

Rare Disorders of the MAPK pathway; Current status/ future directions

A workshop preceding the ESHG meeting Barcelona, 2008

Organisers: Bronwyn Kerr, Martin Zenker, Didier Lacombe, Nicole Philip, Alain Verloes, Mike Patton, Marco Tartaglia.

Contacts for enquiries or registration: bronwyn.kerr@cmmc.nhs.uk,
mzenker@humgenet.uni-erlangen.de

Registration limit: 170 persons.

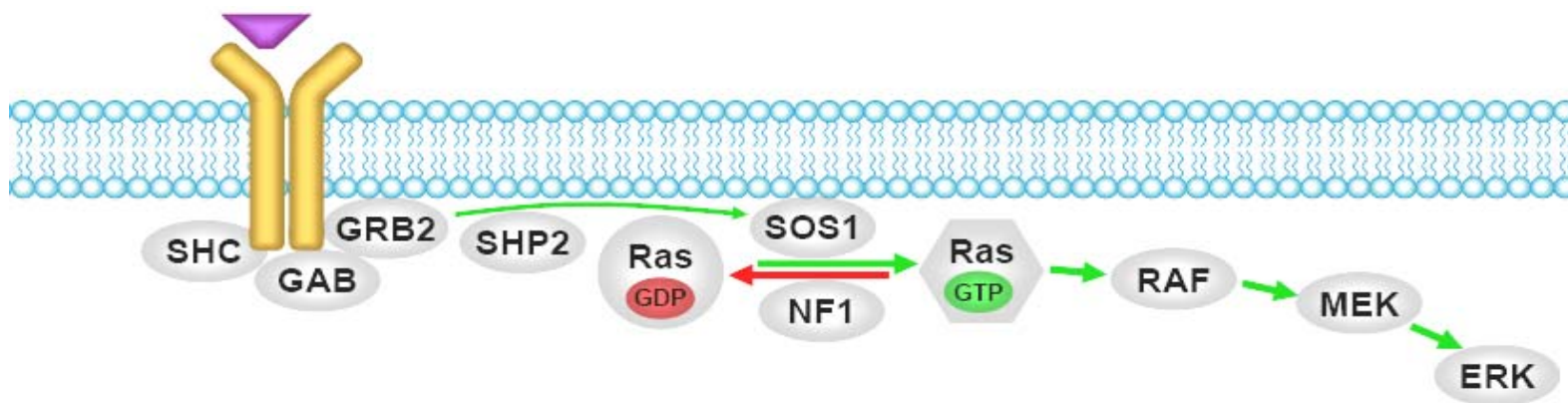
Registration cost: 75 Euros payable in advance or on the day.

Location: Room 124 and 125, ESHG venue, Centre Convencions Internacional, Rambla Prim, 1-17, 08019, Barcelona.

Aims:

1. To educate interested genetic professionals regarding the clinical features and underlying genetic basis of CFC, CS and Noonan syndromes and other diseases of the Ras-MAPK pathway.
2. To facilitate research collaboration across Europe in understanding the disorders caused by deregulation of the Ras-MAPK pathway.
3. To establish a mechanism for establishing a complete genotype/phenotype dataset for European patients.
4. To determine a mechanism for using this dataset to facilitate clinical research, document long-term natural history, improve management and encourage treatment trials.

It is hoped that attendees will be able to share their phenotype/genotype data with presenters before the meeting to enable the largest possible data set for considering genotype/ phenotype correlation.



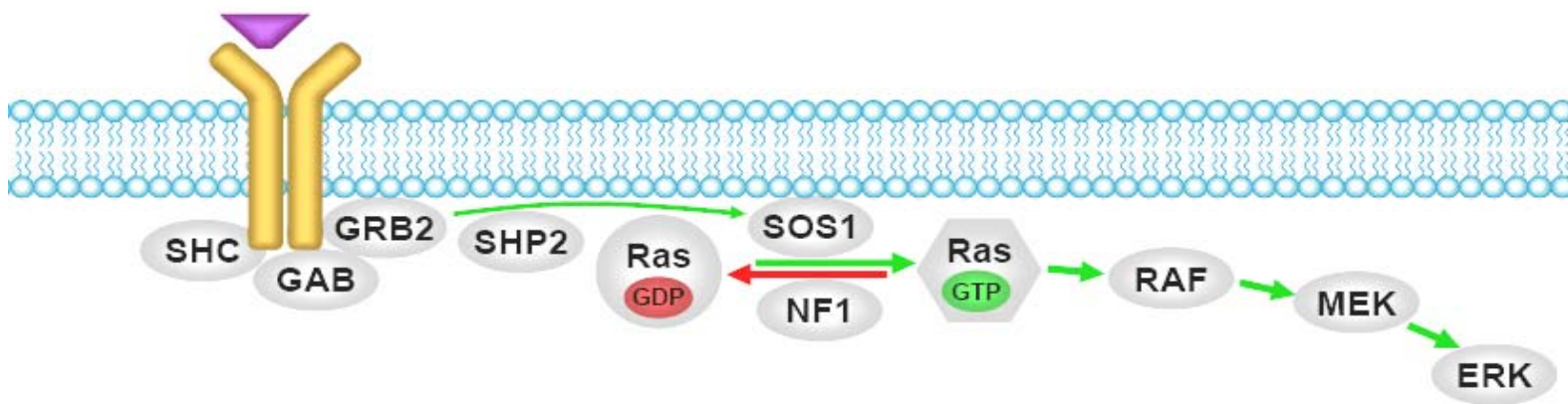
Program

Day 1 (May 30)

- 14³⁰ Registration, Coffee
- 15⁰⁰ Genes, mechanisms and genotype phenotype correlations; are there mutation specific phenotypes?
Chair: Didier Lacombe
- 15⁰⁰ Introduction/ Aims
15⁰⁵ PTPN11
15²⁵ KRAS
15⁴⁵ SOS1
16⁰⁵ RAF1
16²⁵ BRAF and MEK1/2
16⁴⁵ HRAS
17⁰⁰ Unusual phenotypes in Noonan syndrome:
MEK mutations, cherubism, some other puzzling anomalies
17¹⁵ Leukemia and the Noonan syndrome:
not simple as it used to be
17³⁰ Discussion
17⁴⁵ Finish
- Bronwyn Kerr
Mike Patton
Martin Zenker
Marco Tartaglia
Bruce Gelb
Judith Allanson
Yoko Aoki
Alain Verloes
Hélène Cavé

Day 2 (May 31)

- 09³⁰ Lessons from molecular analysis; unexpected insights for phenotypes.
Chair: Martin Zenker
- 09³⁰ Costello syndrome
09⁴⁵ CFC syndrome
10¹⁵ Noonan Syndrome
10⁴⁵ LEOPARD syndrome
11⁰⁰ Coffee break
11¹⁵ Animal models and treatment prospects for MAPK pathway disorders
Chairs: Marco Tartaglia / Bronwyn Kerr
- 11¹⁵ A mouse model for Costello syndrome
11³⁵ Oncogenes on my mind: prospects for treating cognitive deficits in the Ras pathway
12⁰⁵ Discussion / future directions
12⁴⁵ Close
- Nicole Philip
Giovanni Neri
Ineke van der Burgt
Maria Cristina Digilio
Carmen Guerra
Ype Elgersma



Speakers and Chairpersons:

Prof. Judith Allanson MD

Department of Genetics
Children's Hospital of Eastern Ontario
Ottawa, Canada

Prof. Yoko Aoki MD PhD

Department of Medical Genetics
Tohoku University School of Medicine
Sendai, Japan

Dr. Hélène Cavé, PharmD PhD

Département de Génétique
UF de Génétique Moléculaire,
Hôpital Robert Debré
Paris, France

Dr. Maria Cristina Digilio MD

Medical Genetics
Bambino Gesù Hospital
Rome, Italy

Dr. Ype Elgersma

Department of Neuroscience
Erasmus University Medical Center
Rotterdam, The Netherlands

Prof. Bruce Gelb MD PhD

Center for Molecular Cardiology
Department of Pediatrics and Department of
Genetics and Genomic Sciences
Mount Sinai School of Medicine
New York, USA

Dr. Carmen Guerra PhD

Molecular Oncology Programme
Centro Nacional de Investigaciones
Oncológicas (CNIO)
Madrid, Spain

Dr. Bronwyn Kerr MD

University Department of Medical Genetics
Royal Manchester Children's Hospital
Manchester, UK

Prof. Didier Lacombe MD

Service de Génétique Médicale
Hôpital Pellegrin-Enfants
CHU de Bordeaux
Bordeaux, France

Prof. Giovanni Neri MD

Istituto di Genetica Medica
Università Cattolica del S. Cuore
Rome, Italy

Prof. Michael Patton MD

Medical Genetics
St Georges Hospital Medical School
London, UK

Prof. Nicole Philip

Department of Medical Genetics
Hôpital d'Enfants de la Timone
Marseille, France

Prof. Marco Tartaglia PhD

Department of Cell Biology and
Neurosciences
Istituto Superiore di Sanità
Rome, Italy

Dr. Ineke van der Burgt MD

Department of Human Genetics
University Medical Center St Radboud
Nijmegen, The Netherlands

Prof. Alain Verloes MD PhD

Département de Génétique
UF de Génétique Clinique
Hôpital Robert Debré
Paris, France

Dr. Martin Zenker MD

Institute of Human Genetics
University of Erlangen-Nuremberg
Erlangen, Germany