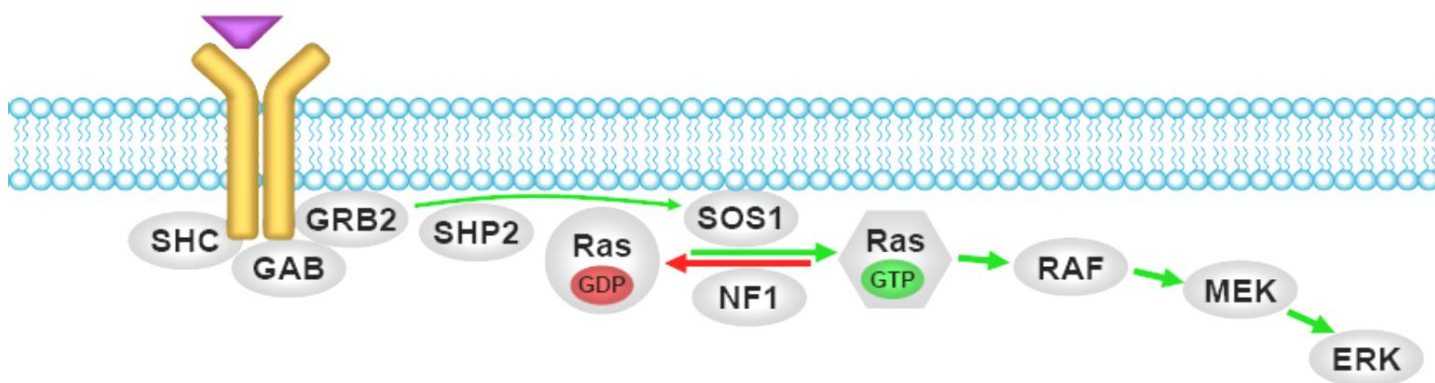




10th International Meeting on Rare Disorders of the RAS-MAPK Pathway

Scientific Board: Marco Tartaglia, Alain Verloes, Emma Burkitt-Wright, Martin Zenker

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Organized by: ERN-ITHACA

Supported by: SEDRA Foundation



Date: 12-13 October 2026

Registration limit: 150 participants

Location: Courtyard by Marriott Rome Central Park, Via Giuseppe Moscati, 7, 00168 Rome, Italy

<https://www.marriott.com/en-us/hotels/romcp-courtyard-rome-central-park/>

Programme - Day 1 (October 12)	
8:30	Registration
	RASopathies nosology consensus & Noonan guidelines meeting Chairs: Emma Burkitt-Wright, Bruce Gelb, Marco Tartaglia, Alain Verloes, Martin Zenker, PAG representatives
9:00	Guidelines: Status, critical pending issues & preparation of the manuscript – Emma Burkitt-Wright (Manchester)
10:30	Nosology – Martin Zenker (Magdeburg) Impulse talk: Toward a hierarchical nosology of RASopathies: integrating molecular coherence, clinical utility and stakeholder perspectives – Dagmar Tiemens (Nijmegen)

11:30	Presentation of ILIAD – Alain Verloes (Paris)
12:00	<i>Light Lunch</i>
13:00	Opening & Welcome : (greetings, logistics & Thanks to sponsors)
	First Session: 25 Years of <i>PTPN11</i> and Noonan Syndrome Chairs: Giovanni Neri, Michael Patton
13:15	Chairs' introductory remarks (15 min)
13:30	Invited talk: <i>PTPN11</i> & Noonan syndrome – Marco Tartaglia & Bruce D. Gelb
14:00	Invited talk: <i>Exploring SHP2 mutations from the molecular to the cellular scale</i> – Neel H. Shah
14:30	Selected abstract: New insights into the mechanism of RAS activation by SHP2 – Jeroen den Hertog
14:45	Selected abstract: Peptide-based inhibitors of SHP2 protein-protein interactions – Gianfranco Bocchinfuso
	Second Session: Genes & Pathways Chair: Marco Tartaglia, Yoko Aoki
15:00	Invited talk: <i>Exploring the role of downstream MAP kinases in the cognitive symptoms of Rasopathy syndromes</i> - J. Elliott Robinson
15:30	Invited talk: <i>Phenotypic heterogeneity of dysregulated YWHAZ and ERF function</i> - Marco Tartaglia
16:00	Selected abstract: Clinical spectrum, gen-phen correlations and functional characterization of pathogenic <i>NF1</i> variants affecting Lys1436 – Alessandro De Luca
16:15	Selected abstract: Ala13Asp in RAC1 enhances MAPK signalling and dysregulates cytoskeletal function, causing a neurodevelopmental condition with features resembling NS – Simona Coppola
16:30	<i>Coffee Break</i>
	Third Session: Cancer Chairs: Helene Cavé, Martin Zenker
17:00	Invited talk: <i>Recurrent FGFR1 hotspot variants drive low-grade brain tumors in PTPN11-related Noonan syndrome</i> - Karen W. Gripp
17:30	Invited talk: <i>LZTR1 and Cancer: penetrance matters</i> - Hélène Cavé
18:00	Selected abstract: Tumor predisposition in Noonan syndrome and related RASopathies – Adeline A Bonnard
18:15	Selected abstract: Solid tumors in RASopathies – Valentina Trevisan

20:00	Dinner
21:30	Evening Poster Session: Ideas, Insights & Connections
	Program – Day 2 (October 13)
	Fourth Session: Cardiovascular system Chairs: Bruce D Gelb, Giuseppe Limongelli
9:00	Invited talk: <i>Pathophysiology of pulmonary valve stenosis in NS</i> - Bruce D. Gelb
9:30	Invited talk: <i>From Cellular Crosstalk to Gene Correction: Patient iPSC Models as a Roadmap to Targeted Therapies in Noonan Syndrome Cardiomyopathy</i> - Lukas Cyganek
10:00	Invited talk: <i>HCM & targeted therapies</i> - Cordula Wolf
10:30	Selected abstract: RASopathy gene mutations in a cohort of patients with congenital heart defects – Christina Lissewski
10:45	Coffee break
	Fifth Session: Neurocognitive/neuromuscular aspects Chair: Yves Sznajer, Anna Fejtova
11:15	Invited talk: <i>Impacts of dysregulated RAS-MAPK signalling on functional development of nervous system</i> - Anna Fejtova
11:45	Invited talk: <i>Psychological Landscape in NS</i> - Paolo Alfieri
12:15	Selected abstract: Muscle ultrasonography in Costello syndrome - Germana Viscogliosi
12:30	Selected abstract: Forebrain GABAergic interneuron-specific <i>Brnf^{Q241R}</i> expression produces behavioural and hippocampal circuits alterations in a mouse model of CFCS – Jeimmy MV Ceron Gonzalez
	Patient Perspectives Session Chair: Alain Verloes, Keir Shiels
12:40	Selected abstract: NS: measuring the unmeasurable – Keir Shiels
12:50	Round table discussion with PAG representatives
13:30	Light lunch
	Sixth Session: Therapies Chair: Emma Burkitt-Wright, Sahar Mansour
14:30	Invited talk: <i>Pathogenesis and therapeutic approaches in mouse models of Noonan syndrome</i> – Yoko Aoki

15:00	Invited talk: Novel therapeutic perspectives for short stature in NS – Thomas Edouard
15:30	Selected abstract: Trametinib for multiple non-ossifying fibromas due to KRAS mosaic mutations: long-term follow-up – Nicolò Tesio
15:45	Selected abstract: Beneficial effects beyond growth of rGH therapy in children with NS and related disorders – Janne CP van de Ven
Seventh Session: Open topics	
Chair: Giuseppe Zampino, Thomas Edouard	
16:00	Invited talk: <i>Breaking the rules: selfish selection and transmission ratio distortion in RASopathies</i> – Anne Goriely
16:30	Selected abstract: Perinatal mortality in RASopathies – Yline Capri
16:45	Selected abstract: Mitochondrial dysfunction drives systemic metabolic alterations in CS - Cristina Nuevo-Tapioles
17:00	Selected abstract: NS-associated hypoleptinemia arises from central leptin hypersensitivity and contribute to increased energy expenditure – Armelle Yart
17:15	Selected abstract: Towards perspective controlled MEK inhibition: MIRACLES study, an AIFA-funded phase II multicentre, randomized, double-blind, placebocontrolled crossover clinical trial – Alessandro Mussa
17:30	Closing remarks
17:45	End of the meeting
18:00 – 19:30	Research-oriented networking (open to attendees)

Speakers and Chairpersons:

Yoko Aoki, MD, PhD, Department of Medical Genetics, Tohoku University School of Medicine, Sendai, Japan

Paolo Alfieri, MD, PhD, Ospedale Pediatrico Bambino Gesù, IRCCS, Rome, Italy

Gianfranco Bocchinfuso, PhD, Department of Chemical Sciences and Technologies, University of Rome Tor Vergata, Rome, Italy

Adeline A. Bonnard, MD, Department of Genetics, Robert Debré University Hospital, APHP, Paris, France

Emma Burkitt-Wright, MD, Manchester Centre for Genomic Medicine, Manchester University NHS Foundation Trust and University of Manchester, Manchester, UK

Yline Capri, MD, Department of Genetics, Robert Debré University Hospital, APHP, Paris, France

Hélène Cavé, PhD, INSERM UMR_S1131, Institut de Recherche Saint-Louis, Université de Paris & Assistance Publique des Hôpitaux de Paris AP-HP, Hôpital Robert Debré, Département de Génétique, Paris, France

Jeimmy M.V. Ceron Gonzalez, PhD, Department of Genetics and Molecular Neurobiology, Research Group “Synapto-Oscillopathies”, Otto von Guericke University Magdeburg, Magdeburg, Germany

Simona Coppola, PhD, Istituto Superiore di Sanità, Rome, Italy

Lukas Cyganek, PhD, Stem Cell Unit, Clinic for Cardiology and Pneumology, University Medical Center Göttingen & DZHK (German Center for Cardiovascular Research), partner site Göttingen, Göttingen, Germany

Alessandro De Luca, PhD, Fondazione IRCCS Casa Solievo della Sofferenza – Istituto CSS-Mendel, Rome, Italy

Jeroen den Hertog, PhD, Hubrecht Institute-KNAW and University Medical Center Utrecht, Utrecht & Institute Biology Leiden, Leiden University, Leiden, The Netherlands

Thomas Edouard, MD, PhD, Toulouse University Hospital, Toulouse, France

Anna Fejtova, PhD, Department of Psychiatry and Psychotherapy, Universitätsklinikum Erlangen, Friedrich-Alexander-Universität Erlangen-Nürnberg, Erlangen, Germany

Bruce D. Gelb, MD, PhD, Mindich Child Health and Development Institute, Icahn School of Medicine at Mt. Sinai, New York, NY, USA

Anne Goriely, PhD, MRC Weatherall Institute of Molecular Medicine, Radcliffe Department of Medicine, University of Oxford, Oxford, UK

Karen W. Gripp, MD, Nemours Children’s Health, Wilmington, DE, USA

Giuseppe Limongelli, MD, PhD, University of Campania “Luigi Vanvitelli”, Naples, Italy

Christina Lissewski, PhD, Institute for Medical Genetics, Carl von Ossietzky University Oldenburg, Oldenburg, Germany

Sahar Mansour, MD, PhD, St George’s University Hospitals NHS Foundation Trust, London, UK

Alessandro Mussa, MD, PhD, Department of Public Health and Pediatric Sciences, University of Torino & Pediatric Clinical Genetics Unit, Regina Margherita Children’s Hospital, Città della Salute e della Scienza, Torino, Italy

Giovanni Neri, PhD, MSc, MPharma, Università Cattolica del Sacro Cuore, Rome, Italy

Cristina Nuevo-Tapióles, PhD, NYU School of Medicine, New York, NY, USA

Michael Patton, MD, St. Georges University of London, UK

J. Elliott Robinson, MD, PhD, Cincinnati Children’s Hospital Medical Center, Cincinnati, OH, USA

Neel H. Shah, PhD, Columbia University, New York, NY, USA

Keir Shiels, MB BChir, Great Ormond Street Hospital & University College London, London, UK

Yves Sznajer, MD, PhD, Center for Human Genetic Cliniques Universitaires Saint Luc, Brussels Belgium

Marco Tartaglia, PhD, Molecular Genetics and Functional Genomics, Ospedale Pediatrico Bambino Gesù, Rome, Italy

Nicolò Tesio, MD, Department of Sciences of Public Health and Pediatrics, University of Turin, Turin, Italy

Dagmar Tiemens, MD, Department of Pediatrics, Radboud Institute for Health Sciences, Amalia Children’s Hospital, Radboud University Medical Center, Nijmegen, The Netherlands

Valentina Trevisan, MD, Centre for Rare Diseases and Birth Defects, Department of Woman and Child Health and Public Health, Fondazione Policlinico Universitario “A. Gemelli” IRCCS; and Genomic Medicine, Department of Life Sciences and Public Health, Catholic University of the Sacred Heart, Rome, Italy

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Germana Viscogliosi, MD, Fondazione Policlinico Universitario “A. Gemelli” IRCCS & Università Cattolica del Sacro Cuore, Rome, Italy

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Armelle Yart, PhD, RESTORE Research Center, INSERM, Université de Toulouse, Toulouse, France

Giuseppe Zampino, MD, Centre for Rare Diseases and Birth Defects, Department of Woman and Child Health and Public Health, Fondazione Policlinico Universitario "A. Gemelli" IRCCS & Department of Life Sciences and Public Health, Catholic University of the Sacred Heart, Rome, Italy

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

Patient Advocacy Representatives:

Serge Arnoulet, President of the French Costello and Cardio-Facio-Cutaneous Syndromes Association, France

Tammy Bowers, Noonan Syndrome Foundation, USA

Jeanette Dyer, Noonan Syndrome Association, UK

Antonella Esposito, Angeli Noonan, Associazione Italiana Sindrome di Noonan, Italy

Angelo Petroni, Angeli Noonan, Associazione Italiana Sindrome di Noonan, Italy

Leonie Ran, MEK2 Research Foundation, The Netherlands

Andrea Reid-Kelly, Deputy Chair, Noonan Syndrome Association, UK

Lisa Schoyer, MFA, President of RASopathiesNet, USA

Gretchen Spiess, CFC International, USA

Beth Stronach, RASopathies Network, USA

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Paolo Velonà, Associazione Nazionale Sindrome di Noonan, Italy

Justyna Walczuk, MEK2 Research Foundation, Poland