

## 5<sup>th</sup> International Meeting on Rare Disorders of the RAS-MAPK Pathway

A workshop preceding the 2014 ESHG meeting, Milan, May 30<sup>th</sup> 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**
- 1540 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 <i>RRAS</i> underlie a phenotype within the RASopathy spectrum and contribute to leukemogenesis - <b>Francesca Pantaleoni</b>
16:40	A novel <i>PAK3</i> mutation alters the RAS/MAPK pathway causing an X-linked syndromic phenotype – <b>Pamela Magini</b>
17:00	Functional and computational models of MEK1 mutations - Fabian Glaser
17:20	Pharmacological rescue of behavioural phenotypes in a <i>Spred1</i> mouse model for Legius syndrome - <b>Hilde Brems</b>
17:40	Characterization of a neuronal phenotype in a mouse model of Noonan syndrome - Franzeska 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

Supported by:

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