SCIENTIFIC PROGRAMME

Monday, 11th March 2013

10.00 – 14.00 *Registration and poster setting up*

14.15 – 14.30 **Welcome** ***Sala 1000***

14.30 – 15.00 **Opening address**
Lucia Monaco (Fondazione Telethon)

15.00 – 15.30 **LECTURE**

An introduction to the International Rare Diseases Research Consortium (IRDiRC)
Irene Norstedt (European Commission, Brussels, Belgium) *(Talk 1)*

15.30 – 17.00 **PLENARY SESSION 1** – **Genes and mechanisms of Intellectual Disabilities: paving the way for potential therapies**

Chairpersons: Han Brunner, Maria Passafaro

Intellectual Disability: lessons from 500 diagnostic exomes
Han Brunner (Nijmegen, The Netherlands) *(Talk 2)*

The X-linked Intellectual Disability protein TSPAN7 regulates excitatory synapse development and AMPAR trafficking
Maria Passafaro (Milan, Italy) *(Talk 3)*

FMR1 expression and FMRP function as possible targets of pharmacotherapy in the Fragile X Syndrome
Giovanni Neri (Rome, Italy) *(Talk 4)*

Molecular bases and in vitro modeling of Cdkl5 dependent infantile neurological disorders
Vania Broccoli (Milan, Italy) *(Talk 5)*

17.00 – 17.30 *Coffee break*

17.30 – 19.00 **POSTER SESSION 1**

19.00 – 20.00 **O.Ma.R. Journalism Award** ***Sala 1000***

Organized by the *Osservatorio Malattie Rare*, *Fondazione Telethon* and *Orphanet Italia* with the collaboration of the *National Rare Diseases Centre (CNMR)*.
http://www.premiomalattierare.it/

20.00 – 21.00 *Welcome cocktail*

Tuesday, 12th March 2013

08.30 – 09.00 *Registration and poster setting up*

09.00 – 10.30 **PLENARY SESSION 2 – Therapeutic strategies for Mendelian disorders** ***Sala 1000***

Chairpersons: Luis Galietta, Ennio Ongini (Nicox - Milan, Italy - and Rare Partners)

Pharmacological strategies to rescue chloride transport in Cystic Fibrosis
Luis Galietta (Genoa, Italy) *(Talk 6)*

Exploiting artificial nucleases for targeted transgene integration in human Hematopoietic Stem Cells and induced Pluripotent Stem Cells derived from normal donors and SCID-X1 patients
Angelo Lombardo (Milan, Italy) *(Talk 7)*

High Content Screening Facility at TIGEM: pharmacological approaches to treat rare genetic diseases
Diego Medina (Naples, Italy) *(Talk 8)*

A computational strategy to discover pharmacological chaperones with therapeutic potential for Retinitis Pigmentosa
Francesca Fanelli (Modena, Italy) *(Talk 9)*

10.30 – 11.00 *Coffee break*

11.00 – 12.30 **POSTER SESSION 2**

12.30 – 13.00 **PICTURE OF TELETHON SCIENTISTS**
taken by Filippo Sbalchiero *gathering point: registration desk*

13.00 – 14.00 *Buffet Lunch*

14.00 – 15.00 **KEYNOTE LECTURE** ***Sala 1000***

TIGEM goes translational
Andrea Ballabio (Naples, Italy) *(Talk 10)*

Imagining new therapies for patients with Rare Diseases: Shire HGT and TIGEM translate the future
Arthur Tzianabos (Shire - Lexington, MA, USA) *(Talk 11)*

15.00 – 16.00 **PLENARY SESSION 3 – Advances in clinical trials**

Chairpersons: Robertson Parkman (Los Angeles, CA, USA), Luigi Naldini (Milan, Italy)

The development of Givinostat for the treatment of Duchenne Muscular Dystrophy: from the animal to the clinic
Paolo Bettica (Italfarmaco – Cinisello Balsamo, Italy), *(Talk 12)*
Eugenio Mercuri (Rome, Italy)

Phase I/II clinical trial of Hematopoietic Stem Cell gene therapy for the treatment of Metachromatic Leukodystrophy
Alessandra Biffi (Milan, Italy) *(Talk 13)*

Clinical trial of Hematopoietic Stem Cell gene therapy for Wiskott-Aldrich Syndrome
Alessandro Aiuti (Milan, Italy) *(Talk 14)*

16.00 – 17.00 **ROUND TABLE – Navigating the Valley of Death**
An interactive session engaging all Telethon scientists in tackling the hurdles of translating laboratory research to the clinic

Moderators: Robertson Parkman (Los Angeles, CA, USA),
Helen Heslop (Houston, TX, USA)

Speakers: Luigi Naldini (Milan), Alberto Auricchio (Naples), Grazia Valsecchi (University of Milano-Bicocca, Milan), Alessandro Aiuti (Milan), Paolo Bettica (Italfarmaco – Cinisello Balsamo)

17.00 – 17.30 *Coffee break*

17.30 – 19.00 **POSTER SESSION 3**

19.00 – 20.00 **SPECIAL EVENT** ***Sala 300***

The Executive Director of the American Society for Cell Biology meets young investigators

Stefano Bertuzzi (ASCB - Bethesda, MD, USA)

Wine and Cheese

Wednesday, 13th March 2013

09.00 – 10.30 **PARALLEL SESSIONS A-B**

A- Cardiomyopathy - failure and success ***Sala 1000***

Chairpersons: Daniel Garry, Silvia Priori

Emerging therapies for DMD Cardiomyopathy
Daniel Garry (Minneapolis, MN, USA) *(Talk 15)*

Channelopathies: moving toward gene therapy
Silvia Priori (Pavia, Italy) *(Talk 16)*

Melusin gene therapy: an innovative approach to prevent Familial Cardiomyopathies
Guido Tarone (Torino, Italy) *(Talk 17)*

Inherited Cardiomyopathies: from phenotype-based to genetic-based nosology
Eloisa Arbustini (Pavia, Italy) *(Talk 18)*

B- Decoding an engineering marvel: insights from genetic Renal Diseases

Chairpersons: Gregory Germino, Antonella De Matteis ***Sala 300***

On the edge of glory? Polycystic Kidney Disease research as a model for moving from bench to bedside
Gregory Germino (Bethesda, MD, USA) *(Talk 19)*

Uromodulin and chronic diseases of the kidney
Luca Rampoldi (Milan, Italy) *(Talk 20)*

Cell biology and pharmacology of Lowe Syndrome
Antonella De Matteis (Naples, Italy) *(Talk 21)*

CLC-5, an endosomal chloride – proton exchanger mutated in Dent's Disease: a biophysical perspective
Michael Pusch (Genoa, Italy) *(Talk 22)*

10.30 – 11.00 *Coffee break*

11.00 – 13.00 **PARALLEL SESSIONS C-D**

C- Trial readiness in Neuromuscular Diseases ***Sala 1000***

Chairpersons: Jeffrey Chamberlain, Rossella Tupler

Therapeutic potential of AAV-microdystrophin vectors for gene therapy of Duchenne Muscular Dystrophy
Jeffrey Chamberlain (Seattle, WA, USA) *(Talk 23)*

Facioscapulohumeral Muscular Dystrophy: a walk on the dark side of the (epi)genome
Davide Gabellini (Milan, Italy) *(Talk 24)*

From the bench to the clinic: what we have learnt from the Italian National Registry for Facioscapulohumeral Muscular Dystrophy
Rossella Tupler (Modena, Italy) *(Talk 25)*

Trial readiness in Peripheral Neuropathies: the Charcot-Marie-Tooth Disease pathway
Davide Pareyson (Milan, Italy) *(Talk 26)*

Trial readiness in Peripheral Neuropathies: developing a unifying treatment strategy
Carla Taveggia (Milan, Italy) *(Talk 27)*

D- Blood Disorders: from genetics to genetic therapies ***Sala 300***

Chairpersons: Punam Malik, Giuliana Ferrari

Mutations in the 5'UTR of ANKRD26 result in a "new" form of Inherited Thrombocytopenia that predisposes to Leukemia and is characterized by the presence in platelets and megakaryocytes of a "new" cell structure
Carlo Balduini (Pavia, Italy) *(Talk 28)*

Hepcidin at the crossroad between Hemochromatosis and genetic Iron Deficiency
Clara Camaschella (Milan, Italy) *(Talk 29)*

RNA-based therapeutic approaches for Blood Coagulation Factor Deficiencies caused by splicing mutations
Mirko Pinotti (Ferrara, Italy) *(Talk 30)*

Gene therapy for non-lethal disorders: the paradigm of Beta-Thalassemia
Giuliana Ferrari (Milan, Italy) *(Talk 31)*

Genetic therapy for Hemoglobinopathies: from the bench to the bedside
Punam Malik (Cincinnati, OH, USA) *(Talk 32)*

13.00 – 13.30 **LATE BREAKING NEWS**

A highly secreted sulfamidase engineered to cross the blood-brain barrier corrects brain lesion of mice with mucopolysaccharidoses type IIIA
Alessandro Fraldi (Naples, Italy)

Acylated and unacylated ghrelin impair skeletal muscle atrophy in mice
Andrea Graziani (Novara, Italy)

Molecular function of Ubiad1, a gene associated to Schnyder Crystalline Corneal Dystrophy (SCCD)
Vera Mugoni (Torino, Italy)

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