

# 5<sup>th</sup> International RASopathies Symposium: When Development and Cancer Intersect

Renaissance Orlando at SeaWorld, Orlando, Florida ~ Friday, July 28 to Sunday, July 30, 2017

## AGENDA

Chairs: Katherine A. Rauen, MD, PhD and Frank McCormick, PhD, FRS

Co-Organizers: RASopathiesNet - Lisa Schoyer, MFA, Lisa Schill, BS, Beth Stronach, PhD

### DAY 1 – FRIDAY 7/28/17

8:00-10:00 pm	<b>Dessert and Scientific Poster Session</b> (Scientists and advocacy/family groups) Goal: Encourage collaboration between researchers and families in a nonclinical setting
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### DAY 2 – SATURDAY 7/29/17

7:00-8:00 am	<b>Breakfast</b>
9:00 am-5:00 pm	<b>Family Photo Shoot</b> in and around meeting areas - Rick Guidotti
8:00-10:00 am	<b>Session 1: What Defines a RASopathy?</b> Moderator: <b>Martin Zenker, MD</b> Goals: Hear from patients and families about their experiences living with a RASopathy. Lay the scientific foundation for a comprehensive and accurate understanding of the current definition of a RASopathy.
8:00-8:30 am	<b>Four Individuals/Caregivers:</b> Perspectives from the Home Front
8:30-9:00 am	<b>Frank McCormick, PhD, FRS:</b> Molecular primer on the Ras signaling pathway
9:00-9:30 am	<b>Katherine A. Rauen, MD, PhD:</b> The RASopathies
9:30-10:00 am	<i>Discussion</i> (Clinical Diagnosis, Phenotype-Centered vs. Molecularly Defined)
10:00-10:10 am	<i>Break</i>
10:10 am -12:00 pm	<b>Session 2: Syndromic and Sporadic Cancers of the Ras Pathway</b> Moderator: <b>Karen Gripp, MD</b> Goal: Examine cancer types and mechanisms in individuals with RASopathies
10:10-10:50 am	<b>KEYNOTE: Nancy Ratner, PhD:</b> Preclinical studies to guide NF1 clinical trials
10:50-11:10 am	<b>Corinne Linardic, MD:</b> Pediatric sarcomas, rhabdomyosarcoma
11:10-11:30 am	<b>Hélène Cavé, PharmD PhD:</b> Myoproliferative neoplasms (JMML) in RASopathies
11:30-11:50 am	<b>Brigitte Widemann, MD:</b> Clinical trial updates, NF1 Plexiforms and MPNSTs
11:50 am – 12:00 pm	<i>Discussion</i>
12:00 – 1:00 pm	<b>Lunch and Learn: Rick Guidotti: Positive Exposure</b>
1:00-4:30 pm	<b>Session 3: Human Development: Effects on Organ Systems</b> Moderator: <b>Bruce Korf, MD, PhD</b> Goal: Learn how RASopathy mutations affect developing tissues, organs, and body systems
1:00-1:40 pm	<b>KEYNOTE: Marco Tartaglia, PhD:</b> RASopathy genetics, new mutations, congenital defects
1:40-2:40 pm	<b>NERVOUS SYSTEM</b>
1:40-2:00 pm	<b>Erika Yeh, PhD (Weiss lab):</b> From skin cells to neurons
2:00-2:20 pm	<b>Erik Ullian, PhD:</b> How do astrocytes affect brain function in RASopathies?
2:20-2:40 pm	<b>Giuseppe Zampino, MD:</b> Penn MDBR 2015 Grantee - Pain in RASopathies: new investigative techniques and treatments
2:40-2:50 pm	<i>Discussion</i>
2:50-3:00 pm	<i>Break</i>
3:00-3:40 pm	<b>CIRCULATORY SYSTEMS</b>
3:00-3:20 pm	<b>Bruce Gelb, MD:</b> Human inducible pluripotent stem cells for the study of heart defects
3:20-3:40 pm	<b>Max Itkin, MD:</b> Imaging of the central lymphatic system in patients with Noonan syndrome using Dynamic Contrast Enhanced MR Lymphangiography
3:40-4:00 pm	<b>GASTROINTESTINAL SYSTEM</b> <b>Cheng Sun, PhD (Kontaridis lab):</b> Penn MDBR 2014 Grantee - Using human inducible pluripotent stem cells to delineate the cause of gastrointestinal abnormalities in RASopathy disorders
4:00-4:10 pm	<i>Discussion</i>
4:10-4:20 pm	<i>Break</i>

**5<sup>th</sup> International RASopathies Symposium: When Development and Cancer Intersect** Agenda, continued

<b>4:20-5:50 pm</b>	<b>Session 4: Developmental Perspective: Modeling RASopathies in Animals and in Silico</b> Moderator: <b>Suma Shankar, MD, PhD</b> Goal: Discuss nonhuman model experimental systems and how they are used to study RASopathies
4:20-4:40 pm	<b>Stanislav Shvartsman, PhD:</b> Quantitative biology of RASopathies
4:40-5:00 pm	<b>Ethan Perlstein, PhD (Perlara):</b> Rare disease drug discovery using whole animal disease models
5:00-5:20 pm	<b>Edward Stites, MD, PhD:</b> Computational analysis of pathological Ras mutants
5:20-5:40 pm	<b>Annette Schenck, PhD:</b> Habituation learning in <i>Drosophila</i> - a high-throughput platform to identify drugs that ameliorate cognitive and behavioral problems in Rasopathies
5:40-5:50 pm	<i>Discussion</i>
5:50-6:30 pm	<i>Break</i>
<b>6:30-8:30 pm</b>	<b>Networking Dinner with Cash Bar</b>

**DAY 3 – SUNDAY 7/30/17**

<b>7:00-8:00 am</b>	<b>Breakfast Meeting: How Do You Define a RASopathy?</b>
<b>8:00-9:30 am</b>	<b>Session 5: Ras Pathway Mechanics</b> Moderator: <b>Martin McMahon, PhD</b> Goal: To discuss molecular mechanisms of Ras pathway signaling: How do crystal structures and dynamic imaging shed new light?
8:00-8:20 am	<b>Jay Groves, PhD:</b> Membrane signaling dynamics
8:20-8:40 am	<b>John Albeck, PhD:</b> Quantifying the cellular effects of Ras pathway mutations with live-cell imaging
8:40-9:00 am	<b>Deborah Morrison, PhD:</b> Divide and Conquer: Targeting Raf Regulatory Interactions
9:00-9:20 am	<b>Marc Therrien, PhD:</b> Allosteric control of RAF activation by dimerization
9:20-9:30 am	<i>Discussion/Break</i>
<b>9:30-11:00 am</b>	<b>Session 6: Potential Therapeutics</b> Moderator: <b>David Stevenson, MD</b> Goal: Discuss promises and challenges of Ras pathway therapeutic drug development
9:30-9:50 am	<b>Brage Andresen, PhD, FRCPATH:</b> Splice switching oligonucleotides (SSOs) for HRAS exon 2 skipping
9:50-10:10 am	<b>Novartis:</b> Shp2 inhibitor
10:10-10:30 am	<b>Christopher Gibson, MD, PhD (Recursion Pharmaceuticals):</b> Image-based high throughput screens for rare disease therapeutics
10:30-10:50 am	<b>Philip Stork, MD:</b> Penn MDRB 2016 Grantee - What can Ras-dependent cancers teach us about Rasopathies?
10:50-11:00 am	<i>Discussion/ Break</i>
<b>11:00-12:30 am</b>	<b>Session 7: Next Generation: Junior Investigator Poster Session Abstracts and Closing Keynote</b> Moderator: <b>Bronwyn Kerr, MBBS</b> Goal: Attract and highlight research on RASopathies from junior investigators
11:00-11:30 am	<b>Junior Investigator Poster Finalists:</b> Three 10-minute presentations, winner announced
11:30-12:20 pm	<b>KEYNOTE: Frank McCormick, PhD, FRS:</b> Targeting Ras and NF1-related malignancies
12:20-12:30 pm	<i>Discussion and Next Steps</i> <b>Frank McCormick, PhD, FRS, Katherine A. Rauen, MD, PhD, Lisa Schoyer, MFA</b>
<b>12:30-1:30 pm</b>	<b>Breakout Sessions</b> <b>For RASopathy Family Groups:</b> CFC: Katherine A. Rauen, MD, PhD CS: Karen Gripp, MD, David Stevenson, MD, and Bronwyn Kerr, MBBS NS: Bruce Gelb, MD, and Amy Roberts, MD  NIH Q&A: William C. Timmer, PhD, NCI Program Director
<b>12:30-6:00 pm</b>	<b>NF Network's Post-Symposium Family Meeting</b>
<b>2:30-6:00 pm</b>	<b>Post-Symposium Events</b> for CFC and NS families