

# Third International Meeting on Genetic Syndromes of the Ras/MAPK Pathway: Towards a Therapeutic Approach

August 2-4, 2013  
Renaissance Orlando at SeaWorld

## AGENDA

FRIDAY ~ August 2, 2013		
20:00 – 22:00	Dessert and Poster Session For Symposium Attendees and Advocacy/Family Support Groups •• <b>ATRIUM C/D</b>	
SATURDAY ~ August 3, 2013		
7:00 – 8:15	Breakfast •• <b>OCEANS 5</b>	
Meeting •• <b>OCEANS 6</b>		
8:15 – 8:30	Welcoming Comments and Introduction	<a href="#">Bruce Korf</a> University of Alabama, Birmingham <a href="#">Alcino Silva</a> University of California, Los Angeles
8:30 – 9:00	Keynote Presentation: TRND Program at NIH and possibilities for drug discovery in the Ras/MAPK disorders	<a href="#">Sittampalam Gurusingham</a> , NCATS
9:00 – 9:30	Advocates' Panel	<a href="#">Advocates</a> Representing CFC, Costello, Neurofibromatosis type I and Noonan syndromes
9:30 – 10:50	Ras/MAPK Pathway Phenotypes and Therapeutic Goals Moderators: <a href="#">Karen Gripp</a> , DuPont Hospital for Children and <a href="#">Judith Allanson</a> , Children's Hospital, Eastern Ontario	
	Therapeutic endpoints in NFI	<a href="#">Scott Plotkin</a> Massachusetts General Hospital
	Cardio-facio-cutaneous syndrome	<a href="#">Emma Burkitt Wright</a> University of Manchester
	Costello syndrome	<a href="#">Bronwyn Kerr</a> University of Manchester
	Noonan syndrome	<a href="#">Amy Roberts</a> Children's Hospital, Boston
10:50 – 11:10	Break	
11:10 – 12:30	Genomic Approaches Moderator: <a href="#">Ludwine Messiaen</a> , University of Alabama, Birmingham	
	Molecular analysis of NFI	<a href="#">Ludwine Messiaen</a> University of Alabama, Birmingham
	Structural and functional characterization of RAS mutants	<a href="#">Reza Ahmadian</a> Heinrich-Heine University
	Use of antisense morpholino oligomers as a gene-therapeutic approach to restore normal splicing	<a href="#">Conxi Lazaro</a> IMPPC Barcelona
	Ras pathway gene mutations in human genetic disorders	<a href="#">Yoko Aoki</a> Tohoku University
12:30 – 13:35	Lunch	
13:35 – 14:20	Presentation of Submitted Abstracts by Young Investigators Moderators: <a href="#">Bruce Korf</a> , University of Alabama, Birmingham and <a href="#">Alcino Silva</a> , University of California, Los Angeles	

**SATURDAY ~ August 3, 2013 - Continued**

14:20 – 15:40	Ras Pathway Biology and Identification of Therapeutic Targets Moderators: <a href="#">Ype Elgersma</a> , Erasmus University and <a href="#">Benjamin Neel</a> , Ontario Cancer Institute	
	Mechanism and treatment for the cognitive deficits associated with animal models of Noonan syndrome	<a href="#">Alcino Silva</a> University of California, Los Angeles
	Modeling RASopathies with hiPSC	<a href="#">Sonia Mulero-Navarro</a> Mt. Sinai School of Medicine
	Learning difficulties in the RASopathies: Pathogenesis and treatment	<a href="#">Ype Elgersma</a> Erasmus University
15:40 – 16:00	Break	
16:00 – 17:20	Preclinical Drug Development and Testing Moderator: <a href="#">Bruce Gelb</a> , Mt. Sinai School of Medicine	
	Therapeutic approaches to HCM in Noonan syndrome with multiple lentiginos	<a href="#">Maria Kontaridis</a> Harvard Medical School
	Mouse models of Noonan syndrome and other RASopathies	<a href="#">Benjamin Neel</a> Ontario Inst. Can. Res.
	Drug screening for Noonan syndrome with Drosophila	<a href="#">Bruce Gelb</a> Mt. Sinai School of Medicine
18:00	<b>Dinner</b> for Registrants who signed up for: <ul style="list-style-type: none"> <li>• CFC International family forum dinner •• <a href="#">Crystal Ballroom D, E</a></li> <li>• Noonan Syndrome Foundation family forum dinner •• <a href="#">Crystal Ballroom A, B, C</a></li> </ul>	
18:30	<b>Dinner</b> for Registrants who signed up for Costello syndrome family forum dinner •• <a href="#">Discovery Ballroom</a>	

**SUNDAY ~ August 4, 2013**

7:00 – 8:00	Breakfast •• <a href="#">Crystal B</a>	
	Meeting •• <a href="#">Discovery Ballroom</a>	
8:00 – 9:20	Clinical Trials Moderator: <a href="#">Katherine Rauen</a> , University of California, San Francisco	
	NF Clinical Trials Consortium	<a href="#">Bruce Korf</a> University of Alabama, Birmingham
	Clinical trials for Fragile X syndrome	<a href="#">Reymundo Lozano</a> University of California, Davis
	Use of Raf inhibitors for treatment of melanoma	<a href="#">Chao Zhang</a> Plexxicon
	The NS clinical trial	<a href="#">Calum MacRae</a> Brigham and Women's Hospital
9:20 – 10:20	Infrastructure to Support Therapeutic Development Moderator: <a href="#">Amy Roberts</a> , Children's Hospital, Boston	
	The CTF therapeutic development pipeline	<a href="#">Annette Bakker</a> Children's Tumor Foundation
	Tissue banking in pediatric oncology	<a href="#">Corrine Linardic</a> Duke University School of Medicine
	Patient-driven disease registries	<a href="#">Pat Furlong</a> Parent Project Muscular Dystrophy
10:20 – 10:35	Break	

**SUNDAY ~ August 4, 2013 - Continued**

10:35 – 12:15	Break-out Sessions with RASopathies Families	
	CFC syndrome families •• <b>Atlantis A</b>	<a href="#">Rebecca Josowitz</a> Mt. Sinai School of Medicine <a href="#">Katherine Rauen</a> University of California, San Francisco
	Costello syndrome families •• <b>Atlantis B</b>	<a href="#">Karen Gripp</a> DuPont Hospital for Children <a href="#">Bronwyn Kerr</a> University of Manchester <a href="#">Angela Lin</a> Harvard Medical School
	Neurofibromatosis type I families •• <b>Odyssey A</b>	<a href="#">Bruce Korf</a> University of Alabama, Birmingham <a href="#">David Viskochil</a> University of Utah
	Noonan syndrome families •• <b>Odyssey B</b>	<a href="#">Judith Allanson</a> Children’s Hospital, Eastern Ontario <a href="#">Bruce Gelb</a> Mt. Sinai School of Medicine <a href="#">Amy Roberts</a> Children’s Hospital, Boston <a href="#">Alcino Silva</a> University of California, Los Angeles <a href="#">Md. Abdur Razaque</a> Cincinnati Children’s Hospital
12:15 – 12:30	Final Comments and Discussion •• <b>Discovery Ballroom</b>	<a href="#">Bruce Korf</a> University of Alabama, Birmingham <a href="#">Alcino Silva</a> University of California, Los Angeles <a href="#">Lisa Schoyer</a> RASopathies Network USA
1:15-4:00	NF Network Break-out •• <b>Odyssey A</b>	<a href="#">Ludwine Messiaen</a> University of Alabama, Birmingham <a href="#">Alcino Silva</a> University of California, Los Angeles <a href="#">David Viskochil</a> University of Utah