THINK RARE

Diseases
Research
Funding
Cooperation
Europe

2014
E-RARE
SCIENTIFIC MEETING & STRATEGIC WORKSHOP
ATHENS
JANUARY 20-22

St GEORGE
LYCABETTUS BOUTIQUE HOTEL

Hellenic Center for Disease Control & Prevention
Ministry of Health
DAG 1
MONDAY, 20th JAN
E-RARE
SCIENTIFIC MEETING
ATHENS, GREECE

SPECIAL SESSION DEDICATED TO PROJECT REPORTING & NETWORKING
Session Chairs: Natalia MARTIN, ANR, France and Ralph SCHUSTER, DLR, Germany

09:00 - 09:30 Presentation of E-Rare reporting procedures, Ignacio BAANANTE, ISCIII, Spain
09:30 - 11:00 European Infrastructures - A one-stop-shop for your projects on rare diseases

EATRIS The European Infrastructure for translational research, Giovanni MIGLIACCIO, Scientific Director of EATRIS, The Netherlands

ECRIN European Clinical Research Infrastructure Network, Jacques DEMOTES, ECRIN Coordinator, France

EU-OPENSSCREEN Novel chemical tools to study rare and neglected diseases. Ronald FRANK, EU-OPENSSCREEN Coordinator, Germany

BMBRI Biobanking and biomolecular resources research infrastructure addressing rare diseases research needs, Luca SANGIORGI, BMBRI National Coordinator, Italy

11:00 - 12:00 Round table discussion: Questions & share of experience
12:00 - 13:00 LUNCH

OPENING
13:00 - 13:15 Welcome word by Greek Official Representative
13:15 - 13:30 Presentation of E-Rare, Daria JULKOWSKA, E-Rare Coordinator, France
13:30 - 13:45 European Commission statement, Anders COLVER, European Commission
13:45 - 14:00 E-Rare and IRDiRC: Key partners in funding rare diseases research, Paul LASKO, Chair of IRDiRC, Canada
14:00 - 14:15 Involvement of rare diseases patients in research, Maria MAVRIS, EURORDIS, France

08:00 - 09:00 Registration
## SESSION I: DIAGNOSING RARE DISORDERS USING HIGH THROUGHPUT SEQUENCING

**Session Chairs:** André REIS, Friedrich-Alexander University Erlangen-Nuremberg, Germany & Jacques S. BECKMANN, University of Lausanne, Switzerland

<table>
<thead>
<tr>
<th>Time</th>
<th>Event</th>
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<tr>
<td>14:15 - 14:45</td>
<td><strong>Keynote speaker:</strong> Whole exome sequencing - A game changer in the diagnosis of rare disorders, Orly ELPELEG, Hadassah - Hebrew University Medical Center, Jerusalem, Israel</td>
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<td>14:45 - 15:05</td>
<td><strong>CRANIRARE-2:</strong> An integrated clinical and scientific approach for craniofacial malformations, Bernd WOLLNIK, University of Cologne, Germany</td>
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<td>15:05 - 15:25</td>
<td><strong>NsEuroNet:</strong> European network on Noonan syndrome and related disorders, Marco TARTAGLIA, Istituto Superiore di Sanità, Italy</td>
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<td>15:25 - 15:45</td>
<td><strong>TUB-GENCODEV:</strong> The wide spectrum of malformations of cortical development caused by mutations of tubulins and MT-related proteins, Jamel CHELLY, Institut Cochin, France</td>
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<td>15:45 - 16:05</td>
<td><strong>COFFEE BREAK</strong></td>
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<td>16:05 - 16:25</td>
<td><strong>Euro-Scar:</strong> Nasology and molecular diagnosis of the degenerative recessive ataxias, Michel KOENIG, IGBMC, France</td>
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<td>16:25 - 16:45</td>
<td><strong>NEUTRO-NET:</strong> Inherited inhibition of innate immunity - an integrated molecular genetic approach to discover novel human gene defects, Christoph KLEIN, University of Munich, Germany</td>
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<td>16:45 - 17:05</td>
<td><strong>EuroGeBeta:</strong> European network on genetics, pathophysiology and translational research into rare pancreatic beta-cell insufficiency diseases, Martine VAXILLAIRE, European Genomic Institute for Diabetes, France</td>
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<td>17:05 - 17:25</td>
<td><strong>GENOMIT:</strong> Mitochondrial Disorders - Connecting Biobanks. Empowering Genetic Diagnostics and Exploring Disease Models, Thomas MEITINGER, University of Munich, Germany</td>
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<td>17:25 - 17:45</td>
<td><strong>ELA-2-CN:</strong> Congenital neutropenia with ELA-2 mutations (ELANE-CN): Evaluation of genetic co-factors and molecular pathways with respect to the heterogeneity on phenotype, Cornelia ZEIDLER, Hannover Medical School, Germany</td>
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DAY 1
MONDAY, 20th JAN
E-RARE
SCIENTIFIC MEETING
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18:00 - 18:15  Assessment of E-Rare funding - Analysis of JTC 2007 & 2009 projects, Arnaud GOOLAERTS, FNRS, Belgium
18:15 - 19:00  Feedback & discussion with researchers on E-Rare research funding strategy & calls

POSTER SESSION

Session Chair: Helen MICHELAKAKIS, Institute of Child Health, Greece
19:30 - 21:00  TALK & EAT poster session with BUFFET DINNER

Presentation of posters for the E-Rare Poster Session Prize
### SESSION II: FROM CLINICS TO FUNCTIONAL STUDIES

**Session Chairs:** Virginia SYBERT, University of Washington, USA & György KOSZTOLANYI, University of Pécs, Hungary

- **08:30 - 09:00** **Keynote speaker:** The Podonet experience: exploring inherited diseases of the podocyte, Franz SCHÄFER, Heidelberg University Hospital, Germany

- **09:00 - 09:20** **HMA-IRON:** Towards improved diagnosis and treatment of rare inherited microcytic hypochromic anemias related to iron metabolism, Martina MUCKENTHALER, Heidelberg University Hospital, Germany

- **09:20 - 09:40** **RHORCOD:** Comprehensive analysis of rod-cone photoreceptor degeneration associated with rhodopsin gene mutations, Isabelle AUDO, Institut de la Vision, France

- **09:40 - 10:00** **ERMION:** European research project on mendelian inherited optic neuropathies, Dominique BONNEAU, University of Angers, France

- **10:00 - 10:20** **EDEN:** Eugène Devic European Network: establishment and use of an European database and biological bank for research and treatment in acute neuromyelitis optica and related disorders, Romain MARIGNER, Lyon University Hospital, France

- **10:20 - 10:40** **EuPAPNet:** European pulmonary alveolar proteinosis network: molecular determinants of causes, variability and outcome, Maurizio LUISETTI, Policlinico San Matteo, Italy

- **10:40 - 11:00** **IPF-AE:** Acute Exacerbation of Idiopathic Pulmonary Fibrosis: Mechanism and biomarkers, Antje PRASSE, Freiburg University Medical Center, Germany

- **11:00 - 11:20** **COFFEE BREAK**

### SESSION III: TRANSLATIONAL STUDIES: PATHOPHYSIOLOGY, CELLULAR AND ANIMAL MODELS, THERAPEUTIC APPROACHES

**Session Chairs:** Domenica TARUSCIO, ISS, Italy & Leo NijTMANS, NCMLS, The Netherlands

- **11:20 - 11:50** **Keynote speaker:** Real time functional genomics in neonates, Nicholas KATSANIS, Duke University Medical Center, Durham, NC, USA

- **11:50 - 12:10** **EMINA:** European Multidisciplinary Initiative on Neuroacanthocytosis syndromes, Adrian DANEK, Ludwig-Maximilian University of Munich, Germany
SESSION III: TRANSLATIONAL STUDIES: PATHOPHYSIOLOGY, CELLULAR AND ANIMAL MODELS, THERAPEUTIC APPROACHES
Continues

Session Chairs: Domenica TARUSCIO, ISS, Italy & Leo NJITMANS, NCMLS, The Netherlands

12:10 - 12:30 MLC-Team: Molecular pathogenesis of MLC, a rare disease affecting brain water homeostasis, Raul ESTEVEZ, University of Barcelona, Spain

12:30 - 12:50 NEMMYOP: Functional characterization of nemaline myopathy in a murine model with nebulin mutation: moving from basic understanding towards therapeutic interventions. David BENDAHAN, CRMBM, France

12:50 - 13:50 LUNCH

13:50 - 14:10 Euro-CDG: A European research network for a systematic approach to CDG and related diseases, Nathalie SETA, Xavier Bichat-Claude Bernard Hospital, France

14:10 - 14:30 MTM-Pathies-2: MTM1 and MTMR2 myotubularins: biochemical activity and the regulation of membrane trafficking in health and disease, Alessandra BOLINO, Scn Raffaele Institute, Italy

14:30 - 14:50 RARE-G: The Epidermal growth factor system in rare glomerular disease: From molecular mechanisms to therapeutics, Marcus MOELLER, University Hospital of the RWTH Aachen, Germany

14:50 - 15:10 WHIM-ThemeT: WHIM syndrome: Pathological basis and development of therapeutic molecules, Françoise BACHELERIE, University Paris XI, INSERM, France

SESSION IV: INNOVATIVE THERAPIES & PRODUCTS

Session Chairs: Erik TAMBUYZER, ABCconsult, Belgium & Jacques DEMOTES, ECRIN, France

15:10 - 15:40 Keynote speaker: Translational research towards therapies in progeria and defective prelamin A processing associated syndromes, Nicolas LEVY, Hôpital d’Enfants La Timone and INSERM UMR_S 910, Marseille, France

15:40 - 16:00 Cure-FXS: New therapeutic agents enhance cognition in FMR1 hemizygous mice, Mara DIERSSSEN, Center for Genomic Regulation, Spain

16:00 - 16:20 COFFEE BREAK
SESSION IV: INNOVATIVE THERAPIES & PRODUCTS
Continues

Session Chairs

Erik TAMBUYZER, ABConsult, Belgium & Jacques DEMOTES, ECRIN, France

16:20 - 16:40  CAV-4-MPS: Understanding and treating neurodegeneration caused by mucopolysaccharidoses, Assumpcio BOSCH, Universitat Autonoma de Barcelona, Spain

16:40 - 17:00  HEMO-iPS: Use of patient-specific induced pluripotent stem cells to improve diagnosis and treatment of hemophilia A, Jordi BARQUINERO, Vall d’Hebron Research Institute, Spain

17:00 - 17:20  TRANSPOSTSMART: An innovating platform using transposon and SMAR for von Willebrand disease gene therapy, Daniel SCHERMAN, University Paris Descartes - Chimie Paris Tech, France

17:20 - 17:40  GETHERTHAL: Improvements of vector technology and safety for the gene therapy of thalassemia, Nicholas ANACNOU, University of Athens, Greece

17:40 - 18:00  EURO-CGD: Genetics and pathogenesis of chronic granulomatous disease and development of new gene transfer therapeutic approaches, Manuel CREZ, Institute for Tumor Biology and Experimental Therapy, Georg-Speyer-Haus, Germany

18:00 - 18:20  EB: Identification of revertant mosaicism in epidermolysis bullosa and subsequently using the revertant keratinocytes in a pre-clinical mouse model suitable to test revertant cell therapy, Marion PASMOOIJ, University of Groningen, The Netherlands

18:20 - 18:40  SkinDev: In vitro and in vivo models of congenital rare skin diseases, Hans HENNIES, Cologne Center for Genomic, Germany

18:40 - 19:00  CLOSING and Announcement of the E-Rare Poster Session Prize

19:30 - 21:00  OFFICIAL CLOSING - VISIT AND DINNER AT THE NATIONAL ARCHEOLOGICAL MUSEUM OF ATHENS