



Costello Syndrome Family Network

2006-2007

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Newsletter:

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Letter from the President

April 2007

Hi All,

As we move toward our conference at Doernbecher Children's Hospital in Portland, Oregon July 19-22, 2007, amazing progress keeps us constantly on our toes to keep our balance, adjusting to new and intense information. But we shouldn't be too surprised. It's what our children do – push us to learn something new so we can give them the best care we can muster. Somehow, each time, we rise to the challenge.

While the Hras gene find has been thrilling, it's taken me a year and a half to get a passable understanding on what it means for our children. These mutations are along the Ras-MAP kinase pathway, one of a few well-known information pathways from outside the cell to the nucleus. It helps me if I think of it as one of the famous roads that lead to Rome. For Costello, Noonan and CFC syndromes, all along the same pathway, it's like the traffic light at those particular intersections stay on too long ("gain-of-function"). This causes overflow and back-ups in the path ahead, and spilling into side streets in attempts to bypass the snarl. No wonder our children appear to have storage disorders!

Fortuitously, much is already known about Hras! For example, there are already medicines that stop Hras gain-of-function, so it makes sense to encourage researchers to see if they might help our children. This is by no means a quick process – the ideal would be for someone to design a mouse model to test the medications on, and that in itself takes a lot of work and resources. We're encouraging researchers in a number of efforts, and at our next conference, we hope to share their progress. Your presence and participation will do much to encourage them further. A research symposium on Saturday, July 21, will nurture dialog at the researchers' level of discussion. Parents and others interested in our children's wellbeing are welcome to audit these workshops.

This year, we're expanding our conference. Joining the workshops about our children's medical issues will be a full day of workshops about our children's non-medical issues, such as a round-table discussion on Individualized Education Plan strategies (what works, what to watch out for, resources for support), adolescent transition, estate planning, a sibshop session, and putting together a fabulous care notebook. This day of non-medical workshops will be on Saturday, concurrent to the research symposium.

Above all, bringing your child with Costello syndrome gives you and your child an opportunity to be around a bunch of people who have walked the same path (way). Our children will have a chance to spend time with people just like them, a truly rare treat.

To help make travel less of a barrier, our Conference Chair, Dawn Macready-Santos (mom of Marcella), has secured a 10% discount for plane tickets on Alaska Airlines and its sister airline, Horizon Air. For those who are financially fragile, Mercy Medical Airlift can help a parent and a child fly to the conference from anywhere in the US and parts of Canada. You'll find more about how to get to the conference inside this issue.

For those who need help with hotel expenses, Colin Stone, founder of all our support groups, webmaster and Executive Director of the International Costello Syndrome Support Group, has again offered to help some families defray hotel costs. This is on a first-come, first-served basis.

I appeal to those of you who have resources, please donate or help us fundraise! The more help we get raising funds for the conference, the more certain we'll be able to help bring families to the conference, families who we would otherwise not be able to meet.

I look forward to meeting you in Portland!

Ciao,

Lisa Schoyer

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IN THE SPOTLIGHT

Angela E. Lin, MD

Angela E. Lin, MD grew up in Philadelphia where she attended St. Joseph's University and Thomas Jefferson Medical College. After completing a residency in pediatrics (Children's Hospital of Pittsburgh), a fellowship in pediatric cardiology (UCLA) and fellowship in medical genetics (West Penn Hospital, Pittsburgh), she moved to Boston in 1990. Dr. Lin had always been fascinated by the type and pattern of heart problems in children with genetic syndromes, a lifelong interest which she pursues now as a fulltime clinical researcher. Dr Lin recalls the 1999 David Smith Meeting when she heard Dr Ginny Proud discuss Costello Syndrome for the first time. "I was so inspired, I had to learn more about the heart in this syndrome. I think I followed Ginny out of the lecture room and asked if anyone else in the group was working on this topic." Using data from the 1999 First Costello Syndrome Conference, she published a review about the heart with Dr Paul Grossfeld and several other geneticists and cardiologists, and presented it at the second conference in 2001 in Toronto. She continues to collect data on heart problems, providing updates at the biannual meetings. With Dr Karen Gripp, she has co-authored articles trying to correlate the presence of the HRAS gene to cardiac abnormalities in Costello Syndrome. Her interest extends to other syndromes in the "ras/MAPK family".



Dr Lin has worked closely with Dr. Gripp, and more recently, Dr Kate Rauen, and other geneticists to develop the medical advisory board. This was re-named the Professional Advisory Board in recognition of the broader membership. Dr Lin's career in medical genetics is currently in the field of birth defects surveillance, rather than clinical care. Since she is not actively seeing Costello syndrome patients, she sees her role on the Board as a facilitator between the parent leadership and professional board, helping parents and doctors network and providing professional peer education. On an ongoing basis she has tried to outline research goals for the group, and compliments her outstanding colleagues (Drs Gripp and Rauen) for their success in developing projects, publishing and grant funding. Dr Lin has worked closely with President Lisa Schoyer and members of the parent leadership. She feels strongly about the vibrancy of advocacy groups, and throughout her career has worked with several groups at both the local and national level. She has tried to study the role of professionals who interact with advocacy groups, and hopes to pursue that as future research. One of her goals is to encourage trainees in genetics to have some participation in a conference during fellowship.

"It's hard to explain to a medical colleague who has never participated in a group just what is the 'group experience'. To say this is a valuable experience is an understatement. Words like doctor and patient have been replaced by professional and consumer. What really matters are the relationships, learning, sharing and hopes. And don't forget the fun."

The 5th International Costello Syndrome Conference and 1st International Research Symposium

Doernbecher Children's Hospital at OHSU July 19-22, 2007
Portland, Oregon, USA



When:

July 19: Workshops
July 20: Genetic visits*, CSFN Board development (tentative),
Relax and get to know other families, and Family Fiesta Dinner
July 21: Workshops and 1st International Research Symposium
July 22: Workshops

Why:

Meet other families going through what you're going through!
An opportunity for your child to hang out with others just like him or her!
Learn the latest about Costello Syndrome!
Meet researchers who have been helping us get answers!

RSVP:

Dawn Macready-Santos or Lisa Schoyer
2737 SE 60th Avenue 244 Taos Road
Portland OR 97206 Altadena CA 91001-3953
(503) 788-8109 (daytime) (626) 569-6086 (daytime)
macready@pdx.edu taos@earthlink.net

Hotel:

University Place, Portland State University
310 SW Lincoln Street
Portland, OR 97201
(866) 845-4647
www.uplace.pdx.edu

Remember to mention Room Block Title: **Costello Kids**
and Room Rate (per night): **\$69+tax** Double Double (two double beds)

Seasonal heated pool; Pets are allowed, \$25 fee; refrigerator \$15; Rollaway bed \$15. Fees are per stay, *not* per night. Overnight parking is available for \$12 per night.

Air Travel:

Alaska Airlines / Horizon Air will provide a 10% discount. Go to <http://alaskaair.com> and input "ECCMR9341" in the eCert Code box when filling out the Flights Quick Search box. The discount will be applied to any flights that meet the criteria of the prearranged meeting code.

Mercy Medical Airlift can help provide one adult and one child with Costello syndrome to the conference by using donated frequent flyer miles. Contact Gene Smelser at mercymedicalpac@erols.com More information at <http://www.mercymedical.org>

* Supported by Doernbecher Children's Hospital, OHSU, and the Costello Syndrome Professional Advisory Board

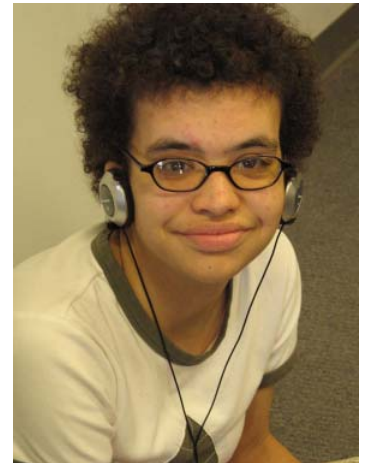
IN THE NEWS!!!!!!!!!!!!!!

Julie Cameron celebrated her 24th birthday on February 6th. Her only request for the day was to go to a local car dealership to meet her two favorite hockey players from the OHL Barrie Colts. She got her wish. The fellow on the right is our Team Captain and most recently played for Team Canada, Brian Little. The other fellow in the left is Julie's favorite - Hunter Tremblay, Assistant Captain. She went to High School with both of them. Needless to say, she was one excited girl on her birthday!



Keynote Speakers

for the First International Costello Syndrome Research Symposium: Kristin Carillo (below), Erin Hefner (middle), and Laure Messier (right).



On Saturday, three of our young adults with Costello Syndrome, Erin, Laure and Kristin, will speak about the syndrome from their viewpoint. Each is working on a short video tape that answers a few questions. Such as: When did you know you had Costello Syndrome? What bothers you about living with Costello Syndrome? How can others include you better? What do you want in life? What is your dream for your life?



Danielle Slawitschek raised \$200 for Jump Rope for Heart.

Photos of Erin, Laure and Danielle are courtesy of Rick Guidotti,
www.positiveexposure.org



more IN THE NEWS

We have had an interesting and fantastic year; Helaina was invited to Number 10 Downing Street for a tea party with the wife of the Prime Minister, Cheri Blair. Helaina is standing on the steps of the famous house. Never one to pass a photo opportunity she is smiling for the press at the other side of the road.

We had a fantastic party, with clowns, and magic, and presents and more presents, and food, and chocolate. Helaina loves chocolate. It was a day to remember, which ended with Helaina switching on the lights of the great Downing Street Christmas tree, and gathering with the other children under the glow of the frosty night sky singing Christmas carols with joy and happiness.



January turned out to be even more special than our trip to London. Helaina joined the scouts, and we looked in pride as Helaina joined the scouts.

Helaina in the old scout hut, with her amazing smile, standing proud and true.



Helaina may be shorter than the rest of the group, but her huge personality makes up for the limitations of height. She just loves to join in, and it rounds out her activities, from dance to drama, and now scouts. She has grown into the most amazing person, who lights up any room and has touched many hearts.

Lots of love,
Colin, Cath and Helaina

ASK CK

Welcome to ASK CK. ASK CK is our new Question and Answer forum. You can email questions to Lori Gerhardt at lori.gerhardt@costellokids.org.uk. Answers will be researched and answered by the CSFN Board.

Question:

Our doctor has diagnosed our child with Costello syndrome. We now need to know where to send her blood sample for the definitive diagnosis. Can you help us with this?

Answer:

You can access www.genetests.org for places people can get their child tested for HRAS. Click the “Gene Reviews” button. Type in “Costello syndrome” into the box. Click on “TESTING” to get a list of clinical labs that test for Costello syndrome. You’ll get a table that describes the services that each lab provides. Some labs can do prenatal testing. Take the information to your child’s doctor, who should be able to contact the lab that your insurance plan works with.

Another list of labs, research labs, can also do the gene testing, and will do it for free – if the lab agrees with your doctor that a Costello diagnosis is likely. On the Costello Syndrome, click the “RESEARCH” button to get to the next webpage, and then click on “Costello syndrome” near the bottom of the page. This lists the research laboratories who have contacted GeneTests.org.

The difference between clinical labs and research labs is that clinical labs are paid to provide the client the information. Research labs are funded to focus on the research study. Researchers aren’t paid to provide further explanation than if the sample tests positive for Hras.

Good news: you can either provide blood or a cheek swab for this test!

Nemours molecular diagnostic lab also offers confirmation of research results for Costello and CFC genes. The cost is \$100. For more information, please contact Lisa Schoyer at taos@earthlink.net.

Question:

What is the difference between Costello syndrome and CFC syndrome?

Answer:

Costello syndrome and CFC syndrome can look so much alike that sometimes it comes down to molecular testing. The mutation found in CFC is found in BRAF, MAP2K1, MAP2K2, or KRAS genes whereas in CS it is on the HRAS gene.

Dr. Gripp and Dr. Lin have a fantastic description you can access all about Costello syndrome at www.genetests.org. On the same page that has “TESTING,” “RESEARCH,” “REVIEWS” and “RESOURCES,” Click on “REVIEWS” and you’ve got it!

Dr. Rauen talks all about CFC and Costello Syndrome their similarity differences. Click on “GeneReviews” at the top of the page, and enter “CFC syndrome” in the box. Then choose “REVIEWS” to get to Dr. Rauen’s description of CFC. Scroll down to “Differential Diagnosis.”

Question:

I really feel isolated. I would really like to meet other families with a child with Costello Syndrome. Do you have any suggestions?

Answer:

The best place, and most fun would be to attend the 5th International Costello Syndrome Conference in Portland, Oregon July 19-22. The flyer is in this newsletter with all the information you need to come. Feel free to call or email Dawn Macready-Santos or Lisa Schoyer (info on the flyer) if you have questions. Also, join the listserve if you haven’t already. You can use it as much or as little as you want. Contact Colin Stone at c.stone@costellokids.org.uk or go to www.costellokids.org.uk to join! We welcome all question and concerns. 6

**We would like to thank and recognize everyone that has donated. The
Following gave donations from July 2005 to March 2007**

Under \$50 = amino acid

June Dargle in honor of Hunter Arvel
Michelle Foote
Chad & Gina Henrickson
Gail Herbert in honor of Hunter Arvel
David & Susan Lane
Ourso Flower Shop in honor of Hunter Arvel
Anna Sanchez in honor of Hunter Arvel
Daniel and Fran Zubik

\$50 to \$99 = nucleotide

Wendy & Jim Applegate
Adrienne Beck
Thomas & Karen Driessen
Karen Johnson
Susan Mentzer
Jerome Zubik

\$100 to \$499 = codon

William Caldera in honor of Luke Lydiksen
Casey & Molly Driessen
Gerald P. Driessen
Kathy and Bob Hefner in honor of Erin
International Costello Syndrome Support Group
Tammy Moore
Driessen Water I Inc.
Cyrus Miller & Lisa Barsa
Sue Sullivan

\$500 to \$999 = exon

Cintia Cuperman
Lori Gerhardt
Dawn Macready-Santos
Lisa Schoyer

\$1000 to \$2499= allele

Arvel Family
Driessen Water I Inc.
Cyrus Miller & Lisa Barsa

\$2500 to \$4999 = gene

Carla & Mark Morrison

\$5000 to \$9999= chromosome

\$10,000 to \$24,999 = DNA

Lee Johnson & Lisa Schoyer, in memory of Doreen
Weir Schoyer

\$25,000 and above = Nucleus

AI duPont Hospital for Children
St. Louis University

Thank You!



Costello Syndrome Family Network
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Altadena, CA. 91001
Phone: (626) 794-7343
taos@earthlink.net

Yes, I wish to support the Costello Syndrome Family Network. Enclosed is my contribution of:

Amino Acid under \$50____ **Nucleotide** \$50____ **Codon** \$100____ **Exon** \$500____
Allele \$1000____ **Gene** \$2500____ **Chromosome** \$5000____ **DNA** \$10,000____
Nucleus \$25,000____

This contribution is made in honor of _____.

Name_____

Address_____

City_____ State_____ Zip_____

Phone () _____

CSFN is a 501(c)(3) nonprofit organization
Thank you for your **TAX-DEDUCTIBLE** contribution!
A copy of our financial statement is available upon request.