



2nd International Meeting on Rare Disorders of the RAS-MAPK pathway

A workshop preceding the ESHG meeting Vienna, 2009

Organisers: Bronwyn Kerr, Martin Zenker, Marco Tartaglia, Judith Allanson, Ineke van der Burgt, Didier Lacombe, Michael Patton, Nicole Philip, Alain Verloes.

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Registration limit: 100 persons. Registration cost: 100 €

Location: Austria Trend Hotel Donauzentrum, Room "Lobau", Wagramer Str. 83-85, Vienna

Program

Day 1 (Friday, May 22)

- 14³⁰ Registration, Coffee
- 15⁰⁰ Welcome and Introduction
Bronwyn Kerr, Manchester
- 15¹⁰ **First Session: Genotype and Phenotype**
Chair: Didier Lacombe, Bordeaux / Michael Patton, London
- 15¹⁰ Overview and Update on Phenotypes and Genes Involved
Giovanni Neri, Rome
- 15⁴⁰ The Face in Noonan Syndrome and Related Disorders from a Clinical Point of View
Judith Allanson, Ottawa
- 16⁰⁰ 3D Face Analysis in RAS-MAPK Pathway Disorders
Peter Hammond, London
- 16³⁰ Sleep Disorder in Costello Syndrome
Giacomo Della Marca, Rome
- 16⁴⁵ Noonan Syndrome Above 16: Fertility and Neuropsychological Profile
Ineke van der Burgt, Nijmegen
- 17⁰⁵ Is there a behavioural phenotype in Noonan syndrome?
Laura Freeman, Jeremy Turk and Michael Patton, London
- 17²⁰ Phenotypic Spectrum Associated with *BRAF* Gene Mutations
Anna Sarkozy, Newcastle
- 17⁴⁰ Molecular and Clinical Aspects of Legius Syndrome
Eric Legius, Leuven
- 18⁰⁰ Supper
- 19³⁰ **Informal Evening Session: Short Presentations and Unusual / Unique Phenotypes and Other Brief Contributions**
Chair: Nicole Philip, Marseille / Alain Verloes, Paris
Attendees are invited to give brief presentations on unusual cases, novel mutations or study results. Please contact the organizers.

Day 2 (Saturday, May 23)

- 8³⁰ **Second Session: New Insights in Genes and Pathogenesis**
Chair: Marco Tartaglia, Rome and Martin Zenker, Erlangen
- 8³⁰ A New Gene for Noonan Syndrome
Martin Zenker, Erlangen
- 8⁴⁵ A new gene and a novel distinctive condition in the neurocardiofacialcutaneous syndrome family
Marco Tartaglia, Rome
- 9⁰⁰ Molecular mechanisms underlying SHP2 functional dysregulation
Marco Tartaglia/Simone Martinelli, Rome
- 9¹⁵ Drosophila models of Ras Pathway Disorders: Functional Gains and Losses
Bruce Gelb, New York
- 9³⁰ Consequences of Noonan syndrome and LEOPARD syndrome causing SHP2 mutations in zebrafish development
Jeroen den Hertog, Utrecht
- 9⁴⁵ *BRAF* mutations in zebra fish
Elizabeth Patton, Edinburgh
- 10⁰⁰ Modeling genetic syndromes on the Ras/MAPK pathway in mice
Carmen Guerra, Madrid
- 10¹⁵ Synaptic Long Term Potentiation in Patients with Noonan Syndrome and Neurofibromatosis Type 1
Volker Mall, Freiburg
- 10⁴⁵ **Coffee break**
- 11¹⁵ **Third Session: Treatment Perspectives**
Chair: Judith Allanson, Ottawa and Patrick Raynal, Toulouse
- 11¹⁵ Induced Pluripotent Stem Cells for Ras Pathway Disorders: Backing Up into the Future
Bruce Gelb, New York
- 11⁴⁰ Growth Hormone and Growth Hormone Treatment in Patients with Noonan Syndrome
Jovanna Dahlgren, Gothenborg
- 12⁰⁵ Developing Treatment Trials for Rare Disorders: Lessons from the Progeria Trial
Annachiara De Sandre-Giovannoli, Marseille
- 12³⁰ Developing Guidelines for Diagnosis and Treatment in RAS-MAPK Pathway Disorders
NF1: The Manchester experience, **Sue Huson, Manchester**
Noonan guidelines (precirculated) for discussion, **Ineke van der Burgt**
- 13⁰⁰ Discussion
- 13³⁰ **Close**

Speakers and Chairpersons:

Prof. Judith Allanson, MD

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Ottawa, Canada

Prof. Jovanna Dahlgren, MD PhD

Inst. of Clinical Sciences, Dept. of Pediatrics
The Queen Silvia Children's Hospital
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