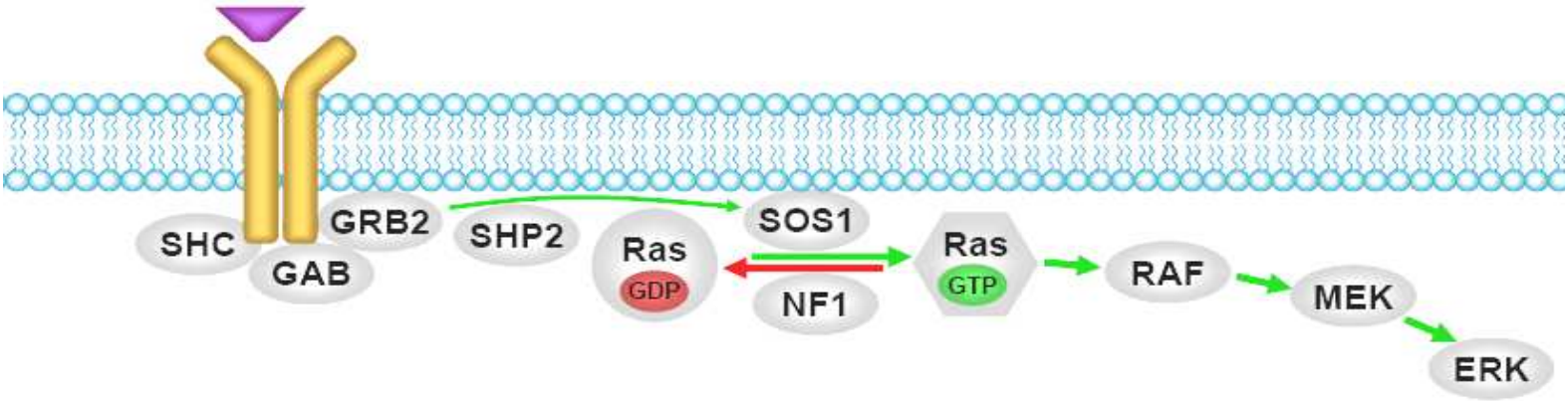

5th International Meeting on Rare Disorders of the RAS-MAPK Pathway

A workshop preceding the 2014 ESHG meeting, Milan, May 30th 2014

Organisers: Bronwyn Kerr, Marco Tartaglia, Martin Zenker.

Contacts for enquiries or registration: bronwyn.kerr@cmft.nhs.uk, marco.tartaglia@iss.it, martin.zenker@med.ovgu.de,

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 *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 - **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**

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21:30 Closing remarks

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