6th International Meeting on Rare Disorders of the RAS-MAPK Pathway
A workshop preceding the ESHG meeting Barcelona, 2016

Organisers: Bronwyn Kerr, Marco Tartaglia, Martin Zenker.
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Registration limit: 100 participants.
Registration cost: 100 € (including buffet dinner on May 20), students 50 €
Location: HOTEL BARCELONA PRINCESS, Avinguda Diagonal 1, 08019 Barcelona, Spain
Date: Beginning: Friday, May 20, 2016, 1 p.m. – End: Saturday, May 21, 2016, 1 p.m.

Program - Day 1 (Friday, May 20)

900 Noonan syndrome guidelines consensus meeting
(guidelines consensus group members only)
Organisation and chair: Bronwyn Kerr

1200 Registration
1300 Welcome and Introduction
1310 First Session: Cancer risk in RASopathies and Mosaic RASopathies
Chair: Marco Tartaglia and Martin Zenker

1310 RASopathies and cancer: insights from the NSEuroNet database
Christina Lissewski

1330 Molecular genetics of JMML
Helene Cavé

1400 Structural an dynamic effects of MEK1 mutations causing cancer and/or CFC syndrome
Juan Fernandez-Recio

1420 Tumor risk in LEOPARD syndrome / Noonan syndrome with multiple lentigines
Ludwine Messiaen

1440 Cancer in Costello syndrome
Karen Gripp

1500 Oculoectodermal syndrome and Mosaic RASopathies
Martin Zenker

1520 Coffee Break

1550 Second Session: Molecular mechanisms and RASopathy models
Chair: Bruce Gelb and Marco Tartaglia

1550 PTPN11 (15th anniversary lecture)
Marco Tartaglia & Bruce Gelb
Second session continued:

16:20 Interaction between SPRED1 and neurofibromin implicated in Legius syndrome and NF1
Hilde Brems

16:35 SHOC2 trapping at different subcellular sites differentially impacts on ERK signaling output
Marialetizia Motta

16:50 Transcriptional hallmarks of RASopathies
Enzo Medico

17:10 Consequences of Noonan syndrome-causing SHP2 mutants on growth plate development and
longitudinal bone growth
Thomas Edouard

17:30 Dissecting the “RASopathy” signaling network using C. elegans
Simone Martinelli

17:50 iPSC modeling of RASopathies
Bruce D. Gelb

18:20 Prospects for targeting RAS signal transduction
Reza Ahmadian

18:40 Dinner Buffet

20:10 - New HRAS mutation affecting transcript processing (F. Pantaleoni)
20:20 - A novel mutation cluster in MEK1 (E. Flex)
20:30 - Clinical and molecular spectrum of NRAS mutations (C. Lissewski)
20:40 - Clinical and molecular spectrum of RIT1 mutations (B. Kerr)
20:50 - SOS2, LZTR1, RRAS, RASA2 & other RASopathy genes: an update on prevalence, mutation spectrum and genotype-phenotype correlations. Participants are invited to share their experience dealing with rare/new mutations and phenotypes. A template for one powerpoint slide per patient/cohort to be presented in three minutes.
Fourth session continued:

950 News from growth and GH therapy studies
(12 min each, 6 min final discussion)
- GH sensitivity and genetics (Jovanna Dahlgren)
- Growth, puberty and GH therapy in RASopathies (Laura Mazzanti)

1020 News from bone metabolism and energetic metabolism studies
(12 min each, 6 min final discussion)
- Energy expenditure and bone metabolism in RASopathies (Chiara Leoni)
- Bone mineral density in Noonan Syndrome (Giovanni Battista Ferrero)

1050 Coffee break

1120 Fifth Session: RASopathy family support group session
Chair: Bronwyn Kerr and Martin Zenker
- Associazione Italiana Sindrome Costello (Italy)
- Angeli Noonan (Italy)
- Noonan Syndrome Association (UK)
- Association Française des syndromes Costello et CFC (France)
- RASopathies Network (USA)
- Free speech (associations/parents/patients)

1150 GenIDA project (Jean-Louis Mandel)

1210 Report from the guideline session (Bronwyn Kerr)

1230 Closing remarks

1330 NSEuroNet – Kick-Off Meeting
(NSEuroNet partners and external collaborators only)
Chair: Marco Tartaglia

1330 Overview of the Consortium and major goals (M. Tartaglia)
1345 Identification of novel disease genes (M. Tartaglia/M. Zenker)
1400 Molecular mechanisms of pathogenesis (R. Ahmadian/J. de Hertog)
1415 Translational research (A. Yart/J. den Hertog)
1430 Molecular epidemiology and gen-phen correlation studies (M. Zenker/H. Cavé)
1445 Discussion and closing remarks

Supported by:

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Speakers and Chairpersons:

Prof. Reza Ahmadian, PhD, Institute of Biochemistry and Molecular Biology II, Heinrich-Heine University, Düsseldorf, Germany

Dr. Paolo Alfieri, MD, Child Neuropsychiatry, Department of Neurosciences, Ospedale Pediatrico Pambino Gesù, Rome, Italy

Dr. Hilde Brems, MD, PhD, Department of Human Genetics, Catholic University of Leuven, Leuven, Belgium

Dr. Giulio Calcagni, MD, Department of Pediatric Cardiology and Cardiac Surgery, Ospedale Pediatrico Bambino Gesù, Rome, Italy

Prof. Hélène Cavé, PhD, - Institut Universitaire d'Hématologie, Département de Génétique - UF de Génétique Moléculaire, Hôpital Robert Debré, Paris, France

Prof. Jovanna Dahlgren, MD, Inst. of Clinical Sciences, Dept. of Pediatrics, The Queen Silvia Children's Hospital, Gothenborg, Sweden

Dr. Thomas Edouard, MD, PhD, Endocrine, Bone Diseases, and Genetics Unit, Children's Hospital, University Hospital Center of Purpan, Toulouse, France

Dr. Juan Fernandez-Recio, PhD, Life Sciences Department, Barcelona Supercomputing Center, Barcelona, Spain

Prof. Giovanni Battista Ferrero, MD, Department of Pediatric and Public Health Sciences, University of Turin, Torino, Italy

Dr. Shruti Garg, MD, Institute of Brain Behaviour and Mental Health, University of Manchester, UK

Prof. Bruce D. Gelb, MD, PhD, Mindich Child Health and Development Institute, Icahn School of Medicine at Mt. Sinai, New York, New York, USA

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Dr. Giuseppe Limongelli, MD, Cardiologia SUN, Ospedale Monaldi (Azienda dei Colli), Seconda Università di Napoli, Napoli, Italy.

Christina Lißewski, Institute of Human Genetics, University Hospital of Magdeburg, Germany

Prof. Jean-Louis Mandel, MD, Translational Medicine and Neurogenetics Dept., Institut de Génétique et de Biologie Moléculaire et Cellulaire (IGBMC), University of Strasbourg, Illkirch, France

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