Taveggia**
Partners: Alessandra Bolino, Stefano Carlo Previtali, Maurizio D'Antonio

Duration (N. Years): **4** *Starting year:* **2015** *Project ending year:* **2019**

Telethon Project (nr): GGP15012

Disease Name:
Charcot-Marie-Tooth Hereditary Neuropathies

Poster 21

GENE THERAPY AND LONG TERM EVALUATION OF DIFFERENT DIETARY REGIMENS IN A GLYCOGEN STORAGE DISEASE TYPE III KO MOUSE MODEL

Terapia genica e valutazione nel lungo periodo di diversi tipi di dieta in un modello murino KO di Glicogenosi di tipo III

Coordinator: **Giacomo Comi**
Partner: Federico Mingozzi

Duration (N. Years): **4** *Starting year:* **2015**

Telethon Project (nr): GGP15051

Disease Name:
Glycogen Storage Disease Type III

Poster 22

MITMED CONSORTIUM: FROM THE IDENTIFICATION AND CHARACTERIZATION OF NUCLEAR GENES RESPONSIBLE FOR HUMAN MITOCHONDRIAL DISORDERS TOWARDS POTENTIAL THERAPEUTIC APPROACHES IN EXPERIMENTAL MODELS

Consorzio mitmed: dall'identificazione e caratterizzazione di geni nucleari responsabili di malattie mitocondriali verso potenziali approcci terapeutici in modelli sperimentali

Coordinator: **Daniele Ghezzi**
Partners: Rodolfo Costa, Claudia Donnini

Duration (N. Years): **3** *Starting year:* **2015** *Project ending year:* **2019**

Telethon Project (nr): GGP15041

Disease Name:
Mitochondrial Disease

Poster 23

CLINICAL EFFICACY OF NIV AND MODAFINIL ON EXCESSIVE DAYTIME SLEEPINESS: LESSONS LEARNED FROM A MULTICENTER, RANDOMIZED, DOUBLE-BLIND, PLACEBO-CONTROLLED CLINICAL TRIAL IN DM1

L'efficacia della ventilazione non-invasiva (NIV) e del modafinil sull'eccessiva sonnolenza diurna: esperienza acquisita da uno studio multicentrico, randomizzato, doppio-cieco, placebo- controllato nella Distrofia Miotonica di tipo 1 (DM1)

Coordinator: **Valeria Sansone**
Partners: Lino Nobili, Roberto Massa, Fabio Placidi

Duration (N. Years): 2 *Starting year:* **2016** *Project ending year:* **2019**

Telethon Project (nr): GUP15004

Disease Name:
Myotonic Dystrophy Type 1, Steinert Disease

Poster 24

PRE-CLINICAL IDENTIFICATION OF DRUGS TARGETING POLG DISORDERS BY USING A ZEBRAFISH/YEAST TRANS-SPECIES APPROACH (ZIPPY)

Identificazione di farmaci per le patologie POLG tramite test su sistemi lievito-zebrafish (ZIPPY)

Coordinator: **Francesco Argenton**
Partner: Enrico Baruffini

Duration (N. Years): **3** *Starting year:* **2019**

Telethon Project (nr): GGP19287

Disease Name:
Polg-Related Diseases

Poster 25

REGISTRY FOR TRIAL READINESS IN SPINAL AND BULBAR MUSCLE ATROPHY

Registro per l'atrofia muscolare bulbo-spinale

Coordinator: **Caterina Mariotti**
Partners: Davide Pareyson, Giovanni Sorarù, Mario Sabatelli

Duration (N. Years): **3** *Starting year:* **2016**

Telethon Project (nr): GUP15009

Disease Name:
Spinal and Bulbar Muscular Atrophy (SBMA)

Poster 26

PHOSPHORYLATION-MEDIATED CHANGES OF ANDROGEN RECEPTOR STRUCTURE AND FUNCTION IN SPINAL AND BULBAR MUSCULAR ATROPHY PATHOGENESIS

Modifiche post-traduzionali del recettore degli androgeni ne influenzano la struttura e funzione e hanno ripercussione sulla malattia di Kennedy

Coordinator: **Maria Pennuto**

Duration (N. Years): **5** *Starting year:* **2013** *Project ending year:* **2017**

Telethon Project (nr): TCP12013

Disease Name:
Spinal and Bulbar Muscular Atrophy (SBMA)

Poster 27

DEVELOPMENT OF A PREDICTIVE BODY FAT EQUATION FOR SPINAL MUSCULAR ATROPHY TYPE I CHILDREN

Sviluppo di una equazione predittiva per la massa grassa di bambini affetti da Atrofia Muscolare Spinale

Coordinator: **Simona Bertoli**
Partners: Enrico Bertini, Giovanni Baranello, Marina Pedemonte, Caterina Agosto

Duration (N. Years): **3** *Starting year:* **2016** *Project ending year:* **2019**

Telethon Project (nr): GUP15014

Disease Name:
Spinal Muscular Atrophy

Poster 28

IDENTIFICATION OF NEW DRUGGABLE TARGETS AND POTENTIAL THERAPEUTIC COMPOUNDS FOR SPINAL MUSCULAR ATROPHY, USING A C. ELEGANS MODEL OF NEURODEGENERATION

Coordinator: **Elia Di Schiavi**

Duration (N. Years): **3** *Starting year:* **2017**

Telethon Project (nr): GGP16203

Disease Name:
Spinal Muscular Atrophy

Poster 29

CELL PENETRATING PEPTIDE-CONJUGATED MORPHOLINO FOR TREATMENT OF SMA SYMPTOMATIC CASES

Morfolino coniugato a peptide per il trattamento dei casi sintomatici affetti da SMA

Coordinator: **Monica Nizzardo**

Duration (N. Years): **3** *Starting year:* **2014** *Project ending year:* **2018**

Telethon Project (nr): GGP14025

Disease Name:
Spinal Muscular Atrophy

Poster 30

ANTHROPOMETRIC STANDARDS IN NAÏVE PATIENTS WITH SPINAL MUSCULAR ATROPHY TYPE 1

Standard di crescita in pazienti affetti da atrofia muscolare spinale di tipo 1

Coordinator: **Simona Bertoli**
Partners: Enrico Bertini, Giovanni Baranello, Marina Pedemonte, Caterina Agosto

Duration (N. Years): **3** *Starting year:* **2016**

Telethon Project (nr): GUP15014

Disease Name:
Spinal Muscular Atrophy Type 1

Polyneuropathies

Poster 31

KNOCKDOWN AND REPLACEMENT OF MFN2 FOR TREATMENT OF DOMINANTLY INHERITED PERIPHERAL NEUROPATHY CMT2A PATIENTS

Silenziamento e ri-espressione del gene MFN2 come trattamento per i pazienti affetti da CMT2A, una neuropatia periferica ereditaria

Coordinator: **Stefania Corti**

Duration (N. Years): **3** *Starting year:* **2020**

Telethon Project (nr): GGP19002

Disease Name:
Charcot-Marie-Tooth Disease 2A

Poster 32

TTR-FAP ITALIAN REGISTRY: A COLLABORATIVE NETWORK FOR DEFINITION OF NATURAL HISTORY, PSYCHOSOCIAL BURDEN, STANDARDS OF CARE AND CLINICAL TRIALS

Polineuropatia amiloidosica familiare, transtiretina, storia naturale, carico psicosociale, standard di cura, bisogni di cura

Coordinator: **Giuseppe Vita**
Partners: Giampaolo Merlini, Lorenza Magliano, Mario Sabatelli, Marina Grandis, Gian Maria Fabrizi, Davide Pareyson, Lucio Santoro, Alessandro Mauro

Duration (N. Years): **2** *Starting year:* **2017**

Telethon Project (nr): GUP15010

Disease Name:
Familial Amyloidotic Polyneuropathy

NEUROLOGICAL DISORDERS

Genetic neurological disorders

Poster 33

FINDING NEW TARGETS TO COUNTERACT BRAIN PROGENITOR CELLS DYSREGULATION IN AGC1 DEFICIENCY HYPOMYELINATION: A MULTIDISCIPLINARY APPROACH

Alla ricerca di nuovi bersagli terapeutici per contrastare l'ipomielinizzazione nell'AGC1-deficiency: uno studio multidisciplinare sui precursori delle cellule cerebrali

Coordinator: **Barbara Monti**
Partner: Francesco Massimo Lasorsa

Duration (N. Years): **3** *Starting year:* **2019**

Telethon Project (nr): GGP19067

Disease Name:
AGC1 Deficiency

Poster 34

THE AICARDI-GOUTIÈRES SYNDROME – FROM NUCLEIC ACID SENSING TO DISEASE MODELLING

Il sindrome di Aicardi-Goutières – dal riconoscimento degli acidi nucleici ad un modello di malattia

Coordinator: **Anna Kajaste-Rudnitski**

Duration (N. Years): **5** *Starting year:* **2016**

Telethon Project (nr): TGT16D03

Disease Name:
Aicardi-Goutières Syndrome

Poster 35

IMPROVING DEVELOPMENTAL TRAJECTORIES IN 22Q11.2 DELETION SYNDROME BY OXYTOCIN: FOCUS ON ANTI-INFLAMMATORY EFFECTS

Ossitocina nella sindrome da delezione 22q11.2: implicazione di possibili effetti anti-infiammatori

Coordinator: **Francesco Papaleo**
Partner: Bice Chini

Duration (N. Years): **3** Starting: **2020**

Telethon Project (nr): GGP19103

Disease Name:
Chromosome 22q11.2 Deletion Syndrome

Poster 36

ENHANCED THALAMOCORTICAL SYNAPTIC TRANSMISSION AND DYSREGULATION OF THE EXCITATORY-INHIBITORY BALANCE AT THE THALAMOCORTICAL FEED-FORWARD INHIBITORY MICROCIRCUIT IN A MOUSE MODEL OF FAMILIAL HEMIPLEGIC MIGRAINE

Aumentata trasmissione sinaptica talamo-corticale e disregolazione del bilancio eccitazione-inibizione nel microcircuito talamo-corticale in un modello animale di emicrania emiplegica familiare.

Coordinator: **Daniela Pietrobon**

Duration (N. Years): **3** *Starting year:* **2015** *Project ending year:* **2019**

Telethon Project (nr): GGP14234

Disease Name:
Familial Hemiplegic Migraine

Poster 37

A NOVEL COMPREHENSIVE STRATEGY FOR THE STUDY OF THE MOLECULAR BASIS OF FAMILIAL HEMIPLEGIC MIGRAINE 3

Una nuova strategia integrata per lo studio delle basi molecolari dell'Emicrania Emiplegica Familiare 3

Coordinator: **Paola Gavazzo**

Duration (N. Years): **3** *Starting year:* **2017**

Telethon Project (nr): GGP17178

Disease Name:
Familial Hemiplegic Migraine 3

Poster 38

ROLE OF ER-PHAGY IN HEREDITARY SENSORY AXONOPATHIES

Ruolo dell'autofagia selettiva del reticolo endoplasmatico nelle assonopatie sensoriali ereditarie

Coordinator: **Paolo Grumati**

Duration (N. Years): **1** *Starting year:* **2019**

Project ending year: **2019**

Telethon Project (nr): TIGEM

Disease Name:
Hereditary Sensory Axonopathies

Poster 39

TRPML1 LINKS LYSOSOMAL CALCIUM TO AUTOPHAGY INITIATION

TRPML1 collega il calcio lisosomiale all'inizio dell'autofagia

Coordinator: **Diego Luis Medina**

Duration (N. Years): **2** *Starting year:* **2017** *Project ending year:* **2019**

Telethon Project (nr): Tigem - Granted by Mucolipidosis Type 4 Foundation

Disease Name:
Mucolipidosis Type 4 (MLIV)

Ataxias

Poster 40

EXCITATORY/INHIBITORY UNBALANCE IN ATAXIA TELANGIECTASIA AND NEW THERAPEUTHICAL INTERVENTIONS

Sbilanciamento tra eccitazione ed inibizione nell'A-T e nuovi approcci terapeutici

Coordinator: **Flavia Antonucci**
Duration (N. Years): **2** *Starting year:* **2017**
Telethon Project (nr): GGP16015
Disease Name:
Ataxia Telangiectasia

Poster 41

REGULATION OF ALTERNATIVE SPLICING OF VOLTAGE-GATED CA2+ CHANNELS BY CRISPR/CAS9-MEDIATED GENOME EDITING AS POTENTIAL GENETIC THERAPY FOR EPISODIC ATAXIA TYPE 2

Regolazione dello 'splicing' alternativo dei canali Ca2+ mediante 'genome editing' di tipo CRISPR/Cas9 come potenziale terapia genica per l'atassia episodica di tipo 2

Coordinator: **Lorenzo Cingolani**
Partner: Federico Zara
Duration (N. Years): **3** *Starting year:* **2020**
Telethon Project (nr): GGP19181
Disease Name:
Episodic Ataxia Type II

Poster 42

RNA THERAPEUTICS FOR FRIEDREICH'S ATAXIA

Terapia A base di RNA per l'Atassia di Friedreich

Coordinator: **Stefano Gustincich**
Partners: Ivano Condò, Antonello Mallamaci
Duration (N. Years): **2** *Starting year:* **2015** *Project ending year:* **2018**
Telethon Project (nr): GGP15004
Disease Name:
Friedreich's Ataxia

Poster 43

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

Coordinator: **Enrico Bertini**
Partners: Giangiacomo Consalez, Enza Maria Valente
Duration (N. Years): **3** *Starting year:* **2014** *Project ending year:* **2018**
Telethon Project (nr): GGP13146
Disease Name:
Joubert Syndrome

Epilepsy and Seizures

Poster 44

DELINEATING THE MOLECULAR PATHWAY AND PATHOGENIC MECHANISM UNDERLYING AUTOSOMAL DOMINANT LATERAL TEMPORAL EPILEPSY (ADLTE)

Coordinator: **Carlo Nobile**
Partner: Federico Zara
Duration (N. Years): **3** *Starting year:* **2015** *Project ending year:* **2019**
Telethon Project (nr): GGP15229
Disease Name:
Autosomal Dominant Lateral Temporal Epilepsy

Poster 45

PROTEIN SUBSTITUTION THERAPY: A PROMISING TREATMENT FOR CDKL5 DEFICIENCY DISORDER

Terapia proteica sostitutiva: un trattamento promettente per il disordine CDKL5

Coordinator: **Elisabetta Ciani**
Partners: Maurizio Giustetto, Charlotte Kilstrup-Nielsen, Tommaso Pizzorusso
Duration (N. Years): **3**
Telethon Project (nr): GGP15098
Disease Name:
CDKL5 Deficiency Disorder

Poster 46

TOWARD GENE THERAPY FOR DRAVET SYNDROME: UNCOVERING DYNAMICS OF REVERSIBILITY AND MECHANISMS OF SCN1A GENE MODULATION

Verso la terapia genica per la sindrome di Dravet: reversibilità della malattia e meccanismi di modulazione del gene Scn1a

Coordinator: **Gaia Colasante**
Duration (N. Years): **3** *Starting:* **2019**
Telethon Project (nr): GGP19249
Disease Name:
Dravet Syndrome

Poster 47

RESCUING EPILEPSY ASSOCIATED WITH SYN1 AND SCN1A GENE MUTATIONS BY INHIBITING EEF2K/EEF2 PATHWAY

Recupero dalla patologia epilettica indotta da mutazioni dei geni SYN1 e SCN1a tramite inibizione dell'attività della chinasi eEF2K

Coordinator: **Carlo Sala**
Duration (N. Years): **3** *Starting year:* **2018**
Telethon Project (nr): GGP17176
Disease Name:
Epilepsy, Intellectual Disability

Poster 48

SOLVING THE PUZZLE OF PROTOCADHERIN-19 MOSAICISM TO UNDERSTAND THE PATHOPHYSIOLOGY OF PCDH19 FEMALE EPILEPSY (PCDH19-FE)

Studio del ruolo del mosaicismo nella patofisiologia dell'epilessia femminile PCDH19-Dipendente

Coordinator: **Silvia Bassani**

Duration (N. Years): **3** *Starting year:* **2018**

Telethon Project (nr): GGP17260

Disease Name:
Epileptic Encephalopathy Early Infantile 9 (EIEE9)

Poster 49

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

Analisi della funzione del gene Aristaless-related Homeobox nell'Epilessia e identificazione di bersagli molecolari a scopo terapeutico

Coordinator: **Maria Giuseppina Miano**

Duration (N. Years): **3** *Starting year:* **2014** *Project ending year:* **2018**

Telethon Project (nr): GGP14198

Disease Name:
Infantile Spasms (ISSX1); West Syndrome

Poster 50

GENOTYPE-PHENOTYPE CORRELATIONS, NOVEL PATHOGENETIC MECHANISMS, AND PILOT CLINICAL STUDIES IN NEONATAL EPILEPSIES ASSOCIATED TO MUTATIONS IN THE KCNQ2/3 POTASSIUM CHANNEL GENES

Coordinator: **Maurizio Taglialatela**

Duration (N. Years): **3** *Starting year:* **2016**

Telethon Project (nr): GGP15113

Disease Name:
KCNQ-related Diseases

Poster 51

INTERACTION OF PRRT2 WITH SODIUM CHANNELS: PATHOGENETIC BASIS AND NEW TARGETS FOR THE CURE OF PRRT2-ASSOCIATED PAROXYSMAL DISORDERS

Interazioni di PRRT2 con i canali sodio: basi patogenetiche e nuovi bersagli terapeutici per le malattie parossistiche associate a mutazioni nel gene PRRT2

Coordinator: **Fabio Benfenati**

Duration (N. Years): **3** *Starting year:* **2019**

Telethon Project (nr): GGP19120

Disease Name:
Paroxysmal Kinesigenic Dyskinesia

Intellectual Disabilities

Poster 52

NLG3 SHAPES EXCITATION/INHIBITION RATIO IN NEURONAL CIRCUITS OF ASD MURINE MODELS: IMPLICATIONS OF THE CA2 HIPPOCAMPAL CIRCUIT IN SOCIAL DEFICITS

La neuroligina 3 controlla l'equilibrio tra eccitazione e inibizione nei circuiti neuronali: coinvolgimento della regione ippocampale CA2 nei deficit sociali osservati in modelli murini di disordini dello spettro autistico

Coordinator: **Enrico Cherubini**
Partner: Andrea Barberis

Duration (N. Years): **3** *Starting year:* **2017**

Telethon Project (nr): GGP16083

Disease Name:
Autism Spectrum Disorders

Poster 53

ROLE OF INTRACELLULAR CHLORIDE ACCUMULATION IN DOWN SYNDROME PHYSIOPATHOLOGY IN MICE

Ruolo della accumulazione di cloro intracellulare nella fisiopatologia della Sindrome di Down

Coordinator: **Laura Cancedda**

Duration (N. Years): **5** *Starting year:* **2016**

Telethon Project (nr): TCP15021

Disease Name:
Down Syndrome

Poster 54

NEUROTROPHIC-MIMETIC STRATEGY TO RESCUE SYNAPTIC PLASTICITY AND COGNITIVE FUNCTIONS IN A MOUSE MODEL OF DOWN SYNDROME

Coordinator: **Andrea Contestabile**

Duration (N. Years): **3** *Starting year:* **2015** *Project ending year:* **2019**

Telethon Project (nr): GGP15043

Disease Name:
Down Syndrome

Poster 55

DROSOPHILA MELANOGASTER AS A MODEL TO STUDY THE ROLE OF FMRP PROTEIN, INVOLVED IN THE FRAGILE-X SYNDROME, IN THE PIRNA-MEDIATED GENOME STABILITY

La Drosophila melanogaster come modello per lo studio del ruolo della proteina FMRP, coinvolta nella sindrome della X-fragile, nella stabilità genomica mediata dai piRNA.

Coordinator: **Maria Giuseppina Bozzetti**

Duration (N. Years): **3** *Starting year:* **2014** *Project ending year:* **2018**

Telethon Project (nr): GGP14181

Disease Name:
Fragile-X Syndrome

Poster 56

SETD5 REGULATES CHROMATIN METHYLATION STATE AND PRESERVES GLOBAL TRANSCRIPTIONAL FIDELITY DURING BRAIN DEVELOPMENT AND NEURONAL WIRING

SETD5 regola la metilazione della cromatina e preserva la fedeltà trascrizionale durante lo sviluppo cerebrale e il funzionamento neuronale

Coordinator: **Alessandro Sessa**
Partners: Alessio Zippo, Massimiliano Andreazzoli

Duration (N. Years): **3** *Starting year:* **2015** *Project ending year:* **2019**

Telethon Project (nr): GGP15096

Disease Name:
Intellectual Disability, Autism Spectrum Disorders

Poster 57

INTRACELLULAR CHLORIDE DYNAMICS IN AUTISTIC BRAIN: A BETTER UNDERSTANDING IS NEEDED FOR TAILORED CURES

Dinamica della concentrazione del cloro intracellulare in modelli di deficit cognitivi

Coordinator: **Gian Michele Ratto**
Partner: Claudia Lodovichi

Durationg (N. Years): **3** *Starting year:* **2019**

Telethon Project (nr): GGP19281

Disease Name:
Mental Retardation, X-linked; Macrocephaly/Autism

Poster 58

NEURONAL DYSFUNCTIONS UNDERLYING PHELAN–MCDERMID SYNDROME AND THEIRS RESCUE BY GENETIC AND PHARMACOLOGICAL MODULATION OF MGLU5 SIGNALING

Caratterizzazione dell'attività di farmaci attivatori del recettore metabotropo di tipo 5 per migliorare i difetti neurologici della sindrome di Phelan McDermid

Coordinator: **Chiara Verpelli**
Partner: Alessandro Tozzi

Duration (N. Years): **3** *Starting year:* **2017**

Telethon Project (nr): GGP16131

Disease Name:
Phelan McDermid Syndrome

Poster 59

EXPLOITING WHOLE-BRAIN STRATEGIES OF GENE THERAPY AND NOVEL THERAPEUTIC TARGETS IN RETT SYNDROME MOUSE MODELS

Nuove strategie di terapia genica e nuovi obiettivi terapeutici per la sindrome di Rett

Coordinator: **Vania Broccoli**

Duration (N. Years): **3** *Starting year:* **2019**

Telethon Project (nr): GGP19038

Disease Name:
Rett Syndrome

Poster 60

ALTERED L-TYPE CHANNEL GATING, ACTION POTENTIAL FIRING AND EXCITATORY/INHIBITORY SYNAPTIC RESPONSES IN HIPPOCAMPAL NEURONS OF THE AUTISTIC TIMOTHY SYNDROME TYPE-2 MOUSE

Coordinator: **Emilio Carbone**

Duration (N. Years): **3** *Starting year:* **2015** *Project ending year:* **2019**

Telethon Project (nr): GGP15110

Disease Name:
Tymothy Syndrome

Poster 61

MECHANISTIC DISSECTION OF POLYCOMB-DEPENDENT DYSREGULATION IN WEAVER SYNDROME NEURAL LINEAGES

Studio della deregolazione dipendente da Polycomb nella sindrome di Weaver tramite l'uso di tipi cellulari neurali derivati da paziente

Coordinator: **Alejandro López Tobón**

Duration (N. Years): **3** *Starting year:* **2019**

Telethon Project (nr): GGP19295

Disease Name:
Weaver Syndrome

Poster 62

SPOTLIGHT ON LATERAL HABENULA (LHB) FUNCTION IN TETRASPANIN7 (TSPAN7) KNOCK-OUT MICE

Funzionalità della Lateral Habenula (LHb) negli animali knock-out per tetraspanin7 (TSPAN7)

Coordinator: **Maria Passafaro**

Duration (N. Years): **3** *Starting year:* **2018**

Telethon Project (nr): GGP17283

Disease Name:
X-linked Intellectual Disability

Neurodegenerative diseases

Poster 63

ALTERATION OF LYSOSOMES AND OF LYSOSOMAL ACTIVITY IN CHARCOT-MARIE-TOOTH 2B PERIPHERAL NEUROPATHY

Alterazioni dei lisosomi e della funzionalità lisosomale nella neuropatia periferica Charcot-Marie-Tooth di tipo 2B

Coordinator: **Cecilia Bucci**
Partners: Stefano Previtali, Lucio Santoro

Duration (N. Years): **3** *Starting year:* **2017**

Telethon Project (nr): GGP16037

Disease Name:
Charcot-Marie-Tooth Type 2B

Poster 64

FULL ATOMISTIC MODEL OF PRION STRUCTURE AND CONVERSION

Primo Modello Atomistico della Struttura e della Replicazione di un Prione

Coordinator: **Emiliano Biasini**

Duration (N. Years): **5** *Starting year:* **2015**

Telethon Project (nr): TCP14009

Disease Name:
Creutzfeldt-Jakob Disease

Poster 65

MITOCHONDRIAL CA2+ UPTAKE IN THE PATHOGENESIS OF FAMILIAL ALZHEIMER'S DISEASE

L'accumulo mitocondriale di calcio nella patogenesi delle forme familiari della malattia di Alzheimer

Coordinator: **Rosario Rizzuto**
Partner: Tullio Pozzan

Duration (N. Years): **3** *Strating year:* **2017**

Telethon Project (nr): GGP16029

Disease Name:
Familial Alzheimer's Disease

Poster 66

DEVELOPMENT OF EXON SPECIFIC U1 SNRNA-BASED THERAPY FOR FAMILIAL DYSAUTONOMIA

Sviluppo di una terapia basata sugli Exon Specific U1 nella disautonomia familiare

Coordinator: **Franco Pagani**

Duration (N. Years): **3** *Starting year:* **2017**

Telethon Project (nr): GGP17006

Disease Name:
Familial Dysautonomia

Poster 67

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

Coordinator: **Gianluigi Forloni**
Partners: Benedetto Ignazio Roiter, Fabrizio Tagliavini

Duration (N. Years): **3** *Starting year:* **2018**

Telethon Project (nr): GSP18001

Disease Name:
Fatal Familial Insomnia

Poster 68

NEUROSERPIN MISFOLDING AND FENIB NEURODEGENERATION: MECHANISM AND INHIBITION PROCESSES

Coordinator: **Martino Bolognesi**

Duration (N. Years): **3** *Starting year:* **2017**

Telethon Project (nr): GGP17036

Disease Name:
FENIB

Poster 69

LYSOSOMAL STORAGE DISORDERS (LSD) - MODELING THE DISEASE COMPLEXITY TO REFINE GENE/CELL THERAPY TREATMENT STRATEGIES

Malattie da accumulo lisosomiale – modellare la complessità della malattia per affinare strategie di terapia genica e cellulare

Coordinator: **Angela Gritti**

Duration (N. Years): **5** *Starting year:* **2016**

Telethon Project (nr): TGT16D02

Disease Name:
GLD, MLD, GM2 Gangliosidosis

Poster 70

MENINGES AS AN OVE RLOOKED PHARMACOLOGICAL TARGET FOR GLOBOID CELL LEUKODYSTROPHY

Coordinator: **Francesco Bifari**
Partners: Angela Gritti, Marco Riva

Duration (N. Years): **2** *Starting year:* **2019**

Telethon Project (nr): GGP19250

Disease Name:
Globoid Cell Leukodystrophy

Poster 71

PLASMALOGEN-BASED THERAPEUTIC STRATEGY FOR THE TREATMENT OF HEREDITARY SPASTIC PARAPLEGIA

Coordinator: **Diana Pendin**

Duration (N. Years): **3** *Starting year:* **2019**

Telethon Project (nr): GGP19304

Disease Name:
Hereditary Spastic Paraplegia

Poster 72

SLUGGISH MITOCHONDRIAL FLICKERING AT THE BASIS OF HEREDITARY SPASTIC PARAPLEGIA (SPG7)

L'alterata apertura del canale di membrana mitocondriale (mPTP) è alla base della paraplegia spastica ereditaria di tipo 7 (SPG7)

Coordinator: **Giorgio Casari**

Duration (N. Years): **3** *Starting year:* **2016**

Telethon Project (nr): TIGEM

Disease Name:
Hereditary Spastic Paraplegia Type 7 (SPG7)

Poster 73

TARGETING NEURONS WITH CHOLESTEROL. HOW CAN IT CHANGE THE FUTURE OF HD PATIENTS

Coordinator: **Elena Cattaneo**

Duration (N. Years): **3** *Starting year:* **2018**

Telethon Project (nr): GGP17012

Disease Name:
Huntington's Disease

Poster 74

DISSECTING THE MOLECULAR FUNCTION OF MUTANT HUNTINGTIN WITH STEM CELLS

Studio della funzione della proteina Huntingtin mutata mediante l'uso di cellule staminali

Coordinator: **Graziano Martello**
Partners: Enrico Moro, Martin Leeb, Vittorio Maglione

Duration (N. Years): **5** *Starting year:* **2014**

Telethon Project (nr): TCP13013

Disease Name:
Huntington's Disease

Poster 75

MIR-181A AND MIR-181B DOWNREGULATION AMELIORATES MITOCHONDRIAL-ASSOCIATED NEURODEGENERATION BY ENHANCING MITOCHONDRIAL BIOGENESIS AND MITOPHAGY

Coordinator: **Brunella Franco**

Telethon Project (nr): TIGEM

Disease Name:
Mitochondrial Disfunction

Poster 76

DISEASE' MECHANISMS AND PHARMACOLOGICAL TARGETING OF BEHAVIORAL SYMPTOMS IN SANFILIPPO SYNDROME

Basi neurobiologiche e trattamenti farmacologici per i sintomi comportamentali nella malattia di Sanfilippo

Project TIGEM (Disease' mechanisms leading to dopaminergic dysfunction underlying behavioural symptoms in MPS-IIIA)

Coordinator: **Elvira De Leonibus**

Duration (N. Years): **2** *Starting year:* **2018**

Project TIGEM (New therapeutic strategies for the treatment of behavioural symptoms in MPS-IIIA)

Coordinator: **Elvira De Leonibus**
Partners: Giancarlo Parenti, Zhifeng Huang

Duration (N. Years): **1** *Starting year:* **2019**

Telethon Project (nr): TIGEM

Disease Name:
Mucopolysaccharidosis Type III A

Poster 77

LYSOSOMAL AMYLOID DEPOSITION IMPAIRS AUTOPHAGY AND IS A DRUGGABLE TARGET FOR THE NEURODEGENERATION IN LYSOSOMAL STORAGE DISEASES

L'aggregazione amiloide lisosomiale danneggia l'autofagia risultando un possibile target per il trattamento terapeutico della neurodegenerazione nelle malattie da accumulo lisosomiale

Coordinator: **Alessandro Fraldi**

Duration (N. Years): **5** *Starting year:* **2016**

Telethon Project (nr): TIGEM

Disease Name:
Mucopolysaccharidosis Type III A

Poster 78

TARGETING LIPIDS IN CLN8-ASSOCIATED NCL DISEASES: STRUCTURAL AND FUNCTIONAL INTERACTION OF CLN8 WITH VESICLE-ASSOCIATED MEMBRANE PROTEIN-ASSOCIATED PROTEIN A (VAPA), AND GENOTYPE-PHENOTYPE CORRELATIONS

Ruolo dei lipidi nelle patologie NCL associate a CLN8: interazioni strutturali e funzionali del CLN8 con la proteina vescicolare di membrana-associata alla proteina A (VAPA), e correlazioni genotipo-fenotipo

Coordinator: **Patrizia Guarneri**
Partner: Alessandro Prinetti

Duration (N. Years): **3** *Starting year:* **2017**

Telethon Project (nr): GGP16277

Disease Name:
Neuronal Ceroid Lipofuscinoses (NCLs)

Poster 79

AGE-DEPENDENT BEHAVIORAL DEFICITS AND PROTEIN AGGREGATION IN LRRK2 HG2019S MICE

Deficit motori e aggregati proteici in un modello murino di Morbo di Parkinson

Coordinator: **Giovanni Piccoli**

Duration (N. Years): **5** *Starting year:* **2015**

Telethon Project (nr): TCP14005

Disease Name:
Parkinson's Disease

Poster 80

IMPLEMENTATION OF HUMAN NEURONAL CULTURES AND MOUSE MODELS OF PANTOTHENATE KINASE 2 DEFICIENCY TO INVESTIGATE PATHOGENIC MECHANISMS OF IRON-RELATED NEURODEGENERATION AND EVALUATE COENZYME A THERAPEUTIC EFFICACY

Sviluppo di modelli di cellule nervose umane e di modelli murini affetti da carenza di pantotenato chinasi-2 utili per lo studio dei meccanismi patogenetici di neurodegenerazione da accumulo di ferro e per valutare l'efficacia terapeutica del coenzima A

Coordinator: **Sonia Levi**
Partners: Vania Broccoli, Stefano Taverna, Valeria Tiranti

Duration (N. Years): **3** *Starting year:* **2017**

Telethon Project (nr): GGP16234

Disease Name:
PKAN and CoPAN

Poster 81

A NEW EXPLOITATION OF A PORPHYRIN WITH ANTI-PRION PROPERTIES: CHARACTERIZATION OF THE MECHANISM OF ACTION AND PRECLINICAL STUDIES IN MOUSE MODELS OF GENETIC PRION DISEASE

Una nuova applicazione di una porfirina ad attività antiprionica: caratterizzazione del meccanismo d'azione e studi preclinici in modelli murini di malattie da prioni di origine genetica

Coordinator: **Roberto Chiesa**
Partners: Stefano Banfi, Giovanna Musco

Duration (N. Years): **3** *Starting year:* **2015** *Project ending year:* **2019**

Telethon Project (nr): GGP15225

Disease Name:
Prion Genetic Diseases

Poster 82

ALTERNATIVE TRANSLATION INITIATION AS A NOVEL STRATEGY TO BLOCK TOXICITY OF THE MUTANT ANDROGEN RECEPTOR IN SBMA

Inizio alternativo della traduzione come nuova strategia per bloccare la tossicità del recettore degli androgeni mutato nell'atrofia muscolare spinale e bulbare (SBMA)

Coordinator: **Angelo Poletti**
Partner: Maria Pennuto

Duration (N. Years): **3** *Starting year:* **2019**

Telethon Project (nr): GGP19128

Disease Name:
Spinal and Bulbar Muscular Atrophy (SBMA)

Poster 83

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

La morte del motoneurone nell'atrofia muscolare spinale e bulbare: approcci molecolari per contrastare la tossicità del recettore degli androgeni mutato

Coordinator: **Angelo Poletti**

Duration (N. Years): **3** *Starting year:* **2014** *Project ending year:* **2018**

Telethon Project (nr): GGP14039

Disease Name:
Spinal and Bulbar Muscular Atrophy (SBMA)

Poster 84

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

Dal meccanismo patogenetico alla terapica in SCA38, una nuova forma di atassia spinocerebellare

Coordinator: **Barbara Borroni**
Partners: Alfredo Brusco, Donatella Caruso, Loredana Boccone, Filippo Tempia

Project ending year: **2019**

Telethon Project (nr): GGP14225

Disease Name:
Spinocerebellar Ataxia 38 (SCA38)

OTHER GENETIC DISORDERS

Inborn errors of metabolism

Poster 85

OXIDATIVE LIPIDOMICS IN BARTH SYNDROME

Fenomeni ossidativi nella sindrome di Barth: focus sulla Cardiolipina

Coordinator: **Angela Corcelli**

Duration (N. Years): **1** *Starting year:* **2019**

Telethon Project (nr): GGP19091

Disease Name:
Barth Syndrome

Poster 86

CREATINE DEFICIENCY SYNDROME: NOVEL INSIGHT INTO BRAIN FUNCTION AND THERAPEUTIC STRATEGIES

Coordinator: **Laura Baroncelli**
Partner: Alessandro Gozzi

Duration (N. Years): **3** *Starting year:* **2020**

Telethon Project (nr): GGP19177

Disease Name:
Creatine Transporter Deficiency

Poster 87

CIRCULATING ANTI-GB3 ANTIBODY AS BIOMARKER OF MYOCARDIAL INFLAMMATION IN PATIENTS WITH FABRY DISEASE CARDIOMYOPATHY

Anticorpi antiGB3 circolanti come biomarkers di infiammazione miocardica nella cardiomiopatia di Fabry

Coordinator: **Cristina Chimenti**

Duration (N. Years): **1** *Starting year:* **2019**

Telethon Project (nr): GGP19171

Disease Name:
Fabry Disease

Poster 88

METABOLIC REPROGRAMMING OF T REGULATORY CELLS AS THERAPEUTIC TOOL TO DAMPEN THE IMMUNO-INFLAMMATORY RESPONSE ASSOCIATED TO ATHEROSCLEROSIS IN PATIENTS AFFECTED BY FAMILIAL HYPERCHOLESTEROLAEMIA

Riprogrammazione metabolica di cellule T regolatorie come trattamento della risposta immuno-infiammatoria associata all'aterosclerosi in pazienti affetti da ipercolesterolemia familiare

Coordinator: **Giuseppe Danilo Norata**

Duration (N. Years): **3** *Starting year:* **2019**

Telethon Project (nr): GGP19146

Disease Name:
Familial Hypercholesterolaemia

Poster 89

EXPLOITING TARGETED EPIGENOME EDITING FOR THERAPEUTIC APPLICATIONS AND TO UNCOVER NOVEL GENE REPRESSION MECHANISMS

Sviluppo di una piattaforma per il silenziamento genico mirato e suo utilizzo per identificare nuovi meccanismi di repressione genica

Coordinator: **Angelo Lombardo**
Duration (N. Years): **5** *Starting year:* **2016**
Telethon Project (nr): TGT16F01; TGT16F05
Disease Name:
Familial Hypercholesterolemia, Memoglobinopathies

Poster 90

INCREASED AUTOIMMUNITY RISK IN GLYCOGEN STORAGE DISEASE TYPE 1B IS ASSOCIATED WITH ALTERATION OF REGULATORY T CELLS

L'aumento del rischio di sviluppare malattie autoimmunitarie nella glicogenosi di tipo 1b è associato con l'alterazione delle cellule T regolatorie

Coordinator: **Giuseppe Matarese**
Duration (N. Years): **2** *Starting year:* **2018**
Telethon Project (nr): GGP17086
Disease Name:
Glycogen Storage Disease Type 1B

Poster 91

EXPLOITING A BACTERIAL REDOX CYCLER AGAINST MITOCHONDRIAL DISEASE LINKED TO RESPIRATORY COMPLEX III DYSFUNCTION

Una molecola di origine batterica migliora i difetti causati da una disfunzione del complesso III della catena respiratoria presente in alcune malattie mitocondriali

Coordinator: **Ildiko Szabo**
Duration (N. Years): **3** *Starting year:* **end of 2019**
Telethon Project (nr): GGP19118
Disease Name:
Mitochondrial Complex III Disease

Poster 92

NOVEL THERAPEUTIC APPROACHES FOR COENZYME Q DEFICIENCY

Nuovi approcci terapeutici per il deficit di Coenzima Q

Coordinator: **Luca Scorrano**
Partner: Leonardo Salviati, Valerio Carelli, Paolo Bernardi
Duration (N. Years): **3** *Starting year:* **2014** *Project ending year:* **2018**
Telethon Project (nr): GGP14187
Disease Name:
Mitochondrial Diseases

Poster 93

HEMATOPOIETIC STEM CELL GENE THERAPY FOR MUCOPOLYSACCHARIDOSIS TYPE I, HURLER VARIANT (MPS-IH)

Terapia genica ex-vivo con cellule staminali ematopoietiche per la Mucopolisaccaridosi di tipo I, Hurler

PI: **Alessandro Aiuti**
Co-PI: Maria Ester Bernardo, Bernard Gentner
Telethon Project (nr): TGT16E06
Disease Name:
MPSIH

Poster 94

IN VIVO INDUCTION OF AG-SPECIFIC TOLERANCE BY HEPATOCYTE-TARGETED GENE TRANSFER

Coordinator: **Silvia Gregori**
Partners: Cantore Alessio
Duration (N. Years): **5** *Starting year:* **2016**
Telethon Project (nr): TGT16G02
Disease Name:
Mucopolysaccharidosis I / Type 1 Diabetes

Poster 95

POMPE DISEASE, NEW APPROACHES TO ADDRESS UNMET NEEDS

La malattia di Pompe, nuovi approcci per le problematiche irrisolte associate alla malattia

Coordinator: **Giancarlo Parenti**
Telethon Project (nr): TIGEM
Disease Name:
Pompe Disease

Poster 96

NOVEL THERAPIES FOR UREA CYCLE DISORDERS

Coordinator: **Nicola Brunetti-Pierri**
Duration (N. Years): **5** *Starting year:* **2017**
Telethon Project (nr): TIGEM
Disease Name:
Urea Cycle Disorders

Poster 97

INTEGRATED APPROACHES TO GENE THERAPY OF WILSON DISEASE

Approcci integrati di terapia genica per la malattia di Wilson

Coordinator: **Pasquale Piccolo**
Duration (N. Years): **3** *Starting year:* **2019**
Telethon Project (nr): TIGEM
Disease Name:
Wilson Disease

Chromosomal anomaly

Poster 98

ANALYTICAL METHOD: VALIDATION AND ANALYSES OF STUDY SAMPLES
Metodo analitico: convalida e analisi di campioni di studio

Telethon Project (nr): SR - TIGET

Disease Name:
Genetic Diseases

Genetic bone diseases

Poster 99

AUTOSOMAL DOMINANT OSTEOPETROSIS TYPE 2 (ADO2): CLOSE TO THE CURE. WHAT DO WE MISS?
Ostepetrosi autosomica dominante di Tipo 2 (ADO2): prossimi alla cura. Cosa manca?

Coordinator: **Anna Maria Teti**

Duration (N. Years): **3** *Starting year:* **2019**

Telethon Project (nr): GGP19031

Disease Name:
Autosomal Dominant Osteopetrosis Type 2

Poster 100

EXPANDED CIRCULATING HEMATOPOIETIC STEM/PROGENITOR CELLS AS A NOVEL CELL SOURCE FOR THE TREATMENT OF AUTOSOMAL RECESSIVE OSTEOPETROSIS
Espansione delle cellule staminali ematopoietiche per un innovativo trattamento dell'osteopetrosi autosomica recessiva

Coordinator: **Anna Villa**

Duration (N. Years): **5** *Starting year:* **2016**

Telethon Project (nr): TGT16C05

Disease Name:
Autosomal Recessive Osteopetrosis

Poster 101

EMERGING ROLES OF ER-PHAGY IN MAINTAINING CELLULAR FITNESS AND FUNCTION IN CHONDROCYTES
Ruoli Emergenti di ER-phagy nel mantenimento del benessere cellulare in condrociti

Coordinator: **Carmine Settembre**

Telethon Project (nr): TIGEM

Disease Name:
Chondrodysplasias

Poster 102

PROTEOSTASIS IN THE EARLY SECRETORY COMPARTMENT AS A PATHOGENETIC MECHANISM AND THERAPEUTIC TARGET: ALTERED COLLAGEN BIOSYNTHESIS AND BONE DEVELOPMENT IN THE ABSENCE OF ERP44, A ZINC-REGULATED CHAPERONE

Coordinator: **Roberto Sitia**

Starting year: **2017**

Telethon Project (nr): GGP15059

Disease Name:
Ehlers Danlos Disease

Poster 103

FIBROUS DYSPLASIA: A ROADMAP TO TREATMENT ENABLED BY DISCOVERY OF UNPREDICTED MECHANISMS IN FIRST-IN CLASS MOUSE MODELS

Coordinator: **Mara Riminucci**

Telethon Project (nr): GGP15198

Disease Name:
Fibrous Dysplasia

Poster 104

TMEM16E / ANO5 MUTATIONS RELATED TO BONE DYSPLASIA OR MUSCULAR DYSTROPHY CAUSE OPPOSITE EFFECTS ON LIPID SCRAMBLING

Coordinator: **Anna Elisabetta Boccaccio**

Duration (N. Years): 1 *Starting year:* **2016** *Project ending year:* **2017**

Telethon Project (nr): GEP15078

Disease Name:
Gnathodiaphyseal Dysplasia

Poster 105

OSTEOPETROSIS AND BARTTER SYNDROME: STRUCTURAL-FUNCTIONAL INVESTIGATION OF MUTATIONS CAUSING DISEASES
Analisi struttura-funzione di singole mutazioni che causano Osteopetrosi e sindrome di Bartter

Coordinator: **Alessandra Picollo**

Duration (N. Years): **5** *Starting year:* **2015**

Telethon Project (nr): TCP14008

Disease Name:
Osteopetrosis and Bartter's syndorme

Genetic cardiac diseases

Poster 106

OXIDIZED LDL/CD36/PPARΓ CIRCUITRY IS A TRIGGER OF ADIPOGENESIS IN ARRHYTHMOGENIC CARDIOMYOPATHY

Contributo dello stress ossidativo e dei lipidi ossidati nella patogenesi della Cardiomiopatia Aritmogena

Coordinator: **Giulio Pompilio**
Partners: Alessandra Rossini, Alberto Corsini

Duration (N. Years): **3** *Starting year:* **2017**

Telethon Project (nr): GGP16001

Disease Name:
Arrhythmogenic Cardiomiopathy

Genetic developmental defect during embryogenesis

Poster 107

MUTATIONS OF THE SUBCORTICAL MATERNAL COMPLEX AND IMPRINTING DISORDERS: HOW THE GENOTYPE INTERACTS WITH THE EPIGENOTYPE

Ruolo delle mutazioni materne nei disordini dell'imprinting: come il genotipo della madre influenza l'epigenotipo dei figli

Coordinator: **Andrea Riccio**

Durationg (N. Years): **3** *Starting year:* **2016**

Telethon Project (nr): GGP15131

Disease Name:
Beckwith-Wiedemann Syndrome/Siver-Russell Syndrome

Poster 108

A NOVEL SEMA3G MUTATION IN TWO SIBLINGS AFFECTED BY HYPOGONADISM, DEVELOPMENTAL DELAY AND FACIAL MALFORMATIONS

Coordinator: **Cariboni Anna**

Duration (N. Years): **3** *Starting year:* **2013** *Project ending year:* **2018**

Telethon Project (nr): GGP13142

Disease Name:
Hypogonadotropic Hypogonadism

Genetic eye diseases

Poster 109

INHIBITION OF AUTOPHAGY CURTAILS VISUAL LOSS IN A MODEL OF AUTOSOMAL DOMINANT OPTIC ATROPHY

L'inibizione dell'autofagia blocca la perdita di vista in un modello di atrofia ottica dominante

Coordinator: **Luca Scorrano**

Duration (N. Years): **3** *Starting year:* **2019**

Telethon Project (nr): GGP19089

Disease Name:
Autosomal Dominant Optic Atrophy

Poster 110

CONE DYSTROPHIES AND RETINAL DEGENERATION FROM PROTEIN STRUCTURES TO BIOLOGICAL NETWORKS: TOWARD THE DESIGN OF THERAPEUTIC MOLECULES

Coordinator: **Daniele Dell'Orco**
Partners: Mario Milani, Lorenzo Cangiano

Duration (N. Years): **3** *Starting year:* **2017**

Telethon Project (nr): GGP16010

Disease Name:
Cone Dystrophy

Poster 111

ENZYMATIC PHENOTYPE AND RESPONSE TO VITAMIN B6 OF ORNITHINE AMINOTRANSFERASE VARIANTS ASSOCIATED WITH GYRATE ATROPHY OF THE CHOROID AND RETINA

Coordinator: **Barbara Cellini**
Partner: Leonardo Salviati

Duration (N. Years): **3** *Starting year:* **2016** *Project ending year:* **2019**

Telethon Project (nr): GPP15114

Disease Name:
Gyrate Atrophy of the Choroid and Retina

Poster 112

MODULATION OF MICRORNA EXPRESSION: A NEW THERAPEUTIC AVENUE FOR INHERITED RETINAL DISEASE?

Modulazione dell'espressione di microRNA: una nuova strategia terapeutica per le malattie retiniche ereditarie?

Coordinator: **Sandro Banfi**

Telethon Project (nr): TIGEM

Disease Name:
Inherited Retinal Disease

Poster 113

THERAPEUTIC TARGETING OF MIR-211/EZRIN AXIS PREVENTS RETINAL DEGENERATION IN THE RHOP23H MOUSE MODEL

Il targeting terapeutico dell'asse miR-211/Ezrin previene la degenerazione retinica nel modello di topo RhoP23H

Coordinator: **Ivan Conte**

Telethon Project (nr): TIGEM

Disease Name:
Retinitis Pigmentosa

Poster 114

PIGMENT EPITHELIUM-DERIVED FACTOR (PEDF) PEPTIDES AS THERAPEUTIC AGENTS FOR INHERITED RETINAL DEGENERATION

Peptidi derivati da Pigment Epithelium-derived Factor (PEDF) come agenti terapeutici per la degenerazione retinica ereditaria

Coordinator: **Valeria Marigo**

Duration (N. Years): **3** *Starting year:* **2019**

Telethon Project (nr): GGP19113

Disease Name:
Retinitis Pigmentosa

Poster 115

INTEIN-MEDIATED PROTEIN TRANS-SPLICING EXPANDS ADENO-ASSOCIATED VIRUS TRANSFER CAPACITY IN THE RETINA

Il trans-splicing proteico mediato dalle inteine espande la capacità di trasferimento del virus adeno-associato nella retina

Coordinator: **Alberto Auricchio**
Duration (N. Years): **5** *Starting year:* **2016**
Telethon Project (nr): TIGEM
Disease Name:
Stargardt Disease, Leber Congenital Amaurosis 10

Genetic gastroenterological diseases

Poster 116

DISCOVERING MOLECULAR DEFECTS OF SEVERE GUT DYSFUNCTION: NEW ABNORMALITIES UNDERLYING CHRONIC INTESTINAL PSEUDO-OBSTRUCTION (CIPO)

Lo studio dei difetti molecolari nella pseudo-ostruzione intestinale cronica: identificazione di nuove alterazioni genetiche e funzionali

Coordinator: **Elena Bonora**
Partners: Francesca Bianco, Roberto De Giorgio
Duration (N. Years): **3** *Starting year:* **2015**
Telethon Project (nr): GGP15171
Disease Name:
Chronic Intestinal Pseudo-Obstruction

Genetic hematologic diseases

Poster 117

DEFINING HEMATOPOIESIS IN BETA-THALASSEMIA PATIENTS AND AFTER GENE THERAPY

Studio dell'emopoiesi in pazienti talassemici e dopo terapia genica

Coordinator: **Giuliana Ferrari**
Duration (N. Years): **5** *Starting year:* **2016**
Telethon Project (nr): TGT16A04
Disease Name:
Beta-Thalassemia

Poster 118

REGULATION OF HEMATOPOIESIS IN NORMAL AND STRESSED CONDITIONS

Coordinator: **Giuliana Ferrari**
Duration (N. Years): **5** *Starting year:* **2016**
Telethon Project (nr): TGT16A03
Disease Name:
Beta-Thalassemia

Poster 119

GENE THERAPY FOR THE TREATMENT OF ADULT AND PEDIATRIC PATIENTS AFFECTED BY TRANSFUSION DEPENDENT BETA-THALASSEMIA

Coordinator: **Giuliana Ferrari**
Telethon Project (nr): SR-TIGET GENE THERAPY FOR BETA THALASSEMIA
Disease Name:
Beta-Thalassemia

Poster 120

DISSECTING CELL SENESCENCE PROGRAMS IN THE HEMATOPOIETIC COMPARTMENT

Coordinator: **Raffaella Di Micco**
Telethon Project (nr): TGT16E05
Disease Name:
Diseases of Immune-Hematological Lineage

Poster 121

LIVER-DIRECTED GENE THERAPY WITH LENTIVIRAL VECTORS ACHIEVE NORMAL LEVELS OF CLOTTING FACTOR VIII AND IX IN NON-HUMAN PRIMATES

La terapia genica diretta al fegato con vettori lentivirali permette di raggiungere livelli normali di fattore VIII e IX della coagulazione in primati non umani

Coordinator: **Luigi Naldini, Alessio Cantore**
Duration (N. Years): **5** *Starting year:* **2016**
Telethon Project (nr): TGT16D04
Disease Name:
Hemophilia

Poster 122

CONVENTIONAL DCS AND ENDOGENOUS TRYPTOPHAN DERIVATIVES PREVENT THE DEVELOPMENT OF ANTI-FVIII ANTIBODIES IN HEMOPHILIA A MODEL

Le cellule dendritiche convenzionali e derivati del triptofano inibiscono la formazione di anticorpi anti-FVIII in un modello di emofilia A

Coordinator: **Francesca Fallarino**
Duration (N. Years): **3** *Starting year:* **2017**
Telethon Project (nr): GGP17094
Disease Name:
Hemophilia

Poster 123

FROM COAGULATION TO ANGIOGENESIS: NEW ROLES FOR FVIII IN ENDOTHELIAL FUNCTIONALITY

Dalla coagulazione all'angiogenesi nuovi ruoli del FVIII nella funzionalità endoteliale

Coordinator: **Antonia Follenzi**
Partner: Salvatore Oliviero
Duration (N. Years): **3** *Starting year:* **2019**
Telethon Project (nr): GGP19201
Disease Name:
Hemophilia A

Poster 124

GENOMIC MECHANISMS OF HUMAN GRANULOPOIESIS: IMPLICATIONS FOR BONE MARROW RECONSTITUTION AFTER GENE THERAPY

Coordinator: **Renato Ostuni**
Starting year: **2016**
Telethon Project (nr): TGT16F04
Disease Name:
HSC Gene Therapy, HSC Transplantation

Poster 125

MESODERMAL RETINOIC ACID SIGNALING REGULATES THE SPECIFICATION OF HUMAN DEFINITIVE HEMATOPOIETIC PROGENITORS FROM HUMAN PLURIPOTENT STEM CELLS
L'attivazione della via di segnalazione dell'Acido Retinoico regola la specificazione dei progenitori ematopoietici umani dalle cellule staminali pluripotenti umane

Coordinator: **Andrea Ditadi**
Duration (N. Years): **5** *Starting year:* **2016**
Telethon Project (nr): TGT16C04
Disease Name:
inherited Bone Marrow Failure

Poster 126

MODELLING THE EMBRYONIC ORIGIN OF OMENN SYNDROME AUTO-REACTIVE T-CELLS
Studio dell'origine embrionale dei linfociti T autoreattivi nella Sindrome di Omenn

Coordinator: **Andrea Ditadi**
Duration (N. Years): **5** *Starting year:* **2016**
Telethon Project (nr): TGT16C04
Disease Name:
Omenn Syndrome

Poster 127

EX VIVO EXPANSION OF GENETICALLY-ENGINEERED HEMATOPOIETIC STEM AND PROGENITOR CELLS FROM MOBILIZED PERIPHERAL BLOOD

Coordinator: **Bernhard Gentner**
Duration (N. Years): **5** *Starting year:* **2016**
Telethon Project (nr): TGT16C01
Disease Name:
Platform Technology

Poster 128

GENE CORRECTION OF CD40LG GENE IN T CELLS AND HSPC FOR THE TREATMENT OF X-LINKED HYPER-IGM IMMUNODEFICIENCY

Coordinator: **Pietro Genovese**
Duration (N. Years): **5** *Starting year:* **2016**
Telethon Project (nr): TGT16E3
Disease Name:
X-linked Hyper IgM Syndrome

Genetic hepatic diseases

Poster 129

IDENTIFICATION AND THERAPEUTIC TARGETING OF NEW MOLECULAR PATHWAYS IN WILSON DISEASE
Identificazione di nuovi meccanismi molecolari come potenziali target terapeutici nella Malattia di Wilson

Coordinator: **Roman Polishchuk**
Duration (N. Years): **2** *Starting year:* **2017**
Telethon Project (nr): TIGEM
Disease Name:
Wilson Disease

Genetic immune diseases

Poster 130

MODULATION OF LINE-1 RETROTRANSPOSITION BY AICARDI-GOUTIÈRES SYNDROME-RELATED GENES
Regolazione della retrotrasposizione di LINE1 da parte dei geni responsabili della singrome di Aicardi-Goutières.

Coordinator: **Marco Muzi Falconi**
Duration (N. Years): **3** *Starting year:* **2015**
Telethon Project (nr): GGP15227
Disease Name:
Aicardi-Goutières Syndrome

Poster 131

GENE THERAPY AND PATHOGENESIS OF CHRONIC GRANULOMATOUS DISEASE
Terapia genica e patogenesi della malattia granulomatosa cronica

Coordinator: **Alessandro Aiuti**
Duration (N. Years): **5** *Starting year:* **2016**
Telethon Project (nr): SR-TIGET PROJECT GENE THERAPY
Disease Name:
Chronic Granulomatous Disease

Poster 132

SCREENING CVID PATIENTS WITH T CELL DEFECTS FOR PATHOGENIC VARIANTS OF CILIARY PROTEINS IDENTIFIES CCDC28 AS NEW PLAYER IN IMMUNE SYNAPSE ASSEMBLY

Lo screening di pazienti CVID per varianti patogeniche di proteine ciliari identifica CCDC28B quale coordinatore dell'assemblaggio della sinapsi immunologica

Coordinator: **Cosima T. Baldari**
Duration (N. Years): **3** *Starting year:* **2017**
Telethon Project (nr): GGP16003
Disease Name:
CVID (Common Variable Immunodeficiency)

Poster 133

MECHANISMS OF ENHANCED HEMATOPOIETIC STEM CELL TRANSDUCTION AND NUCLEIC ACID SENSING

Meccanismi di efficiente trasduzione e di riconoscimento degli acidi nucleici nelle cellule staminali ematopoietiche

Coordinator: **Anna Kajaste-Rudnitski**
Duration (N. Years): **5** *Starting year:* **2016**
Telethon Project (nr): TGT16C03
Disease Name:
Genetic Disorders

Poster 134

ADVANCED GENETIC ENGINEERING OF HEMATOPOIETIC STEM/PROGENITOR CELLS

Ingegneria Genetica Innovativa di Cellule Progenitrici/Staminali Ematopoietiche

Coordinator: **Luigi Naldini**
Duration (N. Years): **5** *Starting year:* **2016**
Telethon Project (nr): TGT16E04
Disease Name:
HIGM Syndrome

Poster 135

REGULATION OF PATHOGEN-SPECIFIC T-CELL RESPONSES IN PATIENTS WITH HYPER-IGE SYNDROME (HIES)

Coordinator: **Jens Geginat**
Duration (N. Years): **3** *Starting year:* **2019-2020**
Telethon Project (nr): GGP19323
Disease Name:
Hyper - Ige Syndrome (HIES)

Poster 136

EXPLORING THE PATHOGENETIC BASIS OF ICF SYNDROME WITH HUMAN INDUCED PLURIPOTENT STEM CELLS

Coordinator: **Maria R. Matarazzo**
Duration (N. Years): **3** *Starting year:* **2015**
Telethon Project (nr): GGP15209
Disease Name:
ICF Syndrome

Poster137

IDENTIFICATION AND THERAPY OF COMBINED IMMUNODEFICIENCIES AND ADENOSINE DEAMINASE 2 DEFICIENCY

Identificazione e terapia delle immunodeficienze combinate e del deficit di adenosina deaminasi 2

Coordinator: **Alessandro Aiuti**
Duration (N. Years): **5** *Starting year:* **2017**
Telethon Project (nr): TGT16C06
Disease Name:
Immunodeficiencies

Poster 138

TARGETED GENOME EDITING IN RECOMBINATION ACTIVATING GENE 1 (RAG1): A PRECISE CORRECTION OF THE GENETIC DEFECT IN HUMAN SCID

Coordinator: **Anna Villa**
Duration (N. Years): **5** *Starting year:* **2016**
Telethon Project (nr): TGT16E02
Disease Name:
RAG1 Severe Combined Immunodeficiency

Poster 139

NOVEL STRATEGIES TO GENERATE TOLEROGENIC DENDRITIC CELLS FOR ANTIGEN-SPECIFIC IMMUNOTHERAPY

Coordinator: **Silvia Gregori**
Duration (N. Years): **5** *Starting year:* **2016**
Telethon Project (nr): TGT16G01
Disease Name:
T Cell Mediated Diseases

Poster 140

HSPC BIOLOGY: IN VIVO CLONAL TRACKING AND LINEAGE MODELING

Tracking clonale delle cellule staminali e dei progenitori ematopoietici in vivo in esseri umani

Coordinator: **Alessandro Aiuti**
Duration (N. Years): **5** *Starting year:* **2016**
Telethon Project (nr): TGT16B02
Disease Name:
Wiskott Aldrich Syndrome

Poster 141

SAP AND DIACYLGLYCEROL KINASE A RECIPROCALLY REGULATE TCR SIGNALLING

Coordinator: **Andrea Graziani**
Partner: Gianluca Baldanzi
Duration (N. Years): **3** *Starting year:* **2016**
Telethon Project (nr): GGP16252
Disease Name:
XLP-1 (Duncan Syndrome)

Genetic renal diseases

Poster 142

MOLECULAR MECHANISMS OF PATHOGENESIS AND PRECLINICAL TREATMENT IN RENAL DISORDERS ASSOCIATED WITH UROMODULIN MUTATIONS

Meccanismi molecolari di patogenesi e trattamento in modello preclinico della malattia renale associata a mutazioni di uromodulina

Coordinator: **Luca Rampoldi**

Duration (N. Years): **3** *Starting year:* **2014** *Project ending year:* **2018**

Telethon Project (nr): GGP14263

Disease Name:
Autosomal Dominant Tubulointerstitial Kidney Diseas

Poster 143

UNRAVELLING THE ROLE OF PAX2 MUTATIONS IN HUMAN FOCAL SEGMENTAL GLOMERULOSCLEROSIS

Comprendere il ruolo delle alterazioni del gene di pax2 nello sviluppo di glomerulosclerosi focale segmentaria

Coordinator: **Ariela Benigni**

Duration (N. Years): **3** *Starting year:* **2017**

Telethon Project (nr): GGP17221

Disease Name:
Focal Segmental Glomerulosclerosis

Poster 144

DECODING AND TARGETING THE MTORC1-TFEB AXIS IN GENETIC DISEASES

Decodifica e targeting dell'asse mTORC1-TFEB nelle malattie genetiche

Coordinator: **Andrea Ballabio**

Starting year: **2016**

Telethon Project (nr): TIGEM

Disease Name:
Genetic Diseases, Birt-Hogg-Dubé (BHD) Syndrome

Genetic respiratory diseases

Poster 145

COMPUTATIONAL AND QUANTITATIVE BIOLOGY IN RARE GENETIC DISEASES

Coordinator: **Diego Di Bernardo**

Telethon Project (nr): TIGEM

Disease Name:
Cystic Fibrosis, Lysosomal Storage Disorders

Genetic skin diseases

Poster 146

A FUNCTIONAL GENOMICS FRAMEWORK TO INVESTIGATE THE MOLECULAR BASES OF RARE GENETIC DISEASES

Coordinator: **Davide Cacchiarelli**

Duration (N. Years): **5** *Starting year:* **2017**

Telethon Project (nr): TIGEM STARTUP GRANT

Disease Name:
AEC Syndrome

Poster 147

THERAPEUTIC STRATEGIES TO RESCUE SKIN EROSIONS IN AEC SYNDROME

Coordinator: **Caterina Missero**

Duration (N. Years): **3** *Starting year:* **2017**

Telethon Project (nr): GGP16235

Disease Name:
AEC Syndrome or Hay-Wells Syndrome

Poster 148

A THERAPEUTIC APPROACH FOR RARE GENODERMATOSES CAUSED BY ABERRANT CONNEXIN HEMICHANNELS

Un approccio terapeutico per genodermatosi rare causate da emicanali aberranti di connessine espressi nell'epidermide

Coordinator: **Fabio Mammano**

Duration (N. Years): **2** *Starting year:* **2019**

Telethon Project (nr): GGP19148

Disease Name:
Keratitis - Ichthyosisdeafness (KID) Syndrome

Genetic systemic or rheumatologic diseases

Poster 149

NEW PHARMACOLOGICAL TARGETS AND STRATEGIES IN AGEL AMYLOIDOSIS

Nuove strategie e bersagli farmacologici contro l'amiloidosi da gelsolina

Coordinator: **Matteo de Rosa**

Duration (N. Years): **1** *Starting year:* **2016** *Project ending year:* **2017**

Telethon Project (nr): GEP15070

Disease Name:
Agel Amyloidosis

Poster 150

THE ROLE OF TELOMERIC DILNCRNAS AND DDRNAS IN THE HUTCHINSON-GILFORD PROGERIA SYNDROME
Il ruolo dei dilncRNA e DDRNA telomerici nella sindrome di Hutchinson-Gilford

Coordinator: **Fabrizio d'Adda di Fagagna**
Duration (N. Years): **3** *Starting year:* **2018**
Telethon Project (nr): GGP17111
Disease Name:
Hutchinson-Gilford Progeria Syndrome

Poster 151

IDENTIFICATION OF CORRECTORS OF LOWE SYNDROME
Identificazione di correttori della Sindrome di Lowe

Coordinator: **Maria Antonietta De Matteis**
Starting year: **2016**
Telethon Project (nr): TIGEM
Disease Name:
Lowe Syndrome

Genetic vascular diseases

Poster 152

ENDOTHELIAL CELL CLONAL EXPANSION IN THE DEVELOPMENT OF CEREBRAL CAVERNOUS MALFORMATIONS
Il ruolo della espansione clonale delle cellule endoteliali nella formazione delle malformazioni vascolari cavernose

Coordinator: **Elisabetta Dejana**
Duration (N. Years): **3** *Starting year:* **2019**
Telethon Project (nr): GGP19202
Disease Name:
Cerebral Cavernous Malformations (CCM)

Poster 153

CROSSTALK BETWEEN OXIDATIVE STRESS AND INFLAMMATION IN THE PATHOGENESIS OF CEREBRAL CAVERNOUS MALFORMATION (CCM) DISEASE: FROM THE IDENTIFICATION OF BASIC MECHANISMS TO THE DEVELOPMENT OF THERAPEUTIC STRATEGIES
Identificazione del ruolo cruciale dello stress ossidativo e dell'infiammazione nella patogenesi delle malformazioni cavernose cerebrali: dalla scoperta dei meccanismi molecolari allo sviluppo di nuove strategie terapeutiche

Coordinator: **Saverio Francesco Retta**
Partners: Paolo Pinton, Lorenza Trabalzini
Duration (N. Years): 3 *Starting year:* **2015** *Project ending year:* **2019**
Telethon Project (nr): GGP15219
Disease Name:
Carebral Cavernous Malformation (CCM)

Undiagnosed diseases with proven genetic origin

Poster 154

THREE YEARS OF THE TELETHON UNDIAGNOSED DISEASES PROGRAM: DATA AND FINDINGS
Tre anni di Telethon Undiagnosed Diseases Program: dati e risultati

Coordinator: **Giorgio Casari**
Partners: Angelo Selicorni, Nicola Brunetti-Pierri
Duration (N. Years): **4** *Starting year:* **2016**
Telethon Project (nr): GSP15001
Disease Name:
Undiagnosed Diseases

GENETIC BIOBANKS

Poster 155

TELETHON NETWORK OF GENETIC BIOBANKS

Coordinator: **Luca Sangiorgi**
Partners: Marina Stroppiano, Domenico Coviello, Roberto Cilia, Alessandra Renieri, Elena Pegoraro, Monica Sciacco, Marina Mora, Giuseppe Merla, Luisa Politano, Barbara Garavaglia
Duration (N. Years): **3** *Starting year:* **2018**
Telethon Project (nr): GTB18001
Disease Name:
Genetic Diseases in general

INSTITUTIONAL POSTERS

Poster 156

TIGEM INSTITUTE OVERVIEW

Authors: **Staff Tigem**
Disease Name:
Genetic Diseases

Poster 157

SR-TIGET INSTITUTE OVERVIEW
Istituto San Raffele-Telethon per la Terapia Genica (SR-TIGET)

Authors: **Aida Paniccia, Luigi Naldini**
Disease Name:
Genetic Diseases

Poster 158

L'UNIVERSO TELETHON: CHI SIAMO, COSA FACCIAMO E PER CHI LAVORIAMO

Authors: **Fondazione Telethon**

Disease Name:
Genetic Diseases

Poster 159

LA RACCOLTA FONDI: COSA SI FA E CHI CI LAVORA

Authors: **Fondazione Telethon**

Disease Name:
Genetic Diseases

Poster 160

I NOSTRI PRODOTTI DI PIAZZA

Authors: **Fondazione Telethon**

Disease Name:
Genetic Diseases

Poster 161

IO ADOTTO IL FUTURO: IL PROGRAMMA DI DONAZIONE REGOLARE

Authors: **Fondazione Telethon**

Disease Name:
Genetic Diseases

Poster 162

VIAGGIO AL CENTRO DEI SOCIAL TELETHON

Authors: **Fondazione Telethon**

Disease Name:
Genetic Diseases

Poster 163

MONITORING FONDAZIONE TELETHON'S RESEARCH INVESTMENT FOR A STRATEGIC PORTFOLIO MANAGEMENT
Monitoraggio dell'investimento in ricerca della Fondazione Telethon per una gestione strategica del portafoglio

Authors: **Lucia Monaco, Danila Baldessari, Aldo Borrè**

Disease Name:
Genetic Diseases

Poster 164

FROM THE DEFINITION OF RESEARCH INITIATIVES TO THE MONITORING OF FUNDED RESEARCH: THE TELETHON RESEARCH TEAM AT A GLANCE
Dalla definizione di iniziative di ricerca al monitoraggio della ricerca finanziata: il team di ricerca di Telethon a colpo d'occhio

Authors: **Manuela Battaglia, Anna Ambrosini, Elena Bruno, Ermanno Rizzi, Alessandra Zatti**

Disease Name:
Genetic Diseases

Poster 165

BUSINESS DEVELOPMENT OFFICE: FROM THE LAB TO THE MARKET

Authors: **Annamaria Merico, Federica Basilico, Elena Beltrami, Barbara Sanavio, Simona Varani**

Disease Name:
Genetic Diseases

Poster 166

ALLIANCE MANAGEMENT & REGULATORY AFFAIRS - DRIVING INDUSTRIAL COLLABORATIONS TO SPEED UP THE DEVELOPMENT OF ADVANCED THERAPIES AND MAKE THEM AVAILABLE TO PATIENTS
Gestione Alleanze Industriali e Affari Regolatori: Gestione attiva delle collaborazioni industriali per accelerare lo sviluppo di terapie avanzate e renderle disponibili ai pazienti

Authors: **Michela Gabaldo, Giada Farinelli, Claudia Forni**

Disease Name:
Genetic Diseases

Poster 167

CLINICAL DEVELOPMENT AND JUST LIKE HOME - HIGH QUALITY SUPPORT FOR CLINICAL TRIALS AND PATIENTS
Sviluppo clinico e 'Come a casa' – un supporto di alta qualità per le sperimentazioni cliniche e per i pazienti

Authors: **Stefano Zancan, Ambra Corti, Alda Graziano, Margherita Levi, Carmela Acerra**

Disease Name:
Genetic Diseases

Poster 168

ADVANCING RARE GENETIC DISEASE RESEARCH AND THERAPY DEVELOPMENT VIA INTERNATIONAL PARTNERSHIPS AND PROJECTS
Promuovere la ricerca sulle malattie genetiche rare e lo sviluppo di terapie attraverso partenariati e progetti internazionali

Authors: **Stefano Benvenuti, Chiuhui Mary Wang, Lucia Monaco, Michela Gabaldo**

Disease Name:
Genetic Diseases

Poster 169

THE INTERNATIONAL RARE DISEASES RESEARCH CONSORTIUM (IRDIRC)

IRDiRC, il Consorzio Internazionale per la RIcerca sulle Malattie Rare

Authors: **Carla S. D'Angelo, Daria Julkowska, Galliano Zanello, Katie Buchholz, Mary Wang Chiuhui, David Pearce, Lucia Monaco**

Disease Name:
Rare Diseases

Poster 170

THE EUROPEAN JOINT PROGRAMME ON RARE DISEASES (EJP RD)

EJP RD, il Programma Europeo Congiunto sulle Malattie Rare

Authors: **Carla S. D'Angelo, Daria Julkowska**

Disease Name:
Rare Diseases

Poster 171

ARISLA, THE ITALIAN FOUNDATION FOR ALS RESEARCH: MISSION, VISION, AND OUTCOMES OF TEN-YEAR INVESTMENT

AriSLA, la fondazione italiana di ricerca per la sla: mission, vision, e risultati dei primi 10 anni di investimenti

Authors: **Maddalena Ravasi, Anna Ambrosini, Stefania Guareschi, Luca M. Munari, Silvia Pozzi**

Disease Name:
Amyotrophic Lateral Sclerosis

FONDAZIONE

INDEX BY AUTHOR (PI Coordinators and Partners)
INDICE PER AUTORI (Responsabili dei progetti)

INDEX BY PATHOLOGY

INDICE PER PATOLOGIA

POSTER

POSTER

POSTER

POSTER

POSTER

POSTER

POSTER

POSTER

POSTER

POSTER

ASSOCIAZIONI AMICHE

ALBINIT
Lucia Pellegrini

ASSI Gulliver
Gianluca Poddighe, Giovanni Todaro

Associazione AMA.LE IQSEC2
Danila Borio, Giacomo Giordani, Mara Peyrot

Associazione Conto alla Rovescia
Davide Scognamiglio

Associazione Famiglie di soggetti con deficit di ormone della crescita ed altre patologie
Cinzia Sacchetti

Associazione Familiari Insonnia Familiare Fatale
Vladimiro Artuso

Associazione Fibrodisplasia Ossificante Progressiva
Enrico Cristoforetti

Associazione Genitori Monosomie 18
Sarah Russotti

Associazione Italiana Acidemia Metilmalonica Con Omocistinuria Cblc Onlus
Rosa Brindisi

Associazione Italiana Amaurosi Congenita di Leber
Nicola Stilla

Associazione Italiana Angiomi cavernosi Onlus
Massimo Chiesa

Associazione Italiana Delezione del Cromosoma 22
Alfredo Carratello

Associazione Italiana Displasia Setto Ottica e Ipoplasia del Nervo Ottico
Diana Vitali

Associazione Italiana Febbri Periodiche
Paolo Calveri

Associazione Italiana Gaucher
Laura Pastore

Associazione Italiana Glicogenosi Onlus
Vincenzo Mazzotti, Angela Tritto

Associazione Italiana GLUT1 Onlus
Bianca Borri, Monica Lucente, Lorenza Uzzeni

Associazione Italiana Malati di Alcaptonuria
Silvia Sestini

Associazione Italiana Malattia Frontotemporale
Maria Loda, Silvana Morson

Associazione Italiana Mowat Wilson
Greta Barbanti, Diego Zotti

Associazione Italiana Mucopolisaccaridosi e Malattie Affini
Flavio Bertoglio

Associazione Italiana Niemann Pick e malattie affini Onlus
Annalisa Bisconti

Associazione Italiana per la Ricerca sulla Distonia
Beatrice Pozzoli

Associazione Italiana per la Sindrome di Ipoventilazione Centrale Congenita (Sindrome di Ondine)
Alessandro Carcano

Associazione Italiana per la Sindrome di Phelan-McDermid
Michela Colombara

Associazione Italiana Rene Policistico
Sandrini Grazia, Sani Maria Vittoria

Associazione Italiana Sindrome di Beckwith-Wiedemann
Monica Bertoletti, Daniela Valle

Associazione Italiana Sindrome di Costello Cardiofaciocutanea Rasopatie Onlus
Maria Grazia Pungente

Associazione Italiana Sindrome di Ehlers – Danlos Onlus
Laura Traversone

Associazione Italiana Sindrome di Noonan Onlus
Francesco Crudo, Angelo Petroni

Associazione Italiana Sindrome di Pallister-Killian Onlus
Samantha Carletti

Associazione Italiana Sindrome di Poland
Maria Di Matteo

Associazione Italiana Sindrome di Williams Onlus
Maurizio Granato

Associazione Italiana Sindrome Kabuki
Marta Gobbetti

Associazione Italiana Sindrome X Fragile
Alessia Brunetti, Viviana Scannicchio

Associazione Italiana Sindromi Neurodegenerative da Accumulo di Ferro
Natale Scalise

Associazione italiana Vivere la Paraparesi Spastica
Gerardo Carrillo, Maria Piera Pognante, Giorgia Tartaglia, Marina Zapparoli Manzoni

Associazione Italiana Xeroderma Pigmentoso APS
Barbara Accoriniti, Valentina Cagnoni

Associazione Ligure Sindrome X-Fragile Onlus
Daniele Nasi

Associazione Marfan per la sindrome di Marfan e le patologie correlate
Isacco Formentini, Manuela Gandolfo

Associazione Nazionale ALFA 1-AT
Cristina Barbiero, Annamaria Fra

Associazione Nazionale Angioma Cavernoso Cerebrale
Mara Gatti, Francesca Maspero, Rita Treglia

Associazione Nazionale Atassia Telangiectasia Onlus
Sara Biagiotti

Associazione Nazionale Ceroidolipofuscinosi
Lorena Piccinato, Mara Piccinato

Associazione nazionale famiglie di persone con disabilità intellettive e/o relazionali
Biancamaria Lanzetta

Associazione Nazionale Malattia di Wilson onlus
Salvatore Di Lorenzo

Associazione Nazionale Onlus per la Malattia di Charcot Marie Tooth
Marina Garcea

Associazione Nazionale Porpora Trombotica Trombocitopenica ONLUS
Rossana Giannone

Associazione Nazionale Sindrome di Noonan e RASopatie ODV
Bruno Smorto

Associazione p63 Sindrome E.E.C. International network word communication-MALATTIE RARE ONLUS
Cristina Bolzonella

Associazione per la ricerca sulla Sindrome di Rett Onlus
Salvatore Franzé, Roberto Turrini

Associazione per le Immunodeficienze Primitive
Alberto Barberis

Associazione per le malattie reumatiche infantili onlus
Stefania Cortigiano

Associazione Persone Williams Italia Onlus
Paola Risso

Associazione Progetto Mitofusina 2 Onlus
Silvia Mari

Associazione Ricerca Diabete Italia
Aurora Laura Panizzi

Associazione Sclerodermia ed Altre Malattie Rare
Maria Pia Sozio

Associazione Sclerosi Tuberosa
Carmine Lechiara, Francesca Macari

Associazione Sindrome di Klinefelter Italiana Sicilia ONLUS
Vincenzo Grafeo

Associazione Sindrome Sturge Weber Italia
Antonella Perini, Annalisa Valentini, Michela Volpi

Associazione Volontaria per la Lotta lo Studio e la Terapia dell'Angioedema Ereditario
Carlotta Cicardi, Martina Perera

Associazione Xia Gibbs
Baldinetti Carlo, Tino Roberta

Barth Italia Onlus
Paola Cazzaniga

CDKL5 Associazione di volontariato
Emanuela Defranceschi, Antonella Statello

Cometa Associazione Studio Malattie Metaboliche Ereditarie
Graziano Bacco

DADA2 ETS
Annamaria Beretta

Dodò
Ivana Arrisicato

F.A.R.C.E.L.A. con l'autismo e i DGS Onlus
Maurizio Alberton, Monica Mocellin

Famiglie SMA
Rosa Zogno

Federazione Nazionale Prader Willi - ONLUS
Miriam Idome

Fondazione Italiana 'Leonardo Giambrone' per la Guarigione della Thalassemia
Tommasina Iorno

Giovani e Cuore Aritmico
Graziella Marcon, Michele Menin

GNAO1
Massimiliano Tomassi

Gruppo di Sostegno DBA Italia Onlus
Maria Elisabetta Villa

Gruppo Famiglie Dravet Associazione Onlus
Simona Borroni, Rita Rega

Gruppo Familiari Beta-Sarcoglicanopatie Onlus
Ersilia Forneti, Carles Sanchez Riera

International Aicardi-Goutières Syndrome Association
Nadia Cairati

Kool Kids Italia
Lorenzo Ciapparelli, Francesco Volpatti

Lega per la Neurofibromatosi 2 APS
Stefania Mostaccioli

LND Famiglie Italiane Onlus
Paola Mazzucchi

MITOCON - Insieme per lo studio e la cura delle malattie mitocondriali
Cristina Rebagliati

Nonsolo15 associazione per la ricerca e il sostegno di familiari e amici di persone affette da sindrome invdup15 (idic15) onlus
Francesca Cappelletti, Christine Reimann

Parent Project onlus
Cristina Picciolo, Ilaria Zito

PTEN Italia Associazione Italiana per la lotta alle PHTS
Claudio Ales

R.T.S una vita speciale onlus
Sonia Pizzato

Retina Italia Onlus
Roberto Rabito

Retinite Pigmentosa Liguria
Claudio Pisotti

Una Famiglia per il Cuore Onlus
Adele Bigiarini, Claudio Bigiarini

UNIAMO FIMR Onlus
Annalisa Scopinaro

Unione Italiana Lotta alla Distrofia Muscolare
Stefania Pedroni

VOA VOA
Guido De Barros

XLPDR International Association Onlus
Patrizia Gentile

FONDAZIONE

Rome headquarter
Via Varese, 16/B
00185 Rome, Italy
Tel. +39 06 440151
Fax +39 06 44015521

Milan headquarter
Via Carlo Poerio, 14
20129 Milan, Italy
Tel. +39 02 2022171
Fax +39 06 44015600

www.telethon.it mail: telethon@telethon.it

FONDAZIONE Telethon