

## The origins of CFC

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Duke University Medical Center*

Unlike syndromes named for a particular doctor or even for a particular patient Cardiofaciocutaneous syndrome (CFC) gets its name from the parts of the body that were described as having differences: the heart, the face, and the skin. Why does it have this name? Who named it?

CFC has deep roots in the medical specialty of clinical genetics, and so we begin the story with the end of polio. In 1955 a vaccine against poliomyelitis was introduced, with 450 million doses administered in the United States over the next 4 years. A polio vaccine became standard among childhood immunizations, with its total eradication in the Americas in 1994. This work was funded in a remarkable way. President Franklin D. Roosevelt, himself a victim of polio, established the National Foundation for Infantile Paralysis in 1938. A celebrity of the day, Eddie Cantor, coined the phrase “March of Dimes” appealing to Americans to send dimes directly to the White House. This campaign was the birth of the organization we call the March of Dimes Foundation today.

In 1958 the “expanded program” of the March of Dimes was announced. This would re-focus the foundation on birth defects prevention. It was an exciting time when (in 1951) it was shown that altering the diet of a child with phenylketonuria (PKU) could prevent mental retardation. Newborn screening for PKU became possible on a national scale in 1966. There was hope that birth defects might be eradicated too.

In 1968 the first Clinical Delineation of Birth Defects meeting was hosted by the March of Dimes and The Johns Hopkins Hospital. This important meeting brought together doctors interested in preventing birth defects and mental retardation. The first step was to systematically catalogue what they saw in Institutions and in their clinics. Begun in 1966, Dr. Victor McKusick from The Johns Hopkins Hospital published this catalogue, which is now continually updated and publicly available as the Online Mendelian Inheritance in Man. Today we recognize this as

one of the most important foundations upon which the field of clinical genetics would be built.

The doctors who attended the March of Dimes Birth Defects Conferences in the late 1960s – early 1970s became the first generation of clinical geneticists (recognized as a specialty by the American Medical Association in 1991). They spoke frequently with one another, visited each other’s practices, and learned much from their collaborative efforts. From this group, or their disciples, seven doctors would write the first paper coining the term Cardio-Facio-Cutaneous (CFC) syndrome. They were Jim Reynolds (in Helena, Montana), Giovanni Neri (in Rome, Italy), Jürgen Herrmann (German-born, working in Wauwatosa, Wisconsin), Bruce Blumberg (in San Francisco, California), James Coldwell (in Tulsa, Oklahoma), Paul Miles (in Twin Falls, Idaho), and John M. Opitz (German-born, working in Helena, Montana). To be fair, we note that case reports of children with what would be called CFC today were previously published.

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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

2006 was a banner year for CFC International and the scientists and clinicians who service our affected family members. Not only did we all celebrate the gene discoveries but then received the exciting news that the NIH funded R13 grant had been approved to bring even more great minds together to keep the research pace moving along.

As we continue to look forward to even more advances, I am pleased to announce that recently our CFC International BioBank has been transferred to Gene Logic in Gaithersburg, Maryland. Making the move to Gene Logic allowed all the Genetic Alliance BioBank member groups to step up to the next level in research. We will now have the ability to not only track DNA we deposit but Gene Logic has provided a Tissue Repository Information Management System (TRIMS). What this means is that all our medical records can be entered into a computer data entry system and we will have secure and safe storage using over 1,000 fields. This project will take time to complete but eventually researchers with approved IRB approved research projects can access information on specific concerns to better understand CFC and the evolution of each individual's medical conditions. We truly are entering a new remarkable stage in research. I can't begin to thank all our families and donors who have supported us to make this project become a reality! Without all of you, we would not be at the stage to even support research today.

We will continue to add more DNA and clinical data to our CFC International BioBank at the Orlando, Florida conference this July. Even if you cannot attend but wish to enroll your child in the BioBank, we can easily assist you. Please send any requests to [info@cfcsyndrome.org](mailto:info@cfcsyndrome.org)

I look forward to meeting many of our newly diagnosed CFC children and their families this July in Orlando as well as catching up with our other friends from past conferences.

*Brenda Conger*

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



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### 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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## A PERFECT UNION OF THREE!

Jay and Jenn Merithew (cousins of Luke Lydiksen) were married in October and chose to make CFC Syndrome a part of their special day. In lieu of giving traditional wedding favors to each guest, a generous donation was made to CFC International. A beautiful scroll was at each seat and upon opening the scroll, a handsome picture of Luke was on it and a detailed description of CFC Syndrome and what our organization does. Jay and Jenn felt that this was a great way to spread the word to all their guests and invite them to visit the website to learn more and hopefully inspire some to donate to the organization.



Newlyweds Jenn & Jay

## 2nd Annual CFC International Golf Tournament

We waited in anticipation for the day of our 2nd Annual CFC International Golf Tournament to arrive. The weather report called for rain and possible flooding. They could not have been more wrong. The weather was absolutely perfect!



Ronnie and dad at the registration table.

Our players arrived at 7:30 am to hot coffee provided by Port City Java and tasty baked goods donated by Great Harvest Bread Company. The event, which was

held on Saturday, October 28th, at Olde Point Country Club, raised over \$8,500.00 to benefit CFC International.

Thank you to all of the volunteers that assisted in the golf tournament. This would include Sarah Benton, who was our event photographer. Our “beverage girls” were Angie Thomason and Rachael Riebe. Daryl Stahl helped with sponsors and prize donations. My sisters helped with organization and made our signs: Kayla Stein came all the way from Michigan and Jo Cassidy from Alaska! Clare Weingarden made calls from Toledo, OH to prize sponsors.

As always I would like to offer a special thank you to Joyce and Edwin Benton. Edwin was our AV coordinator and honey-doer and Joyce is...remarkable. Joyce Benton is Ronnie’s caretaker and certainly his best friend. Without her I am not exactly sure how this event or my life would turn out. Thank You Joyce!!! I would also like to thank all of our sponsors. I extend a special thank you to our family sponsors. Your children’s pictures made such an impact on or participants.

After a wonderful meal that was prepared by Capt’n



Ronnie cruising around with Aunt Kayla

Bill’s Backyard Grill, we watched a Power Point Presentation that enlightened many of our participants on the details of CFC Syndrome. Following our presentation,

we awarded prizes to the winners as well as individual prizes: Men’s and Women’s closest-to-the-pin, Men’s and Women’s Longest Drive and a putting contest prize.

Thank you to all of our participants!  
The Zeunen family, Wilmington, North Carolina

## Parents Raise Money with Pillowcases

Della McVay and her friend Jeanne Marie Trottier from Toledo, Ohio made pillowcases to sell to raise money for CFC International. Della donated the fabric and Jeanne did the sewing. They set up their displays at local church craft sales, offices where friends and family worked and also promoted their handmade pillowcases through word of mouth. Della and Jeanne raised a total of \$410. Thank you crafty ladies for your clever fundraiser and the donation!

## Sun Coast Cruiser's Raise Funds for CFC International

We would like to thank Wilmington, NC Sun Coast Cruisers!!! Due to a simple request from Ronnie Zeunen's best friend Joyce Benton, we were put on their list for a fundraising event in honor of Ronnie to raise money for CFC International.

The event was held on Saturday, October 28, 2006, in a local Arby's parking lot. The event had a Halloween theme, which is a tradition of the Sun Coast Cruisers. Car enthusiasts came from all around to attend the event.

Prizes were awarded for creative costume as well as the best dressed automobile. There was music, food, dancing, and of course a 50/50 raffle that had many participants. The Wilmington Sun Coast Cruisers raised a whopping \$242.00 in only a few short hours.

We would also like to thank Paula Clearwater who generously donated her portion of the 50/50 raffle back to CFC International. Paula has had some medical issues to deal with and without a blink of an eye handed us her \$242.00. She asked that we combine it with the Wilmington Sun Coast Cruisers contribution to make a total of \$484.00. Thank you Paula, and our prayers go out to you for a full recovery.

We had a wonderful time and hope everyone who either attended or helped put the event together enjoyed it as well. We appreciate you all!!

Diana Zeunen  
Wilmington, North Carolina



Ronnie and Paula Clearwater

## Grandparents With a Goal

Bobbie and David Olsen became involved with CFC International after attending the 2003 Maryland conference along with their CFC grandson Daniel Hess and his parents. The Olsens will be attending their 3rd conference this coming July in Orlando, FL. As annual support of CFC International they have proposed a unique challenge for this coming conference. They will match dollar for dollar any money raised through the conference silent auction. What a great way to encourage all of us to raise money to offset this conference. Even if you are not attending the conference you can ship an item to be auctioned off. Thank you Bobbie and David to getting us all off to a great start!

## Chili Feed Fundraiser a Winning Event in Wisconsin!

The Lindgren family hosted a Chili Feed fundraiser in honor of their daughter Sarah and CFC International on November 12, 2006. Chili, crackers, cheese, bread and cake were served to 125 guests. Door prizes were given away to people who bought tickets for the meal. In addition, friends and business in the area donated items for the raffle. Family friend Kit Riewestahl played a major role in organizing this event as well as submitting matching funds through her company Excel Energy. Everyone had a wonderful time at the event that raised a total of \$2,000 for CFC International. Fifteen year old Sarah had such a great time that it took her a couple of days to come back down from the clouds!



Sarah Lindgren

## Brady Scoggins Heart of a Champion Foundation supports CFC International

In 2005 CFC International lost a remarkable young man named Brady Scoggins. Brady had attended his first CFC Conference in Rockville, Maryland in 2003. He was anxiously awaiting the 2005 conference in Orlando, FL when he passed away the month prior. Brady's legacy lives on through the fund his family has established in Canada. The Brady Scoggins Heart of a Champion Foundation generously awarded CFC International \$15,560. Funds were used to support the November NIH Scientific Symposium and plans are already underway to fund our newly created Parent folder and package that is shipped out to new families who contact us. In addition to these two worthy projects, money has been set aside to help fund the upcoming Orlando conference Child Care Rooms run by Kid's Nite Out. A special thank you goes out to Brady's family who has kept Brady's memory alive and supported our programs.

## Doyle's 4th CFC International chili cook-off fundraiser

Well, the Doyle's 4th CFC International chili cook-off fundraiser is in the books. It was held at the Medina Eagles banquet hall again this year. We sent out more flyers this year and encouraged people to bring friends; and they did! We estimate attendance was about 220 people. Giant Eagle was a new sponsor this year. They donated most of the extra food and beverages. They also donated a huge cake. What a wonderful job Judy in the bakery did recreating our "Jack" Chili Bowl logo. Our friend, Jim



Kaci Smith

Shields covered the evening's beer tab and Pete Effinger supplied enough cheese to feed everyone and then some. We covered the rest of the incidental expenses so the total amount raised by sponsors, donations, admissions, chili entries, big screen TV raffle, raffle prizes, 50/50 ticket sales and silent auction items went directly to CFC International. We exceeded all previous

years totals and raised \$10,124!

This year's chili competition turned out to be a battle between two returning champs. Lisa Kellogg, who placed second last year, pushed Patty Alber to second place. Rich Doyle made it to the podium for the first time and took home a third place finish. Congratulations to first place winner Lisa who won the Chili Bowl trophy (now proudly hung in her kitchen), a blue ribbon, commemorative apron



Jack draws winning ticket for the 42" TV LCD

and \$100 in gold coins (which she donated back!). Thanks to all 20 of our cooks. Remember, it's not too early to

start working on your recipe for next year. (Side note; only 4 votes for Tim's chili this year, time to revamp the recipe?)

We appreciate everyone who donated raffle prizes and silent auction items. The spur of the moment 50/50 raffle was a great success. Judy's brother Brian, nephew Jake and our friend Rob collected a huge pot. Carrie Nunnari was the lucky winner of \$309. Tim's idea to raffle off a 42" LCD TV turned out well. Mike Campbell was glad he brought his pickup truck and took home his huge grand prize. Congratulations Mike!

We really appreciate all that everyone did to make this a successful event. Many of our friends and family volunteered their time so the evening would run smoothly. We could not have done any of this without their help. Plans are already in the works for next year. It is never too early to start planning the family vacation in beautiful Ohio.....in February!

***"You may never know what results come from your action. But if you do nothing, there will be no result."***

**Mahatma Gandhi**

## The origins of CFC (Continued)

The CFC story itself begins in the same year as the first gathering of doctors to discuss birth defects. In 1968 an 11-week old boy at the Wisconsin Orthopedic Hospital for Children, part of the University of Wisconsin, was sent to Dr. Opitz referred from the cardiology service. This baby was large, severely hypotonic, developmentally delayed, with sparse curly hair, ptosis, an abnormal EEG, possible heart defect, and hyperkeratosis of the skin which became worse with age. Then, in 1969 Dr. Opitz saw a 4½-month-old girl referred for a large cavernous hemangioma. She was thought to represent a variant of Noonan syndrome, although her appearance and constellation of congenital anomalies reminded Dr. Opitz of the boy he had seen the year before. In 1971 a third child came to the Wisconsin clinic; this was a 3¾-year-old girl referred for developmental delays, heart defects, cavernous hemangioma, cleft palate, eye abnormalities, skin abnormalities, and spasticity. At this point Dr. Opitz suggested putting together a paper to share the similarities of these children with other doctors interested in clinical genetics, however this was not accomplished.

In 1976 Dr. Phil Pallister saw a 2-year-old girl with the same condition in Montana (where Dr. Opitz would soon move his practice). This girl was more significantly delayed initially, although it is heartening to know that as an adult today she is living self-sufficiently and considers herself “perfectly normal.” In 1979, a separate group of geneticists led by Dr. Bruce Blumberg made a presentation at the March of Dimes Birth Defects Conference describing two children with “a new mental retardation syndrome with characteristic facies, ichthyosis, and abnormal hair.” Dr. Jim Reynolds, working with Dr. Opitz in Montana, started to put this together into a publication. This paper was submitted to a medical journal in 1985, was presented at an invitation-only meeting for leading clinical geneticists called the David W. Smith Workshop on Morphogenesis and Malformations in 1986, and then published in the same year. At the David Smith meeting 6 cases were presented; the publication contained 8 cases because cases were added following the presentation, including a 7-year old boy with this condition seen by Dr. James Coldwell of Oklahoma in 1983.

Following this publication Dr. Giovanni Neri and his Italian colleagues published a report with the first two cases outside the United States, an 18-month old boy

seen in 1985 and a 22-month old boy seen 1986. This was followed by a 1988 publication by Dr. Verloes and his Belgian and French colleagues on a girl of Greek descent ascertained at 8½-years and a girl seen at 3-years of age. These authors also pointed out that another 1986 publication by Drs. Baraitser and Patton in England called “A Noonan-like short stature syndrome with sparse hair” ought to be taken together with the Reynolds paper as further description of CFC syndrome. Five more cases were published in 1989, another two in 1990, and by 1991 debate was breaking out asking whether CFC should be considered a distinct syndrome. Some doctors advocated that it was the same as Noonan syndrome, others that it was a separate syndrome.

Once the CFC syndrome was published in the late 1980s doctors around the world were able to start diagnosing children with the condition. Unfortunately, when parents would ask their geneticist for another family to talk to, there was no network available among the parents to offer support and hope. Around 1990 Mrs. Nancy Carlson wrote a letter to “Exceptional Parent” magazine, searching for another parent. Nancy’s daughter, Kristen, was born in 1987, diagnosed with Noonan when she was 2 years old, but then Nancy did her own research at the college library and saw that Noonan was not a perfect fit. Kristen’s cardiologist went to a medical conference where he learned of CFC syndrome and subsequently changed her diagnosis. Kristen’s geneticist apparently agreed that the diagnosis fit. Nancy started a correspondence with the families who responded to her letter in “Exceptional Parent.” Nancy describes some of those early telephone calls in which they joked that CFC might stand for “constantly facing challenges” or perhaps “children, funny children.” With this birth of a CFC syndrome family network Nancy began to regularly answer calls from newly diagnosed families and send out newsletters starting around October 1991. The cost of postage was getting expensive for this kitchen table non-profit, and so a listserv on YahooGroups was a welcome development. This made sharing information and support fast, easy, and free.

While this was going on from Nancy’s home in New Jersey, in New York Brenda and Cliff Conger were searching for a diagnosis for their son, born in 1993. Three years of investigation finally brought them to the diagnosis of CFC. Brenda then contacted Nancy through an ad in “Exceptional Parent” magazine, going on to take over the CFC support group from Nancy in 1997; at that time the family list included 21 names and addresses.

Brenda attended advocacy training both hosted by the National Organization for Rare Diseases and by the Genetic Alliance to learn how to obtain more awareness for CFC syndrome. She incorporated the CFC Family Network in 1999 and created a vision for the organization. The vision included: a syndrome brochure, website, newsletters, Parent's Guide, medical advisors, and eventually a conference.

The first CFC conference took place in July 2000 in Salt Lake City, Utah with 37 families attending, nearly the entire medical advisory board (Drs. Allanson, Carey, Kavamura, Neri, Noonan, and Opitz), rehabilitative medicine staff, local geneticists, and the leaders of the Noonan and Costello support organizations (Wanda Robinson and Colin Stone, respectively). While discovering the genetic cause of CFC seemed a long way off at the time, the medical advisors were quite excited by this unique opportunity to hone their abilities to make a clinical diagnosis of CFC syndrome. Thus among all the discussions, lectures, examinations, blood draws, and other (fun) activities perhaps the most exciting was the opportunity to take photos of the children and then study them side by side. This parsing of CFC vs. Noonan vs. Costello vs. "other" was a key step forward for physicians making a clinical diagnosis of the condition.

By 2004 the support organization grew to close to 100 families representing nine different countries. Leadership by then encompassed an active Board of Directors. The Board changed the name from CFC Family Network to CFC International and introduced their new logo. After attending a training program in Washington, DC on advocacy organization biobanking, CFC International boldly moved forward to bank their own DNA through the jointly owned Genetic Alliance BioBank Program. This venture captured the attention of researchers from around the globe. By the fall of 2005, CFC International partnered with a team of researchers led by Dr. Katherine Rauen at the UCSF Comprehensive Cancer Center. Two years worth of clinical data and DNA storage paid off as the collaborative team efforts quickly located three of the genes responsible for CFC syndrome. Publication of the gene discovery occurred in one of the most highly visible formats in all of science and medicine in March 2006. Additional publications followed quickly, along with an NIH grant to host the first International Symposium on CFC and Noonan syndromes in November 2006. CFC International continues to collect clinical data and DNA with the hope to move research forward into treatment programs to help the next generation of affected children.

## Fleur's story

Hello, we are Peter and Thea Hoedjes from the Netherlands. We live in Oosthuizen, a small village near Amsterdam. Four years ago, on the tenth of April 2003, our daughter Fleur was born. Thea's pregnancy went well until the 7th month. Then she began holding up much fluid what eventually was the first sign that something was "wrong" with Fleur. Thea was sent to the hospital for an ultrasound check. The gynecologist thought that Fleur had excessive fluid in her stomach and he also saw only one kidney. He wanted us to go to the Academic Medical Centre in Amsterdam for an excessive ultrasound check as soon as possible. After two days of stress we could go for the examination. They checked all organs, also the heart but they did not find any physical disorders at that time. The only thing they could find was that Fleur was above average (large), but nothing to worry about. Relieved we went home.



**Fleur Hoedjes**

From that moment on Thea had ultrasound checks on a weekly basis. She held more and more fluid and at the end she could hardly walk anymore. In the end of week 36 Thea felt that labour started and we went to the hospital. There she turned out to have high blood pressure and she had to stay in the hospital. Labour did not go on until the next day when it started again. In the evening at nine o'clock they decided to break the waters and that showed that Thea had extremely much amniotic fluid. After a long night Fleur was born with a forceps delivery at 5:56 AM. She weighted 3700 grams. We were in love immediately.

Because Thea had lost a lot of blood we had to stay in the hospital an extra night. In those two days in hospital, Fleur lost weight of about 500 grams and did not drink well. Despite that Fleur did not drink well, we were allowed to go home. All bagged and packed we were exited to go home, but at the final check the doctor heard a fairly severe heart murmur. We had to stay for a couple of extra days. That was a big disappointment for us.

Fleur had to stay in the child observation unit in an open incubator. Little by little Fleur was able to drink and she gained some weight. Finally, after 6 days of ups and downs with feeding problems, we were allowed to go home.

The first weeks at home Fleur still did not drink well because she became very tired while breastfeeding. With a lot of efforts we luckily managed to feed Fleur enough and she grew steadily.

Within a couple of weeks the first heart scan was done. It showed a defect that is typically for the Noonan and CFC syndrome, which we did not know at that particular moment. The defect wasn't that severe that immediate action was needed. Periodic scans of the heart were sufficient at that moment, but after 11 months Fleur needed a percutaneous angioplasty because the stenosis of the blood vessel was increasing. The surgery was successful but the stenosis did not decrease but stabilized. Luckily, Fleur's heart is still stable today.

Three months after Fleur's birth, our paediatrician told us that they suspected that Fleur had the Noonan Syndrome. Back home we immediately searched on the Internet for information about Noonan Syndrome. Every one of us has been through this same moment somehow. We don't have to describe what you are going through at that moment and the period thereafter. For both of us it came sort or less unexpected as we did not see any different face or body features. We thought Fleur was the most beautiful baby girl in the world, and she was just off course. Our love for Fleur became only greater than it already was!

After the diagnosis of Noonan Syndrome we gathered as much as information about it as we could. We joined the Noonan Foundation in the Netherlands and we attended the Noonan Family day. After that day we both felt that Noonan wasn't the right diagnosis. Some features of the other children corresponded with Fleur, others didn't, in particular Fleur's mental development. It was less developed than most of the Noonan children. In the period thereafter we started to inform ourselves about other syndromes together with our clinical geneticist. Amongst others, CFC and Costello came up as possible syndromes. After further investigation of these syndromes it became clear that Fleur had most likely the CFC syndrome. It took a total of 3 years before we came to this diagnosis. Officially it is not yet confirmed by a DNA-test, but the test is currently in progress.

In the first year of Fleur's life she needed to go to the hospital a number of times, usually because of dehydration. Other causes were intestinal disorders, pneumonia and the RS-virus. During these hospitalizations, she was tube fed and needed extra oxygen. These periods lasted mostly one to two weeks.

Besides the hospitalizations, abdominal cramps, regular inflammation of the ear and bad sleeping, Fleur was a sweet and quiet baby.

From about 1.5 to 2 years Fleur got sometimes really frustrated because we could not yet communicate with her. She wanted to express herself and she wanted to be understood. She also wasn't mobile yet, she was only able to sit with our help and could not walk or crawl. Her frustration expressed in bad behaviour like pulling her own hair, hitting herself, or biting her self. We could also be victim of that behaviour. We felt really powerless and sad with that situation.

After Fleur started with occupational therapy, the frustrated behaviour slowly improved. Fleur started to crawl and she could sit by herself. At a certain point she also dared to walk with our help. Since 8 months now, Fleur is going to a therapeutical playgroup. In a group of 6 children with all different disorders, she gets physio-, occupational-, speech therapy and normal supporting care. Fleur has gone through a tremendous improvement of her development since then. She is able to speak several words now and she is learning to express herself through sign language. Her frustrated behaviour is almost gone now!



**The Hoedjes Family**

Since December 2005 Fleur has got a little brother Daan. She loves him but she is also really jealous.

At this moment Fleur is doing really great. Besides the eating and sleeping problems that still occur, Fleur is really our CFC Angel, our little sunshine. We love her very much and everybody who Fleur knows does.

Peter, Thea, Fleur and Daan Hoedjes

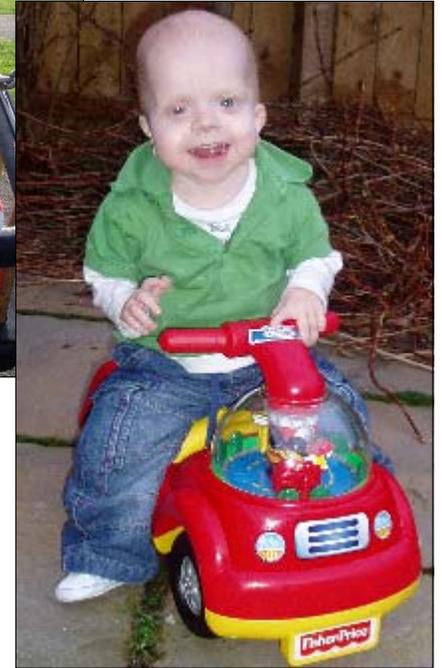
# Photo Gallery



**Esther Robinson (4 years old) with her walker.**



**Marcus Weston  
(2 years old)**



**Lauren Wallace (3 years old) and her sister Karlee all dressed up for Halloween.**

**Brenan Wilson  
(4 years old) on the kids roller coaster in the basement.**



**Lauren Wallace (3 years old) at Therapeutic Riding**

**Brianna Hinojosa eating her Birthday cake at her 4th Birthday!**





**Nathaniel Epstein (6 years old) at the beach with his brother Theo.**



**Avery Clark (21 months)**



**Jared on his 10<sup>th</sup> birthday in his Abraham Lincoln top hat!**



**Felix (2 years old), Louise (3 years old) and Fleur (4 years old) at the first Dutch CFC Family meeting in Capelle aan den IJssel, the Netherlands.**



**Jenna Braun (7 years old)**



**Megan (13 years old)**

## A tribute to Anthony John

Anthony John Verrino came into this world on November 18, 2004, 2 months early and unexpected, with a failure to thrive, and with a heart the size of mountain. Yes, medical savvy parents, he also had hypertrophic-cardio-myopathy, but you know that is not what I really mean. He came into the world to two parents who could not love him any greater, and to a family of aunts and grandparents and cousins who were ready to meet him and help him on his journey. At the time, none of us, including our doctors, had any idea what kind of journey it would be, but after 3 months in the NICU, many problems, and no diagnosis, we realized that we would be different kinds of



**Nevada and Anthony**

parents for a baby with a very different kind of life ahead.

The onset of Anthony's seizures were very early, even during the week of his birth, and they continued

to progress

unrelentingly throughout his short life. His seizures were frequent, severe, and never controlled. They were impervious to medications, of which we tried everything available from here to Canada, as well as diets, and nutrition. In the end, we worked only to sedate him out of the discomfort caused by the refractory seizures that would ultimately weaken his body and lead to his passing.

Inside this skinny little body, was a spirit that had so much strength to offer. Inside a body that never spoke a word, that couldn't make a sound, was a wise voice with so much to say. We will never be able to put into words how blessed we feel to say we are the parents of such an amazing person. There are a lot of things that I do not understand about the way this world works, but I know that I am in awe of the strength of spirit that my son carried with him in that tiny, seizure ridden body for 26 months.

My son taught my husband and me a lot, and we hope to continue to learn from him for the rest of our lives. Because of baby Ant., we appreciate what we have in our love for each other, our love for our families and friends,

and in the blessing of good health. Anthony grounded us, and made our world slow down, and because we were so focused on his care, we had time for only the most important things, like loving each other and those who cared for us. Anthony brought out the best in us, and also in everyone who had the chance to hold him or hear his story. His spirit reached beyond the little body that confined him, and he inspired so many others to be better, to appreciate their own blessings, and to do more to help others less fortunate.

It takes a village. Our families, friends, community, doctors, hospice care team, along with the support given to us by the CFC International support group families and the teams of researchers and doctors working on their behalf, allowed us to be the kinds of parents that little Anthony needed. The day that we got a firm diagnosis was the first day in 18 months that I could answer the question in the back of my head that kept saying, "What



**Anthony John Verrino**

happened?" I cannot tell you how much that helped me. It was second only to the support of other CFC families, who for lack of a better word, just "got it."

After Anthony's death, I find that I am actually lighter than I imagined I would feel. Instead of sad days or weeks, I find that we have sad moments or hours in the day. We have been grieving with the help of Angela Hospice since Anthony was 12 months old. We knew that the severity of his seizure disorder and the weakness of his heart was a combination that would not allow for a long life, and they helped us to begin to come to terms with that while he was still with us. I can only sing the praises of the people at Angela, who allowed us to see dying as a process, and who would help us walk that path with our little boy. With their support, I was able to come to the point where I could give him permission to pass. And I am so glad that we could make the choices to keep him comfortable and out of the hospital, so that he could die peacefully and privately, in our arms. And when he passed, in our bed, between my husband and me, I was blessed to watch him take his last breaths, with my hand on his chest, and my mind saying, "Go."

It was time for him to rest, and now he truly is a CFC angel.

Thank you for being there to help us in these tough times, and our best wishes to you as you continue to navigate the hard decisions involving the best care for your special children.

With love,  
Nevada and Anthony Verrino.

### Where Can I Find Information on CFC Genetic Testing?

GeneTests is a publicly funded medical genetics information resource developed for physicians, other healthcare providers, and researchers, available at no cost to all interested persons. At this site are online publications of expert-authored disease reviews, an international directory of genetic testing laboratories, an international directory of genetics and prenatal diagnosis clinics and other educational materials. For more information on genetic testing for CFC Syndrome search here:

<http://www.genetests.org>

## 4th International CFC Conference & Clinic Program

### Make Time For The Magic! at the 4th International CFC Conference



**July 12-14 2007**  
**Rosen Centre Hotel**  
**Orlando Florida**

The Registration form can  
be downloaded from:  
[www.cfcsyndrome.org](http://www.cfcsyndrome.org)

# Cardiofaciocutaneous syndrome and Noonan syndrome Scientific Symposium

*November 17-19, 2006*

*Potomac, Maryland*

*Meeting Report by: Pilar L. Magoulas, MS, CGC*

In November 2006 an outstanding group of scientists and clinicians gathered from around the world to attend the first International CFC/NS Scientific Symposium held in Potomac, Maryland with the common goal of advancing knowledge and promoting interest in these two connected syndromes.

The Conference was supported by a generous R-13 grant from the National Institute of Health (NIH) Office of Rare Diseases, Heart, Lung and Blood Institute and the National Institute of Child Health and Development. While there is not enough space available to list all of the remarkable speakers and topics that were presented over the course of the weekend, this article will capture some of the highlights from the conference.

## **Making the diagnosis**

One theme that was discussed throughout the conference was the challenge and difficulty of differentiating some individuals with NS, CFC, and Costello syndromes from one another. These conditions often share many of the same physical and facial features, as well as similar cardiac defects and health complications.

Dr. Maria Ines Kavamura addressed this topic by presenting the CFC index, a diagnostic tool that she created to aid in the diagnosis and recognition of distinct features in the CFC syndrome. She extracted 82 characteristics from descriptions of CFC patients in the literature and ranked their incidence and occurrence in 54 patients with CFC. The CFC index is calculated by summing the value, which is the frequency of the trait in the CFC population, for each characteristic present in a given patient. She applied the CFC index to Noonan and Costello patients and they had lower scores, indicating that the CFC index had good specificity and was an objective way to measure the phenotypic characteristics in CFC syndrome. There was some concern about the

practicality of using a measure of this size in a clinical setting and some considered the possibility of decreasing the number of characteristics to make it more accessible for others to use. Tremendous progress over the past few years in delineating the genetic basis of these conditions, however, has helped in the classification of patients and has sparked new interest in potential treatment and therapeutic options.

## **Treatment and management of adults with NS and CFC**

While we have made considerable progress in understanding these conditions, there are still many questions that remain unanswered such as the health risks and medical concerns of adults with NS or CFC syndrome. Long-term prognosis in NS and CFC remains difficult to predict due to the scarcity of natural history studies in these populations. Dr. Mary Ella Pierpont addressed this issue by presenting data that she had collected on adult manifestations and developmental outcomes in Noonan syndrome, while Dr. Karen Gripp addressed similar concerns in the context of CFC syndrome.

Individuals with Noonan syndrome typically present with some type of cardiac characteristic and can even present for the first time to medical attention in their 50's and 60's. This warrants the need for lifelong cardiac care and management. Physicians and practitioners need to be able to recognize individuals with these conditions because of the risks associated with cardiac complications, as well as risks secondary to other organ system involvement such as vision abnormalities, hematologic disorders, autoimmune disorders, lymphedema, and hypothyroidism. While only about 50 adults with Noonan syndrome have been followed, there does not appear to be an increase in cancer incidence above baseline for NS patients. Adult women with Noonan syndrome may be at an increased risk for pregnancy complications and should be monitored accordingly.

Dr. Gripp highlighted management and care issues in CFC syndrome. Feeding concerns, as addressed by Dr. Allanson (see below), can be mediated by medications and/or consideration of tube feeding. All individuals with CFC should have full cardiac and ophthalmologic evaluations, while skin abnormalities should be treated symptomatically. Children with CFC should be referred to early intervention services and/or special education programs at the time of diagnosis to facilitate attainment of developmental milestones and promote cognitive abilities. Areas of future study include assessment of

sleep abnormalities, mental health issues, and the possible risks and benefits of growth hormone therapy for short stature. Further research into the natural history of CFC syndrome will help elucidate care and management of the adult with CFC syndrome.

### Clinical features in CFC syndrome

#### Cardiac

Dr. Angela Lin summarized and compared the incidence and type of cardiac abnormalities in Noonan, LEOPARD, Costello, and CFC syndromes. The incidence of any cardiac abnormality is very similar between these conditions ranging from 75-80%. There is much overlap in type and frequency of defects between these conditions, however characteristic patterns do exist. For example, individuals with CFC are most likely to have combination of ASD and PS, whereas individuals with NS are likely to have isolated PS, and individuals with Costello more likely to have arrhythmias or conduction defects. Pulmonic stenosis is most common in NS, and occurs in approximately 40-50% of individuals with CFC syndrome. Involvement of other valves is less common in CFC and NS. Cardiac hypertrophy is reported in 45% of individuals with CFC syndrome. Risk for sudden death possibly related to severe hypertrophic cardiomyopathy and/or arrhythmias may be slightly increased and has been reported in a few patients with Costello, LEOPARD, and in two adults with CFC. At this time, it is too difficult to make genotype-cardiac phenotype correlations with CFC syndrome since the numbers are too small. Future research should aim at better classification of the abnormalities and better reporting by avoiding duplicate reporting of patients. Post-mortem examination may help further elucidate cardiac findings in these patients.

#### Ophthalmologic

Dr. Terri Young addressed the ophthalmologic findings in individuals with CFC syndrome. One of the most common findings is strabismus, which is found in 75% of individuals. Nystagmus and optic nerve hypoplasia have been reported in 54% and 33% of individuals, respectively, however the incidence of optic nerve hypoplasia may be higher given that earlier reports may not have examined patients for this finding. Other ocular findings include exotropia and esotropia. Many individuals with CFC have decreased vision and acuity; however, early diagnosis can lead to correction and improvement with visual aids.

#### Gastrointestinal and neurological manifestations

Dr. Judith Allanson discussed the gastrointestinal and neurological aspects of CFC syndrome. Approximately

66% of individuals with CFC have feeding problems. Failure to thrive (FTT) is very common in newborns and infants with CFC syndrome and can be attributable to poor suck, hyperemesis (vomiting), aspiration, oral aversion, and dysmotility. It can be severe enough to necessitate prolonged tube feeding. Similarly, severe gastroesophageal reflux may require surgical intervention with fundoplication. One significant discrepancy between NS and CFC is that the failure to thrive in Noonan syndrome may resolve, however in CFC syndrome, it can last for several years.

The neurological features in CFC can include structural brain abnormalities and hydrocephalus/ventriculomegaly in ~40%. Seizures have been reported in 18/37 (49%) patients. Hypotonia is a common and almost universal finding present in over 90%. The developmental milestones of 27 individuals over 4 years of age is summarized in the table below. In general, receptive language was better than expressive language within the group.

Developmental skill	Mean age	# of ind.
Babbling	14m	
First words	28m	
Combining words		4/27
Uses full sentences		7/27
Uses sign language		12/27

Developmental skill	Mean age	# of ind.
Rolling over	9m	
Sitting unassisted	14m	
Crawling	2y	
Standing	28m	
Walking	3y	
Running	4y	
Ride bicycle		12/22

### Genetic updates and the Ras/Raf pathway

More than half of the conference was devoted to the genetic and molecular basis of CFC and NS, and the Ras/Raf pathway, the pathway in which the genes are located. Dr. Marco Tartaglia and Dr. Amy Roberts reported, for the first time, the identification of a new gene that causes Noonan syndrome. This gene, called SOS1, is thought to account for about 10% of all NS cases. PTPN11, KRAS and SOS1 mutations account for ~60% of all NS patients. Dr. Roberts reported that in her patient population, there were more reports of pulmonary stenosis

with SOS1 mutations compared to PTPN11, while atrial septal defects were more common in PTPN11 patients compared to SOS1 variants.

Dr. Kate Rauen reviewed her research and discovery of the genes that cause CFC syndrome. She studied 23 individuals with CFC and identified a mutation in the BRAF gene in 18 (78%). Three of 23 individuals (12-15%) had mutations in either MEK1 or MEK2, which are in the same pathway as BRAF. Two of the 23 individuals studied were found to have deletions of BRAF.

### Conclusions

This symposium was a huge step in promoting awareness and interest of CFC and NS by bringing together collaborators from around the world to discuss recent advances in the clinical and molecular basis of these conditions. There are still many areas that need further study, particularly neurodevelopmental and cognitive outcomes, genotype-phenotype correlations, natural history studies, the role of these genes in development of cancers, and the possibility and feasibility of a centralized data collection repository/registry for CFC, Noonan, and Costello syndromes. The difficulty lies in establishing a confidential, accessible, collection of accurate data that can be used for future research.

Several genes have been implicated in CFC and Noonan syndrome, however, there was some controversy as to the clinical phenotype of individuals who were found to have mutations in KRAS. KRAS mutations have been shown in both CFC and NS, so the question arose, should

patients with KRAS be placed into a separate group? And are there common features for these patients? Once more individuals with KRAS mutations are identified, we will be better able to characterize the associated phenotype.

Summary findings from the weekend meeting:

- The genes for CFC, NS, and Costello syndromes are within the Ras/Raf pathway. This helps explain the considerable overlap in clinical features between these conditions.
- We are beginning to understand the structure, function, and role of these genes, which may enable development for future treatment options.
- The long-term effects and prognosis for individuals with NS and CFC is unclear at this time. Therefore, there is a need for the development of collaborative phenotypic databases and detailed documentation of these genetic disorders.

Brenda Conger, President of CFC International would like to thank the organizing committee and especially Dr. Amy Roberts, Dr. Bruce Gelb, and Wanda Robinson for their roles in the grant application process as well as organizing this meeting to bring together researchers and clinicians to better understand and treat patients with CFC, Costello and Noonan Syndromes. Most in the audience agreed that another meeting focusing on these cellular signaling pathways would be helpful. Hopefully, as we all gain a better understanding, therapies might be devised to best help patients.



## Board of Directors announces new addition!

The Board of CFC International is very pleased to announce that Pilar Magoulas has accepted our invitation to join the Board of CFC International.



**Pilar and her husband Demetri**

Pilar attended the 2005 CFC conference and medical clinic and assisted with the clinic program. She became interested in CFC Syndrome during her graduate training when she saw a child who was being evaluated for possible CFC, and became more invested

after meeting Jared Stowell and his family

during a genetics appointment at Primary Children's Medical Center in Salt Lake City, Utah in 2003 where she was employed as a genetic counselor.

Pilar's qualifications are well suited to CFC International. She designed a Power Point presentation on CFC Syndrome during her graduate training. This presentation was also used by Dr. John M. Opitz and was shown at the 2003 Rockville, Maryland conference. She has recently created the fact sheet for CFC Syndrome that our families have shared with their family members, medical and school providers.

Pilar completed her undergraduate work at the University of Florida and received her master's degree in genetic counseling at Northwestern University in Chicago, Illinois. Shortly after graduation, she moved to Salt Lake City, where she worked in the Division of Medical Genetics at the University of Utah. After spending three years in Salt Lake, she moved to Houston, Texas where she currently works as the inpatient consult coordinator for the Department of Molecular and Human Genetics at Texas Children's Hospital and Baylor College of Medicine.

Pilar lives in Houston, Texas with her husband, Demetri who is a financial analyst, and their dog, Charlie, a black lab. Pilar and Demetri are huge sports fans and can often be found cheering for the Florida Gators sports teams or enjoying the sports scene in Houston by watching a baseball, football, or basketball game!

## CFC Scientific Advisory Board

Since establishing the CFC International BioBank the need for approval for all research projects involving our DNA or clinical data became apparent over the past few years. The CFC International Scientific Advisory Board is comprised of well known scientists and clinicians who have no vested interest in researching CFC Syndrome. CFC International appreciates their confidential review of proposals for research!

### **John C. Carey, M.D., M.P.H., F.A.A.P**

*Professor of Pediatrics at University of Utah*

Dr. John C. Carey is the Editor of the American Journal of Medical Genetics and also a Professor of Pediatrics at the University of Utah.

### **Eric W. Johnson, Ph. D.**

*Vice President, Prevention Genetics*

Dr. Eric W. Johnson is the Vice President for Clinical Affairs, BioBanking, and Marketing at Prevention Genetics in Marshfield, WI.

### **Richard I. Kelley, M.D., Ph.D.**

*Director, Division of Metabolism at Kennedy Krieger Institute*

Dr. Richard I. Kelley is the Director of the Clinical Mass Spectrometry Laboratory at Kennedy Krieger Institute. He is also a Professor of Pediatrics at Johns Hopkins University.



### **CFC Awareness bracelet**

**\$25 plus shipping**



Jewelry designer Susanne Fedor used magenta and blue Swarovski pearls and arori borialis Swaravski crystals with an arori borialis Swaravski Heart charm as the focal point for this beautiful CFC awareness bracelet. The bracelet has a magnetic heart clasp for ease of application. The colors were selected to match the CFC logo and the heart is a symbol of life and hope for our very heroic children born with CFC Syndrome. Bracelets will be on sale at the Orlando conference. To place an order please contact Angie Lydixsen at 203-881-9186 or [aLydixsen@adelphia.net](mailto:aLydixsen@adelphia.net) for more details.



# CFC International Annual Report 2006

## *Caring, Facilitating & Connecting*

### **Our Vision**

A world in which no one will be isolated from appropriate diagnosis and treatment.

### **Our Mission**

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

### **Our Programs:**

#### **Patient Registry**

Cardio-Facio-Cutaneous (CFC) Syndrome is a very rare condition. To better understand the characteristics and natural course of CFC syndrome, an International Registry has been established. The Registry offers essential resources for the study of CFC Syndrome by providing centralized information and medical records on CFC individuals from around the world. Confidentiality of personal information regarding incidence, genetics, clinical course, and prognosis is provided to professionals and families. The Registry also serves to improve communication of ideas among interested researchers, and to ensure rapid distribution of any new information that may benefit patients or their families.

#### **BioBank**

CFC International is a founding member of the Genetic Alliance BioBank, which was established in 2004, and now holds the world's largest collection of blood and tissue from people affected by CFC and their immediate relatives. It is the only centralized repository in the world. Access to a large number of samples, combined with comprehensive clinical data and photographs, is a key resource for CFC researchers. These resources played an integral role in the research work associated with the CFC gene discoveries and will continue to do so for future research programs, including development of treatment programs.

#### **Family Conference with Clinical Evaluations**

Every two years CFC International brings physicians, clinical and scientific researchers and families together. Clinics are offered at no cost to affected children. Families hear about research advancements directly from the researchers. Resource materials are provided for affected families to better assist them in caring for a person with this rare disorder.

#### **Family Support**

##### ***Welcome Packet***

Upon registration and completion of the CFC registry, a welcome packet is mailed to families. This includes contact information, past newsletters, CFC brochure, Parent's Guide and other resource materials. There is no fee to join CFC International and support of our members is sponsored through the kindness and generosity of our donors.

##### ***Matching***

CFC International has Regional Ambassadors! Upon registration with CFC International our Regional Ambassadors contact new families. This regional matching helps our group grow stronger. While Internet support has served us well it is important to create alliances between families and encourage a network to increase awareness about this little known disorder in communities around the world.

##### ***List Serve***

CFC International hosts a computerized list serve, which offers immediate distribution of a message to the entire CFC International community. These messages and questions are considered and answered by fellow family members of CFC syndrome children and adults. While CFC International does not endorse opinions as medical advice, the list serve offers support and informed opinions by people who know this disease firsthand.

### Newsletter

Three editions of “The CFC Chronicle” were published in 2006 (March, August & December) and mailed to more than 725 families, friends, doctors and researchers. The newsletter is also available online through the CFC website. It offers the most up to date news about research, family issues, educational concerns, events and issues about CFC syndrome.

### 2006 Highlights

- CFC Genes discovered!!! CFC International BioBank played a crucial role in this discovery.
- The National Institute of Health (NIH) Office of Rare Diseases, Heart, Lung and Blood Institute and the National Institute of Child Health and Development funded a grant totaling \$32,000 for CFC and Noonan Syndromes. The grant funded 28 key speakers from eight countries to attend a Scientific Symposium on these two rare and similar conditions.
- The CFC International Biobank holds samples from 65 affected individuals and their biological parents.
- CFC International now serves over 125 members from 14 countries around the world: Australia, Canada, Denmark, England, France, Germany, Greece, Malaysia, Mexico, Netherlands, Samoa, Scotland, South Africa, and USA.
- In 2006, welcome packets were shipped to twenty-one new families who registered with our organization.
- During 2006, two research proposals were presented to the CFC International Biobank Scientific Advisory Board for access to DNA and clinical data. Both projects were approved.
- Throughout 2006, three outreach talks were presented at Atascadero State Hospital, California by CFC International Vice-President Molly Santa Cruz.
- President Brenda Conger served as an “ambassador” for The National Human Genome Research Institute and presented at Binghamton High School on National DNA Day.
- President Brenda Conger participated in Roberson Museum’s “A Celebration of the Face” by speaking about CFC Syndrome.
- CFC International funded \$38,794 for research programs (BioBank) in 2006.
- Twelve family fundraisers were held during 2006 **raising a total of \$47,229:**
  - \$7,453 4th Annual Toast the Angels wine tasting & dinner hosted by the Conger family NY
  - \$500 Binghamton, NY Cruisin’ Buddies car club
  - \$2,175 Jewelry sales hosted by Dana Kline, MD
  - \$5,320 Macy’s Shop for a Cause supported by CFC International members and their friends
  - \$10,000 Chili Bowl hosted by the Doyle family OH
  - \$2,718 Rally in the Alley for CFC hosted by the Doyle family OH
  - \$1,060 Swim-a-thon with assistance from the Doyle family OH
  - \$1,103 Doctors with a Heart sponsored by Dr. Brian Anderson
  - \$750 Horse Riding Competition Fundraiser hosed by the Ohio Horseman’s Council Club & the Farmer family
  - \$5,597 3rd Annual CFC Dance hosted by the Young family MO
  - \$8,553 2nd Annual CFC Golf Tournament in honor of Ronnie Zeunen, Jr. NC
  - \$2,000 Chili Birthday Celebration in honor of Sarah Lindgren

*Several families have made pledges to provide financial help on a yearly basis. We are very grateful for their generosity and continued support!*

### CFC International Income and Expenses 2006:

INCOME 2006		EXPENSES 2005	
General donations	\$ 32,671	Research	\$ 38,794
Family fundraisers	\$ 47,229	Training	\$ 2,477
Friendship Campaign	\$ 14,993	Newsletters printing & postage	\$ 2,698
Items sold	\$ 150	Office expenses	\$ 2,965
		Advertising	\$ 131
		Salary	\$7,500
		Miscellaneous	\$ 265
<b>Total Income</b>	<b>\$ 95,043</b>	<b>Total Expenses</b>	<b>\$ 54,830</b>



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## CFC International Has a New Website. Check us Out! [www.cfcsyndrome.org](http://www.cfcsyndrome.org)



**CFC**international  
Cardio-Facio-Cutaneous Syndrome

Caring,  
Facilitating &  
Connecting



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

	In The News	What is CFC?
<ul style="list-style-type: none"> <li>CFC International</li> <li>Information &amp; Resources</li> <li>News &amp; Events</li> <li>Research</li> <li>Donate</li> <li>Links</li> <li>Home</li> </ul>	<p><b>A Community United by Cardio-Facio-Cutaneous Syndrome</b></p> <p>Welcome to the official website for Cardio-Facio-Cutaneous International. CFC International was incorporated in 1999 as a support group for families and others whose lives are touched by this very rare syndrome. Our goal is to be a source of information so that we can raise awareness and educate the public, the medical community as well as families about Cardio-Facio-Cutaneous Syndrome. We invite you to take a closer look at who we are, what we do, and how you can support our efforts</p> <p><b>News Flash!</b> Gene for Rare Syndrome Found with Help of Families CFC genes linked to cancer <a href="#">more</a></p>	<p><b>What is Cardiofaciocutaneous (CFC) syndrome?</b></p> <p>CFC syndrome is a rare genetic condition that typically affects the heart (cardio-), facial features (facio-) and skin (cutaneous). It is seen with equal frequency in males and females and across all ethnic groups. Children with CFC syndrome may have certain features that suggest the diagnosis, such as relatively large head size, down-slanting eyes, sparse eyebrows, curly hair, areas of thickened or scaly skin, and small stature. Most will also have a heart defect. <a href="#">Read more...</a></p>