# 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	Dessert and Scientific Poster Session (Scientists and advocacy/family groups)	
8.00-10.00 pm	Goal: Encourage collaboration between researchers and families in a nonclinical setting	

## **DAY 2 - SATURDAY 7/29/17**

Session 1: What Defines a RASopathy? Moderator: Martin Zenker, MD 8:00-10:00 am 8:00-8:30 am 8:00-8:30 am 8:00-8:30 am 8:00-9:30 am 9:30-10:00 am 9:30-10:00 am 10:00-10:10 am 8:00-10:00 am 10:10-10:50 am 11:10-11:30 am 11:30-11:50 am 11:30-11:	7:00-8:00 am	Breakfast
Session 1: What Defines a RASopathy? Moderator; Martin Zenker, MD 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 Foun Individuals/Caregivers: Perspectives from the Home Front 8:30-9:00 am Frank McCormick, PhD, FRS: Molecular primer on the Ras signaling pathway 9:00-9:30 am Katherine A. Rauen, MD, PhD: The RASopathies 9:30-10:00 am Sizeussion (Clinical Diagnosis, Phenotype-Centered vs. Molecularly Defined) 10:00-10:10 am Break  Session 2: Syndromic and Sporadic Cancers of the Ras Pathway Moderator; Karen Gripp, MD Goal: Examine cancer types and mechanisms in individuals with RASopathies 10:10-10:50 am KEYNOTE: Nancy Ratner, PhD: Preclinical studies to guide NF1 clinical trials 10:50-11:10 am Hélène Cavé, PharmD PhD: Myoproliferative neoplasms (JMML) in RASopathies 11:30-11:50 am -12:00 pm Discussion 11:00-1:00 pm Lunch and Learn: Rick Guidotti: Positive Exposure  Session 3: Human Development: Effects on Organ Systems Moderator: Bruce Korf, MD, PhD Goal: Learn how RASopathy mutations affect developing tissues, organs, and body systems 1:00-1:40 pm KEYNOTE: Marco Tartaglia, PhD: RASopathy genetics, new mutations, congenital defects 1:40-2:40 pm Discussion 1:40-2:40 pm Discussion 2:20-2:20 pm Erik Ullian, PhD: How do astrocytes affect brain function in RASopathies: new investigative techniques and treatments 2:20-2:40 pm Discussion 3:00-3:40 pm Discussion 3:00-3:40 pm CIRCULATORY SYSTEMS  Bruce Gells, MD: Human inducible pluripotent stem cells for the study of heart defects Max Ikkin, MD: Imagling of the central lymphatic system in patients with Noonan syndrome using Dynamic Contrast Enhanced MR Lymphangiography CASTROINTESTINAL SYSTEM Cheng Sun, PhD (Kontaridis lab): Penn MDBR 2014 Grantee - Using human inducible pluripotent stem cells to delineate the cause of gastrointestinal abnormalities in RASopathy disorders	9:00 am-5:00 pm	Family Photo Shoot in and around meeting areas - Rick Guidotti
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Key: Family Sessions 042017

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

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

### **DAY 3 - SUNDAY 7/30/17**

DAY 3 – SUNDAY 7	•
7:00-8:00 am	Breakfast Meeting: How Do You Define a RASopathy?
8:00-9:30 am	Session 5: Ras Pathway Mechanics  Moderator: Martin McMahon, PhD  Goal: To discuss molecular mechanisms of Ras pathway signaling: How do crystal structures and dynamic imaging shed new light?
8:00-8:20 am	Jay Groves, PhD: Membrane signaling dynamics
8:20-8:40 am	John Albeck, PhD: Quantifying the cellular effects of Ras pathway mutations with live-cell imaging
8:40-9:00 am	Deborah Morrison, PhD: Divide and Conquer: Targeting Raf Regulatory Interactions
9:00-9:20 am	Marc Therrien, PhD: Allosteric control of RAF activation by dimerization
9:20-9:30 am	Discussion/ <mark>Break</mark>
9:30-11:00 am	Session 6: Potential Therapeutics  Moderator: David Stevenson, MD  Goal: Discuss promises and challenges of Ras pathway therapeutic drug development
9:30-9:50 am	Brage Andresen, PhD, FRCPath: Splice switching oligonucleotides (SSOs) for HRAS exon 2 skipping
9:50-10:10 am	Pascal Fortin, PhD (Relay Therapeutics): Allosteric inhibition of the SHP2 phosphatase: suppressing RAS-ERK signalling in RTK-driven cancers
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 MDBR 2016 Grantee - What can Ras-dependent cancers teach us about Rasopathies?
10:50-11:00 am	Discussion/ Break
11:00-12:30 am	Session 7: Next Generation: Junior Investigator Poster Session Abstracts and Closing Keynote  Moderator: Bronwyn Kerr, MBBS  Goal: Attract and highlight research on RASopathies from junior investigators
11:00-11:30 am	Junior Investigator Poster Finalists: Three 10-minute presentations, winner announced
11:30-12:20 pm	KEYNOTE: Frank McCormick, PhD, FRS: Targeting Ras and NF1-related malignancies
12:20-12:30 pm	Discussion and Next Steps Frank McCormick, PhD, FRS, Katherine A. Rauen, MD, PhD, Lisa Schoyer, MFA
12:30-1:30 pm	Breakout Sessions with RASopathy Family Groups: 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
12:30-6:00 pm	NF Network's Post-Symposium Family Meeting
2:30-6:00 pm	Post-Symposium Events for CFC and NS families

Key: Family Sessions 042017