



COSTELLO SYNDROME FAMILY NETWORK

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Letter From The President

Dear Families,

By now, you've probably learned that there's a gene test for a mutation for Costello syndrome! The mutation was first published by Japanese researchers on September 19, 2005. It turns out to be the gene that regulates the function of H-ras in our children's cells, a link to the way our cells bring information from the outside of the cell to the inside of the cell and the nucleus. Luckily, this is a well-known gene to cancer researchers. Our children hitched a ride with a very big bus!

Researchers are not yet sure if this is the only gene mutation associated with Costello syndrome. We are confident that a positive test for the mutation confirms that your child has Costello syndrome, but a negative test result doesn't rule out CS as a possibility.

For those who don't test positive for CS H-ras mutation, the mutations for a CFC syndrome gene as well as a second Noonan syndrome gene (although rarely affected in most cases) have been identified. Interestingly, the Japanese were originally looking for a second Noonan syndrome gene when they fell upon the CS mutation! The samples of CS genes were supposed to be a "control" for the Noonan gene they were looking for. Noonan syndrome had one already-known gene, but that accounted for only half (50%) of the children who were clinically diagnosed with Noonan syndrome. Now that we have a CS (HRAS), a CFC (BRAF, MEK1 and MEK2) and a rare second Noonan gene mutation (KRAS), we can untangle the confusion and identify MOST of our children with one of these mutations.

For all our children clinically diagnosed with Costello syndrome (including "probable" - a prefix Quin had until I came back from our first conference in Birmingham, Alabama!), I encourage you to get your child tested for the H-ras mutation. It's very important to know if your child has the CS mutation, the CFC or the newly found Noonan mutation, or none of them. Knowing for certain that your child has a specific mutation will lead to appropriate treatment that is not a matter of "if" but "when" - though exactly what can be treated is not yet clear.

If your doctor needs help to find a lab to test your child, www.genetests.org can help. It's organized by the NIH ((US) National Institutes of Health). These folks can definitely guide US families and may be able to help international families. The GeneTests.org site divides the labs into two categories:

- 1) Research
- 2) Clinical

There are pro's and con's for each.

1) Research Lab:

A researcher will provide the test for free, but we need to understand that their primary focus is to meet their research goals, based on the contractual obligation they have to the organization that funded their research. The focus will be for the greater good of all our children.

Research labs are not obligated to explain the implications of the results. That depends on what their "IRB-approved" research project is <http://www.hhs.gov/ohrp/irb/irb_guidebook.htm>, and what their agreement with you is. Fortunately for us, we have been working with researchers who have proven repeatedly to be exceptionally considerate of us parents over a number of years!

2) Clinical Lab:

A clinic does focus on your child. It can do the test and provide you with genetic counseling when the results come in. But your insurance plan may not pay for the cost. Fortunately, I believe we have strong resources to help a parent appeal if the insurance company denies the test - though we can't guarantee that it will lead to success.

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GUIDELINES FOR DIAGNOSIS

As reported by Dr Angela Lin, Dr Virginia Proud and CSFN

Major Features

- Dysphagia/feeding difficulty/gastrostomy tube (g-tube) (95%)
- Postnatal short stature (97%)
- Characteristic facial features (98%)
- Thick Lips (95%)
- Loose skin (94%)
- Abnormal palmar skin creases (99%)
- DD (developmental delay)/MR (mental retardation) (100%)

Unique Features

- Congenital heart problems (65%)
 - pulmonic stenosis (20%),
 - hypertrophic cardiomyopathy (40%)
 - atrial tachycardia (30%)
- Benign (44%) and malignant tumors (16%)
- Characteristic facial features with large mouth (78%)
- Stretchy skin with hyperpigmentation
- Kyphoscoliosis
- Engaging personality
- Curly hair
- Normal head circumference

Other Features

- Polyhydramnios (62%)
- Birth weight >50%
- Hernias (50%)
- Vision problems-ptosis and strabismus

"My Little Sister Maggie Has Costello Syndrome"

by Callie Ernst

My little sister who is two and a half has a syndrome called Costello. A syndrome is a set of symptoms and signs that indicate a presence of a disease, mental disorder, or other abnormal condition. My sister does not have a mental disorder, she is really just different. In the syndrome there is cancer, but my sister does not have it. This doesn't mean she will never have it, it means she doesn't have it at this time. On the internet my mother found Costello Syndrome. There is a girl in England who has the same syndrome. The girl's name is Helaina. Her family sends our family post cards. Our family gets a lot of data from them.

In the syndrome there is something called cardiomyopathy. The word cardio means heart. Cardiomyopathy is when the wall around the heart thickens which isn't a very good thing. Now we know it's in the syndrome so the doctors can look for it when my sister goes to the doctor's office.

Sometimes my sister has to have surgery, in fact just recently Maggie had to have surgery on her legs and had to wear casts on both legs for six weeks. The casts are all the way up to her thighs. Then when six weeks have passed the doctors can put on walking casts. Walking casts are casts that you can walk in.

My sister Maggie has had other surgery also. She had to have surgery on her stomach because she wasn't drinking enough formula. What they did was cut open her stomach and put in something like a balloon. Then the doctors put a tube in her stomach with a little plug you can see.

At night we have an electronic machine. Every week Chartwell Company drops off small packages of tubes in boxes and every night my mom or dad hooks the tube to the plug. Then there is a little bag that hooks up to the tube. My mom or dad put Pediasure into Maggie's stomach.

The syndrome affects our family in many different ways. We've had sad, happy and frustrating times. Sometimes I get frustrated myself because Maggie can't talk. Then I think Maggie can't talk but then I come up with an idea like I can still talk to her even though she can't talk to me. Before my sister was born I had four brothers, but now I have four brothers and one sister. My sister Maggie needs to know some sign language. Because of the syndrome she might not learn how to talk until she's four or older but the doctors don't know. It's really just a guess. The things Maggie can do in sign language are "turtle," "cow," "duck," "bird," "rooster," "teddy-bear," "horsy," "bunny," "doggy," "Barney," "sleep," "telephone," "fish," "no," "and "yes" which are just a shake and a nod of the head.

I've learned a lot from my sister. For example I've learned sign language, and that there are a lot of people out in the world who have syndromes like Costello that can't be treated because they don't have enough money or don't even have a home. So they can't pay for treatment. There are some people who don't have homes and their child has a syndrome that the parents don't know about and that can cause bad things to happen in the syndrome is not treated. Maggie is different and the same if you think about it because of the syndrome she is different, but Maggie as a child just like others and just because she has this syndrome it doesn't make her bad or strange. It just make her different and different is GOOD!!!!!!!!!!!!!!

We keep in our hearts
the family of *Fraser Francis Bryce*
May 6, 2003-February 5, 2004



Michael Dalpiaz lobbies for the right to receive waiver services at a rally at the Louisiana State Capitol in Baton Rouge. With him is his mom (Lois) and Representative Diane Winston. Michael, along with almost 15,000 other individuals, is on the Louisiana waiting list for services.

Bret Gerhardt showing off his self portrait at the county wide art show "Many People, Many Faces."

IN THE NEWS



Francis Collins (Founder of Human Genome Project & current head of NIH Human Genetics Research Institute), Dawn Macready-Santos, Lisa Schoyer at the American Society of Genetics meeting October 2005.



The 5th International Costello Syndrome Conference

Doernbecher Children's Hospital • Portland, Oregon, USA

When: July 18-21, 2007

- Wednesday, July 18: Clinics
- Thursday-Saturday, July 19 - 21: Conference

Why:

- Meet other families going through what you're going through!
- An opportunity for your child to hang out with others just like him/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.

**Maternal and Child Health Bureau
Division of Services for Children with Special Health Needs**

DEFINITION OF FAMILY-CENTERED CARE

Family-centered care assures the health and well-being of children and their families through a respectful family-professional partnership. It honors the strengths, cultures, traditions and expertise that everyone brings to this relationship. Family-centered care is the standard of practice, which results in high quality services.

PRINCIPLES OF FAMILY-CENTERED CARE FOR CHILDREN

The foundation of family-centered care is the partnership between families and professionals. Key to this partnership are the following principles:

- ◆ Families and professionals work together in the best interest of the child and the family. As a child grows, s/he assumes a partnership role.
- ◆ Everyone respects the skills and expertise brought to the relationship.
- ◆ Trust is acknowledged as a fundamental.
- ◆ Communication and sharing are open and objective.
- ◆ There is a willingness to negotiate.

Based on this partnership, family-centered care:

1. Acknowledges the family as the constant in a child's life.
2. Builds on family strengths.
3. Supports the child in learning about and participating in his/her care and decision-making.
4. Honors cultural diversity and family traditions.
5. Recognizes the importance of community-based services.
6. Promotes an individual and developmental approach.
7. Encourages family-to-family and peer support.
8. Supports youth as they transition to adulthood.
9. Develops policies, practices, and systems that are family-friendly and family-centered in all settings.
10. Celebrate successes.

Sources: National Center for Family-Centered Care. Family-Centered Care for Children with Special Health Care Needs (1989). Bethesda, MD: Association for the Care of Children's Health.

Bishop, Woll and Arango (1993). Family/Professional Collaboration for Children with Special Health Care Needs and their Families. Burlington, VT: University of Vermont, Department of Social Work.

Family-Centered Care Projects 1 and 2 (2002-2004). Bishop, Woll, and Arango. Algodones, NM; Algodones Associates

CSFN: FINANCIAL REPORT

By Katie Slawitschek: Treasurer

Current Balance: \$16,303.32

We would like to thank the following people for their generous donations:

Katie Slawitschek

Cyrus Miller

The following people made their donation in honor of **Luke Lydiksen**:

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Letter From the President

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While a positive test for CS (or CFC or the KRAS Noonan gene) finally puts to rest that last vestige of doubt for most of us, I ask that we all remember what it was like to go through this the first time as well as your wait period for the gene mutation test to be completed -- for the families who don't yet have a definitive gene-test answer. I'd like to reassure our families that CSFN will never turn away a family who benefits from our support. We'll be here for you until you find a better fit for your child's needs.

And finally, I hope you can come to our next conference, July 18-21, 2007 in Portland, Oregon! With all this new information, it's sure to be rich with information-sharing. Also, if you haven't had the pleasure of seeing your child hanging out with others like your child, it's priceless. How often does that happen?

Colin Stone, our international support founder, director of the International Costello Syndrome Support Group (ICSSG) and I can help you with fundraising and resources to come to our conference.

With excitement and wishing you patience and serenity,

Lisa Schoyer

Mom of Quin

President, CSFN



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

\$25 \$50 \$100 \$200 \$250 \$500

Other: \$ _____

This contribution is made in honor of _____.

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A copy of our financial statement is available upon request.