



# 7th International RASopathies Symposium: Pathways to Understanding – Expanding Knowledge, Enhancing Research and Therapeutic Discovery

July 23-25, 2021  
VIRTUAL MEETING

## Chairs

Maria Kontaridis, PhD, Masonic Medical Research Institute  
Amy Roberts, MD, Boston Children's Hospital

*Honorary Chairs: Marco Tartaglia, PhD, Bambino Gesù Children's Hospital, and Martin Zenker, MD, Universitätsklinikum Magdeburg*

Duration	Schedule (in EDT)	Description	Presenter
<b>DAY 1 – FRIDAY, JULY 21, 2021 – 10am to 2pm EDT (7am to 11am PDT)</b>			
<b>50m</b>	<b>10:00-10:50am</b>	<b>OPENING SESSION</b>	
2m	10:00-10:05am	Welcome Remarks	Maria Kontaridis PhD, MMRI, Utica, NY and Amy Roberts, MD, Boston Children's Hospital, MA
15m	10:05-10:20am	Celebrating 20 <sup>th</sup> Anniversary of the Discovery of PTPN11, the First RASopathy Gene	Marco Tartaglia PhD, Bambino Gesù Hosp, Rome, Italy and Bruce Gelb MD, Mount Sinai, NYC, NY
20m	10:20-10:40am	Advocate Keynote: Finding a Diagnosis & Our Voice as Advocates	Elisabeth Parker BS, Yoga Instructor, Move2Advocate
10m	10:40-10:10:50am	Q&A moderated by Amy Roberts, MD, Boston Children's Hospital, MA	
<b>1h</b>	<b>10:50-11:50am</b>	<b>SESSION 1: Genes, Pathways and Genocopies</b> Moderator: Katherine A. Rauen MD PhD, UC Davis, CA	
2m	10:50-10:52am	Introduction	Katherine A. Rauen MD PhD, UC Davis, CA
12m	10:52-11:04am	CDC42, RASA2, FBXW11, ZNF426, YWHAZ, TRAF7: Novel RASopathy genes or not?	Martin Zenker MD, University of Magdeburg, Germany
12m	11:04-11:16am	Novel disease gene identification for RASopathies: where are we now?	Marco Tartaglia PhD, Bambino Gesù Hosp, Rome, Italy
12m	11:16-11:28am	Noncanonical GTPases: RRAS2, RRAS, MRAS, RIT1	Yoko Aoki MD PhD, Tohoku Univ, Sendai, Japan
12m	11:28-11:40am	RASopathy genetic variation in biobanks	Bruce Gelb MD, Mount Sinai, NYC, NY
10m	11:40-11:50am	Q&A moderated by Katherine A. Rauen MD PhD, UC Davis, CA	
<b>10m</b>	<b>11:50am-12:00pm</b>	<b>BREAK</b>	
<b>1h</b>	<b>12:00-1:00pm</b>	<b>SESSION 2: Novel Strategies and Mechanisms in RASopathies</b> Moderators: Pau Castel PhD, NYU Langone; Annette Bakker PhD, Children's Tumor Foundation, NYC, NY	
2m	12:00-12:02pm	Introduction	Pau Castel PhD, NYU Langone, NY
12m	12:02-12:14pm	Intronic CRISPR repair in a preclinical model of LZTR1-associated cardiomyopathy	Lukas Cyganek PhD, University Med Center, Göttingen, Germany
12m	12:14-12:26pm	Using iPSCs to study syndrome-specific properties	Maria Kontaridis PhD, MMRI, Utica, NY
12m	12:26-12:38pm	How to temper Shoc2-ERK1/2 signals - molecular insights	Emilia Galperin PhD, University of Kentucky, Lexington, KY
12m	12:38-12:50pm	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
10m	12:50-1:00pm	Q&A moderated by: Pau Castel PhD, NYU Langone, NY	



Duration	Schedule (in EDT)	Description	Presenter
<b>1h</b>	<b>1:00-2:00pm</b>	<b>SESSION 3: Lymphatic and Cardiovascular Manifestations</b>	
		Moderators: Sahar Mansour MD, St Georges Hospital, London, UK; David Stevenson MD, Stanford Univ CA	
2m	1:00-1:02pm	Introduction	Sahar Mansour MD, St Georges Hospital, London, UK
12m	1:02-1:14pm	Lymphatic problems and lymphedema in RASopathies	Kristiana Gordon MD, St. Georges Hosp, London, UK
12m	1:14-1:26pm	Patterns and therapeutic options in Noonan syndrome-related lymphedema	Maxim Itkin MD, Penn Medicine, Philadelphia, PA
12m	1:26-1:38pm	Therapeutic strategies for the treatment of hypertrophic cardiomyopathy in Noonan syndrome with multiple lentiginos	Anton Bennett PhD, Yale School of Medicine, New Haven, CT
22m	1:38-2:00pm	Q&A moderated by: David Stevenson MD, Stanford Univ CA	

**DAY 2 – SATURDAY, JULY 22, 2021 – 10am to 2pm EDT (7am to 11am PDT)**

<b>2h50m</b>	<b>10:00-11:50</b>	<b>KEYNOTE &amp; POSTER SESSION</b>	
		Moderator: Beth Stronach PhD, RASopathies Network, Pittsburgh PA	
2m	10:00-10:02am	Introduction	
23m	10:02-10:25am	<b>Opening Keynote:</b> 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
5m	10:25-10:50am	Q&A moderated by Beth Stronach PhD, RASopathies Network, Pittsburgh, PA	
<b>1h20m</b>	<b>10:30-11:10am</b>	Poster Session – 6 Posters Presented by Families	
10m	10:30-10:40am	1) Child with CFC/YWHAZ	Amanda Monty, Mother
10m	10:40-10:50am	2) Adult with Attenuated Costello syndrome	Amaris Figueroa, Self-Advocate, w/Yadira Figueroa, Mother
10m	10:50-11:00am	3) Child with NF1 on MEK inhibitor	Hilary Berens, Mother
10m	11:00-11:10am	Poster Q&A moderated by Beth Stronach PhD, RASopathies Network, Pittsburgh PA	
10m	11:10-11:20am	4) Child with NS/RIT1 on MEK inhibitor	Samantha Craig Spires, Mother
10m	11:20-11:30am	5) Adult with NS/BRAF	Katie Ryan, Self-Advocate
10m	11:30-11:40am	6) Child with NS/SHOC2	Lauren Throop, Mother
10m	11:40-11:50am	Poster Q&A moderated by Beth Stronach PhD, RASopathies Network, Pittsburgh PA	
<b>10m</b>	<b>11:50am-12:00pm</b>	<b>BREAK</b>	
<b>1h</b>	<b>12:00-1:00pm</b>	<b>SESSION 4: Neurocognitive Associations</b>	
		Moderators: Tamar Green MD, Stanford University, A; Marni Axelrad PhD, Texas Children’s, Houston, TX	
2m	12:00-12:02pm	Introduction	Marni Axelrad PhD, Texas Children’s, Houston, TX
12m	12:02-12:14pm	Non-NF1 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
12m	12:14-12:26pm	Neurological and neurodevelopmental features of CFC syndrome: a multinational study	Rene Pierpont PhD, University of Minnesota, Minneapolis, MN
12m	12:26-12:38pm	Epilepsy and BRAF mutations: phenotypes, natural history, and genotype-phenotype correlations	Domenica Battaglia MD, Fondazione Policlinico Universitario A Gemelli IRCCS, Catholic University, Rome, Italy
12m	12:38-12:50pm	Behavioral phenotypes across the RASopathies	Shruti Garg MBBS MRCPsych PhD, University of Manchester, UK
10m	12:50-1:00pm	Q&A Moderated by: Marni Axelrad PhD, Texas Children’s, Houston, TX	
<b>1h</b>	<b>1:00-1:200pm</b>	<b>SESSION 4: Clinical Manifestations</b>	
		Moderator: Jeroen den Hertog PhD, Hubrecht Institute, The Netherlands	
2m	1:00-1:02pm	Introduction	Jeroen den Hertog PhD, Hubrecht Institute, The Netherlands
12m	1:02-1:14pm	Mosaicism in RASopathies: HRAS in cancer and Costello syndrome	Karen Gripp MD, Nemours/DuPont Hospital, Wilmington, DE
12m	1:14-1:26pm	Modeling skeletal myopathy in CFC	Katherine Rauen MD PhD, UC Davis, CA
12m	1:26-1:38pm	Skin and hair manifestations in CS and CFC	Maija Kiuru MD PhD, UC Davis, CA
12m	1:38-1:50pm	Ocular manifestations in RASopathies	Suma Shankar MD PhD, UC Davis, CA
10m	1:50-2:00pm	Q&A Moderated by: Jeroen den Hertog PhD, Hubrecht Institute, The Netherlands	



Duration	Schedule (in EDT)	Description	Presenter
<b>DAY 3 – SUNDAY, JULY 25, 2021 – 10am to 4:15pm EDT (7am to 11am PDT)</b>			
<b>1h</b>	<b>10:00-11:00am</b>	<b>SESSION 6: Selected Junior Investigator Presentations</b> Moderators: Amy Roberts MD, Boston Children’s, MA; William Timmer PhD, NCI CTEP, Bethesda, MD	
3m	10:00-10:03am	Introduction	Amy Roberts MD, Boston Children’s, MA
14m	10:03-10:17am	Junior Investigator Finalist Presentation #1: A Collection of Drosophila Models to Identify RASopathy Subtype Specific Differences	Tirtha Das PhD, Mount Sinai, NYC, NY
5m	10:17-10:22am	Presentation 1 Q&A moderated by: Amy Roberts MD, Boston Children’s, MA	
14m	10:22-10:36am	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
5m	10:36-10:41am	Presentation 2 Q&A moderated by: Amy Roberts MD, Boston Children’s, MA	
14m	10:41-10:55am	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
5m	10:55-11:00am	Presentation 3 Q&A moderated by: Amy Roberts MD, Boston Children’s, MA	
<b>2h</b>	<b>11:00am-1:00pm</b>	<b>SESSION 7: Learning from Each Other: Perspectives on Diagnosis and Therapeutic Interventions</b>	
40m	11:00-11:40am	<b>Professional Panel: Therapeutic interventions: What's in the Pipeline?</b> Small molecules, RNA therapies, NF1 gene therapy	<b>Moderator:</b> Stas Shvartzman PhD, Princeton University, NJ; <b>Panelists:</b> Stephanie Ware MD PhD, Indiana University PJ Brooks PhD, NCATS/NIH Bruce Korf MD PhD, UAB Jeff R. Livingstone PhD, Igia Pharmaceuticals
15m	11:40-11:55am	Q&A moderated by Stas Shvartzman PhD, Princeton University, NJ	
<b>10m</b>	<b>11:55am-12:05pm</b>	<b>BREAK</b>	
40m	12:05-12:45pm	<b>Family Panel: Compassionate use and clinical trials of MEK inhibitors</b>	<b>Moderators:</b> Pilar Magoulas MS CGC, Texas Children’s Hosp, Houston, TX Lisa Schill, BS, RASopathies Network, Williamstown, NJ <b>Panelists:</b> Theresa Milner, Mother of child with CFCS Sarah and Kevin Chamness, Parents of child with CS Shane Smith, Father of child with NF1 Kevin Plouffe, Father of child with NS
15m	12:45-1:00pm	Q&A moderated by Pilar Magoulas MS CGC, Texas Children’s Hosp, Houston, TX and Lisa Schill, BS, RASopathies Network, Williamstown, NJ	
<b>10m</b>	<b>1:00-1:10pm</b>	<b>BREAK</b>	
<b>2h5m</b>	<b>1:10-4:15pm</b>	<b>SUMMARY OF THE SYMPOSIUM FOR FAMILIES</b>	
45m	1:10-1:55pm	<b>For CFC syndrome Families</b>	David Stevenson MD Suma Shankar MD, PhD Pilar Magoulas MS, CGC NCI RASopathies Study Team
45m	1:55-2:40pm	<b>For Costello syndrome Families</b>	Karen Gripp MD Bronwyn Kerr MD NCI RASopathies Study Team
45m	2:40-3:25pm	<b>For Noonan Syndrome Families</b>	Amy Roberts MD Bruce Gelb MD Tamar Green MD NCI RASopathies Study Team
45m	3:25-4:40pm	<b>For NF1 Families</b>	Bruce Korf MD PhD Annette Bakker PhD
5m	4:40-4:45pm	Discussion and Closing Comments Lisa Schoyer MFA, Rasopathies Network, Altadena, CA	

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