

7th International RASopathies Symposium: Pathways to Understanding – Expanding Knowledge, Enhancing



Research and Therapeutic Discovery

Chairs Maria Kondaridis PhD, and Amy Roberts MD
and Honorary Chairs Marco Tartaglia PhD, and Martin Zenker MD

Lisa Schoyer MFA, Principal Investigator
Lisa Schill BS, and Beth Stronach PhD Co-Investigators

July 23-25, 2021
VIRTUAL MEETING

AGENDA
as of July 16, 2021

Note: The EDT - Eastern Daylight Time of the original agenda <https://rasopathiesnet.org/2021-symposium-agenda/> has been “translated” into **Central European Summer Time (CEST)/ Mitteleuropäische Sommerzeit (MESZ)** (Karola Zenker)

Duration	Schedule (CEST)	Description	Speaker/Moderator
DAY 1 – FRIDAY, JULY 23, 2021 — 16:00 bis 20:00 Uhr (CEST/ MESZ)			
50 min	16:00-16:50	Opening Session	
	16:00	Welcome Remarks	Chair: Maria Kondaridis PhD, MMRI, Utica, NY
	16:05	Celebrating 20th Anniversary of the Discovery of PTPN11, the First RASopathy Gene	Marco Tartaglia PhD, Bambino Gesù Hospital, Rome, Italy, and Bruce Gelb MD, Mount Sinai, NYC, NY
	16:20	Advocate Keynote: Finding a Diagnosis & Our Voice as Advocates	Elisabeth Parker BS, Yoga Instructor, Move2Advocate (Parent of Child with NS/JMML)
	16:40	Q&A Moderated by Amy Roberts MD, Boston Children’s Hospital, Boston, MA	
1h	16:50-17:50	Session 1: Genes, Pathways, and Genocopies Moderator: Katherine Rauen MD PhD, UC Davis, CA	
	16:50	Introduction	Katherine Rauen MD PhD, UC Davis, CA
	16:52	CDC42, RASA2, FBXW11, ZNF426, YWHAZ, TRAF7: Novel RASopathy genes or not?	Martin Zenker MD, University of Magdeburg, Germany
	17:04	Novel disease gene identification for RASopathies: where are we now?	Marco Tartaglia PhD, Bambino Gesù Hospital, Rome, Italy
	17:16	Noncanonical GTPases: RRAS2, RRAS, MRAS, RIT1	Yoko Aoki MD PhD, Tohoku University, Sendai, Japan
	17:28	RASopathy genetic variation in biobanks	Bruce Gelb MD, Mount Sinai, NYC, NY
	17:38	Q&A Moderated by Katherine Rauen MD PhD, UC Davis, CA	
10m	17:50	Commercial Break	

Duration	Schedule (CEST)	Description	Speaker/Moderator
1h	18:00-19:00	Session 2: Novel Strategies and Mechanisms in RASopathies Moderators: Pau Castel PhD, NYU Langone; Annette Bakker PhD, Children's Tumor Foundation, NYC, NY	
	18:00	Introduction	Pau Castel PhD, NYU Langone
	18:02	Intronic CRISPR repair in a preclinical model of LZTR1-associated cardiomyopathy	Lukas Cyganek PhD, University Medical Center, Göttingen, Germany
	18:14	Using iPSCs to study syndrome-specific properties	Maria Kontaridis PhD, MMRI, Utica, NY
	18:26	How to temper Shoc2-ERK1/2 signals – molecular insights	Emilia Galperin PhD, University of Kentucky, Lexington, KY
	18:38	Novel findings and expansion of phenotype in a mosaic RASopathy caused by somatic KRAS variants	Caitlin Chang MD, British Columbia Women & Children's Hospital, Vancouver, BC, Canada
	18:50	Q&A Moderated by Pau Castel PhD, NYU Langone	
1h	19:00-20:00	Session 3: Lymphatic and Cardiovascular Manifestations Moderators: Sahar Mansour MD, St. Georges Hospital, London, UK; David Stevenson MD, Stanford University, CA	
	19:00	Introduction	Sahar Mansour MD, St. Georges Hospital, London, UK
	19:02	Lymphatic problems and lymphedema in RASopathies	Kristiana Gordon MD, St. Georges Hospital, London, UK
	19:14	Patterns and therapeutic options in Noonan syndrome-related lymphatic disorders	Maxim Itkin MD, Penn Medicine, Philadelphia, PA
	19:26	Therapeutic strategies for the treatment of hypertrophic cardiomyopathy in Noonan syndrome with multiple lentigines	Anton Bennett PhD, Yale School of Medicine, New Haven, CT
	19:38	Q&A Moderated by David Stevenson MD, Stanford University, CA	
DAY 2 – SATURDAY, JULY 24, 2021 — 16:00 bis 20:00 Uhr (CEST/ MESZ)			
30m	16:00-16:30	Keynote & Poster Session Moderator: Beth Stronach PhD, RASopathies Network, Pittsburgh, PA	
	16:00	Introduction	Beth Stronach PhD, RASopathies Network, Pittsburgh, PA
	16:02	Opening Keynote: Update on MEK and mTOR inhibition for RASopathy-associated cardiac disease	Gregor Andelfinger MD PhD, CHU Ste-Justine, Montreal, Canada, 2019 Penn Medicine MDBR Grant Recipient
	16:25	Q&A Moderated by Beth Stronach, PhD, RASopathies Network, Pittsburgh, PA	
1h20m	16:30-17:50	Poster Session	6 Posters Presented by RASopathy Individuals and Families – Patients in their Natural Environments
	16:30	1) Child with CFC-YWHAZ	Amanda Monty, Mother
	16:40	2) Adult with Attenuated Costello Syndrome	Amaris Figueroa, Self Advocate, with Yadira Figueroa, Mother
	16:50	3) Child with NF1 on MEK Inhibitor	Hilary Berens, Mother
	17:00	Poster Q&A Moderated by Beth Stronach, PhD, RASopathies Network, Pittsburgh, PA	
	17:10	4) Child with NS-RIT1 on MEK Inhibitor	Samantha Craig Spire, Mother
	17:20	5) Adult with NS-BRAF	Katie Ryan, Self Advocate
	17:30	6) Child with NS-SHOC2	Lauren Throop, Mother
	17:40	Poster Q&A Moderated by Beth Stronach, PhD, RASopathies Network, Pittsburgh, PA	

Duration	Schedule (CEST)	Description	Speaker/Moderator
10m	17:50	Commercial Break	
1h	18:00-19:00	Session 4: Neurocognitive Associations Moderators: Tamar Green MD, Stanford University, CA; Marni Axelrad PhD, Texas Children's Hospital, Houston, TX	
	18:00	Introduction	Marni Axelrad PhD, Texas Children's Hospital, Houston, TX
	18:02	Non-Neurofibromatosis Type 1 RASopathies, particularly Noonan syndrome, are associated with multi-focal low-grade gliomas which commonly harbor FGFR1 mutations	Alberto Broniscer MD MS, University of Pittsburgh Medical Center (UPMC), Children's Hospital of Pittsburgh
	18:14	Neurological and neurodevelopmental features of CFC syndrome: a multinational study	Rene Pierpont PhD, University of Minnesota, Minneapolis, MN
	18:26	Epilepsy and BRAF mutations: phenotypes, natural history, and genotype-phenotype correlations	Domenica Battaglia MD, University of Agostino Gemelli, Rome, Italy
	18:38	Behavioral phenotypes across the RASopathies	Shruti Garg MBBS MRCPsych PhD, University of Manchester, UK
	18:50	Q&A Moderated by Marni Axelrad PhD, Texas Children's Hospital, Houston, TX	
1h	19:00-20:00	Session 5: Clinical Manifestations Moderator: Jeroen den Hertog PhD, Leiden University, Utrecht, The Netherlands	
	19:00	Introduction	Jeroen den Hertog PhD, Leiden University, Utrecht, The Netherlands
	19:02	Mosaicism in RASopathies: HRAS in cancer and Costello syndrome	Karen Gripp MD, Nemours/duPont Hospital, Wilmington DE
	19:14	Modeling skeletal myopathy in CFC	Katherine Rauen, MD PhD, UC Davis, CA
	19:26	Skin and hair manifestations in CS and CFC	Maija Kiuru MD PhD, UC Davis, CA
	19:38	Ocular manifestations in RASopathies	Suma Shankar MD PhD, UC Davis, CA
	19:40	Q&A Moderated by Jeroen den Hertog PhD, Leiden University, Utrecht, The Netherlands	
DAY 3 – SUNDAY, JULY 25, 2021 — 16:00 bis 22:15 Uhr (CEST/ MESZ)			
1h	16:00-17:00	Session 6: Selected Junior Investigator Presentations Moderators: Amy Roberts MD, Boston Children's Hospital, MA; William Timmer PhD, NCI CTEP, Bethesda, MD	
	16:00	Introduction	Amy Roberts MD, Boston Children's Hospital, MA
	16:03	Junior Investigator Finalist Presentation #1: A Collection of Drosophila Models to Identify RASopathy Subtype Specific Differences	Tirtha Das PhD, Mount Sinai, NYC, NY
	16:17	Q&A Moderated by: Amy Roberts MD, Boston Children's Hospital, MA	
	16:31	Junior Investigator Finalist Presentation #2: Abnormal Myelin Differentially Impairs Learning in Models of the RASopathies Neurofibromatosis Type 1 and Costello Syndrome	Alejandro Lopez-Juarez PhD, UT Rio Grande Valley, TX
	16:45	Q&A Moderated by: Amy Roberts MD, Boston Children's Hospital, MA	

Duration	Schedule (CEST)	Description	Speaker/Moderator
	16:50	Junior Investigator Finalist Presentation#3: Biochemical analysis of RIT1 reveals preferential interaction with RAF1 and provides evidence for therapeutic intervention of cardiac hypertrophy	Richard Van BS, UCSF, CA, and Morgan Wagner BS, NCI RAS Initiative, Frederick, MD
	16:55	Q&A Moderated by Amy Roberts MD, Boston Children's Hospital, MA	
2h	17:00-18:00	Session 7: Learning from each other: Perspectives on Diagnosis and Therapeutic Interventions	
	17:00	PANEL: Therapeutic interventions: What's in the Pipeline? Small molecules, RNA therapies, NF1 gene therapy	Moderator: Stas Shvartzman PhD, Princeton University, NJ Panelists: Stephanie Ware MD PhD, Indiana University; PJ Brooks PhD, NCATS/NIH; Bruce Korf MD PhD, UAB; Jeff R Livingstone PhD, Igia Pharmaceuticals
	17:40	Q&A moderated by Stas Shvartzman PhD, Princeton University, NJ	
	17:55	Commercial Break	
	18:05	Introduction: Compassionate Use and Clinical Trials in the RASopathies	Moderators: Pilar Magoulas MS CGC, Texas Children's Hospital, Houston, TX; Lisa Schill BS, RASopathies Network, Williamstown, NJ
	18:10	FAMILY PANEL: Compassionate use and clinical trials of MEK Inhibitors	Advocates Affected by a RASopathy: CFC, CS, NS, NF1
	18:45	Q&A Moderated by Pilar Magoulas MS CGC, Texas Children's, Houston, TX; Lisa Schill BS, RASopathies Network, Williamstown, NJ	
	19:00	Commercial Break	
3h5m	19:10-22:15	SUMMARY OF THE SYMPOSIUM FOR FAMILIES – FREE SEPARATE REGISTRATION – go here to register: https://rasopathiesnet.org/2021-symposium-registration/	
45m	19:10-19:55	for CFC Families	Pilar Magoulas MS, CGC; Suma Shankar MD, PhD; David Stevenson MD; NCI RASopathies Study Team
45m	19:55-20:40	for CS Families	Karen Gripp MD; Bronwyn Kerr MD; NCI RASopathies Study Team
45m	20:40-21:25	for NS, NSML Families	Bruce Gelb MD; Tamar Green MD; Amy Roberts MD; NCI RASopathies Study Team
45m	21:25-22:10	for NF1 Families	Annette Bakker PhD; Bruce Korf MD PhD; NCI RASopathies Study Team
	5m	Discussion and Closing Remarks	Lisa Schoyer, MFA, RASopathies Network, Altadena, CA