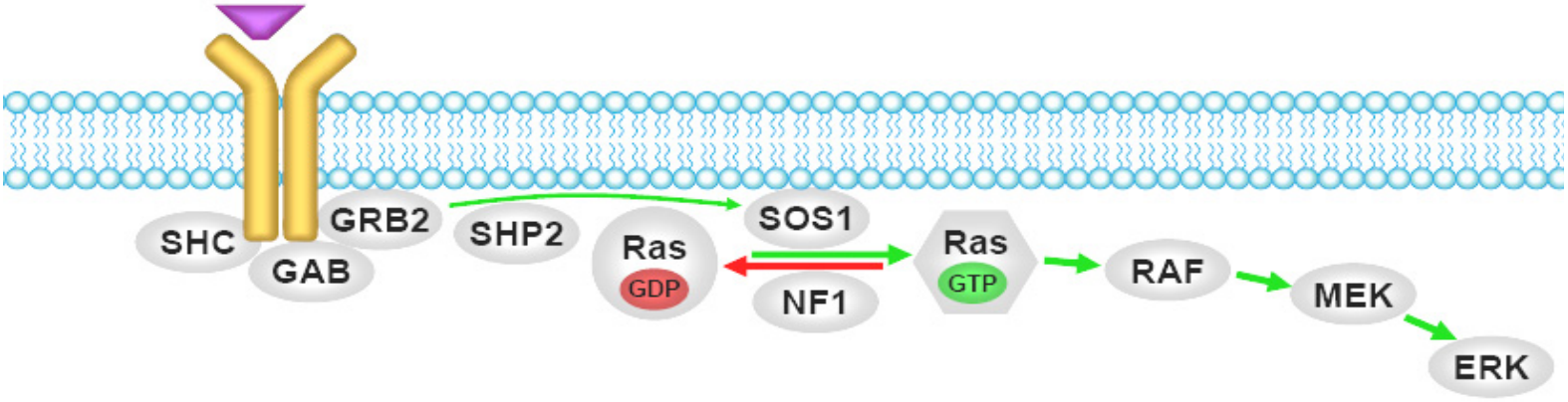

4th International Meeting on Rare Disorders of the RAS-MAPK Pathway
A workshop preceding the ESHG meeting Nuremberg, 2012

Organisers: Martin Zenker, Marco Tartaglia, Bronwyn Kerr.

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

Registration limit: 100 persons. Registration cost: 100 €.
Location: Hotel "Arvena Park", Görlitzer Str. 51, 90473 Nürnberg (2 km from ESHG conference venue)

Program

Day 1 (Friday, June 22)

14^{30} Registration, Coffee

15^{00} Welcome and Introduction
Martin Zenker, Magdeburg

15^{10} First Session: Clinical Aspects
Chair: Martin Zenker, Magdeburg

15^{10} The NSEuroNet Database
Christina Lißewski, Magdeburg

15^{30} *SOS1* and *RAF1*: Mutation Spectrum and Genotype-Phenotype Correlations
Marco Tartaglia, Rome

15^{50} *SHOC2*: Phenotype Analysis in 75 Patients
Martin Zenker, Magdeburg

16^{10} *NF1* and NFNS: Mutation Spectrum and Genotype-Phenotype Correlations
Alessandro De Luca, Rome

16^{30} *CBL* Mutations, Where Are the Patients?
Hélène Cavé, Paris

16^{50} Coffee Break

17^{10} Update on RASopathy Mutation Analysis and Epidemiology in Japan
Yoko Aoki, Sendai

17^{30} Noonan Guidelines, DYSCERNE and Beyond
Bronwyn Kerr, Manchester

17^{50} National Protocol for Diagnosis and Treatment in France: Noonan / Costello / CFC Syndrome
N.N. (provisional)

18^{10} Experience of a RASopathy Clinic
Giuseppe Zampino, Rome

18^{30} Dinner Buffet

Day 1 (Friday, June 22), continued

19^{30} 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.

Day 2 (Saturday, June 23)

8^{30} Second Session: Genes and Pathogenesis
Chair: Marco Tartaglia, Rome

8^{30} Disorders Caused by MYST4 / KAT6B Mutations – How Much of RASopathy?
Christian Thiel, Erlangen

8^{50} Genes and Mechanisms: What's New?
Marco Tartaglia, Rome

9^{10} Exome Sequencing – the Nijmegen Experience
Ineke van der Burgt, Nijmegen

10^{10} Novel Insights into Pathophysiology of *PTPN11* Mutations Causing Short Stature
Patrick Raynal, Toulouse

9^{30} Structural and Functional Characterization of RAS Mutants
Reza Ahmadian, Düsseldorf

9^{50} Noonan Syndrome Mouse Model: Tumor Predisposition and Therapeutic Strategies
Carmen Guerra, Madrid

10^{50} Coffee Break

11^{10} Third Session: Treatment Perspectives
Chair: Judith Allanson, Ottawa

11^{10} Treatment of Cardiomyopathy in LEOPARD Syndrome
Maria Kontaridis, Boston

11^{30} Treatment of *RAF1* Mutation-Induced Cardiomyopathy
Toshi Araki, Toronto

11^{50} Phase 2 Study on MEK162 in Noonan Syndrome Hypertrophic Cardiomyopathy
N.N. (provisional)

12^{10} Learning Difficulties in RASopathies: Pathogenesis and Treatment
Ype Elgersma, Rotterdam

12^{30} Use of Drosophila Models to Search for Noonan Syndrome Therapies
Bruce Gelb, New York

12^{50} Discussion

13^{10} Close, Lunch Buffet

14^{00} Meeting of the NSEuroNet Group
Chair: Marco Tartaglia, Rome

Speakers and Chairpersons:

Prof. Reza Ahmadian, PhD, Institute of Biochemistry and Molecular Biology II, Heinrich-Heine University, Düsseldorf, Germany

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. Toshi Araki, PhD, Department of Medical Biophysics, University of Toronto, Toronto, Ontario, Canada

Prof. Hélène Cavé, AP-HP, Département de Génétique, Hôpital Robert Debré, Paris, France

Dr. Alessandro De Luca, PhD, Dept. of Hematology. Oncology and Molec. Medicine, Istituto Superiore di Sanità, Rome, Italy

Prof. Ype Elgersma, PhD, Department of Neuroscience, Erasmus University Medical Centre, Rotterdam, The Netherlands

Prof. Bruce Gelb, MD, Center for Molecular Cardiology**,** Dept. of Pediatrics & Dept. of Genetics and Genomic Sciences, Mount Sinai School of Medicine**,** New York, USA

Dr. Carmen Guerra, PhD, Molecular Oncology Programme**,** Spanish National Cancer Research Centre (CNIO)**,** Madrid, Spain

Dr. Bronwyn Kerr, MD, University Department of Medical Genetics**,** Royal Manchester Children's Hospital**,** Manchester, UK

Prof. Maria Kontaridis, PhD, Harvard Medical School, Department of Cardiology, Beth Israel Deaconess Medical Center, Center for Life Sciences, Boston, MA, USA

Christina Lißewski, Institute of Human Genetics**,** University Hospital of Magdeburg, Germany

Prof. Patrick Raynal, PhD, Centre de Physiopathologie**,** Hôpital de Toulouse-Purpan**,** Toulouse, France

Prof. Marco Tartaglia, PhD, Dept. of Hematology. Oncology and Molec. Medicine, Istituto Superiore di Sanità, Rome, Italy

Dr. Christian Thiel, MD, Institute of Human Genetics, University Hospital Erlangen, University of Erlangen-Nuremberg, Erlangen, Germany

Dr. Ineke van der Burgt, MD, Department of Human Genetics**,** University Medical Center St Radboud**,** Nijmegen, The Netherlands

Prof. Guiseppe Zampino, MD, Department of Pediatrics, Catholic University, Rome, Italy

Prof. Martin Zenker, MD, Institute of Human Genetics**,** University Hospital of Magdeburg, Germany

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"*Associazione Italiana Sindromi Costello e Cardiofaciocutanea Onlus*"