



**CFC**international

Cardio-Facio-Cutaneous Syndrome

# The CFC Chronicle

## *Caring, Facilitating & Connecting*

Volume XIII Number II

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### CFC Conference Event

The 6th CFC International Family Conference and Clinic Program was held at the Westin O'Hare in Rosemont, Illinois, from July 28th through July 30th. For the first time we combined forces with both The Noonan Syndrome Family Network and the USA Costello Syndrome Family Network.

I was thrilled to work closely with Chicago area parent and board member, Amy Hess. Many of our CFC family members helped, whether it was before the conference started or during the conference. Without them, we could not have coordinated such a successful event. We also could not have done it without the helping hands of several of our members and their friends who assisted at registration.



Ashlyn, Bryce, and Owen

This year we had 52 families travel to Rosemont from 23 different states and four different countries to attend the conference. Daycare was offered for the younger children and

siblings. We hired Kiddie Corp to run our daycare again and combined CFC, Noonan and Costello syndrome children and siblings for a total of 115 participants. It worked out great. The kids had tons of games to play or crafts to make. The Kiddie Corp workers enjoyed the opportunity to work at the conference and, of course, fell in love with our kids.

On Thursday CFC International and The Noonan Syndrome Support Group hosted clinic exams with our Medical Advisory Staff and also other researchers. Thanks to the efforts of our very own board member and genetic counselor, Pilar Magoulas, detailed schedules were developed with specific appointments for families. Pilar set up ophthalmology and genetic exams for 28 CFC individuals. Dr. Kent Reinker met with children for orthopedic exams with a sign up chart. All clinic station team doctors along with Pilar put in a non-stop day except for a short break for lunch. Researchers from all over were

also excited to meet with the largest group of CFC individuals in CFC International's history.

Friday morning held private general sessions with our CFC families hearing specifics on current research and what the future holds with the potential for clinical trials. The medical staff presenting and also participating in clinic kept mentioning how well versed this whole group of parents were who attended this conference. They were very much on top of their child's specific medical concerns and also very well versed on CFC syndrome.

On Saturday while the younger kids were being entertained by the professionals at Kiddie Corp, there were many courses available for the parents from all three organizations. Four to six sessions were offered for each time slot, so families had many choices. Sessions offered covered a wide range: Assistive Technology, Legal and Future Planning, Government Benefits, Genetic Testing, The GI System, Addressing Behavioral Issues, Growing Up with a RASopathy syndrome, and Parent to Parent discussions.

Saturday was also a special day for the siblings as they had private time with other siblings from all three organizations. Certified Sibshop instructor Winter Noe from the Costello Syndrome Family Network hosted the Sibshop program.

The conference ended with the traditional banquet dinner and silent auction. A fantastic buffet for all was followed by a video production of our family members and doctors engaged in activities throughout the weekend. Some of the kids started grooving to the piano version of Lady Gaga's "Born this Way" song. A suggestion was made that we add a dance party for our next conference and this is on our agenda of changes for 2013.

Brenda Conger

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*Forging a path to improve lives through family support, research and education.*



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### **CFC International**

183 Brown Road

Vestal, NY 13850 USA

(607) 772-9666 (Evenings and weekends)

E-mail: [bconger@cfcsyndrome.org](mailto:bconger@cfcsyndrome.org)

Web page: <http://www.cfcsyndrome.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.

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## Message from the President

With the September 7th start of the school year in the Binghamton, NY area no one could imagine how quickly all our lives would change within 24 hours. The students arrived for their first day that Wednesday but by noon there was a quickly approaching flood warning. The students and staff were dismissed early in what we thought would be a one half day off as the rivers and local creeks rose. I went to bed that night expecting to go into work at Binghamton High School the next day.

At 11:00 PM I heard my husband Cliff coming in to let me know he had gone down to his ski shop to check the basement water levels since his basement had flooded back in 2006 by ground water saturation from the nearby Choconut Creek. He now was fearful that he might lose the whole business if the water crested above the basement and into the showroom. He had come home with his business computers and the safe but was getting the van unloaded for another trip to evacuate what he could haul out. I quickly jumped out of bed and threw pants and a top over my pj's. We both worked side by side despite the police evacuation of the neighborhood at midnight. We finally had a report that the creek would crest and come rushing over the streets down at Front Street, Vestal. By the time we left the shop it was 4:00 AM. We hauled out as much as we physically could into our two vehicles.

After a couple of hours of sleep, Cliff was back up and trying to locate a truck to get out more inventory since the creek did not crest but the ground water had indeed started to rise due to the stalled hurricane Lee that kept pummeling our area with non-stop rain. Mike Schapiro and his son came to the rescue with a truck that even had a hydraulic lift. Brown Rd. neighbors Gregg and Jeanne along with a whole group of our friends also showed up with vans and trailers and worked around the clock to help save our inventory. People came forward and opened up their garages and storage areas to us. We will never forget the generosity shown to us during this time of need and disaster.

When the storm ended, the Cliff House Ski and Snowboard Shop had 1.5 feet of flooding in the first floor showroom as well as the whole basement. The walls and floor have been torn down and construction is currently underway. Two hundred pair of brand new ski boots have been washed and are stored out in our garage. A huge sale will take place once the business is back up and running again. As CFC parents, we have all learned the valuable lesson of how quickly life can change! We feel very lucky since we did not lose our home like many others in our area have. Just like with the CFC International parent group, we have also learned how the support of friends and family is so important. We could never have saved the amount of inventory if it was not for those relentless volunteers. Cliffy continues to pat his dad's back and say, "We're getting there." He has always seen the bright side of life and continues to amaze us with his caring personality.

*Brenda Conger*

E-mail: [bconger@cfcsyndrome.org](mailto:bconger@cfcsyndrome.org)

## 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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## The Doyle Family Cooks Up another Great Event

Once again, the Medina Eagles donated their banquet room. Giant Eagle was a sponsor again this year donating most of the extra food and beverages. Jim Shields covered the evening's beer tab and Pete Effinger supplied the cheese. Matt and Randi Clites donated, made and operated the Margaritarator. Kristine Effinger, Carol Ryland and many other great friends made delicious desserts and cornbread. Our family covered the rest of the incidental expenses, so the total amount from sponsors, donations, admission, chili



**Michelle Smith made good use of the MC Margaritarator by bringing it to the CFC Conference in Chicago**

entries, big screen TV raffle, raffle prizes, Margaritarator raffle and silent auction items went directly to CFC International. This year's total amount was just over \$14,000!

The chili competition was as hot as ever. 25 cooks entered for a chance to win the Chili Bowl's top spot. 1st Place went to first time entry and attendee

Dianna Rom, who graciously donated back her winnings. 2nd Place went to Karen Salmoni and I (Judy) actually won 3rd Place. Congratulations to all of this year's winners!

We appreciate everyone who donated raffle prizes and silent auction items. We had a great selection this year. Emma Ryland was the winner of the 55" LCD and Michelle Smith, mom of Kaci, won the MC Margaritarator. She was excited to bring it to the CFC conference this past summer in Chicago for Mom's Night Out where it was enjoyed by many



**Ava, Kat, Kaci, and Jack**

moms.

Jack had a great night. He was busy this year being our official videographer. Even though there are only approximately 300 known cases of CFC in the world, we were so fortunate to have four kids join us. Kat, age 16, and her family came in from Wisconsin, Stone, age 8, and his family came in from Youngstown, Kaci, age 16, from Massillon joins us every year and Ava, age 1½, and newly diagnosed came with her family from Pemberton, Ohio.

The Doyle Family  
Medina, Ohio

## United Way

Believe it or not, 'tis the season . . . for workplace giving campaigns that is. Many businesses around the country are gearing up for their annual giving campaign by teaming with an umbrella organization such as United Way (UW) or Children's Charities of America (CCA) to fundraise for many deserving causes. As a result, you, your family, friends, and co-workers may be able to support a specific mission (such as finding and helping children with CFC Syndrome) through a regular or one-time workplace donation.

If your workplace giving campaign (United Way) does not have an identification number for CFC International, you may usually "write us in" on the campaign giving form. If you are unsure whether your gift will be directed to us, speak with your local workplace campaign coordinator for assistance.

Workplace giving campaigns can be a great opportunity to promote awareness of CFC Syndrome and the programs and services our organization offers. Since many businesses create fundraising goals and offer incentives, much of the motivation and publicity for this fundraising event is done for you. If your employer has set aside space for the campaign, you may be able to display a photo of the person with CFC in your life along with information for co-workers. You might even volunteer to help coordinate your workplace giving program so interested co-workers would come to YOU for information about how they can give!

Workplace giving campaigns create a great giving atmosphere, just as you would want at a benefit dinner or tournament. A campaign focuses everyone's attention on giving and how they can take part. As a result, the "table is set." Drop a few words in the ear of your co-workers about how their workplace gift can help children with CFC Syndrome and you can raise some serious support.

A \$10 gift in every bi-weekly paycheck is a gift of \$260 per year; a gift that would make a huge difference to children with CFC Syndrome.

## Swimming the English Channel for Marcus and CFC International

Joe Hunter of North Yorkshire, England has a gorgeous 6 year old godson, Marcus, who was born with CFC syndrome. Marcus has been on an incredible journey so far, familiar to some, and has greatly benefitted from CFC International. Joe decided he would like to celebrate and recognize Marcus' strength, and thank CFC International, by attempting to swim the English Channel to raise money for the CFC charity. After 8 months of intensive training in local lakes, rivers and the freezing cold North Sea, Joe was given the date of 6th September 2011. Unfortunately the unpredictable English weather took a turn for the worse around this date and with the help of hurricane winds Joe's swim was moved to Friday 23rd September. Greased up with Vaseline and Sudocrem Joe made entry into the English Channel waters at 6am sharp on 23rd September. The water was cold at around 16:C (about 61 degrees F) but the sun was shining and the water was flat. His support crew of 4, in a tiny boat, sympathetically encouraged, fed/watered, and plotted the 22 mile way for Joe. After 14 hours and 10 minutes, lots of jelly fish, tankers and excrement, Joe touched the rocks of



Swimmer Joe

France. He had done it!!!... and he had done it 2 hours quicker than he had expected. A huge achievement for Joe and a very proud moment for him and Marcus to know they raised \$530.00 for CFC International.

## 9th Annual Toast the Angels

Despite horrific flooding in the Binghamton, NY region, dedicated supporters of the annual Toast the Angels wine tasting and dinner pleaded with the Conger family to not cancel the event. The event went on but with a much smaller crowd and less fanfare and excitement than in the past. This year the video production about CFC syndrome was eliminated and the silent auction was much smaller with local businesses trying to clean up from the flood or support



Clifford and the event volunteers

some of their best products. Chateau Lafayette Reneau and McGregor Vineyards both provided our guests with excellent wines with each course. The Conger family raised \$5568.00.

## Garlic Fest/CFC fundraiser

The Iacobelli family in Breinigsville, Pennsylvania love garlic. They love their cousin Nola too! So they put these two things together to have a stinking good time as they raised \$300 for CFC International in honor of Nola Iacobelli.



(From left to right) Logan, Owen, Haley, Benjamin, and Lyndsey Iacobelli made home made bead bracelets, conducted a 50/50 raffle, sold iPod raffle tickets, and sold "guess the garlic" jar tickets to their 150 garlic fest guests.

those who lost their homes. Still, the wine and food were excellent. Orlando's Catering put on a great six course spread and two wineries featured



## CREATIONS FOR A CURE

My name is Samantha Colleran. I live in Rockaway Beach, NY and two summers ago I had a shell sale on the 4th of July to make some spending money for myself. It was more successful than I thought it would be and I made about 65 dollars. That winter I saw a show on tv about kids doing some really great things to help other people and I thought that maybe if I had another sale I could do something good with the money I raised.



**Samantha with her shell display**

My aunt and cousin were visiting from England and they spent one of their afternoons here helping me collect and paint shells as well. My neighbor taught me how to make bracelets so I had more options to offer people on the day of the sale. My granny made a special trip to the craft store and bought me some other items like magnets and picture frames to paint. I also made little cards that explained what CFC was so I could give them out to people at school before it was over for the summer so they would remember to come. My brother Justin helped me with that and gave them out to everyone in his class.

My neighbor has a huge party every 4th of July that draws a pretty big crowd, which is why I have the sale that day. They very generously let me set up my table right at their party to make it easier for the guests to shop. I put some information out about CFC and made it available for people who had questions. I like to do this because not a lot of

people know what CFC is and I think it is important for them to learn about it. Everyone loved the pictures of Nola and I am hoping that one of these years she will make a guest appearance!



**A table full of bracelets**

This year was the best year yet! I made a sign that said CFC for CFC. It stands for Creations for a Cure. My neighbors Billy and Taryn started me off with a very generous donation. After that my table had a crowd all day. I ended up making a lot more people aware of CFC and raising a little over \$600! I am already looking forward to next year. I love you Nola!

**"When we can no longer change the situation, we are challenged to change ourselves."**

**Viktor Emily Frankel**



## Photo Gallery



Kinley Greenhaw & Meg Young



The Chapman family



Melanie, Shyanna, & Robert Trimm



The Santa Cruz family



The Thompson Family



The ladies group: Sarah, Jessica, & Rachel



The CFC kids club

## Greetings to our CFC Family from North Dakota

Let me introduce myself. My name is Cheryl Murch. My husband, Marty and I have 3 children. Megan, who is 19 1/2 yrs old; Casey, who is 16 1/2 yrs old; and Mandy, who is 15 yrs old. Mandy is our CFC child. She was my biggest baby at birth. From birth, she had no interest in eating and at one week of age, she started gagging and throwing up what little she did eat. So by 4 months, she was diagnosed with failure to thrive. At 3 months, an ASD was heard, which grew shut by 5 yrs old. At 6 months, I took her to the Mayo Clinic in Rochester, MN, to find out what was going on because by now she was noticeably delayed. I was told we would be at Mayo for 4 days, instead we were there for 10 days. I had to say that was enough testing at that point because they were not finding anything. Mandy did sit by herself at 11 months and walked at 3 yrs. Her gagging and appetite gradually got better over time. She has had PT, OT and speech over the years. Now she is just getting OT and speech.

She was thought to have Noonan Syndrome as a preschooler and the geneticist thought the same in early elementary age. She tested negative for Noonan Syndrome in 2008. So the geneticist moved on to test for CFC and that was positive in September 2009. It was such a relief to finally have a name for her issues.

I was disappointed that I had to wait to go to a conference until 2011. So I was happy when July 2011 finally arrived. It was such an undescrivable feeling when I walked into the motel, knowing there were other CFC families there also. After waiting for 13 years for a name with a diagnosis for her and then waiting for 2 more years to meet everyone, I was so glad to finally be in Chicago at our 1st conference. On July 28, Mandy and I attended ophthalmology, orthopedic, and genetic/cardiology exams. I wish we had more time for the genetic/cardio exam because there was so much information on both sides to cover. Mandy was also able to participate in the studies that were being conducted on Thursday. It was a very full day.

It was amazing to go to the Dessert/Welcome Reception on Thursday evening, to see the families that were there as part of the CFC, Costello, and Noonan syndromes. It was the first time I was able to talk with parents face to face that are dealing with the same issues that we have had with Mandy. I felt like I finally fit in somewhere in this puzzle of health issues called CFC.

July 29 started off with breakfast and then taking the kids to childcare that was set up in the basement of the motel. Mandy enjoyed her time in the childcare. Then we started with sessions that told us of the history of CFC, cardiology

research and what we have learned, physical activity and the musculoskeletal in CFC, the tissue bank, and moving forward and making progress. All of these were very informative especially since I had not attended a conference previously.

Mealtime was the best time to connect with other parents and meet their children. It was such a good feeling to be able to do this after so long without a diagnosis. I am envious of the children that were diagnosed at a young age because those parents have had the chance to connect to others and have support when it is needed for the difficult times and someone to share the joy when a goal is met. Starting Friday afternoon and continuing on Saturday, there were mini sessions to go to. It was hard to choose one which to go to, but as a plus, my parents came so we usually divided up to be able to hear more information. Then we compared notes later.



**Cheryl & Mandy Murch**

tools out there to help people with special needs. Or perhaps, part of it also, was the enthusiastic way the speaker presented the material. The one I learned the most from, was the session about special needs legal and future planning.

There was a group picture of the kids wearing their conference t-shirts on Saturday afternoon. It was so good to see them together as a group.

After the banquet on Saturday evening, I was finally able to connect with a couple of parents that have daughters Mandy's age. It felt good to visit with them.

Overall, the conference time went very fast, but I was thankful to be able to attend and connect with other parents. I can't wait for the next one.

See you in 2013!  
Cheryl Murch

While I got information from each session, I think the one I got the most from was the session on assistive technology. I didn't realize that there were so many



## International Meeting on Genetic Syndromes of the Ras/MAPK Pathway

July 29-31, 2011  
Chicago, IL

<http://ras-pathway-syndromes.com/symposium/>

By J. Jasper, Ph.D.

Research professionals and research physicians from around the world met concurrently with the Noonan Syndrome Family Meeting to discuss the latest findings in the cellular signaling pathway called the “Ras and Mitogen-activated protein kinase (MAPK) pathway” and put these findings into context with specific syndromes associated with alterations in this cell signaling. The syndromes include Noonan syndrome (NS), LEOPARD syndrome (LS), Costello syndrome (CS), cardio-facio-cutaneous (CFC) syndrome and neurofibromatosis (NF1). All of these syndromes share certain features such as alterations in the cardiovascular system, and skeletal and skin changes. The family groups for CFC Syndrome, NS, CS and NF-1 were represented in a presentation by J. Jasper that included videos and slides. On the second day of the meeting there was a panel of discussants from each of the groups: Representing CFC were Molly Santa Cruz and Rosemarie Pavilonis while Martha Goodwin and Darcie Robinson represented the NS group. The researchers asked various questions of the panel ranging from medical questions to how the economy and political atmosphere might affect their families in the future.

Below are brief summaries of some of the scientific presentations.

**Leslie G. Gordon, MD, PhD** was the keynote speaker: **“Progeria, aging and translational medicine: From Obscurity to Treatment Trials and Beyond.”** Dr. Gordon is an Associate Professor at Brown University and also co-founder of the Progeria Research Foundation. Although Progeria is not a disease of the Ras/MAPK pathway, the story of how Progeria became well-known in the scientific community and now has potential medical treatments is relevant to our groups. Dr. Gordon described Progeria as a rare premature aging syndrome that affects about 200-250 children worldwide. It is now known that the condition is the result of a congenital mutation of the LMNA gene. Children with Progeria typically die of atherosclerosis by an average age of 13 years. Because of recent discoveries, there

are now ongoing clinical trials for these children which offer hope.

**Helene Cave, PharmD, PhD: “Germline Mutations of the CBL Gene: A New RASopathy With Predisposition to Juvenile Myelomonocytic Leukemia (JMML).”** The CBL gene codes for a protein that modulates growth factor receptor signaling in cells. CBL gene mutations were known to account for a number of patients having JMML. We now know that germline (before birth) alterations in this gene affect fetal development resulting in features reminiscent of NS or NF1. Thus, this might be considered as a newly discovered gene responsible for about 1% of people with NS.



Dr. Jeff Jasper and Dr. Jacqueline Noonan

four unrelated patients (out of over 900 individuals with NS but no identified gene alteration). Thus, NRAS mutations account for less than about 1% of NS. These subjects seem to have a relatively mild clinical expression, but there are too few people diagnosed to date to make many conclusions about the ‘genotype/phenotype’ correlations (that is, what gene alteration causes what kind of changes in a person’s body).

**Amy E. Roberts, MD: “Ras/MAPK Disorders: Genotype Phenotype Correlations.”** Dr. Roberts gave a nice overview of the different syndromes in the Ras/MAPK pathway and the corresponding gene changes that are known, to date. Alterations in the proteins coded for by these genes can change the way cells work and can lead to numerous health problems. Thus far, eight genes have been shown to cause NS (or closely related conditions): PTPN11, SOS1, KRAS, NRAS, RAF1, BRAF, SHOC2 and CBL. By knowing which gene alteration a patient has, one can sometimes predict their “phenotype” (e.g. cardiovascular issues, growth, development, skin and blood features). This can aid in risk assessment and management of these health problems.

**Martin Zenker, MD: “NRAS Mutations- A Rare Cause of Noonan Syndrome.”** Dr. Zenker described an international research collaboration that led to the discovery of NRAS

mutations in

CFC Syndrome includes a number of issues such as failure to thrive and short stature, congenital heart defects and a characteristic facial appearance. There is a lot of overlap between characteristics of people with CFC syndrome and NS. CFC is due to mutations in four known genes: BRAF, KRAS, MEK1 and MEK2.

Costello Syndrome has features of skin abnormalities, short stature and heart problems – similar to CFC and NS (but possibly more severe). Mutations of the gene HRAS have been identified as the cause of about 85% of the cases of CS.

**Rene Pierpont, PhD: “Neurodevelopmental Profiles for RASopathies.”** A great deal of work has gone into establishing neuropsychological profiles for people with Ras/MAPK pathway syndromes. Cognitive function can vary from significant intellectual disability to merely mild delays in specific areas or no significant learning or behavior problems. Research using animal models is helping researchers understand the neurobiological basis for differences in learning and memory processes in the “RASopathies” and may suggest potential ways to minimize/treat these differences. A lot of different factors influence how a person might respond to treatments including the age at intervention, presence of medical risk factors and the baseline level of functioning. Work is ongoing to validate different neuropsychological measurements to help with clinical trials. Dr. Pierpont was optimistic that there will be ways to positively help individuals with these various syndromes.

**Katherine A. Rauen, MD, PhD: “Skeletal Muscle Pathology in Costello and Cardio-facio-cutaneous Syndromes: Developmental Consequences of Germline Ras/MAPK Activation on Myogenesis.”** Essentially all of the RASopathies have some characteristic hypotonia (low muscle tone), but CS and CFC syndrome are typically more severe. Dr. Rauen’s group is investigating the molecular mechanisms for these issues by studying skeletal muscles. People with these conditions have abnormal muscle fiber size and variability; they also have a predominance of ‘type 2 muscle fibers’ (these type 2 muscle fibers are the so-called ‘fast fibers’ because they help your body make quick movements, but use more energy and get tired more easily). Using some special muscle cells that grow in a Petri dish (called C2C12 cells) they found that if they induced CS and CFC mutations in these cells that the cells did not grow the same way. The group is now starting to look at special CS and CFC mouse models to help to better understand the changes in skeletal muscle with the hope that potential treatments might be possible.

**Ben Neel, MD, PhD: “Noonan Syndrome-associated Raf1 Mutants With Increased or Decreased Kinase Activity Differentially Activate Erk and Cause Distinct Syndromic Phenotypes.”** Germline mutations (before birth) of the RAF1 gene account for ~3-5% of Noonan Syndrome. Alterations that increase the activity of RAF1 increase the likelihood of Hypertrophic Cardiomyopathy (HCM). They found that mutations that either activate RAF1 or inhibit its biological activity both promote a condition called heterodimerization with BRAF. This heterodimerization appears to lead to many of the NS characteristics. Interestingly, in mouse models, they found that drugs called “MEK inhibitors” (previously designed as anti-cancer agents) could normalize the NS defects in at least one of the mouse models. Their data suggest that knowing a person’s genotype might help to develop a mutation-specific drug therapy.

**Alcino J. Silva, PhD: “Mechanisms Underlying the Cognitive Deficits in Animal Models of Rasopathies.”** Dr. Silva’s lab studies the molecular and cellular mechanisms responsible for learning deficits in the Rasopathies. They found that two common gene mutations found in the PTPN11 gene associated with NS caused deficits in spatial learning and long-term potentiation (memory) in mice. Interestingly, when the researchers treated the NS-like mice with an experimental drug called SL327, a “MEK inhibitor”, the alterations in spatial learning and memory were normalized. These findings open up the possibility that some NS individuals might benefit from certain types of drugs designed to work on the Ras/MAPK pathway. They are still a LONG way off from a therapy for humans, but this is encouraging.

**Bruce Gelb, MD: “Induced Pluripotent Stem Cells.”** Dr. Gelb described that there are many techniques for generating animal models to study Rasopathies including CFC and NS (such as fruit flies, zebrafish and mice) but there are potential advantages to studying diseases in human cells. Dr. Gelb’s lab has developed “induced pluripotent stem cells” (iPSCs) from human skin fibroblast cells from individuals with Ras/MAPK mutations. These cells might be great tools for better understanding what is happening in human cells with these gene alterations.

Also, Dr. Gelb’s lab and Dr. Ross Cagan’s lab have been working together to set-up fruit fly models of Ras pathway diseases. With these special fruit flies, the researchers are even doing high-throughput screening to search for new drugs for these conditions.



## Exciting New Study ~ Looking for Adults with CFC

Cardio-Facio-Cutaneous syndrome was first classified as an official syndrome in 1986. At that time, information was gathered regarding the common characteristics of 8 affected individuals. That initial study, along with others, helped to define a set of characteristics that are now used to clinically diagnose individuals with CFC. Since that time, both the number of individuals diagnosed with CFC and our understanding of what causes this syndrome have grown significantly.

Twenty-five years has passed since CFC syndrome was first described. Many of our children and loved ones have transitioned or are beginning to transition into adulthood. While much is known about how CFC manifests in newborns and young children, far less is known about the characteristics common to adults with CFC syndrome. You may be asking yourself, "What can I expect as my child enters their adult years?".

A team of researchers from the University of California San Francisco (UCSF) Comprehensive Cancer Center is interested in answering this very important question. This research group, led by Dr. Katherine Rauen (medical geneticist and CFC Medical Advisory Board member) is the same group that discovered the genes responsible for CFC syndrome back in 2006. As part of their long standing commitment to CFC research, they are currently interested in identifying the changes that occur as CFC individuals transition into adulthood. Dr. Brandi Thompson (cellular and molecular biologist), a member of the Rauen laboratory, is currently conducting a study to further define the common characteristics of adult CFC individuals. They hope that this study will not only add to the natural history of CFC syndrome, but also serve as a guide for future patient care.

Parents/caregivers who are interested in this study will be asked to complete a short survey for their child. The survey will be given in the form of a phone interview and should take less than 30 minutes to complete. You may also be asked to provide copies of some medical records. Because this is a study focusing on older individuals, the UCSF team asks that those that are interested in participating be 16 years of age or older. If you would like to participate or would like more information, **please contact Dr. Brandi Thompson by email ([thompsonb@peds.ucsf.edu](mailto:thompsonb@peds.ucsf.edu))**. Please contact her at your earliest convenience.

## CFC Says Goodbye to a Founding Board Member

Hi Everyone,

Effective at the end of the 2011 Chicago CFC Conference I stepped down as a CFC Board Member. My ski business is consuming more of my attention and I am enjoying doing more things with Cliff so I wasn't able to fulfill my duties as a board member. I have been involved with CFC from the beginning of our nonprofit incorporation in 1999 and this decision was not taken without a lot of thought and was discussed at length with Brenda. I am confident that CFC International is in good hands as we march on trying to improve lives of those born with CFC syndrome. I will still be visible and active at the CFC Conferences and helping out wherever needed.

Thank you for the privilege of meeting and working with you all.

Sincerely yours,  
Cliff Conger

## CFC International announces new Board Member

**Jenny Verrine Iacobelli** 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 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 3). Nola was diagnosed relatively early with a BRAF mutation in 2009 at the Children's Hospital of Philadelphia after 9 months of uncertainty and struggle. Fortunately 6 months later the Iacobelli family was able to attend their first CFC conference in Berkeley, CA where they met amazing families who let them know they were not alone. It was an overwhelming but magical experience and she is looking forward to serving CFC International as a board member.



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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;
- maintain the CFC Tissue and Biobank, which is critical to future research.

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

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

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