



CFC International
Cardio-Facio-Cutaneous Syndrome

The CFC Chronicle

Caring, Facilitating & Connecting

Volume XVI Number II

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CFC International 2015 Conference

- in conjunction with The Costello Syndrome Family Network & Rasopathies Network USA Meetings -

Seattle, Washington
July 15 - 19, 2015
DoubleTree by Hilton - Seattle Airport

To learn more about the exciting city of Seattle visit:
<http://www.visitseattle.org/Home.aspx#video>

See also Conference **Frequently Asked Questions** on page 10.

Local attractions:

- Visit Seattle's famous Pike Place Market, home of the flying fish
- View a 360-degree panorama of the city from the top of the Space Needle
- Shop at over 180 stores and over 50 restaurants at Westfield's Southcenter Mall, just minutes away
- Take in a Seattle Mariners game
- Explore the piers and boardwalks of Seattle's Waterfront



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To improve the quality of life through family support, research and education.

CFC International

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Computer Listserv: Judy Doyle

Newsletter Editor: Peter Hoedjes, The Netherlands

CFC International

183 Brown Road

Vestal, NY 13850 USA

(607) 772-9666

E-mail: bconger@cfc syndrome.org

Web page: <http://www.cfc syndrome.org>

We offer information, support, newsletters, an address directory, brochure and Parent's Guide. Our mission is to assist those whose lives are touched by CFC Syndrome and to improve lives through family support, research and education. The group is self-funded. Contributions are gratefully accepted and will help the next family to receive information about CFC Syndrome.

CFC International and any associated parties will not be held responsible for any actions readers take based on their interpretation of published or disseminated materials. Please review medical treatments and decisions with your own physician.

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Message from the Executive Director

Twenty-one years ago this month we started our trek to Philadelphia to find out what rare condition our son had been born with. It took our team three years to uncover CFC syndrome. At that time Clifford had a g-tube for feeding and many serious medical impairments throughout his body. We never thought he would be able to walk, talk, eat orally or sleep through the night. He has come such a very long way in life.

Cliff and I attended Clifford's graduation on Tuesday, June 3rd. Clifford had attended a public elementary school special education class up until middle school. At that point we enrolled him in the local special education school. At graduation he was given the Lifetime Learning Award for his interest in always asking and learning about things. Once he took off talking, he has never stopped and despite his lower level IQ his vocabulary is now up to a 10 year old child's. Our days of sign language, picture board and augmentative communication devices are long gone but I will never forget how we looked at any modality to get our son communicating.

Yesterday Clifford had his cardiology check up and remains stable. When done with the two hours of testing and an interview he told the echo technician "lady, I'm out of here!". As his sister Paige has said, "be careful what you wish for". I know that life is not easy for all of you parents and many of you grieve for what life should have been like. Keep your faith and even though your child's accomplishments may be small compared to their siblings or others, celebrate those little achievements since you and your child have worked hard. In a few weeks we will



celebrate Clifford's 21 years of life and his achievements with a graduation party. Of course the theme of this graduation party will be John Deere since this is what brings him such joy. I never in a million years could have imagined a tractor graduation theme party but life delivers many special surprises.

Brenda Conger

Donations

Thank you!

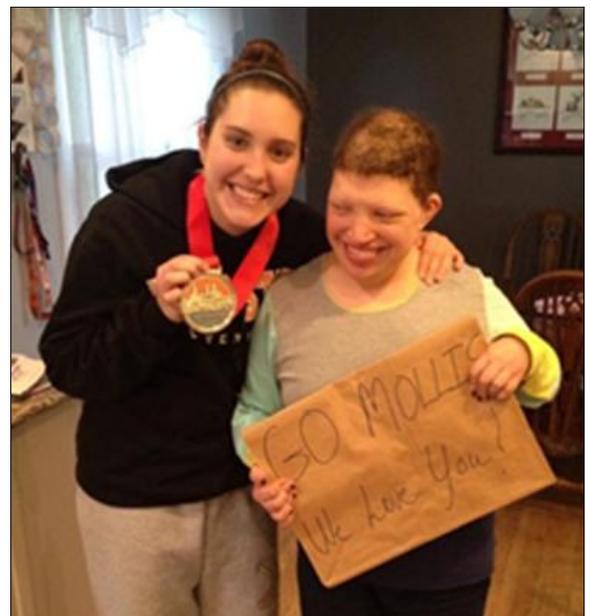
CFC International is grateful to its supporters for their generosity. We extend our deepest thanks to the contributors listed below for their kind donations.

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Sister Runs Half Marathon and Raises \$1,000 for CFC International

Mollie Scholl, sister to Maddie, age 24, ran in the Capital City Half Marathon on May 3, 2014 in Columbus, Ohio. Mollie raised \$1,000 with sponsorship funds directed to CFC International. Mollie attended the 2013 CFC conference in Orlando, FL to learn more about the rare syndrome that affects her older sister Maddie.

The 11th Annual Capital City Half Marathon attracted a record field of 14,500 runners and walkers. The Half Marathon, Quarter Marathon and 5K all started at the Town and Front streets in downtown Columbus and finished at the Columbus Commons, site of the Byers Xtra Mile Finish Line Party and Michelob Ultra Post-Race Concert. Way to go Mollie!



Mollie and Maddie Scholl celebrating the finish!

The Million Dollar Bike Ride

By Lisa Schill, RASopathies Network USA

A new rare disease research center, the Center for Orphan Disease Research and Therapy (CODRT), was created at the University of Pennsylvania to study rare diseases and help decrease the time it takes to get treatments from bench to bedside. CODRT created an event called the Million Dollar Bike Ride to raise money for rare disease research grants. They chose 15 different rare syndromes to participate and the RASopathies, which includes CFC, was chosen. CODRT will match what each rare team raises to create a grant that any researcher in the world studying the RASopathies can apply for.

Our RASopathies Network team was able to raise over \$30,000! CFC International and families raised \$3,544 of that. Ben from the CFC group and Matt from the Costello group graciously donated their pictures and stories and were each matched with a cyclist who rode in their honor.

The team captain, Lisa Schill, Vice President of the RASopathies Network USA, also participated in the event. When my son Max was diagnosed, I felt so alone. A doctor introduced Kelly Hudak and I via email since our sons were

diagnosed with the same rare disease. That was three years ago. Her son just went through open heart surgery. My son was about to have spinal surgery. We have stayed in touch with each other via Facebook, we have never met in person. We met each other for the first time yesterday. Our children were able to meet yesterday. Both of us don't have a lot of biking experience but decided to do the 77 miles together. It took us 7 hrs and 23 minutes. I wouldn't have been able to get through it without her and all those affected with the syndromes who sometimes go through so much more. I wouldn't have been able to through it without my team, some who I knew and others who I did not but all were willing to lend a hand and help. The Million Dollar Bike Ride raised 1.4 Million Dollars for rare disease research. Our team raised over \$30,000 that will be matched to create a RASopathies research grant. I will remember this day for the rest of my life. It is an experience that will always fill my heart with joy. Thank you to all who contributed. Thank you for taking the time to care. I am forever grateful.

We will be keeping you updated on the progress of this RASopathies research grant. The RASopathies Network USA is also going to be participating in the Million Dollar Bike Ride next year! Stay tuned on ways you can help make next year's event even better!!



Kohler Family Establishes Conference Scholarship Fund in Memory of Daughter Stephanie

Stephanie was our second child. She came in a hurry on a cold December day in 1986, three weeks early and just 45 minutes after we made it to the hospital. She just couldn't wait to start her life. That girl loved life, no matter what she was going through physically. She lived her life with zeal. She faced each and every day with a smile and that contagious giggle. She had the same challenges as many of the kiddos with this syndrome. She was startled by certain sounds, she had pulmonary stenosis and later developed a thickening of the mitral valve, she wore glasses, developed seizures when she was about eight years old, had nephrocalcinosis, and of course, constipation. Our lives revolved around poop, more than I had ever imagined was possible. If we weren't discussing poop, we were worrying about poop. If we weren't worrying about poop, I was up to my elbows in poop. I actually miss those days, go figure. Her heart never required treatment; it was the least of her problems, as her cardiologist used to say. We never protected her from the sounds that scared her but that most people wouldn't even notice. She went where we went. With time she got used to most sounds, she reached a point where very little frightened her. Her brothers helped her out in this aspect. The one thing that always made her cry was the sound of a child crying. Our girl worried about all the little children. She wanted everyone to be happy, as happy as she was. Her biggest medical challenge came from her kidneys. She developed lots, and I mean LOTS, of kidney stones. She went through one memorable and horrific six month stretch during which she passed 60 kidney stones. Of course, she passed many more stones throughout her years. She also would get dehydrated very easily and quickly, which turned our many travels into what we called the Great Hospitals of the United States Tour. As Stephanie taught us, you have to go with the flow.

As a family we liked to be on the go; we took many family trips. Stephanie "hiked" on her mommy's back in the mountains of Colorado, swam in the Gulf of Mexico in Florida, rode on the back of her dad's bike in Canada, fished and rode on a boat in Wisconsin (many times), went to a fish boil in Door County, Wisconsin, saw bison and Mount Rushmore in South Dakota, dug for diamonds in Arkansas, and camped many times in Missouri and Illinois. She was an adventurous girl. She loved to go fast. She knew one sign, which actually means "more" but to her it meant, "go".

She was quite bossy with her "go" sign. Sometimes I wondered what she'd have to say to us if she could speak. I'm sure it would have been bossy and so very funny. What a sense of humor she had! When she would go for a ride in her go-cart with her dad, he would strap her in her three-point harness in the passenger seat and climb into the driver's seat. She'd get a devious grin on her face and give him the "go" sign and off they'd go. Speeding down the bumpy gravel road, her hair blowing in the wind, that beautiful smile on her face and the sound of her laughter drowned out by the noisy engine. She LOVED a bumpy ride. It seems to me there's a lesson in her love of a bumpy ride and the great joy she derived from each and every bump. When life's road is bumpy and rough, don't focus on how hard or scary things are...maybe we need to be able to find the joy in the ride and reasons to laugh. Next time you find yourself going through a tough time, throw your hands in the air and laugh!...and think of our girl.



The Kohler Family

One of Stephanie's greatest joys was riding horses. She participated in a hippotherapy program for many years. The place where she rode was right alongside some train tracks. The train would go by during her lesson and, of course, blow it's horn. She quickly became accustomed to the noise and eventually didn't react in any way other than to put her hands over her ears while she continued riding. When Stephanie started classes, she could not get herself into a sitting position from laying down without help. One morning, we were amazed to go into her room and find her sitting up and laughing. She was so proud of herself. I know parents of children with CFC syndrome know what I mean, as they have seen that same pride on their child's face too. Stephanie benefitted in so many ways from riding that horse.

I would encourage all of you to find a hippotherapy program if there is one available near you. There really is some magic there.

Stephanie gave us the gifts of empathy, humor, and appreciation. After Stephanie, we had two more boys. Our three boys are all much more loving and empathetic having had Stephanie as a sister. We all love each other and appreciate each other more having had this experience.

Every day of Stephanie's life gave us a paradoxical combination of joy and challenge. At the end of the day we were tired, but content that we had survived another day. One day at a time.

This poem was written by Jeff after Stephanie passed away:

Imperfection?

Our precious girl was born, with too many challenges for one girl,
but she never thought she had them, with her cute smile and wavy curls.

What is perfection? Is it the beauty of a model or glitz of a star?

Or is it her capacity to love every human near and far.
Is it the perfect piece of art, the writing of a book?
Or the smile when you are hurting, because of mommy's worried look.

Is it the strength of the powerlifter, the boxer's crushing blow?

Or the loving caress to daddy's face, when he is feeling low.
Is it the Nobel prize, Oscar or the Mensas oh so bright?
Or the model for your brothers to know wrong from right.

Steph was an angel from the start, an angel without sin,
an angel that without fail could make the world grin.

She was so strong, so tough, she never showed her pain,
she got close to heaven when she stroked the horse's mane.

Our precious girl's body was frail, but her spirit was so strong,
God needed her back in heaven, he only loaned her for so long.

Imperfection is most likely what some might see at first,
an angel is what we saw, and now our hearts have burst.

"Imperfection" is not the word for her, but an apostrophe and space help change and link it.
"I'm perfection" is so much better, but of course our Stephanie would never think it.

Because being perfect is being humble, putting others ahead of you.

Being perfect is reserved by God for the paradoxically imperfect few.

Two years after we lost our sweet girl, Jeff and I attended our first CFC conference, in Orlando in 2013. We were anxious and unsure. Would it be hard to be around all the children and young adults who look so much like Stephanie? Children who look more like her than her own brothers do? We were expecting to shed many tears. We were pleasantly surprised. It was incredibly healing to be around the kids. It felt like being close to Stephanie again. We volunteered in the teen room. We thought we were there to help the teens but let me tell you, it was the other way around. What a fun group of teens and young adults we got to spend the day with!

The conference is a great mix of family medical resources, meeting other families and individual family fun. There are so many great families on this CFC journey. It feels good to be around people on the same journey with similar issues and challenges, with the same love of their precious kids. We wish we had been able to attend a conference with Stephanie, even if only for her to have an opportunity to meet other kiddos like her. We want every family to have the opportunity to experience all that the CFC Conference has to offer. That is why we have established Stephanie's Conference Scholarship Fund, in memory of our sweet girl.

Cooking up the Chili in Ohio



We hosted our 9th CFC Chili Bowl in Medina, Ohio on Saturday, March 1. It was the most successful one yet! We had 332 people, seven CFC families showed up, 40 chili entries, a ton of cool silent auction items and many raffle prizes. **We raised just over \$20,000!** So many CFC families helped out by sending items from their state like Renee Melvin (Harley's mom) sent a delicious Community Coffee basket that you can only get in Louisiana. Mimi Foster (Callie's mom), who is an author, donated 10 copies of her new book for a book club basket. Rosemarie Pavilonis (Michael's mom) drove all the way from Chicago and donated a Surface tablet. Danielle Goedel (Bryce's

mom) did a great job rounding up a ton of prizes, Coke products and friends to attend. We couldn't have done it without these wonderful women (and their husbands too). Since 2002 we have raised almost 100,000 dollars for CFC, mostly schlepping chili. It isn't very hard, just find something that your family and friends like to do and make a fundraiser out of it. I would be happy to help you out in any way that I can. I will even come to your event and help you! Not sure if you have noticed but if we the families don't do it, no one will.



We would like to see the funds from this year's Chili Bowl to be used for research on CFC syndrome.

To recap the evening we added a Restaurant Division and asked two of our favorite Akron restaurants, the infamous Sky Way and downtown's Lockview to compete. Lockview was the big winner and David Basone will be coming back next year to defend the title against a new competitor. In the Amateur Crockpot Division we had 40 entries. First place winner, Donna Schuett (aunt to Ava Hoy (CFC) age 4) got to take home the new traveling trophy, Karen Salmoni took 2nd place and 3rd place went to a combined team effort from Aaron McMahon and Craig Vavrock (first time attendees). We ended the night with music by String Theory (friends Kevin and Jeff) which was a great addition.

We are already making plans for next year. First thing on the to do list is find a bigger venue. Our family and some close friends have all stepped up to help us out. It is clear that this has become too big for the Doyle family to do on our own. We hope you'll come next year, Northeast Ohio is beautiful in March :

MARK YOUR CALENDAR: CFC Chili Bowl 2015 is Saturday, March 7.

It is not too early to start working on your recipe!

The Doyle Family, Medina, OH

CFC International Board Meets for Planning

The eight members of the CFC International Board of Directors convened over the weekend of May 2-4. Treasurer Kayla Stein graciously offered her home for the retreat since she lives outside of Detroit and this was a good central location for members to fly into. We had the added luxury of sharing pet poodle Cooper's lap time as we all worked hard! We addressed necessary document changes to move into compliance in New York State with the new laws coming into effect on July 1.



Back row: Judy Doyle, Kayla Stein with Cooper, Molly Santa Cruz, Jenny Iacobelli
Front row: Shelly Greenhaw, Pilar Magoulas, Luba Djurdjinovic, Brenda Conger

We have now set up different committees with board members heading these up. The following committees have been established: Research, Scholarship for family conference attendance, Social Media, Fundraising/Development committee, Volunteer committee, and Liaison/Welcome committee. If you are interested in helping out and being part of any of the organization committees please contact Brenda Conger at bconger@cfcsyndrome.org and she will put you in touch with the committee chair. We are also looking into a member survey to find out what we are doing great and what we can improve and do better. We have plans to launch this survey around the Seattle conference time. Watch for posts on our Facebook and private listserv for your much needed input.

Photo Gallery



Bredikis girls celebrate graduations- Mom's graduation from our local community college and Brittany cap and gown from high school



Clifford, age 21, graduates from BOCES PALS in NY



Michael, age 22, from Chicago graduates from high school



Izel, age 3, from California enjoying the water



Claudia, age 17, from New York at the playground



Ellyson, age 5, from Texas having a great time on her swing



Jared, age 17, from Utah



Eren, age 5, from Turkey enjoys summer fun



Lucian, age 3, from New Zealand



Kinley, age 12, celebrates her summer birthday in Oklahoma

Michael, age 6, from CT



Jeffrey, age 2, from Canada ready to hit the pool

Logan, age 11, & his sister Ash out on the lake in South Carolina



Alex Hirsch, age 16, with mom and dad posing for a prom picture



Rylee, age 2, from Georgia driving the backhoe at the Fun Factory



Clara, age 5, hanging out at miniature golf in North Carolina

Conference Frequently Asked Questions

Q. What can I expect if I attend the CFC International Family Conference?

Every two years we meet in different cities to listen to speakers, meet with researchers and medical experts, discuss concerns and problems with one another and introduce our families to each other. We learn from educators, researchers and medical professionals and mostly from each other. We strive to better understand our own children and what we can do to help them. We gain knowledge, perspective and hope. We also have a lot of fun, meeting new friends and checking in with old ones from past conferences.

Q. When is the conference and how long does it last?

The conference is July 15-19, 2015 at the DoubleTree by Hilton Airport, Seattle, Washington. We will hold a new family conference orientation on Wednesday evening July 15. We also have a Scientific update session on Sunday July 19, in the morning to hear from some of our medical advisors as to what was presented at the Saturday Scientific Symposium for the RASopathy meeting with all the scientists from around the world who have met on CFC, Noonan, Costello, and NF1. This session is an informal meeting so we are all updated on the latest research. You will also have the opportunity to ask questions.

Q. Who may attend the conference?

Everyone is invited to attend the conference. This includes parents, affected individuals, grandparents, extended relatives, friends and professionals. Many siblings also look forward to attending the conferences.

Q. Will childcare be provided?

Yes, we've always provided professional childcare. This is a considerable cost (\$8,000 for 60 slots) but the staff are insured, dependable and they bring in all the toys and activities. We anticipate a program that the children will enjoy with many activities to keep them busy and happy. We know personally that if parents know their children are happy, they can get the most out of the meetings. We can only allow CFC children and their siblings into this program due to space and cost. Space is limited to 60 slots and once that is

filled the childcare program will be closed out. Childcare will cost \$40.00 per child for two full days of childcare. We look for sponsorship for the majority of the bill so the \$40 per child fee is a small portion with the majority funded by a generous sponsor.

Q. How old are the children in childcare?

We generally have children of all ages - from toddlers to age 16 (CFC individuals). We have both typically developing children and our children who are affected with CFC syndrome in childcare. Our childcare services will combine CFC, and Costello individuals and their siblings as well. Infants are usually with their parents or extended family members and the adults take turns attending the sessions. Many times families bring grandparents or other family or friends to help out. Please note that we cannot and do not provide one-on-one childcare or nursing care. Children under age 12 months are not accepted in the childcare program.

Q. What about older CFC Individuals who need supervision?

We will have an age 16 and over teen and young adult lounge staffed by volunteers. Individuals in the lounge room will have movies, games, arts and crafts. The fee to attend the CFC Lounge is \$20 to cover all day on Friday and Saturday.

Q. What is included in the price of registration?

CFC International recognizes that our bi-annual conference is an extremely important educational and networking tool of our organization. Each family is responsible for their hotel room and parking; it is not a part of registration. The registration fee includes the group meals offered, the facility for our sessions, printed materials, audio video costs, and medical staff and speaker costs. For many years, we have charged each attendee a very nominal registration fee for the conference. It is our desire to make it possible for as many families wishing to attend the conference to do so. We will continue to do our best to make it economically feasible for our members to attend. For the first time we will offer a stipend program to first time attendees who are members. The stipend will cover a partial cost for the family to attend the conference.

Due to the generosity of our members and their friends who organize fundraisers or donate during the year, we are able to put on the conference & medical consult program

every two years (At a cost of approximately \$55,000 per conference).

Early Bird Registration fees have been set at \$110 per adult, \$50 for children ages 6-12; CFC children and all children 5 and under are free. Payment after April 15 is \$180 per adult and \$85 per child. Parking has been discounted from \$25 per day down to \$8 per day and \$10 overnight.

Q. Do I have to stay at the same hotel as the conference?

Being together at the same hotel is so important and lends itself to the very purpose of our conference. Most people secure their hotel room first. We encourage you to do this early. We have secured a set block of rooms at a reduced rate of \$129 per night plus tax. A rollaway bed is also available for a \$15 onetime charge during your stay. The Costello group will also be at this conference as well as the Scientific meeting so there are very few extra rooms to be located. Once the rooms are gone we are not able to obtain more rooms. We encourage you to reserve your room as soon as possible. Please register with DoubleTree by Hilton Seattle Airport by calling 1-800-222-8733 or by calling the hotel direct at 1-206-246-8600. **You must mention CFC International to obtain the reduced rate.**

Q. What is the Meet the Researcher/Expert program and who can attend?

The Meet the Research/Expert day is free of charge to all our registered CFC families who have joined CFC International through the website JOIN button. There is no fee to JOIN CFC International. This day is on Thursday, July 16. In the past only new families have been given slots but we have now expanded this program to all families and you will be able to choose what researchers you would like to meet with depending on how many slots we have open. This day is extremely busy for the medical teams involved and we set up folders on each child to be seen by the doctors. Some researchers wish to review the CFC child's folders the night before the consult. We set up a medical consult schedule for the conference a full two months in advance and only have so many slots for that one day. Medical consult spots are reserved on a first come, first serve basis according to the date we have received your paid in full completed conference registration form.

Q. What topics are included in the general sessions and workshops?

Our general sessions on Friday tend to give an overview of the syndrome and any new research updates. The smaller

breakout sessions on Saturday are joint workshops shared by CFC and Costello family members. You can visit the website and get a feel for what the conference will look like this year. As we get closer to the date, the schedule and more information about the sessions will become available.

Q. How do I register for the conference?

You register for the conference by going to our website (www.cfc syndrome.org) and finding the Conference under "News & Events" and then "Conference Information". The registration form should be printed out and mailed in with your payment in full or you can register online and pay with a credit card. No partial payments will be accepted and no holds on medical consult spots are granted until all paperwork with payment is received. We hope to have the registration available after January 1, 2015.

Q. If I am unable to attend the full conference, can I pay a reduced registration fee?

CFC International does not adjust the fee for days not attended. The registration fee remains the same whether you attend the conference for one, two, or three days.

Q. May I receive a refund if I am unable to attend the conference?

Cancellations received one month or more prior to the conference will be refunded less a \$25 administration fee per registration. No refunds will be given two weeks or less before the conference event. Refunds will be mailed after the conference and may take up to six weeks to process.

Q. A caregiver is coming to watch our kids. Does she pay the registration fee?

Does she plan to join us for any meals? Yes, she pays.

Does she want to sit in on one session? Yes, she pays.

Will she join us for the social on Thursday evening? Yes, she pays.

Does she want to socialize with other families in the conference setting? Yes, she pays.

Q. I am a teacher/therapist/researcher interested in CFC etc. and I would love to attend the conference. What is my fee?

All professionals are expected to pay the full registration fee of \$180.

Q. Will I be able to talk to doctors and specialists about my child/child's condition?

Many of our doctors and specialists are very willing to chat with you about your child, and discuss general aspects of current research. Please take advantage of this conference time to ask questions of the world's most knowledgeable experts on CFC syndrome.

Who do I contact with additional questions? Please e-mail CFC International at: info@cfcysndrome.org

A Conference in Our Own Backyard

Like most couples, Ernie and I definitely wanted children. He had a daughter from his first marriage but I had no kids. It was a long three years before I was able to get pregnant. We were even at the point where we had seen a fertility specialist and started the testing when we found out we were expecting. It was the happiest moment, I remember it like it was yesterday. It was 2:30 in the morning and Ernie had just gotten home from work, I had told him I thought I was pregnant but wanted to wait to take the test when he was with me. When it came out positive we both called and woke up our moms to share the news. My pregnancy was like most, morning sickness, cravings, and an occasional trip to the hospital because I was always dehydrated. You see I do not like water and just like my pregnancy, our son Stephen was born much like any normal child. My water broke at 5 am and we headed to the hospital. Once admitted they discovered he had pooped in my belly and proceeded to "rinse me out" shortly after, even though they told me he hadn't swallowed anything. I started running a fever and stopped dilating. Two days after Stephen was born and while in the hospital he showed signs of respiratory distress and was admitted to the NICU and seen by a cardiologist who noticed a heart murmur. He was also treated for possible sepsis even though tests later came back negative. He was in the hospital for 10 long days before he got to come home. Everything went well from there until we saw the cardiologist.

At a follow up cardiology appointment a couple of months later Stephen was found to have another heart condition called hypertrophic cardiomyopathy. When he was diagnosed with that we were whisked to a geneticist to find out where it came

from. Usually it comes from a parent but in some rare cases it is associated with a disorder or syndrome.

That journey started when he was four months old. At the geneticist we were sent to do blood test after blood test. At this point we really started to examine him and realized all the things that weren't quite right. He had/has a lazy eye (strabismus is the medical term) and he was much delayed and seemed to have not enough strength in his arms and legs. We were able to quickly get him into Early Steps, an early intervention system that offers services to infants and toddlers with significant delays. We were so blessed to also have been able to get him into a program in our area that not only did all the therapies he needed in house but all the caregivers took part in helping him progress.



The Ames Family

Low and behold 23 months after starting our journey the geneticist called with a diagnosis. It is called Cardio-Facio-Cutaneous Syndrome or CFC for short. It was discovered that he had a mutated gene MEK 1/2. Once we had the diagnosis it was both a worry and a relief. We finally had a answers, but also so many questions. How could we help our son? What do we do now? How will this affect his whole life? Ours? We started doing research and were directed to the CFC website. Once we joined it wasn't but two days later and Judy Doyle called me. She shared a bit of her knowledge of CFC and instantly I felt at ease. With one phone call I no longer felt

alone. I knew that there was support out there and that I could get to it. We joined the Facebook page and the parents Facebook group. The support and knowledge we got from these sites was so priceless. I could not give thanks enough to find others that were on this same journey and that I could not only learn from them but with our experiences I could also teach.

In 2012 we moved to Orlando, Florida and just two months after that it was announced that the 2013 bi-annual conference would be in our backyard. Instantly I knew that I had to be part of the group that makes these conferences so amazing. My husband works for a local sports bar and immediately began to gather sports memorabilia for the auction. I reached out to my contacts in the medical field to get as many samples as I could for the parent's bags because I had tried products that I wanted to give other parents access to. I became the local hub for all the shipments and organized all the parent's tote bags that were given out at the conference. Families also were able to ship to my house anything they wanted to donate to the auction. This made it extremely easy for most families as they didn't have to worry about traveling with items. I was also so happy to be able to take a load off the board, they didn't have to worry about getting the bags early days before the conference. I was able to get them ready weeks before.

At the conference I learned sooo much that I still cannot wrap my head around all of it. Since then, when I attended a session on feeding, I realized I needed to start concentrating on getting the nutrition Stephen needs. Rather than trying so hard to get him to eat I made him a special liquid diet that had all the calories he needed. Since then he has started to gain weight and even tried eating more things than before. Another session I attended was on sleeping, with this one I figured out that Stephen has restless leg syndrome. I took him to a sleep specialist and got him diagnosed. He has recently started medicine for this and it's helping but we are in the beginning so adjustments are still needed.

We also applied for an additional grant thru the United Healthcare Children's Foundation. We got approved for help in getting Stephen additional therapy and he now gets an additional hour of physical therapy a week. He is in public school and loves it. He gets one hour each of speech, occupational therapy, and physical therapy a week there. He loves his classmates and is the teacher's biggest helper.

Stephen is a wonderful, loving, talkative child who has a hard time expressing himself due to his delays. By looking at him you would never know the problems he faces. He sees multiple doctors: a neurologist, gastroenterologist,

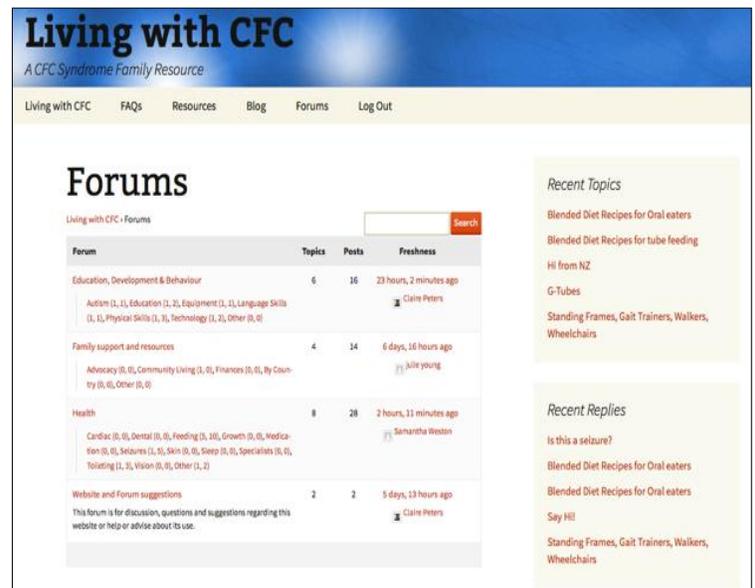
cardiologist, ophthalmologist, dermatologist, orthopedist, and now a sleep specialist. Our poor little guy knows scrubs and doctors coats more than most! And he does not like them. My grandest wish is to see my son walk. This has not been the easiest journey but thankfully we now know what to expect and how to handle it all.

Love,
The Ames family

Living with CFC: An Interactive Bank of Resources

By: *Claire Peters, New Zealand*

Attending our first CFC Conference in 2013 in Orlando, I became aware that many of the specialists didn't have the day to day answers I was after as a parent of a CFC child. These questions where in fact answered by other parents. It became a seed in my mind to create an unofficial library of information regarding living with CFC. While the Facebook group and email listserv work great to get answers to questions, it is hard to keep track of the information.



Living with CFC
A CFC Syndrome Family Resource

Living with CFC FAQs Resources Blog Forums Log Out

Forums

Living with CFC - Forums

Forum	Topics	Posts	Freshness
Education, Development & Behaviour	6	16	23 hours, 2 minutes ago Claire Peters
Auction (1, 1), Education (1, 2), Equipment (1, 1), Language Skills (1, 1), Physical Skills (1, 1), Technology (1, 2), Other (0, 0)			
Family support and resources	4	14	6 days, 16 hours ago Julie Young
Advocacy (0, 0), Community Living (1, 0), Finances (0, 0), By Country (0, 0), Other (0, 0)			
Health	8	28	2 hours, 11 minutes ago Samantha Weston
Cardiac (0, 0), Dental (0, 0), Feeding (3, 10), Growth (0, 0), Medication (0, 0), Seizures (1, 0), Skin (0, 0), Sleep (0, 0), Specialists (0, 0), Toileting (1, 1), Vision (0, 0), Other (1, 2)			
Website and Forum suggestions	2	2	5 days, 12 hours ago Claire Peters
This forum is for discussion, questions and suggestions regarding this website or help or advice about its use.			

Recent Topics

- Blended Diet Recipes for Oral eaters
- Blended Diet Recipes for tube feeding
- Hi from NZ
- G-Tubes
- Standing Frames, Gait Trainers, Walkers, Wheelchairs

Recent Replies

- Is this a seizure?
- Blended Diet Recipes for Oral eaters
- Blended Diet Recipes for Oral eaters
- Say Hi!
- Standing Frames, Gait Trainers, Walkers, Wheelchairs

'Living with CFC' is an interactive bank of resources and experiences for those who care for a child or adult with Cardiofaciocutaneous Syndrome. As CFC is so rare, care providers often have no direct personal experience of caring for those with CFC syndrome. Our aim is to help families connect and share information as well as build a bank of helpful experiences, advice and recommendations to help each other navigate their way through the CFC minefield.

On the previous page is an image taken of the forum website. Once you have registered and completed your login this is what you will see. The forum can only be accessed if you are a parent to an individual with CFC, or if you are an affected individual, creating a private place for us.

To register go to www.cfcfamily.info and click on the register page (top right) and fill out your details. An email will be sent to your email account to activate your registration so you can then login.

You can explore the forums by reading topics and replies to posts by other CFC parents or affected CFC individuals, post your own topics or reply to topics already created. For example: Health (forum) > Feeding (category) > 'Blended diet recipes for oral eaters' 'Reasons for vomiting' 'G-tubes' 'transitioning to solid food' and 'blended diet recipes for tube feeding' are topics created by CFC parents.

CFC International Restructures Board of Directors

Governor Cuomo of New York State has signed the Nonprofit Revitalization Act of 2013. It will enhance oversight efforts and tighten governance measures. It becomes effective July 1, 2014. The Act makes a number of significant changes to the law that will affect the day-to-day operations of both small as well as large not-for-profits operating in New York State.



Jenny Iacobelli

Under this new law, CFC International was required to make restructuring changes to be in compliance with the new law. Brenda Conger will maintain her leadership role as the organization Executive Director. Board member Jennifer Iacobelli was nominated as board President. Jenny has served on the CFC International board for the past two and a half years. Jenny works in the Department of

Global Environmental Affairs and Safety at The Estee Lauder Companies, one of the world's leading manufacturers and marketers of prestige beauty products with a family of 27

brands. As the Executive Director of Retail EHS she is responsible for administration and oversight of all Environmental Health and Safety Activities in Retail Operations including occupational safety and environmental compliance, employee wellness, and sustainability.

Jenny lives in Long Island, NY with her husband Darin and daughter Nola Rose (CFC – age 6).

Another restructure modification to the board is the newly created position for a voluntary Director of Science and Research. Board member Pilar Magoulas fit this position perfectly! With more and more research emerging on CFC syndrome it was evident that a sole person needed to work with researchers and our affected population's families.



Pilar Magoulas

Pilar is an assistant professor and board-certified genetic counselor in the Department of Molecular and Human Genetics at Texas Children's Hospital/Baylor College of Medicine in Houston, Texas. She received her Bachelor of Science degree in Psychology from the University of Florida and her Masters of Science degree in Genetic Counseling from Northwestern University. She is the Genetics Clinic Manager at

Texas Children's Hospital and also works as the in-patient consult coordinator. Pilar developed a particular interest in CFC syndrome during her graduate training after seeing a child with this condition in clinic. She has been involved with CFC International for the past 10 years and serves on the Board of Directors. She has loved being part of this wonderful community of children and families and will continue to work toward improving the lives of all of those affected by CFC syndrome and fulfilling CFC's mission. Pilar is married to Demetri and has two children, Sophia (age 5 ½ years) and Gabriel (age 3 ½ years), and enjoys running, spending time with her family, and watching football (Go Gators!) in her spare time.



A Special 16th Birthday Party Unites Teens who met years ago

Jack celebrated his 16th Birthday on Sunday, June 1st. We really wanted to do something special for him. We decided to throw a big surprise party since we haven't had a big party since he was little. Amazingly enough we pulled it off and he didn't know a thing. We had about 60 family and friends come to celebrate with us. Everyone was so generous and really put a lot of effort and thought into his gifts. He loves to cook so he received cookbooks, a ton of baking supplies and ingredients, a lettuce salad garden, an herb garden and even a golf cart/go cart, as we are still trying to find the best vehicle for him to safely ride.



Jack and Meg

her little sister Ella made the 14 hour trek from Missouri all the way to Ohio. Jack was so excited and had so much fun hanging out with Meg. A funny quote from each of them this weekend, Meg grabs Jack's hand and says "come on Jack, let's go" and Jack went, already doing what she tells him. Jack kept talking about how surprised he was "you distracted me by inviting my girlfriend up to visit". Meg will be turning 16 at the end of June, and we are hoping to return the favor and head down/over Missouri.

Tim had a great idea to invite the Young family from Monett, Missouri. Meg Young and Jack go way back, they have known each other since they were 2. They were both diagnosed in 2001 out in Utah at the first CFC conference by Dr. Opitz. They were so cute and have been sweet on each other ever since. Sherri, her mom, Meg and

What is Cardio-Facio-Cutaneous (CFC) Syndrome?

Cardio-Facio-Cutaneous syndrome is a rare genetic condition that affects approximately 1 in 810,000 people. It causes a range of physical, cognitive, and developmental disabilities that can range from mild to severe. It affects both genders equally and is seen in all races/ethnic backgrounds.

Common features found in CFC syndrome:

- A distinctive facial appearance
- Unusually sparse, brittle, curly hair (not seen in all cases)
- Skin abnormalities
- Heart defects that may be present at birth or develop later
- Growth delays
- Varying degrees of intellectual disability
- Vision abnormalities
- Seizures

What Causes CFC Syndrome?

CFC syndrome is caused by mutations (changes) in one of four different genes called BRAF, MEK1, MEK2 and KRAS. These genetic changes usually occur for the first time in the child, and are rarely inherited from a parent. Therefore, the chances of having another child with CFC syndrome would be very low. There is nothing that either parent did or did not do during the pregnancy to cause these changes in the genes; they just happen by chance.



CFC International
Cardio-Facio-Cutaneous Syndrome



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Our vision is a world in which no one will be isolated from appropriate diagnosis and treatment.

Would you like to contribute to CFC International?

Your donation and support will help:

- families to receive information about CFC syndrome;
- publish our newsletter;
- fund the biannual International CFC Family Conference & Clinic Program;
- fund critical research projects.

Enclosed please find a check in the amount of \$ _____

Gift in Honor of _____

Gift in Memory of _____

Please make checks payable to CFC International & mail to the address above.

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City: _____ State: _____ Zip: _____

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Email: _____