5th International Meeting on Rare Disorders of the RAS-MAPK Pathway
A workshop preceding the 2014 ESHG meeting, Milan, May 30th 2014

Organisers: Bronwyn Kerr, Marco Tartaglia, Martin Zenker.

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Registration limit: 100 participants
Registration cost: 50 € (dinner included)
Location: UNA Hotel Scandinavia, Via G.B. Fauché, 15 – 20154 Milan
(Metro Sempione Arona - Tram 1)

Program

12:30  Registration, Coffee
13:00  Welcome and Introduction
       Martin Zenker, Magdeburg
13:10  First Session: Clinical Aspects
       Chair: Martin Zenker, Magdeburg
13:10  Do feeding problems influence first year growth in Noonan syndrome? - Ineke van der Burgt
13:30  Growth Hormone treatment and growth in Noonan syndrome - Laura Mazzanti
13:50  Cardiac left-sided obstructions in RASopathies - Cristina Digilio
14:10  Outcome of cardiac surgery and interventional procedures in children with RASopathies. Preliminary data of a multicenter study - Giulio Calcagni
14:30  Neuropsychology of adults with Noonan syndrome: strengths and weaknesses in cognitive functioning, and comparison of social cognition in Noonan versus Turner syndromes - Ellen Wingerbermuhle/Renée Roelofs
14:50  Neurocognitive assessment in RASopathies; the problem of heterogeneity - Amy Burns/Ami Brooks
15:10  Electroencephalographic aspects in RASopathies – Domenica Battaglia
15:40  Coffee break
1600  Second Session: Molecular genetics and pathogenetic mechanisms
       Chair: Marco Tartaglia, Rome
16:00  Heterozygous germline mutations in A2ML1 are associated with a disorder clinically related to Noonan syndrome - Helger Ijntema
16:20 Activating mutations in RRAS underlie a phenotype within the RASopathy spectrum and contribute to leukemogenesis - Francesca Pantaleoni

16:40 A novel PAK3 mutation alters the RAS/MAPK pathway causing an X-linked syndromic phenotype – Pamela Magini

17:00 Functional and computational models of MEK1 mutations - Fabian Glaser

17:20 Pharmacological rescue of behavioural phenotypes in a Spred1 mouse model for Legius syndrome - Hilde Brems

17:40 Characterization of a neuronal phenotype in a mouse model of Noonan syndrome - Franceska Altmueller

18:00 RASopathy and cancer predisposition – Helene Cavé

18:30 Discussion

19:00 Dinner Buffet

20:00 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. (10’-15’ each)

- CoQ10 deficiency and BRAF mutations – Leonardo Salviati
- Familial café-au-lait spots and NF1 mutations – Alessandro De Luca
- Three distinct RASopathies in a unique nuclear family - Francesca Lepri
- Developmental disroders in NS: impact and relevance of CNVs – Giuseppina Baldassarre
- Noonan syndrome-like disorder with loose anagen hair (Mazzanti syndrome): A new case with neuroblastoma – Livia Garavelli

21:30 Closing remarks

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