4th International Meeting on Rare Disorders of the RAS-MAPK Pathway
A workshop preceding the ESHG meeting Nuremberg, 2012

Organisers: Martin Zenker, Marco Tartaglia, Bronwyn Kerr.

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Registration limit: 100 persons. Registration cost: 100 €.
Location: Hotel “Arvena Park”, Görlitzer Str. 51, 90473 Nürnberg (2 km from ESHG conference venue)

Program

Day 1 (Friday, June 22)

14:30 Registration, Coffee
15:00 Welcome and Introduction
Martin Zenker, Magdeburg

15:10 First Session: Clinical Aspects
Chair: Martin Zenker, Magdeburg

15:10 The NSEuroNet Database
Christina Lißewski, Magdeburg

15:30 SOS1 and RAF1: Mutation Spectrum and Genotype-Phenotype Correlations
Marco Tartaglia, Rome

15:50 SHOC2: Phenotype Analysis in 75 Patients
Martin Zenker, Magdeburg

16:10 NF1 and NFNS: Mutation Spectrum and Genotype-Phenotype Correlations
Alessandro De Luca, Rome

16:30 CBL Mutations, Where Are the Patients?
Hélène Cavé, Paris

16:50 Coffee Break

17:10 Update on RASopathy Mutation Analysis and Epidemiology in Japan
Yoko Aoki, Sendai

17:30 Noonan Guidelines, DYSCERNE and Beyond
Bronwyn Kerr, Manchester

17:50 National Protocol for Diagnosis and Treatment in France: Noonan / Costello / CFC Syndrome
N.N. (provisional)

18:10 Experience of a RASopathy Clinic
Giuseppe Zampino, Rome

18:30 Dinner Buffet
Day 1 (Friday, June 22), continued

19:30  Evening Session: Presentations Selected from Submitted Abstracts
   Chair: Bronwyn Kerr, Manchester
   Attendees are invited to give brief presentations on unusual cases, novel mutations or study results. Please contact the organizers.

Day 2 (Saturday, June 23)

8:30  Second Session: Genes and Pathogenesis
   Chair: Marco Tartaglia, Rome
   8:30  Disorders Caused by MYST4 / KAT6B Mutations – How Much of RASopathy?
        Christian Thiel, Erlangen
   8:50  Genes and Mechanisms: What’s New?
        Marco Tartaglia, Rome
   9:10  Exome Sequencing – the Nijmegen Experience
        Ineke van der Burgt, Nijmegen
   10:10  Novel Insights into Pathophysiology of PTPN11 Mutations Causing Short Stature
          Patrick Raynal, Toulouse
   9:30  Structural and Functional Characterization of RAS Mutants
        Reza Ahmadian, Düsseldorf
   9:50  Noonan Syndrome Mouse Model: Tumor Predisposition and Therapeutic Strategies
        Carmen Guerra, Madrid
   10:50  Coffee Break

11:10  Third Session: Treatment Perspectives
       Chair: Judith Allanson, Ottawa
   11:10  Treatment of Cardiomyopathy in LEOPARD Syndrome
          Maria Kontaridis, Boston
   11:30  Treatment of RAF1 Mutation-Induced Cardiomyopathy
          Toshi Araki, Toronto
   11:50  Phase 2 Study on MEK162 in Noonan Syndrome Hypertrophic Cardiomyopathy
          N.N. (provisional)
   12:10  Learning Difficulties in RASopathies: Pathogenesis and Treatment
          Ype Elgersma, Rotterdam
   12:30  Use of Drosophila Models to Search for Noonan Syndrome Therapies
          Bruce Gelb, New York
   12:50  Discussion
   13:10  Close, Lunch Buffet
   14:00  Meeting of the NSEuroNet Group
          Chair: Marco Tartaglia, Rome
Speakers and Chairpersons:

Prof. Reza Ahmadian, PhD, Institute of Biochemistry and Molecular Biology II, Heinrich-Heine University, Düsseldorf, Germany
Prof. Judith Allanson, MD, Department of Genetics, Children's Hospital of Eastern Ontario, Ottawa, Canada
Prof. Yoko Aoki, MD, PhD, Department of Medical Genetics, Tohoku University School of Medicine, Sendai, Japan
Dr. Toshi Araki, PhD, Department of Medical Biophysics, University of Toronto, Toronto, Ontario, Canada
Prof. Hélène Cavé, AP-HP, Département de Génétique, Hôpital Robert Debré, Paris, France
Dr. Alessandro De Luca, PhD, Dept. of Hematology, Oncology and Molec. Medicine, Istituto Superiore di Sanità, Rome, Italy
Prof. Ype Elgersma, PhD, Department of Neuroscience, Erasmus University Medical Centre, Rotterdam, The Netherlands
Prof. Bruce Gelb, MD, Center for Molecular Cardiology, Dept. of Pediatrics & Dept. of Genetics and Genomic Sciences, Mount Sinai School of Medicine, New York, USA
Dr. Carmen Guerra, PhD, Molecular Oncology Programme, Spanish National Cancer Research Centre (CNIO), Madrid, Spain
Dr. Bronwyn Kerr, MD, University Department of Medical Genetics, Royal Manchester Children's Hospital, Manchester, UK
Prof. Maria Kontaridis, PhD, Harvard Medical School, Department of Cardiology, Beth Israel Deaconess Medical Center, Center for Life Sciences, Boston, MA, USA
Christina Lißewski, Institute of Human Genetics, University Hospital of Magdeburg, Germany
Prof. Patrick Raynal, PhD, Centre de Physiopathologie, Hôpital de Toulouse-Purpan, Toulouse, France
Prof. Marco Tartaglia, PhD, Dept. of Hematology. Oncology and Molec. Medicine, Istituto Superiore di Sanità, Rome, Italy
Dr. Christian Thiel, MD, Institute of Human Genetics, University Hospital Erlangen, University of Erlangen-Nuremberg, Erlangen, Germany
Dr. Ineke van der Burgt, MD, Department of Human Genetics, University Medical Center St Radboud, Nijmegen, The Netherlands
Prof. Guiseppe Zampino, MD, Department of Pediatrics, Catholic University, Rome, Italy
Prof. Martin Zenker, MD, Institute of Human Genetics, University Hospital of Magdeburg, Germany

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