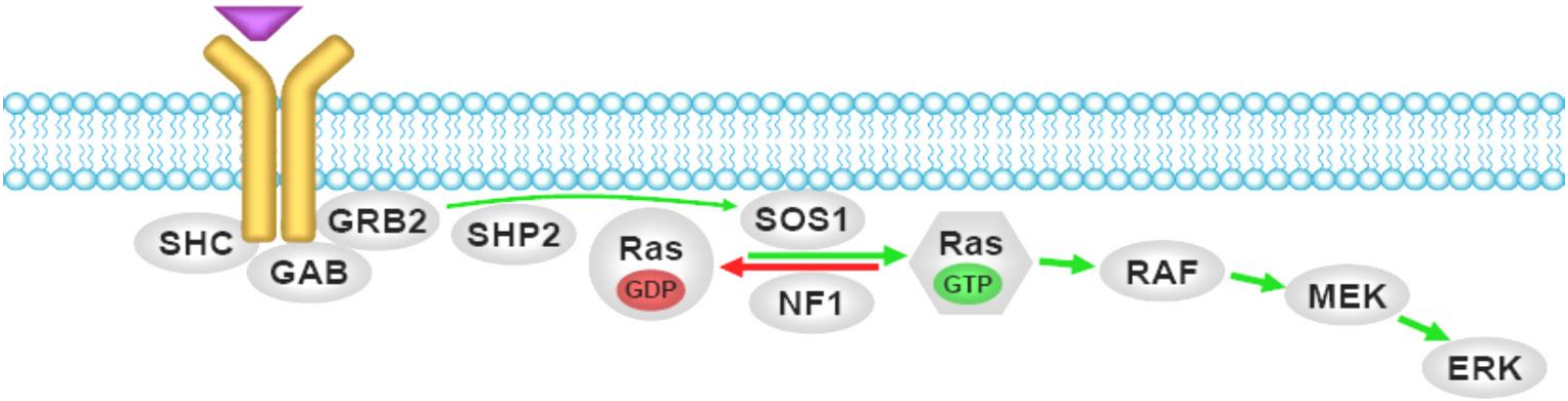

7th International Meeting on Rare Disorders of the RAS-MAPK Pathway

A workshop preceding the ESHG conference in Milan, 2018

Organisers: Bronwyn Kerr, Marco Tartaglia, Martin Zenker.

Contacts: bronwyn.kerr@cmft.nhs.uk, marco.tartaglia@opbg.net, martin.zenker@med.ovgu.de,

Registration limit: 100 participants.

Registration cost: 100 € (including buffet dinner on May 20), one day 50 €, students: free attendance

Location: UNA HOTEL SCANDINAVIA, Via G. B. Fauché, 15, 20154 Milano, Italy

Date: Friday, June 15, 2018 – Saturday, June 16, 2018

Program - *Day 1 (Friday, June 15)*

9^{00} **Noonan syndrome guidelines consensus meeting:**
What can we do to promote further the development of international guidelines? Follow-up of the Barcelona meeting 2016
(for guidelines consensus group members
and interested participants of the main meeting)
Organisation and chair: Bronwyn Kerr

9^{00} Diagnostic criteria for Noonan syndrome
Ineke van der Burgt

9^{30} Suggestions for a nosology of RASopathies
Martin Zenker

10^{00} Should all infants with cardiomyopathy have a RASopathy panel?
Bruce Gelb

10^{20} Prenatal screening: Guidelines for proper molecular screening based on the echographic prenatal assessment
Alessandro de Luca

10^{40} Coffee Break

11^{00} Minimum medical management guidelines for Noonan syndrome and results of a family survey
Bronwyn Kerr

11^{30} Minimum medical management guidelines for cardiofaciocutaneous syndrome and Costello syndrome
Giuseppe Zampino

13^{00} Registration

13^{45} Welcome addresses

14^{00} **First Session: Cancer predisposition**
Chair: Marco Tartaglia

14^{00} RAS signaling and leukemogesis
Giovanni Cazzaniga

14^{30} Cancer surveillance and therapy in children with RASopathies and other cancer predisposition syndromes
Christian Kratz

15^{00} Genomic alterations in Costello syndrome-associated ERMS
Karen Gripp

15^{20} RASopathies: why are these rare spontaneous disorders so common?
Anne Goriely

15^{40} **From the patients' perspective**: Living with with tumor risk.
How to direct research to have a more effective surveillance?

16^{00} Coffee Break

16^{30} **Second Session: Cardiovascular system/hypertrophic cardiomyopathy**
Chair: Bruce Gelb

16^{30} Cardiac defects, morbidity and mortality in patients affected by RASopathies - Update on the CARNET study
Giulio Calcagni

16^{50} Patient specific iPSC-derived cardiomyocytes reveal abnormal signaling pathways underlying hypertrophic cardiomyopathy in Noonan syndrome
Maria Kontaridis

17^{10} Heart defects in a genetic zebrafish model for Noonan Syndrome
Jeroen den Hertog

17^{30} Experiences with MEK inhibitor treatment in patients with RIT1-associated cardiomyopathy
Gregor Andelfinger

17^{50} Outlook on possible strategies of medical treatment for RASopathy-associated cardiomyopathy
Bruce Gelb

18^{10} **From the patients' perspective**: Living with heart defect/cardiomyopathy.
How to direct research to have a more effective management?

18^{30} Dinner Buffet

20^{00} **Late session: new genes & mutations/gen-phen correlations/short reports**
Chair: Marco Tartaglia and Martin Zenker

20^{00} - Functional dysregulation of CDC42 causes diverse developmental phenotypes (**F. Pantaleoni**)
20^{15} - LZTR1 and AD and AR inheritance of Noonan syndrome (**M. Zenker**)
Short presentations (7 min – 8 presentations maximum)
Two additional patients with NS-LAH caused by the recurrent PPP1CB mutation p.Pro490Arg (**M. L. Dentici**)
Phenotypes associated with concomitant mutations (**A. De Luca**)
NIPT assay to detect these selfish point mutations in maternal plasma (**A. Goriely**)
Selected abstracts

Participants are invited to present their data in this session (mainly for interesting case reports and genotype phenotype studies). Please send a brief abstract to the organisers.

Program - *Day 2 (Saturday, June 16)*

9^{00}	**Fourth Session: Nutrition, energetic and bone metabolism** **Chair: Armelle Yart**
9^{00}	Energetic and bone metabolism in Costello and CFC syndromes: an overview **Chiara Leoni**
9^{20}	Chondrocyte differentiation during endochondral growth in Noonan syndrome **Thomas Edouard**
9^{40}	Metabolic abnormlities in a mouse model of Costello syndrome **Ion Cirstea**
10^{00}	The basis of nutritional and metabolic problems in RASopathies: Lessons from mouse models **Shin-Ichi Inoue & Yoko Aoki**
10^{20}	Energetic metabolism in Costello syndrome **Elisabetta Flex**
10^{40}	Metabolic dysfunctions in Noonan and NS-ML syndromes **Armelle Yart**
11^{00}	**From the patients' perspective**: The parents' struggle with nutrition problems in RASopathies. How to direct research to have a more effective management?
11^{20}	Coffee break
11^{50}	**Fifth Session: Nervous system - Lymphatics** **Chair: Martin Zenker**
11^{50}	Epilepsy in RASopathies **Domenica Battaglia**
12^{10}	Behavioral issues and cognitive profiles in RASopathies **Paolo Alfieri & Dany Menghini**
12^{30}	Update on lymphatic problems in RASopathies **Sahar Mansour**
12^{50}	SOS2 mutation-related Noonan syndrome and its association with lymphedema **Christina Lißewski**
13^{10}	**From the patients' perspective :** Living with lymphedema (Judith van de Meereakker). The parents' struggle with epilepsy in cardiofaciocutaneous syndrome.
13^{30}	Closing remarks
14^{00}	**NSEuroNet – Kick-Off Meeting** (NSEuroNet partners and external collaborators only) **Chair: Marco Tartaglia**

Supported by:

Associazione Nazionale SINDROME DI NOONAN RASopatie

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ANGELI NOONAN

ASSOCIAZIONE ITALIANA SINDROME DI NOONAN-ONLUS

Speakers and Chairpersons:

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