Research at the Costello Conference – Friday, July 28, 2017

	Research Title	Researchers/Assistants	Goals	Ages	Participation Activity	Records to Bring
1.	Natural History of Costello Syndrome: Adaptive Skills, Cognitive and Behavioral Characteristics	Dr. Marni Axelrad Dr. Jennifer Katzenstein Dr. Jillian Wise, Asst Dr. Ashley Cooper, Asst	1. Autism evaluation 2. Quality of Life evaluation 3. Develop profiles of individuals with less common mutations. 4. Add new participants	16 months to adult Parents/caregivers also participate.	Questionnaires and specific testing. Depending on age, takes 1-2 hours. Testing on Thursday, Friday and Saturday.	•Copy of the HRAS/genetic test results
2.	Clinical and Molecular Investigations	Dr. Karen Gripp Dr. Kathryn Chatfield Dr. Daniel Doyle Dr. Mihir Thacker Dr. Nicole Weaver Laura Baker, Asst Sarah Donoghue, Asst	To learn more about how Costello syndrome affects persons who have it and learn more about the genetic cause of Costello syndrome. The researchers will talk to the family about the medical history and ask them to complete a questionnaire. They may ask for a cheek swab. You will see each of the doctors separately. They may request medical records.	All ages, except Dr. Weaver will only see individuals with CS who are 16 years of age or older (Note: Dr. Weaver is also helping with research by Dr. Prada.)	Consent, questionnaire, past medical history and may ask to obtain past imaging studies. You will talk briefly with each doctor and they may do a short exam. May do a cheek swab DNA with Dr. Gripp. It will take about 15 minutes with each doctor.	•Copy of the HRAS/genetic test results •CD with brain and spine MR imaging studies •If performed, CD with xrays of spine, hips and other bone x-rays •If performed, copies of results of growth hormone stimulation testing; hormone testing
3a	Developing 3D Craniofacial Morphometry Data and Tools to Transform Dysmorphology	Dr. Ophir Klein (not attending) Nick Mahasuwan, Assistant Clinical Research Coordinator	To better characterize Costello syndrome including facial features, and eventually develop a system to help physicians diagnose these disorders earlier and more accurately. They will gather information and take about 12-15 photos.	3 years old and up	They will explain the study and complete the consent form/ HIPPA release. Fix the hair and take photo. Takes about 15 minutes.	•Copy of the HRAS/genetic test results
3b	Molecular Genetic Investigation of Patients with Congenital Anomalies	Dr. Ophir Klein (not attending) Dr. Amnon Sharir	To learn the structural development of Costello syndrome. They will gather information and copies of CT scans.	All ages	Receive the scans you bring or complete form so they can request them. Should take about 10 minutes.	•Copy of the HRAS/genetic test results •A CD with copy of CT craniofacial (head) scans, if available to give them

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4.	Coping Mechanisms in Rasopathies	Dr. David Stevenson	To study the coping mechanisms and stress of caregivers of individuals diagnosed with Costello syndrome; to study the affect support groups/conferences have on caregivers' coping abilities and stress.	Caregivers (adults) will participate in this research by completing questionnaires.	Questionnaires will be emailed to caregivers 1-3 times over a 1-year period to complete. The may complete the first one and talk to the researcher at the conference. Should take 15-20 minutes.	•Copy of the HRAS/genetic test results **Please note that your name, child's name and email address will be shared with this research team & they may email you the first & subsequent questionnaires.
5.	A Study of Pain in Rasopathies	Dr. Carlos Prada Dr. Nicole Weaver	To evaluate the pain experienced by people with Rasopathies, including Costello syndrome.	Ages 8-40 – will complete a survey/ questionnaire regarding pain.	Complete a questionnaire. Takes about 5 minutes.	•Copy of the HRAS/genetic test results
6a	Genetic Basis of Melanocytic Nevi	Dr. Maija Kiuru Jennifer Urban, Asst	To characterize the clinical features and identify the genetic mutations that drive melanocytic nevi (moles) in Rasopathies. Research includes photography of moles, skin biopsy of mole(s), saliva collection and a questionnaire.	5 years and older	Patients may choose to participate in photography of moles, skin biopsy of moles, saliva collection and a questionnaire. 15-45 minutes.	•Copy of the HRAS/genetic test results •Pathology reports of prior skin biopsies
6b	Characterization of Hair in Rasopathies	Dr. Maija Kiuru Jennifer Urban, Asst	To characterize the clinical and microscopic features of hair changes in Rasopathies. Research includes photographs, a questionnaire, and hair sampling (pulling 5-10 scalp hairs).	5 years and older	Questionnaire, photography of hair, hair sample. About 15 minutes.	•Copy of the HRAS/genetic test results

Meet the Experts

Dr. Katherine Rauen, Clinical Geneticist
Dr. Dawn Siegel, Dermatologist
Dr. Suma Shankar, Ophthalmologist
Dr. Bronwyn Kerr, Clinical Geneticist
with Dr. Emma Burkitt Wright and Dr. Corinne Linardic