Costello Syndrome
FAMILY NETWORK
9th International Family Forum

Seattle, Washington USA July 14-19, 2015
A letter from the President and Forum Coordinator,

Each conference brings an exciting and informative agenda, and the opportunity to learn the latest on Costello syndrome. We come to learn and also to be inspired and supported by the friendships and bonds we make. That gives us hope supported by knowledge, community, awareness and research – all of which are goals of the Costello Syndrome Family Network.

The Costello Syndrome Family Forum is the largest gathering of CS families, individuals, researchers, medical experts and resource professionals of its kind. You will have direct access to numerous networking and social opportunities, as well as the opportunity to attend about 20 learning sessions highlighting almost every medical issue in adults and children, the latest in research, and everyday life issues. You will also have the unique opportunity to participate in research that could impact your child’s life or another child’s life in the future. Perhaps most importantly, you will have face-to-face, personal interaction with other families who are dealing with the very same issues that you face every day, including families who have traveled this road before you and can share their experiences and expertise. I think this is an essential part of your journey.

CSFN has always worked hard to make attending the conference affordable for families. Registration fees are kept low and include all lectures, social events, and meals. A professional childcare company will be caring for your children so you can give your full attention to learning. Two teachers will be hosting the CS Lounge for our adults with CS and special activities are planned. You may attend each medical lecture this year instead of having to choose, and Friday will be a fast-paced day – hang onto your hats! You will be given a binder this year and handouts will be available at most talks for taking notes and insertion into your binder. Saturday will be an interesting mix of talks. Wow! We have come a long way from the first conference in Birmingham, Alabama in 1999 due to the dedication of many. It all began because they had hope and vision.

CSFN is working on a plan for our vision of growth in support of our families. I get excited about the possibilities. You can be part of that excitement too by volunteering a little of your time and expertise. Everyone has something to share, and now is the time to step up. You are needed. We can do so much more together, especially when we share hope and vision.

Welcome to Seattle!

Sandra Taylor
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DONATIONS

A special thank you!

The Family Forum is substantially dependent on fundraising by its families as well as the generous support of Sponsors and Contributors. A special thanks to each of the following families, individuals and companies who have generously donated and fundraised to make the Forum possible.

OUR FUNDRAISING FAMILIES

$10,000 LEVEL
In Honor of Allie Bahnsen
In Honor of Braden Hall
In Honor of Westin Thomas

$7,500 LEVEL
In Honor of Micaela Garcia
In Honor of Gabriella Romero-Vysocky

$5,000 LEVEL
In Honor of Jaylon Flores
In Honor of Ali Kazakoff
In Honor of Brooklyn Polk
In Honor of Luke Stevenson

$2,500 LEVEL
In Honor of Juliet DiMaria
In Honor of Nicholas Eaton
In Honor of Gabriel Kielman
In Honor of Jill Taylor

$1,000 LEVEL
In Honor of Christopher Coates
In Honor of Logan Keiner
In Honor of Asher Pascal
In Honor of Anna-Stella Roberts
In Honor of Pete Scampavia

$500 LEVEL
In Honor of Maximiliano Coca
In Honor of Collin Deckman
In Memory of Bret Gerhardt
In Honor of Erin Hefner
In Memory of Quin Johnson
In Honor of Zane Madaus
In Honor of Joey Tuozzo

thank you!

OTHER SUPPORTERS

In Honor of Shelby Adams
In Honor of Hunter Arvel
In Honor of Makenzie Belanger
In Honor of Arden Benfield
In Honor of Nicole Chamness
In Honor of Jonah Charrabe
In Honor of Fredrick DeNicola
In Honor of Ryan Ellis

In Honor of Maggie Ernst
In Memory of Willa Hunt
In Honor of Hillevi Jagrud
In Honor of Victor Krause
In Honor of Kiley Lanik
In Memory of Robert Mack
In Honor of Dominic Purdy
In Honor of Emily Ray

In Honor of Danielle Slawitschek
In Honor of Rebecca Spooner
In Honor of the Ramos Family
In Honor of Jalisa Sullivan
In Honor of Megan Villarreal
In Honor of Keira Waugh
SPONSORS

Diamond Partner - $6,000...............Saturday Night Dinner/Dance and DJ
Friends of Freescale Semiconductor, Inc.
Austin, Texas
In Honor of Westin Thomas

Silver Partner - $2,500..........................Audiovisual Equipment
Parkview Pediatric Dentistry
Dustin Janssen, DDS
Lubbock, Texas
In Honor of Brooklyn Polk

Garnet Partner - $1,200 ......................Welcome Reception
KCB Painting and Contracting
Quincy, MA
In Honor of Nicholas Eaton

Amethyst Partner - $1,000 .................Cooler Bags for Families
Jason Pascal
New York, NY
In Honor of his son, Asher Pascal

Emerald Partner - $600 .......................Update and Reprint Trifold Brochure
Reliant South Construction Group
Panama City, FL
In Honor of Jill Taylor

Ruby Partner - $450 ..........................Name Badge Wallets
Taylor Architects, Inc.
Panama City, FL
In Honor of Jill Taylor

Peridot Sponsor - $250 ........................Family Cooler Snacks & Drinks
Gaco Western Wall Foam
OR, WA, ID, MT, AK, HI, CA, British Columbia, Alberta, Saskatchewan
In Honor of Shelby Adams

Angel Sponsors

Goodie bag valued at $125 for silent auction in support of all the families.
Assistance with transportation to and from the Family Forum
# FORUM SCHEDULE at a glance

## TUESDAY, JULY 14, 2015
- **2:00pm - 5:00pm**  Consent Forms w/Dr. Gripp by Appt Only  
  Cascade 1-2, 2nd Floor
- **4:30pm - 5:30pm**  New Family Orientation for First Time Attendees  
  Maxi’s Salon, Tower
- **7:00pm - 9:00pm**  Registration & Welcome Reception  
  Grand Ballroom 1, Main Floor

## WEDNESDAY, JULY 15, 2015
- **8:00am - 6:00pm**  Meet the Researchers and Experts  
  Cascade Rooms 1-11, 2nd Floor
- **7:00pm - 10:00 pm**  CSFN, ICSSG, PAC meeting (Board members only)  
  Suite 1370, Tower

## THURSDAY, JULY 16, 2015
- **8:40am**  Group Photo wearing Forum T-shirt  
  Grand Ballroom 1, Main Floor
- **9:00am - 9:45am**  Research by Questionnaire, Ages 1-18  
  Maxi’s, Tower
- **9:45am - 11:00am**  Craft Fair  
  Maxi’s, Tower
- **11:00am - 11:45am**  Town Hall Meeting  
  Maxi’s, Tower
- **6:45pm - 9:00pm**  Guys’ Man Cave  
  Maxi’s Salon, Tower
- **9:15pm - 11:30pm**  Ladies’ Mix & Mingle  
  Maxi’s Salon, Tower

## FRIDAY, JULY 17, 2015
- **8:25am - 5:15pm**  Medical Lectures & Ask the Experts Panel  
  Evergreen 1-2, 2nd Floor
- **8:00pm - 10:00pm**  Symposium Poster Session & Desserts  
  Northwest 1-2, Main Floor

## SATURDAY, JULY 18, 2015
**Rick Guidotti will be taking photos of our children and adults in their activities throughout the day.**
- **9:00am - 10:00am**  Special Session: The Unstoppable Charity  
  Evergreen 1, 2nd Floor
- **10:15am - 3:15 pm**  Workshops CS & CFC  
  Evergreen 1-4, 2nd Floor
- **6:00pm**  Group Photo by the Pool. If Rainy, gather by Grand Ballroom 1.
- **6:30pm - 10:30pm**  Family Banquet – Raffle & Quilt – DJ & Dancing  
  Grand Ballroom 1, Main Floor

## SUNDAY, JULY 19, 2015
- **9:00am - 10:00 am**  Positive Exposure presentation by Rick Guidotti  
  Northwest Ballroom 1, Main Floor
- **10:15am - 11:15am**  CSFN Board Meeting  
  Cascade Rooms 1-2, 2nd Floor
- **11:30am - 12:30pm**  Symposium Wrap-Up  
  Evergreen 1, 2nd Floor
### TUESDAY: welcome reception & registration

<table>
<thead>
<tr>
<th>TIME</th>
<th>TOPIC/ACTIVITY</th>
<th>LOCATION</th>
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</table>
| 1:00pm – 5:00pm | **Information/Welcome Desk Open**  
- Get directions or information.  
- Drop off & sign in your items for the Thursday craft fair or Saturday raffle.  
- Finish up any last minute paperwork.  
- Pick up pre-ordered t-shirts.  
- Sign the Rick Guidotti photo release form.  
- Sign up for Thursday Morning Research Questionnaire. | Grand Foyer,  
Main Level  |
| 2:00pm – 5:00pm | **Consent Forms w/Dr. Gripp by Appt Only**  
Dr. Gripp is getting a jump start on Wednesday by meeting by appointment only with new families to review and sign the consent forms. This is only for families who have been contacted and a specific appointment time has been made. | Cascade 1-2,  
2nd Floor  |
| 4:30pm – 5:30pm | **New Family Orientation for First Time Attendees**  
This orientation is for families who have never attended a conference before.  
We will go over the conference agenda so you will have a better understanding of each day’s activities. You will meet other new families and board members. You will also have time to ask questions. | Maxi’s Salon,  
Tower  |
| 7:00pm – 9:00pm | **Registration & Welcome Reception**  
- Our grand welcome to everyone!  
  Refreshments are served and everyone gathers to meet new families and welcome back friends from past conferences.  
- Pick up the t-shirts you ordered!  
- Pick up your registration packets.  
- Pick up your coolers – one per family.  
- Introductions, Special Announcements, Recognition, Special Prizes & Drawings.  
- Sign the Rick Guidotti photo release form.  
- Drop your name in for the drawings.  
- Finally together again! YAY! | Grand Ballroom 1,  
Main Level  |

Immediately following the Reception, there will be a brief training session for the volunteers working on Wednesday at Meet the Researchers & Experts.
### WEDNESDAY: meet the researchers and experts

<table>
<thead>
<tr>
<th>TIME</th>
<th>TOPIC/ACTIVITY</th>
<th>LOCATION</th>
</tr>
</thead>
<tbody>
<tr>
<td>7:00am – 8:30am</td>
<td>Buffet Breakfast</td>
<td>Grand Ballroom 1, Ground Floor</td>
</tr>
<tr>
<td>8:00am – 6:00pm</td>
<td>Meet the Researchers and Experts <strong>Bring your Passport</strong></td>
<td>Cascade Rooms 1-11, 2nd Floor</td>
</tr>
<tr>
<td>7:00pm</td>
<td>CSFN, PAC, ICSSG Meeting (For Board/Committee Members)</td>
<td>Tower Suite 1370</td>
</tr>
</tbody>
</table>

**please note:**

Due to very limited space, please arrive only at the time assigned in your Passport. Use the elevators to the Tower and come to the 2nd floor. You will see the waiting area and registration desk when you step off the elevators. Sign in at the Registration Desk and you will receive directions. To complete as much research as possible and lessen the waiting time for others, it is important that you keep your appointments brief. There will not be time to review medical records with the Researchers during your appointment. You will have time to talk more with the Experts and ask questions. If we are all considerate, each family will be able to keep their appointments.

You should plan to be here for 4-5 hours, depending on how many researchers and experts you signed up to see. If you have morning appointments, be sure to go to breakfast before you come. If you have afternoon appointments, be sure to eat lunch before you come. You may also want to bring some snacks and drinks with you. The waiting area is small so bring as few people as possible with you.

*Meeting with researchers* means your family is agreeing to learn more about and provide consent to participate in the research studies of your choice. All studies have been approved by an Institutional Review Board (IRB) to ensure your family’s rights and responsibilities as research participants.

*Meeting with experts* means that they will answer questions in the context of what they know about Costello syndrome, not your child, because your child is not officially their patient.

Please see the BIOGRAPHIES section for more information on the Researchers and Experts.

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**STATION 1**

**TOPIC**

*Developing 3D Craniofacial Morphometry Data and Tools to Transform Dysmorphology*

Ophir Klein, MD, PhD (will not be attending)  
Elizabeth Beals and Nick Mahasuwan, Research Coordinators  
University of California, San Francisco, CA, USA

**Goals:** To better characterize genetic syndromes that include facial abnormalities, and eventually develop a system to help physicians diagnose these disorders earlier and more accurately.

**Description:** Brief consent process including collection of demographic information, followed by 3D picture taking (participant will need to sit still for 12-15 minutes). Desired age range is 3+ years.
**Topics and Locations**

<table>
<thead>
<tr>
<th>Topic</th>
<th>Location</th>
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</thead>
<tbody>
<tr>
<td><strong>Costello Syndrome: Clinical and Molecular Investigations</strong></td>
<td>Cascade 2-6</td>
</tr>
<tr>
<td>Dr. Karen Gripp, Lead Researcher - assisted by Laura Baker</td>
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<tr>
<td>AI du Pont Hospital for Children, Wilmington, DE USA</td>
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<tr>
<td>Dr. Dan Doyle, Endocrine and Growth</td>
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<tr>
<td>AI du Pont Hospital for Children, Wilmington, DE USA</td>
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<tr>
<td>Dr. Mihir Thacker, Bone and Joint - assisted by Christina Adler</td>
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<tr>
<td>AI du Pont Hospital for Children, Wilmington, DE USA</td>
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<tr>
<td>Dr. Kathryn Chatfield, Cardiology</td>
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<tr>
<td>University of Colorado School of Medicine, Aurora, Colorado USA</td>
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<tr>
<td>Dr. Nicole Weaver, seeing age 16 and older</td>
<td></td>
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<tr>
<td>Cincinnati Children's Hospital Medical Center, Cincinnati, OH USA</td>
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</table>

**Goals:** To understand how Costello syndrome and related disorders affect the individual. To evaluate the relationship between the specific gene change and the physical, cognitive and social implications.

**Description:**

- Dr. Gripp will talk to the family about the individual’s medical history and ask them to fill out a medical questionnaire. They may ask for spit samples from the individual and parents to analyze for genetic change.
- Dr. Doyle will measure height, weight and head circumference, and ask questions about pubertal development and other hormone related issues.
- Dr. Thacker will ask about bone problems and assess the movement of the patient, and the straightness of the back. He may ask to see copies of X-rays previously done.
- Dr. Weaver will see the older individuals and ask about their medical health and daily living.

**Goals:** Costello patients are at risk to develop numerous types of malignancies due to RAS mutation. We aim to better understand the mechanisms involved in RAS-mediated cancers by examining why cells in epidermal nevi which have the same mutation as we see in some people with Costello syndrome are more proliferative than cells in Costello skin.

**Description:** Each subject will provide two 3mm skin biopsies, 1 saliva sample, and photographs.

Skin biopsies: For pediatric patients and any others who request it, we will first numb the skin with a 1-hour application of EMLA under tegaderm. Subsequently and for all who elect not to do EMLA treatment, we will numb the skin with an injection of lidocaine with epinephrine. After confirming the skin is numb, we will perform the biopsy with a punch tool, and we will place a pressure dressing with gelfoam to prevent bleeding. Subjects will be given supplies to care for sites for 7-10 days. We do not anticipate any pain from the procedure, though a small white scar will result and subjects will need to perform wound care.

Saliva samples: participants will be asked to spit into an oragene collection kit.

Photographs: Four sets of pictures will be taken of the subject’s skin – of the hair, torso, leg and underarm.
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<tr>
<th>STATION</th>
<th>TOPIC</th>
<th>LOCATION</th>
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<tbody>
<tr>
<td>STATION 8</td>
<td>Meet the Experts</td>
<td>Cascade 8</td>
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<tr>
<td></td>
<td>Dr. Katherine Rauen, Clinical Geneticist; and Liga Bivina, Genetic Counselor</td>
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<td></td>
<td>UC Davis MIND Institute, Sacramento, CA USA</td>
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<tr>
<td></td>
<td>Expert on Costello Syndrome</td>
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<tr>
<td>STATION 9</td>
<td>Meet the Experts</td>
<td>Cascade 9</td>
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<tr>
<td></td>
<td>Dr. Dawn Siegel, Dermatologist</td>
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<tr>
<td></td>
<td>Medical College of Wisconsin, Milwaukee, WI USA</td>
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<td></td>
<td>Expert on Costello Syndrome</td>
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<tr>
<td>STATION 10</td>
<td>Meet the Experts</td>
<td>Cascade 10</td>
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<tr>
<td></td>
<td>Dr. Bronwyn Kerr and Dr. Emma Burkitt-Wright, Clinical Geneticists</td>
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<td></td>
<td>Manchester Centre for Genomic Medicine, Manchester, UK</td>
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<td>Expert on Costello Syndrome</td>
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<tr>
<td>STATION 11</td>
<td>Meet the Experts</td>
<td>Cascade 11A</td>
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<tr>
<td></td>
<td>Dr. Suma Shankar, Ophthalmologist</td>
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<td></td>
<td>Emory University, Atlanta, GA USA</td>
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<td></td>
<td>Expert on Costello Syndrome</td>
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<tr>
<td>STATION 12</td>
<td>Meet the Experts</td>
<td>Cascade 11B</td>
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<td></td>
<td>Dr. David Stevenson, Medical Geneticist</td>
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<tr>
<td></td>
<td>Stanford University, Stanford, CA USA</td>
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<tr>
<td></td>
<td>Expert on Costello Syndrome</td>
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notes:
**THURSDAY: family fun day!**

Today is T-shirt day! Wear your Seattle Family Forum T-shirt today. We will be taking group photos immediately following breakfast. Meet outside Grand Ballroom 1 at our Information Table at 8:40am.

<table>
<thead>
<tr>
<th>TIME</th>
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<tbody>
<tr>
<td>7:00am – 8:30am</td>
<td>Buffet Breakfast</td>
<td>Grand Ballroom 1, Ground Floor</td>
</tr>
<tr>
<td>8:40 am</td>
<td>Group photo wearing Family Forum T-Shirts</td>
<td>Grand Ballroom 1 Ground Floor</td>
</tr>
<tr>
<td>9:00am – 9:45am</td>
<td>“Functional Outcomes in Children with Costello Syndrome” Dr. Michael J. Goldberg, Lead Researcher; Viviana Bompadre, PhD</td>
<td>Maxi’s, Tower</td>
</tr>
<tr>
<td></td>
<td>Goals: Determine the functional health for children with Costello syndrome. In 2003 we published a paper based on functional questionnaires given to families during the first International Costello Conference. It is our goal to expand that study with a larger number of participants.</td>
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<tr>
<td></td>
<td>Description: Consent families all at one time, and ask them to fill out the Pediatric Outcome Data collection Instrument (PODCI). The PODCI is a functional and health related quality of life questionnaire. It takes about 10-15 minutes to fill out the questionnaire.</td>
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<tr>
<td></td>
<td>Desired age range of participants is 0-18.</td>
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<tr>
<td>9:45am – 11:00am</td>
<td>Craft Fair - Buy tickets to win the Dr. Seuss Quilt!</td>
<td>Maxi’s, Tower</td>
</tr>
<tr>
<td></td>
<td>Have fun shopping the unique items brought by our families from all over the world. All funds go to the conference expenses. Cash or credit card.</td>
<td></td>
</tr>
<tr>
<td>11:00am – 11:45pm</td>
<td>Town Hall Meeting</td>
<td>Maxi’s, Tower</td>
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<tr>
<td></td>
<td>Ever wonder how we pick a city for a conference? Or how the money is spent that you fundraise? Do you have some good ideas of what you would like to see at a conference? Want to know the right way to go about using a logo? Are you wondering what it takes to become a board member or volunteer? Come and share what’s on your mind, and we may have a few things to share with you too!</td>
<td></td>
</tr>
<tr>
<td>12:00pm – 1:30pm</td>
<td>Buffet Lunch - Dr. Seuss Surprises!</td>
<td>Grand Ballroom 1 Ground Floor</td>
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<tr>
<td></td>
<td><strong>FREE TIME</strong></td>
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<td></td>
<td>Pool anyone? Hang out with friends? Take a nap?</td>
<td></td>
</tr>
<tr>
<td>6:45pm – 9:00pm</td>
<td>Guys’ Man Cave (men only)</td>
<td>Maxi’s Salon, Tower</td>
</tr>
<tr>
<td>9:15pm – 11:30pm</td>
<td>Ladies’ Mix &amp; Mingle (ladies only)</td>
<td>Maxi’s Salon, Tower</td>
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<td></td>
<td>“Come relax and visit in our private lounge light snacks &amp; cash bar”</td>
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<tr>
<td>TIME</td>
<td>TOPIC/ACTIVITY</td>
<td>LOCATION</td>
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<tr>
<td>6:45am – 8:00am</td>
<td>BUFFET BREAKFAST</td>
<td>Grand Ballroom 1</td>
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<tr>
<td>8:00am</td>
<td>Drop off children at Childcare (2nd Floor)</td>
<td>Cascade 1-8 Tower, Suite 1370</td>
</tr>
<tr>
<td>8:20am – 8:25am</td>
<td>Welcome, Announcements</td>
<td>Evergreen 1 2nd Floor</td>
</tr>
<tr>
<td>8:25am – 8:55am</td>
<td>“Costello Syndrome - Understanding Why It Is the Way It Is”, Katherine Rauen, MD PhD</td>
<td>Evergreen 1 2nd Floor</td>
</tr>
<tr>
<td>8:55am – 9:25am</td>
<td>“Orthopedic Manifestations of Costello Syndrome” Mihir Thacker, MD</td>
<td>Evergreen 1 2nd Floor</td>
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<tr>
<td>9:25am – 9:35am</td>
<td>BREAK</td>
<td></td>
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<tr>
<td>9:35am – 10:05am</td>
<td>“The Eye and Vision in Costello Syndrome” Suma Shankar, MD</td>
<td>Evergreen 1 2nd Floor</td>
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<tr>
<td>10:05am – 10:35am</td>
<td>“Skin Care Guidelines for Costello Syndrome - Tips and Tools” Dawn Siegel, MD</td>
<td>Evergreen 1 2nd Floor</td>
</tr>
<tr>
<td>10:35am – 10:45am</td>
<td>BREAK</td>
<td></td>
</tr>
<tr>
<td>10:45am – 11:15am</td>
<td>“Cancer Risks in Costello Syndrome” Bronwyn Kerr, MD</td>
<td>Evergreen 1 2nd Floor</td>
</tr>
<tr>
<td>11:15am – 11:45am</td>
<td>“Costello Syndrome in Adulthood: What You Need to Know” Nicole Weaver, MD</td>
<td>Evergreen 1 2nd Floor</td>
</tr>
<tr>
<td>12:00pm – 1:00pm</td>
<td>Pick up children from Childcare &amp; CS Lounge by 12 noon Both are closed for lunch between 12 noon and 1:00pm</td>
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<tr>
<td>1:00pm – 1:15pm</td>
<td>Drop off children at Childcare &amp; CS Lounge</td>
<td></td>
</tr>
<tr>
<td>1:15pm – 1:35pm</td>
<td>“Finding Hope and Joy in Plan B” Larry, Sandra &amp; Jill Taylor</td>
<td>Evergreen 1 2nd Floor</td>
</tr>
<tr>
<td>1:40pm – 2:10pm</td>
<td>“Update on Clinical Research: Brain Abnormalities, Rare Mutations &amp; Ongoing Work” Karen W. Gripp, MD</td>
<td>Evergreen 1 2nd Floor</td>
</tr>
<tr>
<td>2:10pm – 2:40pm</td>
<td>“The Heart of Costello: Cardiovascular Features and Research Opportunities” Kathryn Chatfield, MD</td>
<td>Evergreen 1 2nd Floor</td>
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<tr>
<td>2:40pm – 2:55pm</td>
<td>BREAK</td>
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<tr>
<td>2:55pm – 3:25pm</td>
<td>“GH use in CS; Are the Benefits Greater than the Risks?” Dan Doyle, MD</td>
<td>Evergreen 1 2nd Floor</td>
</tr>
<tr>
<td>3:30pm – 4:00pm</td>
<td>“Vitamins, Herbs and Supplements: What Do We Know About Alternative Therapies in Costello Syndrome?” David Stevenson, MD; Chiara Leoni, MD</td>
<td>Evergreen 1 2nd Floor</td>
</tr>
<tr>
<td>4:05pm – 5:15pm</td>
<td>Best Practice – Ask the Doctors: Drs. Chatfield, Doyle, Gripp, Kerr, Leoni, Siegel, Stevenson, Weaver</td>
<td>Evergreen 1 2nd Floor</td>
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<tr>
<td>5:15pm – 5:30pm</td>
<td>Pick up children from Childcare &amp; CS Lounge by 5:30pm All close at 5:30pm</td>
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<tr>
<td>8:00pm – 10:00pm</td>
<td>Symposium Dessert Reception and Poster Session</td>
<td>Northwest 1-2, Main Floor</td>
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</table>
FRIDAY: lecture descriptions

8:25am – 8:55am
“Costello Syndrome – Understanding Why It Is the Way it Is”
Katherine Rauen, MD PhD

Costello syndrome is a RASopathy caused by genetic alterations in the HRAS gene. HRas, the protein product of the HRAS gene, is a critical regulator of how a cell communicates with its environment and with other cells. The function of HRas, which is part of a very large Ras Family of proteins, directs cells to grow and divide, to become unique [such as a skin cell or a heart cell or a brain cell] and may also directs cells to migrate. Because of all the critical functions that Ras can perform, it’s very important in the development of both animals and humans. And because it is so important, if Ras does not function properly, it may at times, increase an individual’s susceptibility to cancer.

8:55am – 9:25am
“Orthopedic Manifestations of Costello Syndrome”
Mihir Thacker, MD

Costello syndrome (CS) is a rare genetic condition caused by changes in HRAS. Individuals with CS may have several orthopedic issues which can impact walking and function. These abnormalities can occur in the upper as well as the lower extremities and also involve the spine. Abnormalities affecting the lower extremities can impact ambulation and may vary with age. Typical foot and ankle deformities as well as hip issues are some of the most common concerns in CS. Typical spine deformities are lordosis, kyphosis or scoliosis (spinal curvature). We will discuss some of the orthopedic manifestations and their effect on function. Typical clinical features, early identification and interventional strategies will be discussed. We will also present some of the results of our research and discuss potential new areas of additional research needed to further clarify the evolution of some of the deformities and hopefully improve function of individuals with CS.

9:35am – 10:05am
“The Eye and Vision in Costello Syndrome”
Suma Shankar, MD

The eye is one of the systems that is involved in Costello syndrome. Eye problems include easily correctable ocular problems such as refractory errors (near or far sightedness), amblyopia (lazy eyes) and strabismus (crossing or turning out of the eye). Other problems that may occur include hypoplastic (under-developed) optic nerve, and optic atrophy resulting as a complication of increased intracranial pressure. Recommendations include regular follow-up with a pediatric ophthalmologist, correction of refractive errors, amblyopia, correction of strabismus and surgery to correct elevated intracranial pressure. We will discuss multidisciplinary approach for eye problems in Costello syndrome.

10:05am – 10:35am
“Skin Care Guidelines for Costello Syndrome – Tips and Tools”
Dawn Siegel, MD

Dr. Siegel has an interest and research focus on the skin issues in Costello syndrome and cardio-facio-cutaneous syndrome. The paper on the skin issues CFC was published in 2010 and the Costello skin features paper was published in 2011 in the British Journal of Dermatology. Today Dr. Siegel will share some of findings from this study with you, as well as general skin care tips for children with Costello syndrome.

10:45am – 11:15am
“Cancer Risks in Costello Syndrome”
Bronwyn Kerr, MD

The HRAS gene was first discovered as a gene that is mutated in many human cancers. It is an important gene in cell proliferation and survival. The discovery that germ-line mutations cause CS was surprising, but does partially explain some of the manifestations of CS. The current literature regarding cancer risks in CS and the other Rasopathies will be reviewed and the resulting clinical approach discussed.

(Lecture Descriptions - continued on page 15)
11:15am – 11:45am
“Costello Syndrome in Adulthood: What You Need to Know”
Nicole Weaver, MD

While most published studies about Costello syndrome are skewed towards a younger patient population, in this talk we will review previously published medical literature on adults. We will focus on medical problems, quality of life, and potential areas for future research for our adult population with Costello syndrome.

1:15pm – 1:35pm
“Finding Hope and Joy in Plan B”
Larry, Sandra and Jill Taylor

While struggling to deal with all the medical, emotional and financial issues that come with an unexpected rare diagnosis, it is often difficult to find hope and joy. Larry will give a father’s view, Sandra will share a glimpse of their journey, and Jill will introduce you to some of her friends. They hope to bring you encouragement for your days ahead.

1:40pm – 2:10pm
“Update on Clinical Research: Brain Abnormalities, Rare Mutations & Ongoing Work”
Karen Gripp, MD

Clinical research on Costello syndrome involves the families attending this conference. The purpose of this presentation is to thank all research participants and to update you on the results. Our team has worked with the families since 2001, and together we learned a lot.

Costello syndrome can be caused by many different mutations in HRAS, with the p.Gly12Ser mutation being the most common accounting for about 80%. Some mutations, like the relatively common HRAS p.Gly13Cys and the newly identified p.Gly60Asp, are associated with milder physical and developmental differences. Other mutations, like the very rare Gly12Glu, result in very severe medical problems. Understanding these differences will help us take more appropriate care of the affected individuals. The risk for malignant tumors likely varies by underlying HRAS mutation. We continue to collect data in order to confirm this risk stratification and the need for screening studies.

In the past we studied brain growth and brain structural differences, like a Chiari I malformation. This information should inform imaging studies of the central nervous system and the spinal cord. While our work is ongoing, it has already been cited and validated in a recent research publication.

(Lecture Descriptions - continued on page 16)
Dr. Chatfield will speak on “the Heart of Costello syndrome.” This will include the basics of how the normal heart works, and the problems that are common in Costello Syndrome. These common problems include differences in how the heart develops in fetal life—what we call congenital heart disease (CHD)—and other abnormalities that typically develop after birth. These “acquired” heart problems include cardiomyopathy (or heart muscle disease) and arrhythmias (abnormalities of the electrical system). Outcomes of heart disease in Costello syndrome and the available treatment options will be reviewed.

2:55pm – 3:25pm

“GH Use in CS: Are the Benefits Greater than the Risks?”
Dan Doyle, MD

This talk will review the construction of the existing Costello Syndrome growth curves which go from age 0 to 10 years and the on-going work aimed at constructing curves for ages 10 through 18 years. I will also highlight the possibility of low blood sugar in CS patients who have undergone gastric manipulation such as Nissen fundoplication and the management of low blood sugar in those children. Finally I will talk briefly about other endocrinopathies that have been sporadically reported in some patients with CS.

3:30pm – 4:00pm

“Vitamins, Herbs and Supplements: What Do We Know about Alternative Therapies in Costello Syndrome?”
David Stevenson, MD & Chiara Leoni, MD

Dr. Stevenson will provide an overview of what are alternative therapies and what we know about alternative therapies in Costello syndrome based on current evidence. He has selected a number of substances that have been frequently discussed on chatrooms for the RASopathies and the most common natural products used in the general population in addition to several vitamins. The presentation will end with a more detailed discussion about the use of Vitamin D and effects on bone.

4:05pm – 5:15pm

“Best Practice – Ask the Doctors”
Drs. Chatfield, Doyle, Gripp, Kerr, Leoni, Siegel, Stevenson and Weaver

This is a unique opportunity to ask our panel of experts on Costello syndrome the questions that have come to mind throughout the day as you have listened to the talks, or questions that you haven’t had a chance to ask yet!
### SATURDAY: combined CFC/CS workshop sessions

Rick Guidotti will be in the childcare rooms and CS Lounge taking photographs of our children throughout the day, courtesy of Rasopathies Network USA.

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<tr>
<th>TIME</th>
<th>TOPIC/ACTIVITY</th>
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<tr>
<td>7:00am – 8:30am</td>
<td><strong>BUFFET BREAKFAST</strong></td>
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<tr>
<td>8:30am</td>
<td>Drop off children at Childcare (2nd Floor)</td>
<td>Cascade 1-8 Tower, Suite 1370</td>
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<td>CS Lounge</td>
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<td>9:00am – 10:00am</td>
<td><strong>The Unstoppable Charity</strong> (Costello Only)</td>
<td>Evergreen 1 2nd Floor</td>
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<td></td>
<td>Keegan Johnson</td>
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<td>Jenny Polk, Matt Stevenson and Sue Sullivan – Spotlight Speakers</td>
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<td>10:00am – 10:15am</td>
<td><strong>BREAK</strong></td>
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<tr>
<td>10:15am – 11:15am</td>
<td><strong>Genetics 101 for CS Families</strong> (Costello Only)</td>
<td>Evergreen 1 2nd Floor</td>
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<td>(Choose One)</td>
<td>Deborah Leach-Scampavia, MS</td>
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<td>“Feeding &amp; Sensory Through Occupational Therapy”</td>
<td>Evergreen 4 2nd Floor</td>
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<td>Christiana Schadegg, MOT OTR/L</td>
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<td>11:45am – 12:45pm</td>
<td>Childcare and CS Lounge close for Lunch at 11:45am</td>
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<td>Pick up children promptly</td>
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<td>1:00pm – 2:00pm</td>
<td><strong>Fundraising – You Can Do It!</strong> Keegan Johnson/Judy Doyle</td>
<td>Evergreen 1 2nd Floor</td>
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<td>(Choose One)</td>
<td>“Social Security: SSI and Everything You Have Always Wanted to Know” Kirk Lawson</td>
<td>Evergreen 2 2nd Floor</td>
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<td>“Social Skills &amp; Sexuality: An Open Discussion?”</td>
<td>Evergreen 3 2nd Floor</td>
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<td>Robbie Rigby, MSW</td>
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<td>“Behavior Problems and How to Cope” Melinda Wolford, PhD</td>
<td>Evergreen 4 2nd Floor</td>
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<td>2:00pm – 2:15pm</td>
<td><strong>BREAK</strong></td>
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<td>2:15pm – 3:15pm</td>
<td><strong>Using iPads and Handheld Technology to Offer Solutions</strong></td>
<td>Evergreen 1 2nd Floor</td>
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<td>(Choose One)</td>
<td>Marsha Threlkeld</td>
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<td>“Getting Ready for a Meaningful Life”</td>
<td>Evergreen 2 2nd Floor</td>
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<td>Cathy Murahashi</td>
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<td>Erin Hefner, Self Advocate, Costello Syndrome</td>
<td>Evergreen 3 2nd Floor</td>
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<td>Cathy Holland, Self Advocate, CFC Syndrome</td>
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<tr>
<td></td>
<td>“Feeding &amp; Sensory Through Occupational Therapy”</td>
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<td>“Behavior Problems and How to Cope” Melinda Wolford, PhD</td>
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<td>3:30pm</td>
<td>Childcare and CS Lounge close at 3:30 sharp.</td>
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<td>Pick up children promptly</td>
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<td>6:00pm</td>
<td>Families and doctors, Group Photo by the Pool.</td>
<td>Grand Ballroom 1 Main Floor</td>
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<td>If it is rainy, gather by Grand Ballroom 1.</td>
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<tr>
<td>6:30pm – 10:30pm</td>
<td><strong>BUFFET DINNER, RAFFLES, DJ WITH DANCING &amp; QUILT RAFFLE</strong></td>
<td>Grand Ballroom 1 Main Floor</td>
</tr>
</tbody>
</table>
SATURDAY: workshop descriptions

9:00am – 10:00am

“The Unstoppable Charity”
Keegan Johnson, CEO, Zenzaga
Special Spotlight Speakers from our Costello family will be Jenny Polk, Matt Stevenson and Sue Sullivan.

At this year’s conference you’ll take your first steps towards being unstoppable as Zenzaga CEO, Keegan Johnson, brings you the answers and the attitude it takes to create an unstoppable charity through fund-raising and empowers you to believe in your organization’s mission and share that inspiration with others.

10:15am - 11:15am

“Genetics 101 for CS Families”
Deborah Leach-Scampavia, MS
Are you drowning in an alphabet soup of DNA, G12S, RAS and MAPK? Deborah will help to demystify genetics with a quick tour of “The Basic You”. Talking about how genes work as instruction manuals for our bodies, she will link the tour to the genetics of Costello Syndrome. At the end of the session, parents will have the opportunity to “make-and-take” a Costello DNA coded bracelet, so they can explain genetics to other family members and amaze doctors and friends with their grasp of genetics.

10:15am – 11:15am

“Feeding & Sensory Through OT”
2:00pm – 2:15 pm (repeat)
Christiana Schadegg
Explore Occupational Therapy as a therapeutic intervention for children with sensory regulation and feeding difficulties. Learn how sensory regulation and feeding can go hand in hand and how Occupational Therapy can address the two areas together and separately in order to find the right fit for the many needs children and their caregivers face daily that can be confusing and daunting.

1:00pm – 2:00pm

“Social Security: SSI and Everything You Have Always Wanted to Know”
Kirk Lawson
Kirk Lawson, a Public Affairs Specialist for the Social Security Administration, will explain disability benefits through Social Security and Supplemental Security Income (SSI) programs that are available in the US even for people who have not worked. He will also explain the benefits of working, while receiving benefits, and can answer other questions about Social Security benefits for people with disabilities.

11:15am – 12:15pm

“Behavior Problems and How to Cope”
2:15pm – 3:15pm (repeat)
Melinda Wolford, PhD
We are biologically built to be social individuals. We learn from each other by observing and imitating. If neurology is interrupted, this it only makes interpretation, imitation and learning much more difficult for a child who is developing. It is difficult enough for a child with an adequate neurological make up to maneuver his or her environment and learn effective responses. A child with a deficit has more obstacles to overcome. Understanding the origin of unwanted behavior is par-

1:00pm – 2:00pm

“Social Skills & Sexuality: An Open Discussion?”
Robbie Rigby
The quality of our lives is often measured by the quality of our relationships; in fact, relationships even affect our health and longevity. How do we support individuals with developmental disabilities to develop and maintain meaningful, long-term relationships? How do people learn to participate in mutually respectful, loving, and yes, sexual relationships? This session will provide introductory information to help identify the social skills necessary for building such relationships. The speaker will discuss concrete methods for teaching social skills and providing sexual information that promote inclusion, competence and healthy relationships.

1:00pm – 2:00pm

“Fundraising – You Can Do It!”
Keegan Johnson/Judy Doyle
Keegan and Judy share their fundraising successes to help you build yours.

(Workshop Descriptions - continued on page 19)
amount in our efforts to address and change these behaviors. This session is designed to assist parents and caretakers in identifying the origin of unwanted behaviors and teach basic behavioral interventions to increase desired outcomes. Questions and participation are encouraged.

2:15pm – 3:15pm

“Using iPads and Handheld Technology to Offer Solutions”
Marsha Threlkeld

Learn how individuals can be supported to engage in new experiences and environments, learn new tasks, navigate independently in the community, move from one event to the next, interact and share about themselves with coworkers, neighbors, and friends, have fun and socialize, and have systems to help them be calm, centered, and at their best. See many examples using features and apps in the iPad. Learn how groups of families, self-advocates, and service providers are becoming knowledgeable and putting iOS devices to the test. Hear how staff are coming on board and people are easily using these high tech resources.

SUNDAY:

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<th>TIME</th>
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<tbody>
<tr>
<td>9:00am – 10:00am</td>
<td>Positive Exposure Presentation by Rick Guidotti</td>
<td>Northwest Ballroom 1 Ground Floor</td>
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<td>Sponsored by RASopathies Network USA, this presentation focuses on Rick Guidotti’s mission through Positive Exposure. Please see the BIOGRAPHIES section for more information.</td>
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<tr>
<td>10:15am – 11:15am</td>
<td>CSFN Board Meeting</td>
<td>Cascade 1, 2nd Floor</td>
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<td>Our annual face-to-face board meeting where we review the conference , elect new officers for the next two years, address any current business, and answer questions for anyone attending.</td>
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<tr>
<td>11:30am – 12:30pm</td>
<td>Breakout Session with Researchers from Symposium</td>
<td>Evergreen 1, 2nd Floor</td>
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<td>Come hear our experts Dr. Karen Gripp and Dr. David Stevenson give us an informal presentation of the major insights gained from the Scientific Research Symposium. Sponsored by Rasopathies Network USA.</td>
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2:15pm – 3:15pm

“Getting Ready for a Meaningful Life”
Cathy Murahashi

Cathy discusses things to plan for to get your son or daughter ready for adult life. Meet one adult from the Costello community (Erin Hefner) and one adult from the CFC community (Cathy Holland) who will share their successes with you as well.

Farewell to our friends, and have a safe journey home!
Elizabeth Beals, BS
Research Coordinator
University of California, San Francisco
513 Parnassus Avenue
San Francisco, CA 94143
415-476-2985
Elizabeth.beals@ucsf.edu

Elizabeth Beals graduated with a Bachelor of Science from the University of California, Davis. For the past year she has worked as a Research Coordinator at UCSF, managing research studies with human participants. She plans to go to graduate school to study Nutrition and Metabolic variation, and to eventually work in medical research.

Emma Burkitt-Wright, MBChB PhD
Manchester Centre for Genomic Medicine
St Mary’s Hospital
6th Fl. Central Manchester University Hospitals
NHS Foundation Trust
Oxford Rd, Manchester M139WL
Emma.Burkitt-wright@manchester.ac.uk

Emma Burkitt-Wright graduated in medicine (MBChB with Honours and intercalated MPhil in Clinical Psychology) from the University of Liverpool in 2002, and undertook posts in general internal medicine and paediatrics around Liverpool from 2002-6. She started in clinical genetics in Manchester in 2006, as the first academic clinical fellow in the UK’s integrated clinical academic training scheme. During this, she spent 5 months with Professor Mariano Barbacid’s group at the Spanish National Cancer Centre, helping to characterise the first B-Raf mouse model of cardio-facio-cutaneous syndrome (CFC) (Urosevic et al, PNAS, 2011). She undertook her PhD (awarded 2014) on Manchester Biomedical Research Centre and Wellcome Trust Research Training Fellowships (2009-2013). This focused on de novo germline disorders of the Ras-MAPK pathway, in particular CFC, including clinical and molecular characterisation of patients and further work with mouse models. In collaboration with the diagnostic laboratory of Manchester Centre for Genomic Medicine (MCGM), she piloted the introduction of massively parallel (‘next generation’) sequencing technologies in the molecular diagnosis of Ras-MAPK pathway disorders.

She recently completed her higher specialist training in Clinical Genetics, and is currently a locum consultant clinical geneticist in the MCGM, conducting a Ras-MAPK pathway disorder clinic with Dr Bronwyn Kerr and Dr Sue Huson, amongst others, which runs in conjunction with the nationally commissioned specialist service for complex NF1 based there.
BIOGRAPHIES

Kathryn Chatfield, MD, PhD
Instructor of Pediatrics, Cardiology
University of Colorado School of Medicine
Director, Cardiac Genetics Clinic
Division of Cardiology
13123 East 16th Avenue, B100
Aurora, Colorado 80045
720-777-6820
Kathryn.Chatfield@childrenscolorado.org

Dr. Chatfield is an Instructor of Pediatrics, Cardiology, University of Colorado School of Medicine and the Director of the Cardiac Genetics Clinic. Her degree is from Dartmouth Medical School and she completed her residency at Children’s Hospital of Philadelphia.

Dr. Chatfield’s clinical interests include Noonan-spectrum disorders, Marfan syndrome and related connective tissue disorders, familial congenital heart disease, metabolic and inherited forms of cardiomyopathy. Her research focus is the role of mitochondrial energy metabolism in pediatric cardiomyopathy and heart failure. She is also currently doing research related to the role of cardiac transplantation in children with Noonan-spectrum disorders and hypertrophic cardiomyopathy.

Leoni Chiara, MD
Center for Rare Diseases
Department of Pediatrics
Università Cattolica del Sacro Cuore
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Mobile: (+39) 3387932404
E-mail: leonichia2@gmail.com

Leoni Chiara, MD graduated from Catholic University of Rome in 2007 and obtained her MD in 2008 from Catholic University as well. She trained in pediatrics and dysmorphology as a resident at the Catholic University of Rome, 2009-2014. In between her residency years she spent one year in Medical Genetics Postdoctoral Fellowship Program at the University of Utah, 2012-2013.

Throughout her education and training, Dr. Leoni has always been interested in birth defect syndromes and the care of children with special care needs, in particular individuals affected by RASopathies. Since 2007 she collaborates with Dr. Zampino at the Center for Rare Diseases (Catholic University in Rome) for the diagnosis and clinical management of children with genetic conditions. Dr. Leoni’s research focus has been in congenital malformation, primarily RASopathies, in collaboration with Dr. Zampino and Dr. Tartaglia.
Our group uses tools of human genetics to identify novel pathways relevant to disease processes in the skin and other organ systems. We have established a patient recruiting core to obtain DNA and tissue samples from individuals with genetic skin disease and have developed a system for streamlined patient consent, blood and tissue acquisition, and rapid transport to our laboratory. We maintain a biobank of human specimens including tissue, DNA, and primary cell lines (keratinocytes, fibroblasts, and specialized cell types), and a database with clinical information. We utilize next generation sequencing approaches for disease gene identification and employ human specimens for functional studies. We employ in vitro and ex vivo tissue modeling which recapitulates disease phenotypes and animal models to study the pathobiology of disease states and to understand the role of identified genes in normal epidermal homeostasis. To date, with over 50 clinical collaborators, we recruited over 375 kindreds with genetic skin disease; of these 40 have mosaic/linear disorders. Through our work in such disorders, we have identified that somatic activating RAS mutations cause a spectrum of mosaic phenotypes including keratinocytic epidermal nevi, giant congenital melanocytic nevi, nevus sebaceous, vascular tumors, and the multi-system Cutaneous-Skeletal Hypophosphatemia Syndrome (CSHS). Our other kindreds represent disorders of keratinization (DOK) which are characterized by scaly skin and associated systemic abnormalities. In my research group, undergraduates, medical students, post-doctoral fellows, and MD/PhD students actively participate in translational research projects dedicated to advancing understanding of this group of disorders.

Dan Doyle, MD is an Associate Professor of Pediatrics at Thomas Jefferson University and duPont Children’s Hospital. His interest is in the natural history of growth and the use of growth hormone in Costello syndrome. His previous measurements of the children with CS have resulted in the recent publication of “Normative Growth Curves in Children with Costello Syndrome” in the American Journal of Medical Genetics. He will discussing the use and limitations of these curves as well as the natural history of growth and the use of growth hormone in CS children.
Judy A. Doyle
CFC International, Board Member
Akron Children's Hospital Parent Advisor Coordinator
jdoyle@chmca.org

Judy received her bachelor's in Business Administration, majoring in Marketing. She has three children; daughter Madison 21, Sally 18 and son, Jack 16. It is her medical experience with Jack that brought Judy to her present position at Akron Children’s Hospital to become a board member and active fundraiser for CFC International.

Jack was born with Cardio-Facio-Cutaneous Syndrome (CFC). The first two years were filled with managing Jack's many medical problems and searching for a diagnosis. The Doyles relied on the support of their pediatrician and the internet to understand Jack's problems, searching for a diagnosis and connecting with other parents. They sent Jack's medical history to many of the world's top genetic doctors. Jack was finally diagnosed at the University of Utah in 2000 by Dr. John Opitz. Once diagnosed with CFC Syndrome, the entire Doyle family became involved in raising funds to promote research into this very rare condition. Judy has been an active board member for many years.

Karen Gripp, MD
Professor of Pediatrics, Thomas Jefferson Medical School
Division of Medical Genetics
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Wilmington, DE 19803
302-651-5916, Fax 302-651-5033
kgripp@nemours.org

Karen W. Gripp, MD is the Chief of the Division of Medical Genetics at the AI duPont Hospital for Children in Wilmington, DE, and a Professor of Pediatrics at the Sidney Kimmel Medical College at T. Jefferson University in Philadelphia, PA. She is also an Associate Editor of the American Journal of Medical Genetics. Karen is a Co-Chair of the Professional Advisory Committee for the Costello Syndrome Family Network. She organized and hosted the 3rd International Costello Syndrome meeting in Wilmington, DE in 2003. She was the director of the multidisciplinary Costello syndrome clinic, which opened at the duPont Hospital in Wilmington in 2003 and has now become the “MAP” clinic for all rasopathies. Karen's long standing research interest in Costello syndrome and its underlying genetic cause was recognized in 2003 through the John M. Opitz Young Investigator Award. Dr. Gripp is the author of more than 100 peer reviewed publications, many of these resulting from her ongoing research on Costello syndrome. Karen is the PI (Principal Investigator) on the project "Costello Syndrome: Clinical and Molecular Investigations", which encompasses the various areas presented here today. Our research focuses on the clinical problems seen in individuals with Costello syndrome, and how the different Costello syndrome causing mutations present.
Rick Guidotti
Positive Exposure
43 East 20th Street, 6th Floor
New York, NY 10003
212-420-1931
rick@positiveexposure.org

Rick Guidotti, an award-winning former fashion photographer, has spent the past fifteen years working internationally with advocacy organizations/NGOs, medical schools, universities and other educational institutions to effect a sea-change in societal attitudes towards individuals living with genetic difference; his work has been published in newspapers, magazines and journals as diverse as Elle, GQ, People, the American Journal of Medical Genetics, The Lancet, Spirituality and Health, the Washington Post, Atlantic Monthly and Life Magazine. Rick is the founder and director of Positive Exposure, an innovative arts, education and advocacy organization working with individuals living with a difference or disability. Positive Exposure utilizes the visual arts to significantly impact the fields of genetics, mental health and human rights.

Positive Exposure photographic exhibition premiered at the People’s Genome Celebration, June 2001, at the Smithsonian’s National Museum of Natural History in DC, and continues to exhibit in galleries, museums and public arenas internationally. Rick Guidotti’s Positive Exposure photo and video presentation explores the social and psychological experiences of people living with genetic, physical, cognitive and behavioral conditions of all ages and ethno-cultural heritages. Rick’s presentation celebrates the richness and beauty of human diversity. Positive Exposure provides new opportunities to see individuals living with a physical or cognitive difference, first and foremost as a human being with his/her own challenges rather than as a specific diagnosis/disease entity.

Erin Hefner
Erin Hefner is a 29-year-old woman with Costello syndrome. She was born and raised in Creve Coeur, Illinois (near Peoria). She graduated from East Peoria Community High School in 2005. She works two half-day jobs in childcare and volunteers at her church. Once a week she attends a social group of special needs peers, Heart of Morton, who have aged out of the school system. They go on outings, eat, and just hang out. Erin enjoys riding horses at Central Illinois Riding Therapy and Special Olympics bowling. Erin lives with her parents and has an older brother.
Rachel Holland

Rachel Holland is a 33-year-old woman who was born and raised in Sacramento, California. Rachel and her mother attended the very first CFC conference held in Salt Lake City, Utah in 2000. In 2005, Rachel chose to move into her own apartment in the nearby town of Davis, California. Rachel shares her apartment with one housemate, and a dog named Elvis. Rachel has worked at several different jobs in Davis. She currently works three days a week at Hillel House on the University of California, Davis campus. In addition, Rachel owns her own business called Sparkling Jewels, which allows her to make and sell jewelry and hair accessories. Rachel sells her jewelry online, at local fairs and farmer’s markets, and at various boutiques in Davis. She enjoys many arts, music, cooking, and sports activities in the Davis community, but her favorites activities are walking and running in local races, spending time with her boyfriend, and socializing with friends.

Keegan Johnson

Keegan is the founder and CEO of Zenzaga, a company focused on helping companies, communities and causes around the world, harness the power of "Mass Motivation". Different from other speakers, Keegan's experience in both the for-profit and non-profit worlds, allows him to combine the passion of running a volunteer based charity with the analytics of running an e-Commerce company. Keegan's ability to connect to audiences and actually inspire action is essential to taking your cause, community or corporation to the next level.

Prior to founding Zenzaga, Keegan co-founded Ethoca, a for-profit e-Commerce fraud management company dedicated to eliminating fraud through collaboration. He not only designed Ethoca's flagship product, but also learned the ins and outs of collaboration by setting up deals requiring Visa, Mastercard and over 200 of the largest eCommerce companies and banks including Walmart, Skype, Capital One and Bank of America, to work together.

At the same time, Keegan found out his firstborn son had a rare genetic disorder called Prader-Willi Syndrome (PWS). He co-founded a Canadian charity and became the Executive Director of the US charity with a mission to eliminate the challenges of PWS through research. In this capacity, Keegan learned how to both inspire individual action and bring together organizations from around the world to work together.
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Bronwyn Kerr, MD is a consultant clinical geneticist who studied medicine at the University of Sydney before qualifying in paediatrics and genetics in Australia. After completing a two year research fellowship into X-linked mental retardation with Professor Gillian Turner, she moved to the UK in 1993. She was appointed Consultant Clinical Geneticist in the Regional Genetic Service in Manchester in 1995, based largely at Royal Manchester Children’s Hospital (RMCH). Her main clinical interest is in the causes of developmental disability and congenital abnormality. Her principal research interest has been Costello syndrome, and more recently, other disorders of the RAS/MAPK pathway. She was a founder member of the Medical Advisory Boards of the International Costello syndrome support group, the Association Française des syndromes de Costello and CFC, and the Costello Family Support Network. She has a number of key publications in this area, and is frequently consulted by national and international colleagues for advice on diagnosis and management in this group of disorders. A particular interest is developing an evidence base for management of rare disorders, an endeavor for which international collaboration in the long-term is critical.

Dr. Kerr has a professional interest in medical management, and a strong commitment to improving the patient experience and the quality and safety of healthcare. She was Clinical Director of the Regional Genetic Service in Manchester, one of the largest and most comprehensive genetic services in Europe, from 2003-2010. In April 2010, she was appointed as Associate Medical Director, Central Manchester University Hospitals NHS Foundation Trust, with responsibility for Clinical Effectiveness.

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Dr. Ophir Klein is the Larry L. Hillblom Distinguished Professor in Craniofacial Anomalies and Chair of the Divisions of Craniofacial Anomalies and Orthodontics at the University of California, San Francisco (UCSF). He is also the Medical Director of the UCSF Craniofacial Center and Director of the UCSF Program in Craniofacial Biology. Dr. Klein was educated at the University of California, Berkeley, where he earned a BA degree in Spanish Literature. He subsequently attended Yale University School of Medicine, where he received a PhD in Genetics and an MD degree. He then completed residencies at Yale-New Haven Hospital in Pediatrics and at UCSF in Clinical Genetics. Work in Dr. Klein’s research group centers on organ development and regeneration, with a major focus on understanding the processes
underlying craniofacial development. Dr. Klein is the Lead Researcher for the Research Project, Developing 3D Craniofacial Morphometry Data and Tools to Transform Dysmorphology, and is represented by Elizabeth Beals and Nick Mahasuwan at the Family Forum.

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Kirk Larson is the Western Washington Public Affairs Specialist for the Social Security Administration. He has worked with the agency for over 22 years in both technical and supervisory roles. Kirk has presented Social Security information in both the Seattle and San Francisco Regions. He has had several articles published, and has appeared on TV and radio shows to discuss Social Security issues.

Deborah Leach-Scampavia, MS
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Deborah Leach-Scampavia, MS is the Director of Education Outreach for the Scripps Research Institute, Scripps Florida. Prior to her appointment with Scripps Research and since 1988 Ms. Leach-Scampavia served as the coordinator for the National ESCA and Surface Analysis Center for Biomedical Problems in the Department of Biomedical Engineering at the University of Washington, Seattle. Her research and publication focus has been in the field of surface characterization of biomaterials using ESCA (Electron Spectroscopy for Chemical Analysis) and ToF SIMS (Time-of-Flight Secondary Ion Mass Spectrometry). Throughout her career, she has also been a dedicated science educator. Before her appointment as Director of Education Outreach, she served as a coordinator for the University of Washington Engineered Biomaterials (UWEB) K-12 Science Education Outreach program. Deborah designs and directs outreach programs that allow biomedical researchers to share their understanding of the processes of science and contemporary scientific discovery. A goal of Scripps Research outreach is to contribute directly to greater science literacy in the K-16 Science, Technology, Engineering and Math (STEM) education community.

Nick Mahasuwan
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Nick Mahasuwan graduated from the University of California, Davis in 2013 with a degree in Biological Sciences. He interned at the Center for Excellence in Primary Care before accepting a Study Coordinator position at the Department of Orofacial Sciences at UCSF. He is also pursuing a Master's for Public Health in genetics.
Cathy Murahashi
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Cathy is the Parent Engagement Coordinator for the King County Family Coalition which provides legislative and systems advocacy for families. Previously she has worked as the Parent to Parent Coordinator. She is also the co-creator of the “Start Now” training series for families and students to prepare for future employment.

Her greatest joy and what fuels her passion for families with children with special needs are her 3 children, who are now young adults. Her second daughter, Holly is 26 and happens to have Down Syndrome. She introduced Cathy to the world of disabilities. Her eldest daughter, Laura is a Physical Therapist, and her son, David has his degree in Special Education. Her journey took a twist in the road 2 years ago when David had an unexpected brain bleed. This has provided fresh opportunities to learn and understand what families with disabilities experience.

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Katherine (Kate) Rauen, MD, PhD is a Professor in the Department of Pediatrics, Division of Genomic Medicine at the UC Davis where she currently serves as the Chief of Genomic Medicine and holds the Albert Holmes Rowe Endowed Chair in Human Genetics. She received a MS in Human Physiology and a PhD in Genetics from UC Davis doing research on gene dosage compensation and genetic evolution. She obtained her MD at UC Irvine where she also did research in cancer genetics. Dr. Rauen did her residency training in Pediatrics and fellowship in Medical Genetics at UC San Francisco.

Dr. Rauen is internationally known for her pioneering work in the early application of microarray technology in clinical genetics and as a leader and major contributor to the understanding of the “RASopathies”, the Ras/MAPK pathway genetics syndromes. Her research program involves the clinical and basic science study of cancer syndromes with effort to identify underlying genetic abnormalities affecting common developmental and cancer pathways. Dr. Rauen led the research team, including the CFC International Family Support Group that discovered the genetic cause of cardio-facio-cutaneous syndrome.
Dr. Rauen is committed to academic medicine, medical education, and advancing best practices for patients with RASopathies. She has successfully obtained both intramural and extramural funding for her research activities, and currently holds a 5-year NIH grant studying skeletal myogenesis in Costello syndrome and CFC. She is the innovator of the world-renowned NF/Ras Pathway Clinic which she initiated in 2007 and this clinic has now been emulated around the globe. She serves on the medical advisory board of CFC International, is a Co-Chair of the Professional Advisory Committee of the Costello Syndrome Family Network, and serves on the advisory boards for RASopathies Network USA and Global Genes.

Dr. Rauen was recently awarded the Presidential Early Career Award for Scientists and Engineers (PECASE) on her work for CFC and Costello syndrome. This Award is the highest honor bestowed by the United States Government on science and engineering professionals in the early stages of their independent research careers. This Presidential Award is awarded for innovative and far-reaching developments in science and technology, in an effort to increase awareness of careers in science and engineering, give recognition to the scientific missions of participating agencies, enhance connections between fundamental research and national goals, and highlight the importance of science and technology for the nation’s future. Additionally in June 2014, Dr. Rauen won an International Advocacy Award, the “Global Genes RARE Champion of Hope in Science”.

**Robbie Rigby, MSW**
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Robbie Rigby, MSW, provides private supportive counseling and consultation to individuals with developmental disabilities and their family members regarding social skills and sexuality information. She also has many years of experience providing training and education about sexuality and social skills to teachers/educators, residential and vocational service providers, and to family/community groups. She has worked for over 25 years with people with disabilities in community settings, vocational programs and residential services, and has taught classes on social skills and sexuality in various community settings.

**Christiana A. Schadegg, MOT OTR/L**
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Christiana Schadegg is a pediatric occupational therapist and clinical director at No Stone Unturned Therapeutic Learning Center in Manhattan, Kansas, a pediatric outpatient therapy clinic that is focus on a multidisciplinary team approach for children in central Kansas. The center strives to provide comprehensive therapy services to children with deficits affecting their daily living by optimizing their physical and mental health in one
place. Christiana is a native of Kansas and received a Bachelors of Science degree in Kinesiology from Kansas State University in 2003 and a Masters of Occupational Therapy from Rockhurst University in 2006. Christiana has received clinical training at Children’s Mercy Hospital in Kansas City, Missouri and mentorship study with Lucy Jane Miller at the STAR Center for Sensory Processing in Denver, Colorado. She is also certified in Sequential Oral Sensory Feeding Therapy. Christiana has been trained and certified in Autism Diagnostic Testing and is currently on the Autism Diagnostic Testing Team at No Stone Unturned TLC, working with Kansas University Medical Center. Christiana has worked with pediatrics in both school based and outpatient-based settings in Oklahoma and Kansas. She specializes in treatment and regulation for children on the Autism Spectrum, with sensory processing deficits, ADHD/ADD and with sensory feeding difficulties, following a behavioral treatment approach. She also has experience with neurological disorders, developmental delays, behavioral disorders, Down syndrome, Cerebral Palsy, torticollis and brachial plexus injuries.

**Suma Shankar, MD, PhD**

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Suma P. Shankar MD PhD, Assistant Professor, Department of Human Genetics and Department of Ophthalmology, Emory University, Atlanta, Georgia. Dr. Shankar joined Emory University in January, 2010 with dual appointments in the Department of Human Genetics and the Department of Ophthalmology. She received her medical degree from Bangalore Medical College, India and did her Ophthalmology residency training and obtained the Membership of the Royal College of Ophthalmologists in London and Fellowship of the Royal College of Surgeons in Edinburgh in the United Kingdom. She obtained a PhD, focusing on molecular genetics of eye diseases and completed a fellowship in pediatric ophthalmology at the University of Iowa. She then went to the University of California in San Francisco for another fellowship in medical genetics. Since 2007 she has been involved with the cardio-facio-cutaneous syndrome, Noonan syndrome and Costello syndrome groups. Her research interests include ophthalmic manifestations in Ras/MAPK pathway syndromes and molecular genetic studies of ophthalmic diseases. She has won the Western Scholar and Henry Christian Award for Excellence in Research from American Federation of Medical Research, and the best poster award from Ras/MAPK symposium 2009, for her research on the “ocular features of the Ras/MAPK syndromes”. She is leading the “Ocular Predictive Heath Initiative” project with a goal of establishing a data base and DNA bank for inherited and complex eye diseases with genetic etiology at Emory.
Dawn Siegel, MD is a pediatric dermatologist from Medical College of Wisconsin and Children's Hospital of Wisconsin in Milwaukee. Dr. Siegel received her medical degree from the University of Wisconsin, Madison and then went on to do a pediatric residency at Children's Hospital Oakland, California. Dr. Siegel did her dermatology residency and pediatric dermatology fellowship at the University of California, San Francisco. Dr. Siegel has a particular interest and expertise in genetic dermatologic syndromes, including Costello syndrome, NF and the RASopathies. She is co-director of the Neurofibromatosis and RASopathy clinic at Children's Hospital of Wisconsin. In addition, she has spent the last several years working on gene discovery for a hemangioma syndrome called “PHACE syndrome”. Dr. Siegel has been actively involved in studying the skin issues in Costello syndrome and cardio-facio-cutaneous syndrome since 2006. The paper on CFC was published in 2010 and the Costello paper was published in 2011 in the British Journal of Dermatology. Today Dr. Siegel will teach you about the results of the Costello skin study and share some general skin care guidelines with you.

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Pediatrician and Medical Geneticist, Stanford University. Dr. Stevenson completed his residency in Pediatrics at the University of New Mexico and a 3 year fellowship in medical genetics at the University of Utah. He was on faculty at the University of Utah for 10 years before joining the faculty in the Division of Medical Genetics as an Associate Professor at Stanford University. His initial research focused on neurofibromatosis type 1 (NF1), and since that time he has expanded his research to RASopathies focusing on the musculoskeletal problems and genotype-phenotype correlations. He has received grant funding from the NIH, Doris Duke Charitable Foundation, Thrasher Research Fund, and Department of Defense to investigate the musculoskeletal system in syndromes of the Ras/MAPK pathway. He currently serves as a Co-Chair of the Professional Advisory Committee for the Costello Syndrome Family Network, is the Co-Chair of the Children's Tumor Foundation International NF1 Bone Abnormalities Consortium, and is a member of the National Prader-Willi Syndrome Association (PWSA) USA Scientific Advisory Board. He has published over 80 scientific articles, and 2 GeneReviews focused primarily on Ras/MAPK syndromes. Dr. Stevenson first attended the Costello conference in Portland, Oregon and has attended all the conferences since that time and is excited to visit with the families again in Seattle.
Larry, Sandra & Jill Taylor

Larry and Sandra Taylor grew up in small towns in South Carolina about 70 miles apart. They met in church one Sunday in 1967 and married two years later. After living the first year at Clemson University while he finished his degree in Architecture and she attended Anderson College, they moved to Charlotte, NC where their son Clint and daughter Jill were born. In 1980 they moved to Atlanta, GA and in 1984, to Panama City, FL where Larry has had his own architectural firm since 1986.

Larry has won several architectural design awards, including a NC State AIA award for the design of a Boy Scout Camp. He is an Eagle Scout and was a member of the Rotary Club for 28 years with perfect attendance. Sandra continued her education in Panama City at Gulf Coast Community College, graduating with an AS degree in Legal Assisting in 1998, the same year Jill graduated from Bay High School with a diploma for exceptional students. Sandra has held a variety of interesting positions and recently retired after 13 years at a resort property management company on St. George Island, FL as Director of Human Resources. She held certifications as a NALA Certified Legal Assistant and a SHRM Senior Professional in Human Resources. She has been involved with the Costello Syndrome Family Network for many years, serving as Board President and Conference Coordinator for the past four years. Larry and Sandra are both instrument rated pilots and certified scuba divers. They enjoy travel, cooking, sailing, camping, art and photography, and they have two very spoiled Siamese cats named Bud and Tiramisu. They recently restored a 1953 MG TD named Mabel which was the first car they dated in. They will celebrate 46 years of marriage in August 2015.

Clint spent a tour in Germany in the Army with a brief stint in Saudi Arabia, and attended the Golf Academy of the South graduating with degrees in golf course management and golf instruction. He is currently a deep sea charter boat captain on his boat, the BigFishSGI, off St. George Island. Jill lives in a carriage house behind her parents’ home and volunteers 2-3 days a week at a nursing home. She also takes care of the neighbors’ pets when they are away. She enjoys crafts, cooking, and has a sweet Cavalier King Charles spaniel named Molly.

Mihir Thacker, MD

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Mihir M. Thacker, MD, is an orthopaedic surgeon at the Nemours/Alfred I. duPont Hospital for Children. He went to the Seth G. S. Medical College in Mumbai, India and completed his orthopaedic residency at the Lokmanya Tilak Municipal General Hospital in Mumbai. He has had extensive subspecialty training through pediatric orthopaedic fellowships at the National
University Hospital in Singapore, Hospital for Joint Diseases/New York University in New York, and the duPont Hospital for Children. He also completed a fellowship in musculoskeletal oncology at the Jackson Memorial Hospital/University of Miami.

Dr. Thacker’s major clinical interest is bone and soft tissue tumors. He performs limb salvage surgery for tumors, including using noninvasive expandable (growing) prostheses. He is also interested in hip disorders in children and performs complex hip reconstruction (including surgical hip dislocations and peri-acetabular osteotomies), treatment of complex congenital anomalies, correction of complex deformities and limb lengthening. He directs the clubfoot clinic and practices the Ponseti technique for clubfoot treatment.

Dr. Thacker is active in teaching medical students, residents, fellows and international visiting observers. He is invited to lecture nationally and internationally, and is a member of several societies including the Pediatric Orthopaedic Society of North America, Children’s Oncology Group, Musculoskeletal Tumor Society and Lower Limb Reconstruction Society. He is actively involved in research and has more than 30 peer-reviewed publications, five book chapters and several national and international presentations at scientific conferences.

Dr. Thacker has been selected by Delaware Today 2011 as one of the best surgeons in Delaware among both pediatric surgeons and surgical oncologists. He received the Patient Choice Award and Compassionate Doctor Award and was listed in Best Doctors in America (2009-11).

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Marsha works at WISE and trains on Apple handheld devices and apps, with government agencies, employment specialists, school-to-work transition programs, Voc Rehab, families, and job seekers. She has enjoyed collaborating with Apple Inc., training their retail store staff on accessibility needs and solutions, and leveraging the use of equipment to increase the inclusion, productivity, independence, and communication of individuals experiencing intellectual disabilities. Marsha has built iPad Learning Cohorts across WA State and consulted to cohorts elsewhere; bringing together employment professionals and transition teachers to learn to use and incorporate the equipment. She has a Certificate in Assistive Technology Applications from California State University, Northridge, and is a Subject Matter Expert to the Office of Disability Employment Policy at the U.S. Department of Labor.
K. Nicole Weaver, MD
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K. Nicole Weaver, MD, is a clinical geneticist at Cincinnati Children’s Hospital Medical Center in Cincinnati, Ohio. She is the geneticist for the Cardiovascular Genetics Clinic at CCHMC, which provides diagnostic evaluations and ongoing management for children and adults with connective tissue disorders such as Marfan syndrome, as well as Noonan and related syndromes. Dr. Weaver met Dr. Gripp and became interested in Costello syndrome research through taking care of a very special little girl with Costello syndrome caused by the rare G12E mutation. Dr. Weaver is excited to attend this year’s Rasopathies conference and learn new information to bring back to her clinic patients. She will be conducting research about the problems facing older individuals with Costello syndrome.

Melinda Wolford, PhD, NCSP
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Dr. Melinda Wolford, a specialist in neurological disorders of childhood, has been practicing as a school psychologist since 1994 and in the educational field since 1991. She has a PhD in School Psychology with a minor in Neuropsychology from Texas Woman’s University in Denton, Texas. Dr. Wolford has been active in developing collaborative interactions between families, school personnel, and community agencies to provide supportive partnerships that ensure success for children. She has also been active in identifying and facilitating partnerships with community agencies looking to provide services, support, respite and/or funds for families of children with disabilities. Dr. Wolford has provided numerous training in-service opportunities for educators and parents and is involved in a number of research projects. She is currently working as an Assistant Professor at Youngstown State University in the Counseling Special Education and School Psychology Department.

Dr. Wolford and her husband, Eric Wolford, established the No Stone Unturned Foundation, a nonprofit organization dedicated to the support and research of children with health initiatives and/or disabilities and their families. The Wolfords were inspired by their son Stone who was diagnosed at 2 years of age with Cardio-Facio-Cutaneous Syndrome (CFC Syndrome), a rare genetic syndrome. Currently she is a co-founder and serves on the executive board of the No Stone Unturned Therapeutic Learning Center and Katie’s Way Center in Manhattan, Kansas. To learn more, you may visit her googlesite page at https://sites.google.com/site/melindawolfordphdncsp/home.
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Kara Bahnsen
Sandra Taylor

POSTER SESSION

COSTELLO SYNDROME REPRESENTATIVES

Johnny and Tammy Moore, and their daughter, Kelsi
Birmingham, Alabama USA

Stephanie and Andy Nimmo, and their daughter, Daisy (represented by Stephanie Nimmo)
London, England UK
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Mary Tierney
Jessie Van Der Wel
Lara Weeks
Sarah Weisenbaugh
Becky Winn

Dr. David Stevenson who gathered a list of manuscripts published in the last two years so we could include them for you as a reference.

Cathy Coates for the fabulous “Dr. Seuss Quilt” she created and donated for the Saturday night raffle.

Becky Winn for being our special photographer for our group photograph and snapshots around the conference.

Taylor Architects, Inc. for their support through office supplies, cellphone use, paper and their copier for handouts.

RASopathies Network USA for their support by providing free events for our families.

CSFN MISSION STATEMENT

• To raise awareness about Costello syndrome
• To provide families with information, support and advocacy
• To support and advocate for research

a note of appreciation to...
Costello syndrome with severe nodulocystic acne: unexpected significant improvement of acanthosis nigricans after oral isotretinoin treatment. Case Rep Pediatr 2015
Case report of improvement of acanthosis nigricans and acne after treatment with a drug typically used for acne.

Dysregulation of astrocyte extracellular signaling in Costello syndrome. Sci Transl Med 2015
Study on impact of HRAS on brain cells (astrocytes) and tests on these cells with certain drugs.

A specific HRAS mutation resulting in milder physical features.

Study on frequency of cancer in RASopathies versus the general population rate in Germany. The standardized incidence ratio for Costello syndrome was increased (42.4) showing increased risk for cancer.

Case report of individual with rarer HRAS mutation.

Fatal congenital hypertrophic cardiomyopathy and a pancreatic nodule morphologically identical to focal lesion of congenital hyperinsulinism in an infant with costello syndrome: case report and review of the literature. Pediatr Dev Pathol 2015
Case report of individual with Costello syndrome with unique findings including hyperinsulinism.

Growth hormone replacement therapy in Costello syndrome. Growth Horm IGF Res 2014
Case report of individual with Costello syndrome and experience using growth hormone.

Case report of individual with Costello syndrome with impaired social interactions and non-verbal communications.

Study showing that individuals with Costello syndrome have functional limitations that cause significant disability in daily activity, mobility, and socialization and cognition.

Description of the perinatal findings of 3 individuals with Costello syndrome showing that fetal arrhythmia and neonatal hypoglycemia are relatively specific to Costello syndrome in the RASopathies.

Case report of rare HRAS mutation resulting in G12E. Suggested that disordered elastogenesis may contribute to morbidity in Costello syndrome.

Case report of individual with Costello syndrome with fetal tachycardia and polyhydramnios.

Anesthetic management of Costello syndrome: a case report. AANA 2014
Case report of anesthetic management.

Cardiac events in Costello syndrome: One case and a review of the literature. J Saudi Heart Assoc 2014
Review of the heart findings in Costello syndrome.

Description of the dental and craniofacial features in Costello syndrome in 41 individuals. Common findings included macrocephaly, malocclusion with anterior open bite, enamel hypomineralization, delayed tooth development and eruption, gingival hyperplasia and high palate.

High rates of internalizing and externalizing problems and psychopathological risk in Costello syndrome and the RASopathies.

(Publications - continued on page 38)
Morice-Picard et al., Cutaneous manifestations in Costello and cardiofaciocutaneous syndrome: report of 18 cases and literature review. Pediatr Dermatol 2013
Report of the cutaneous findings of 11 individuals with Costello syndrome. Has a nice table of the summary of the findings. Mentions delayed wound healing in about 50% and has good statement about differences between CS and CFCS.

Giannoulatou et al. Contributions of intrinsic mutation rate and selfish selection to levels of de novo HRAS mutations in the paternal germline. PNAS 2013.
Interesting article in which data have been presented in the past at the symposium by Anne Goriely. They looked at somatic HRAS mutations in sperm in the general population. The data support the role of selfish spermatogonial selection helping explain the occurrence of CS, and differences from the mutation pattern in tumorigenesis.

Dr. Leoni’s data on the Italian cohort in a case: control fashion showing lower BMD. Also showed low 25OH Vit. D concentrations in all the individuals with CS (although no obvious correlation with BMD parameters).

Wey et al., Kinetic mechanisms of mutation-dependent Harvey ras activation and their relevance for the development of Costello syndrome. Biochemistry 2013.
Article estimating kinetics of the various HRas mutations helping to explain the spectrum of the different HRAS mutations in cancer and CS.

Beukers et al., HRAS mutations in bladder cancer at an early age and the possible association with the Costello syndrome. Eur J Hum Genet 2013
States they looked at bladder cancer of young (<20 yrs) compared to older patients. HRAS mutations were more frequent in the younger group and highly uncommon in the older group. Of the whole cohort only one patient had Costello syndrome and there was another patient suspected to be mosaic for HRAS mutation.

Cizmarova et al., Rasopathies – dysmorphic syndromes with short stature and risk of malignancy. Endocr Regul 2013
Could not get this article. From abstract, it looks like a review.

Using screening assessments for autism (SCQ and SRS), autism traits were examined in NF1, NS, CS, and CFC. A total of 44 individuals with Costello syndrome were included with an increase in the SCQ and SRS scores. One individual with Costello syndrome was formally evaluated with diagnostic tools such as the ADOS and ADI-R and diagnosed with Autism and Atypical autism respectively. The individuals with Costello syndrome had the lowest variance in the SRS scores compared to other RASopathies.

Documents that the majority of individuals with CS have hypomineralization of enamel which was also seen in a CS mouse model. They were able to demonstrate the role of Ras signaling in enamel formation.

HRasG12V mice showed locomotor hyperactivity which could be rescued by antioxidant treatment. Some interesting evidence that antioxidants may prove beneficial.

Aytekin and Alyamac, Two new cases with Costello syndrome. Dermatol Online 2013.
Case reports of individuals with Costello syndrome

Verbal recall was impaired whereas recognition memory was relatively intact. Story recognition was highly correlated with listening comprehension, and performance on measures of linguistic ability and academic skills was impaired in CS. Sparring of a cognitive domain (recognition memory).

Concise review of the RASopathies.

Good description of the orthopedic manifestations in CS showing that the musculoskeletal system is frequently affected.

Study with a relatively large number of individuals with CS (N=78), although when breaking into genotype the numbers in subsequent groups became small. The p.G12A (n=4) and p.G12C (n=3) were more severely affected when compared to the G12S group using a severity scoring system. It did not look like severity score increased with age between 2 and 20 years.