CFC international
Cardio-Facio-Cutaneous Syndrome

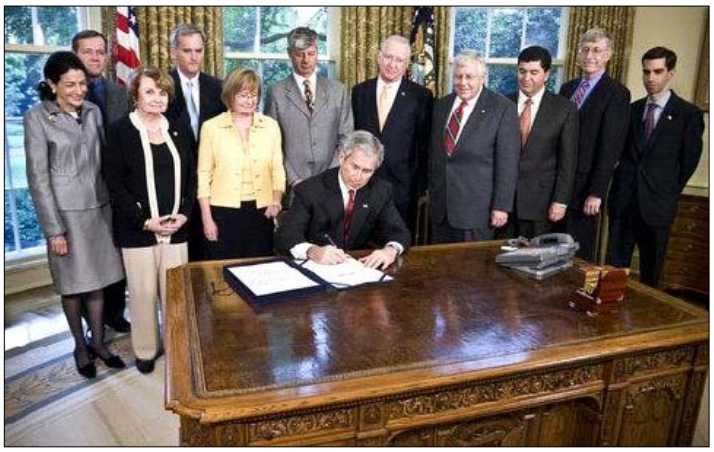
The CFC Chronicle

Caring, Facilitating & Connecting

Volume X Number III *October 2008*

President Bush Signs Landmark Genetic Nondiscrimination Information Act Into Law

Washington, D.C. – May 21, 2008 – The Coalition for Genetic Fairness (http://www.geneticfairness.org/) commends President George W. Bush for signing into law today the first civil rights legislation of the new millennium, the Genetic Information Nondiscrimination Act (GINA). GINA is the first and only federal legislation that will provide protections against discrimination based on an individual's genetic information in health insurance coverage and employment settings.



President George W. Bush signs H.R. 493, the Genetic Information Nondiscrimination Act of 2008, Wednesday, May 21, 2008, in the Oval Office.

"This is a tremendous victory for every American not born with perfect genes – which means it's a victory for every single one us," said Representative Louise Slaughter (D-NY). "Since all of us are predisposed to at least a few genetic-based disorders, we are all potential victims of genetic discrimination."

"Today marks the beginning of a new era in health care," continued Slaughter. "Americans can finally take advantage of the tremendous potential of genetic research without the fear that their own genetic information will be used against them."

Just a few weeks ago, GINA received overwhelming support in both the Senate, with a unanimous vote of approval, and the House of Representatives, where the legislation was passed by a landslide vote of 414-1.

"Individuals no longer have to worry about being discriminated against on the basis of their genetic information, and with this assurance, the promise of genetic testing and disease management and prevention can be realized more fully," stated Sharon Terry, president of the Coalition and CEO of Genetic Alliance (http://www.geneticalliance.org/)."We applaud our champions on the Hill who have worked tirelessly to pass this important legislation. It is now our responsibility to make sure the public knows that these new protections are in place."

The health insurance protections offered by GINA are expected to roll out 12 months after the bill is signed, whereas the employment protections will be fully realized in 18 months.

Continued on page 10.

In this Edition:

CFC International

Executive Director & President:
Brenda Conger, Vestal, New York
Vice President & Family Liaison:
Molly Santa Cruz, Arroyo Grande, California
Secretary: Pilar Magoulas, Houston, Texas
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Amy Hess, Glen Ellyn, Illinois
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Computer Listserv: Judy Doyle
Newsletter Editor: Peter Hoedjes, The Netherlands

CFC International
183 Brown Road
Vestal, NY 13850 USA
(607) 772-9666 (Evenings and weekends)
E-mail: 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.

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

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Message from the President

This summer has been such a sad time for our organization with the loss of CFC mom Angie Lydiksen and also the inevitable deterioration of sweet Harley Melvin. The families connected on the computer list serve have all been providing such outreach and comfort through the miles. Both of these families know that they are not alone and we are all there for them.

September brought about the move of the CFC Office from the attic storage area into the newly remodel former guest room in the front of the Conger house. Cliff has been busy with new walls, electrical and a beautiful Pergo floor. Even Clifford has been caught up in all this excitement and helped his dad out with some of the light construction. If we have any visitors over at the house, Clifford quickly reminds me that maybe I should show them the new CFC office. I am delighted with the bright and beautiful new room that my husband has so generously donated with his remodeling talents!

As we move closer to upcoming new year our board is very excited to move forward with the conference planning for the huge 2009 International event at the Berkley Double Tree Marina in California. With CFC, Noonan, and Costello families all in one location this will be a head-spinning event to see so many similar children with "unique" features. In addition to this event in 2009, CFC International will be celebrating the 10 year anniversary of their Incorporation as a non-profit in the USA. The upcoming year is a great way to make a difference for all our children and plan something special to mark this event and raise funds to meet the demands of more research projects along with a bigger and even better conference next summer!

Brenda Conger
E-mail: 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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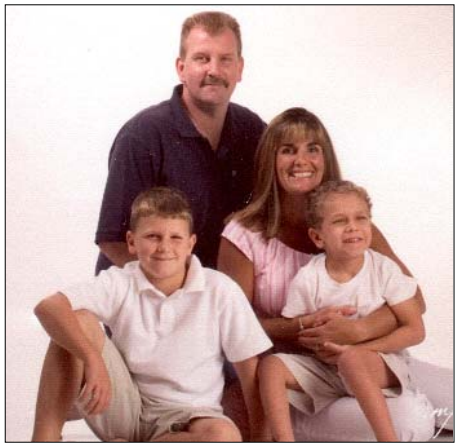
Remembering Angie

Angie Lydiksen, a devoted wife and mother passed away on June 19th after a courageous battle with cancer. Her family came into our organization upon the heels of the 2006 CFC gene discoveries when her 6 year old son Luke tested negative for Costello Syndrome and was then confirmed to be CFC Syndrome.

Angie immediately filled out the CFC Registry, pulled together all of Luke's medical records for research projects and threw herself into learning all about CFC yndrome. She became an active member on our list serve and was very excited to attend the next CFC conference. In the spring of 2006, Angie packed her whole immediate and extended family into two cars and joined the Conger family at the 2006 annual Toast the Angels Fundraiser. Angie wanted to do what ever it took to help Luke and all the other children affected by CFC Syndrome. When Angie returned home she started working on her own community fundraiser where her family all pitched in and raised close to $23,000 for CFC International! It is with great sadness that we said goodbye to Angie. She leaves behind sons Erik and Luke as well as loving husband Erik.



Angie Lydiksen with her sons Luke (CFC), Erik and husband Erik.

Although Angie's illness prevented her from attending the 2007 conference we will remember her dedication and love for all the CFC children and her own family.

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.

6th Annual Toast the Angels Brings Canadian Guests

I have to admit I was a little nervous about taking my 12-year-old son, Walker, to Brenda and Cliff Conger's annual Toast the Angels fundraiser in Vestal, New York. Vestal, after all, is the source of all things CFC, thanks to Brenda Conger, and it sounded like a pretty fancy affair.

Walker isn't the easiest dinner guest, either. Like most children with CFC, he can be unpredictable. His intellectual delay runs to the severe end of the spectrum: he can't speak, and his focus is short. At the same time, he's one of the luckier CFC kids, in that physically, while small, he's quite robust, a kind of miniature linebacker. He is functionally autistic as well, and hits himself, despite our many efforts to prevent it, and I wasn't sure how any of that would go over with a crowd of strangers.

Oh, and did I mention Walker had never met another child with CFC? There are five CFCers in Canada, all in different cities, spread widely across a big country. How was he going to react? Would he even notice?

Then there was the ten hour drive from Toronto. Our nanny and saviour, Olga de Vera, was with me, but my

Clifford Conger, Walker Brown and Victoria Palombo

wife Johanna and our 15-year-old daughter Hayley had other commitments up in Canada. I still remember the time Walker managed to open the car door while I was zooming along a highway at 65 miles an hour.

But this is the strange thing: Walker, as always, seemed to know I was feeling nervous. I noticed it when we stopped to use the bathroom at an interstate rest stop on Highway 90 just west of Batavia. (It was a Friday, and the drive from Toronto to the border alone, normally an hour and a half, had taken three.) Any CFC parent knows what an adventure it can be to visit a public bathroom. We survived anyway, and went to wash our hands—first mine, then his meaty little paws. I was reminded again as I did so—because even these tiny tasks take time with a CFCer, because they force you to concentrate on one thing—how much it touches me to touch him, to feel his skin. And as I washed his hands, which he so simply offered to me, I suddenly realized…how a nervous I was about our trip. How the prospect of the drive and the border and meeting people I didn't know made me nervous. Even in the anonymous bathroom at the side of the road on an anonymous highway where I knew no one, I was afraid—of how Walker might react, get upset, embarrass me, reveal us as a couple of failures. Every parent of a handicapped child knows these feelings, I suspect. I was so concerned about success—or at least an uptight version of it—that even washing my son's hands was setting me off. How, I thought to myself, did I become so caught up in such a conformist view of what is right and wrong?

Chef Russel and Cliff Conger entertain the crowd during the auction

Then I reached for the paper in the dispenser—quickly, so that Walker didn't escape--and there and then, as the dispenser rumbled out its sheet of paper, Walker perked up. He heard the noise, and smiled, and—something he has never done before—extended his hands up into the air for me to dry.

And he smiled as he did it—it gave him pleasure. Was it the noise of the dispenser? Was it the prospect of having his hands rubbed another way? Or did he see how nervous I was, and decided to help me relieve the pressure?

I don't know. But when he laughed, I realized he was telling me we were having fun. I learn something from him every day, far more than I have even been able to teach him.

By the time we got to Vestal, the dinner was starting. There were at least 130 people there, and they all welcomed us. (So much for my nervousness, as Walker predicted). Cliffy Conger met Walker, and they seemed to understand each other in a quick mysterious way I couldn't even begin to understand. Cliffy took an instant interest in Walker's stroller-chair, and fixed the wheel that

has been sticking—the guy's a mechanical whiz—and then took him for a stroll. Walker thanked him in what is often his way of showing enthusiasm, by whacking Cliffy in the leg, which Cliffy took graciously, with a big laugh (and I suspect a bit of a bruise). The other CFCer Walker met was Victoria Palumbo, who looked gorgeous in a red dress. Victoria's mom, Patricia, had provided no fewer than 45 gift baskets for the evening's silent auction. Yes, that's figure is correct. Butler, PA, her hometown, must be the gift basket capital of the country.

And they were just some of the fare at the auction. The dinner itself was spectacular, and a big part of the event's ongoing popularity. Four fine local upstate New York wines were served from Chateau Lafayette Reneau. The four course meal began with appetizers (including delicious peppery meatballs, and soared through beef tenderloin with Bordelaise sauce, complete with Chef Russel Rodrigez describing each course before we ate it. All told, through ticket sales and the auction, The Conger family raised $10,000 bringing the total over the past six years of dinners to more than $35,000. It's a great format that could work anywhere.

I'll never forget it—not the meal, and especially not the new and remarkable feeling of belonging I experienced, as I saw Walker sit for the first time on a couch with other CFCers. Nor will I forget the next morning, when Brenda's husband, Cliff, arranged for Walker to help drive Cliffy's green John Deere lawn tractor.

We had new friends. And thanks to CFC—it is a strange gift, too, some of the time—I know they will be friends always. Thanks for that.

Ian Brown
Toronto, Canada

Dancing for CFC

The Young family from Monett, Missouri held their 4th CFC fundraiser dance on Saturday August 16th. The event started at around 7 PM with visiting and socializing and then Meg and her friends kick started the dance by dancing to Meg's favorite song--Hannah Montana Best of Both Worlds! She had a blast!

Meg Young & Samantha White

Melanie Hills, Kara Tinklepaugh, Terri Briggs, Beth Nation & Sherri Young

Our numbers were down a little this year because there was a local annual German Festival that same night (go figure!:) but we all still had a really good time and we raised around $4,000 (so far; we do still have a few checks coming in from people who couldn't attend). We ask for donations at the door, whatever people are able to give, and we're very fortunate that Meg's friends are kind and generous. We're also fortunate that many people who couldn't come to the dance also sent a donation to us. There were probably about 75 people in attendance. Our town Mayor was kind enough to donate The Monett Park Hall for our dance and we shut it down around midnight! We had a small silent auction, snacks, drinks, and just lots of catching up! Meg has grown up so much, she used to choose songs like 'Bear in the Big Blue House' and she use to want to leave early...now she wanted to stay out late with the grown ups (which of course we let her:) and dance to tween songs. As is tradition at the dance, after Meg kick starts it with her favorite song she dances with those she loves like her daddy and papas and uncles and lots of friends. Everyone loves Meg, she's a precious girl and we are so grateful for her!

Sherri Young and family

Godmother with a Golden Fundraising Idea!

Beth Kaufman, godmother to Clifford Conger came up with a "golden" fundraiser idea. Beth hosted a gold trade-in party at her home in Gainesville, Georgia on July 24th. Twelve friends showed up to trade in their broken gold chains, fillings, and other old scraps of gold for cash. Beth donated her commission along with some donations from her neighborhood friends to raise a total of $429 for CFC International.

Felix's story

At first of all, I have no routine in write down a story in the English language, so apologies for may be using incorrect grammar.

Our family exist 7 persons: Freek (35), Nella (35), the fist son Rudolf (12), a daughter Maralisa (10), the second daughter Elody (6), a boy Felix (3) and the last boy Leander (almost 2 years).

Felix

The first time of Nella's pregnancy from Felix was nice, but Nella was unusual fat. We thought it was due to getting the fourth child. By a control at 30 weeks, they saw on the echo that Felix had some fluid round his lungs. One week later, we had a very comprehensive echo at the university hospital "Erasmus" at Rotterdam (the Netherlands). They saw that all his body had fluid. They did an amniocentesis to look for a chromosome defect. This was on a Tuesday. Early Saturday, Nella got contractions. Felix was born on the 4th of December 2004 in the normal way but 7 weeks too early. Directly, the doctors take him away for artificial respiration and investigation. At 18:00 we were allowed to visit Felix. Felix was very swollen due to the fluid (hydrops) and in a very bad condition. He got morphine and a lot of medicines to get him quiet. He wasn't able to breath, so he still got also artificial respiration. His weight was 116 ounce, it normally should be 77.6 ounce. So the difference was fluid. Monday, the 6th of December, 2004 the blood pressure goes down very hard. So the doctors thought he went to die. Fortunately, at some time the blood pressure raised to normal values.

In the weeks that followed, Felix had better and worse times. I remember that after one week, we must say good bye to Felix with the whole family. For a couple of days, he has had no making water. Again fortunately he made water that night.

This fluid comes from his lymph vessels. The doctors discovered that because the fat of his food come back in the drainpipe. Those drainpipes were in his chest (by his lungs).

Therefore Felix got no food but only foodstuff directly in his blood. Due to this, in the weeks following, he went slowly lesser swollen. During the first weeks, the doctors saw a white matter in his brain. Later on, they found a blockage in his blood vessel from his kidney to his heart. They found also that he has an enlarged heart (hypertrophic cardiomyopathy). With Christmas, Nella was allowed to get Felix in her arms.

In January or February 2005, Felix was negative tested for Noonan. During January he got again hydrops. So the story started again. During the time from January to April, so many things happened that I can't write it down the right way. Again Felix had better and worse times. Some weeks, it seems to us and to the doctors that there was no progress. So the doctors want to stop all the treatments so that Felix could die. But, every time Felix did better before they make the final decision.

Anyway, after all in February 2005 Felix was slowly getting better. Around March 2005, he could be breath on his own! Of course he still needs oxygen. In May of 2005, he came home with lots of medicine and still with oxygen. For all of us a very happy moment.

Freek and Nella Rebel

At this stage, Felix is a cheerful boy. Since June 2006, he is without oxygen. He has coarse skin, sometimes with allergic eruption. Of course he still has the problems around his heart. He doesn't speak any words only some short sort of cries. Since half 2007 we know Felix has CFC. He was positive tested. In November 2007 he was able to walk. February 2008 he scored very low on an intelligence test. Therefore he goes to a sort of education house with more of disabilities (a lot of them have Down Syndrome) on Monday, Wednesday and Friday. The other days he is still on a revalidation house.

Life with Felix is heavy but also very happy. We all see Felix developing in very small steps and we are glorious with all new phases in his growth.

Freek & Nella Rebel, The Netherlands

Photo Gallery

Daniel Hess (7) at his grandparents' house in Connecticut.

Nathaniel Epstein (7) having a swimming lesson in Baltimore, MD

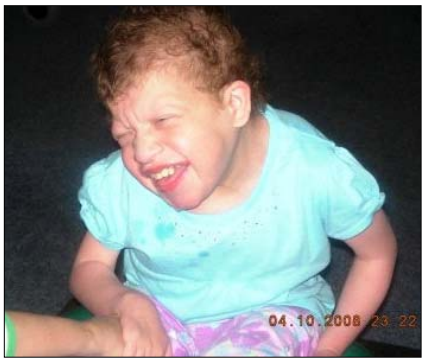
Michael Weber, CFC (9) with tweety bird at Six Flags. Tennessee, USA

Jenna Braun (8) enjoying in her moms flower garden

Lauren Wallace (4) while visiting great-grandparents at the Devil's River, Texas

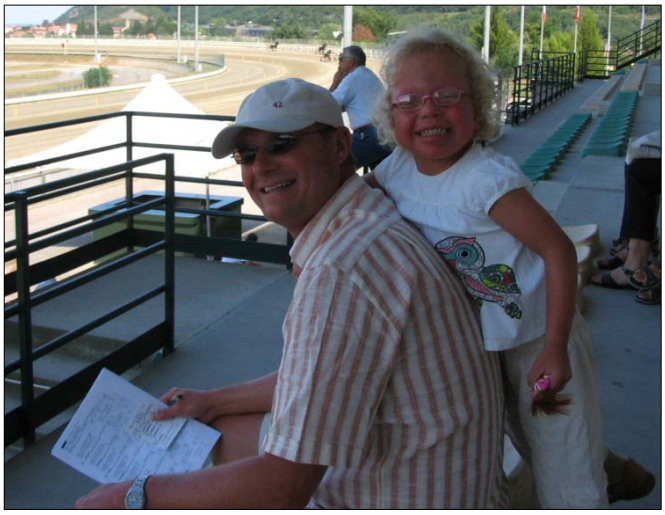
David Jud, Oberwil-Lieli, Switzerland

Princess Aubrey Infinger (4) on her 4th birthday party. Gorham, Maine



Megan Ankeny (14) working with her physical therapist on Therapy Ball

Meg Young (10) on summer vacation to Branson Missouri



Louise (4) and her father on holiday in Italy at a horse race track

The Rebel children, Maralisa, Leander, Elody, Rudolf and Felix, Capelle aan den IJssel, the Netherlands

Regan McCann (2) enjoying her peanut butter and jelly (a favorite), Weymouth, MA

Brenan Wilson at his 6th Birthday Party putting on his party hat

Vaasi (3) from Stockton California

Nancy Newton (22) stealing the blowing of the candles of her niece and nephews party cake. Perth, Australia

Fleur Hoedjes (5), Oosthuizen, the Netherlands

President Bush Signs Landmark Genetic Nondiscrimination Information Act Into Law (Continued)

"Now that GINA has been approved and signed into federal law by the President, American health care consumers and employees will no longer have to fear the adverse effects of being tested to determine their risk status for genetic diseases," said Joann Boughman, Ph.D., executive vice president of the American Society of Human Genetics (http://www.ashg.org/) and a member of the Coalition's executive committee. "Once this legislation has taken effect, clinicians will be able to order genetic tests for patients and their families in a manner that ensures the full realization of the advantages of personalized medicine models, while easing patients' concerns about the risk of genetic discrimination by insurance companies and employers based on this data."

Specifically, the legislation protects against genetic discrimination by health insurers or employers by:

- Prohibiting group health plans and issuers offering coverage on the group or individual market from basing eligibility determinations or adjusting premiums or contributions on the basis of genetic information. They cannot request, require or purchase the results of genetic tests, or disclose genetic information.

- Prohibiting issuers of Medigap policies from adjusting pricing or conditioning eligibility on the basis of genetic information. They cannot request, require or purchase the results of genetic tests, or disclose genetic information.

- Prohibiting employers from firing, refusing to hire, or otherwise discriminating with respect to compensation, terms, conditions or privileges of employment. Employers may not request, require or purchase genetic information, and may not disclose genetic information. Similar provisions apply to employment agencies and labor organizations.

Technology paper: Grade 9 Modular Technology

By: Madison Doyle, 11/26/07

CFC Syndrome, the proper name is Cardio-Facio-Cutaneous Syndrome. CFC is a rare genetic disorder that affects the heart (cardio), face (facio), and skin (cutaneous). It can occur in all races/ethnic groups, and both male and females. A couple things that suggest someone would have CFC would be curly hair, sparse eyebrows, large head, down-slanting eyes, scaly skin and small stature. Most will also have a heart defect. There are many different severities in CFC. Most will have some degree of learning difficulty and developmental delay. Some are in wheelchairs and their case is very severe. Some kids can't talk and use sign language. CFC is very similar to Noonan Syndrome and Costello Syndrome; therefore accurate diagnosis is key to propertreatment. Like I said, CFC is very rare, and there are only about 200 cases in the world. My little brother is one of them. He is 9 years old and in the 1st grade, because of the developmental delay. He is very small for his age, only about 3'5" and 361bs. He has very curly hair and will probably only get to be about 4' or 4'6". When he was a baby they said he would never walk or talk. He never crawled but he can walk on his own pretty well, he takes speech and you can understand him with little difficulty. CFC is caused by a mutation in one of your genes. If there is a change in a gene it can change how we function and how our bodies develop. The genes connected with CFC are B- RAF, MEK-l, and MEK-2 and possibly a few others yet to be discovered. The genes to conclusively diagnose CFC weren't discovered until January, 2006. This was the third time in history that a parent driven organization has co-authored on a gene discovery paper. This was because the parent group collected and stored genetic material from the CFC families that belong to the group, giving them the largest collection of CFC samples in the world. Dr. Kate Rauen, a researcher at University of California, San Francisco (UCSF) Comprehensive Cancer Center became interested in CFC while studying the causes of certain types of cancer. Her discovery highlights a developmental role in the genetic pathway: MAPK, which is more famous for triggering cancerous tumors. Several cancer drugs that target this area are being studied in clinical trials and can be used for potential treatment of CFC symptoms. Previous work figured out that a mutation in a gene called HRAS is related to a more serious condition; Costello Syndrome. Defects in a gene called PTPN11 are the cause of a mild

syndrome called Noonan Syndrome. Some researchers argued that Noonan, Costello and CFC were the same thing. In 2004, 100 CFC patients and their families joined 6 other genetic disease groups and set up a central Biobank to keep track of patient's records and DNA samples. Using the material from the Biobank it took Rauen and her colleagues only a few months to find the mutation in the genes, B-RAF, MEK-l, MEK-2, which explained 21 of the 23 CFC cases in the research cohort. Like the genes that cause Noonan and Costello the three genes are a part of a complex pathway that is a main route by which a cell sends signals from its outside to its nucleus. It also helps tell the cell when to grow and divide. When one of the genes is mutated it results in the gene dividing out of control and creates a tumor. In kids with any of the three syndromes a mutation in the gene can cause heart defects, curly, brittle hair, slow growth, cognitive disabilities, and a variety of skin conditions. When tested in mice MEK-l and MEK-2 mutations have been suggested to cause heart and skin defects, though the facial pattern cannot be predicted. An interest to researchers is that CFC kids do not seem prone to cancer. Since the syndrome was first identified in 1986 the clinical discoveries in the last two years have really made a significant difference, in how quickly children are identified and can start receiving proper therapies and medical treatment. CFC's connection to the cancer can make it possible for a very little known syndrome to receive more attention, which could. lead to treatments and maybe a cure.

Rare Disorders of the MAPK pathway; Current status/future directions

May 30th – 31st 2008 Barcelona, Spain
By: Brenda Conger

Approximately 70 researchers, clinicians, medical students and advocacy group leaders met on May 30th , 2008 in Barcelona prior to the European Society of Human Genetics Conference. The focus of the two day session was to bring researchers together to share and learn more about the rare disorders of the MAPK pathway. This pathway encompasses CFC, Costello, Noonan, Leopard and Neurofibromatosis. Brenda Conger represented CFC International and the Costello Syndrome had representation from the USA with Lisa Schoyer (CSFN) and Colin Stone from England (ICSSG). The aims of the workshop:

1. To educate interested genetic professionals regarding the clinical features and underlying genetic basis of CFC, CS and Noonan syndromes and other diseases of the Ras-MAPK pathway.
2. To facilitate research collaboration across Europe in understanding the disorders caused by deregulation of the RAS-MAPK pathway.
3. To establish a mechanism for establishing a complete genotype/phenotype dataset for European patients.
4. To determine a mechanism for using this dataset to facilitate clinical research, to document long-term natural history, improve management and encourage treatment trials.

CFC International Medical Advisors presented information on our syndrome. Dr. Judith Allanson from Children's Hospital of Eastern Ontario, Ottawa, Canada spoke on the BRAF, MEK 1 and MEK2 genes. She presented the statistical analysis of the many defects found in the cohort she has studied. As noted within the last two years, the seizure disorder found in CFC is now much higher than previously estimated years ago. Approximately 35% of the BRAF and MEK1 and MEK2 individuals experience some type of seizure disorder. Thorough neurological evaluations should be done in all patients with CFC syndrome since over one third of the individuals are noted to experience seizures.

Dr. Giovanni Neri presented the story of CFC Syndrome with the historical perspective, which began in 1983 in Montana when he studied with Dr. John Opitz. As noted by Dr. Neri, there will always be patients who fall in the center of these syndromes (CFC, Noonan and Costello). 2001 was the turning point for the syndromes with the discovery of PTPN11 (a Noonan gene). At this point the bonafide CFC patients were tested for PTPN11 and none were positive for it so it was then official and confirmed that CFC and Noonan Syndromes were two distinct syndromes. Since the 2006 CFC gene discoveries, the accomplishments have been quite impressive in such a short period of time! Dr. Neri feels that the known confirmed cases are presenting the tip of the iceberg. He commented that there are probably many more around the world waiting to be diagnosed.

The Noonan genes (PTPN11, KRAS, SOS1, and RAF1) were explored and it was interesting to note that unusual cases were presented and at times the audience could not determine if the child's case was indeed a Noonan child or a CFC child. When the DNA gene analysis was then shown there were many murmurs in the audience. This discussion of unusual cases was very intriguing and the topic of some individuals never

obtaining a firm diagnosis came up and researchers talked about them falling between the CFC and Noonan Syndromes. As a family support organization we too have told our families that if they feel comfortable with CFC International then they would never be forced to leave our group. We will continue to see more and more of our children rediagnosed with Noonan Syndrome (SOS1 or RAF1). There are still more genes to be discovered with Noonan Syndrome but no matter what transpires, families can benefit from either advocacy organization. The key is to keep educating yourself as a parent and read the literature through both organization newsletters and websites. New medical issues can crop us as the patients age and staying on top of your child's medical concerns can be very crucial!

Standing: Dr. Michael Patton, Colin Stone, Lisa Schoyer, Dr. Maria Ines Kavamura, Dr. Bruce Gelb, Dr. Judith Allanson, Dr. Bronwyn Kerr, Dr. Giovanni Neri, Dr. Martin Zenker, Dr. Alain Verloes, Dr. Didier.
Kneeling: Dr. Helene Cave', Brenda Conger, Dr. Yoko Aoki, Dr. Ineke van der Burght, Dr. Martin Erlangen

I found the talk on Leopard Syndrome by Dr. Maria Cistine Digilio of Portugal very interesting since I knew very little about this syndrome. It is a syndrome that is identified by the PTPN11 mutation. The patients often start out with a Noonan Syndrome diagnosis but as they age, they develop more spots. The patients with Leopard Syndrome have mutations in exons 7, 12, 13.

Another brilliant and exciting talk was presented by Dr. Ype Elgersma who is from The Netherlands. He presented Oncogenes for Treating Cognitive Deficits in the Ras Pathway and specifically targeted the Neurofibromatosis population. His trial study has been using statins that cross the blood/brain barrier to help correct the cognitive barrier. They had 62 children ages 8-16 in their trial drug study for a 3 month period. The results were inconclusive since the placebo group parents reported positive findings as well as the treatment group. The scientists felt that a longer trial period, which could include the children's teachers' observations as well, would have been more helpful.

On Friday evening the meeting presenters and advocacy group leaders headed off to a Barcelona waterfront restaurant for a relaxing evening of seafood fare. I was a bit put off by watching our lobster walk around the table prior to his demise as our dinner. Despite this, the relaxed atmosphere gave all of us time to have more time to chat about our personal lives and connect with each other on a different level. This informal downtime was special and I enjoyed my time talking to Dr. Bruce Gelb who sat across from me, and to Dr. Yoko Aoki.

The ending of the final day of this extremely valuable workshop was a discussion of ongoing workshops every year around the European Society of Human Genetics meeting. The group discussed establishing a directory of uncommon phenotypes, and also setting up a way to track and log-in through a central database all the patients identified. It is evident that research is moving fast and many in the field are hot on the trail of potential treatment programs and collecting more patient studies to learn even more about these linked syndromes. I applaud the scientists I met at this meeting and also give thanks to others working in the field from other regions of the world. It is pretty amazing and exciting that we are all part of this turning point in history with these rare disorders.

Luba Djurdjinovic and Amy Hess Join CFC International Board of Directors

CFC International is pleased to announce the elections of Luba Djurdjinovic and Amy Hess to our Board of Directors. They both bring a wealth of information and leadership to our organization.

Luba Djurdjinovic, MS is the Executive Director of Ferre Institute, Inc., a non-profit organization located in Binghamton, NY that provides services in genetics and repr oductive health. She is also Director of its Genetic Counseling Programs. She is a practicing genetic counselor with clinical interests in issues of infertility, familial cancer histories and most recently, cardiovascular genetics. Luba is credited for developing a unique genetic service delivery model and has promoted its adoption. In her 30 years of practice, she has been active in many state and national genetic professional organizations. She is the past president of the National Society of Genetic Counselors and a recipient of its National Achievement Award. Presently, Luba is co-chair of the NYS Genetic Counselors Group that represents all NYS genetic counselors.


Luba Djurdjinovic

Luba has been a principle investigator on several projects with the National Human Genome Research Institute (Human Genome Project). She has provided public and professional education. In 2000, Luba completed 19 years on the adjunct faculty at the School Social Work in Syracuse, New York. In 2005, she received special recognition from the Alliance of Genetics Supports (an international organization representing genetic support groups). Luba is an author in a leading textbook for genetic counseling students and lectures extensively. She has been an active volunteer with CFC International with recruiting college volunteers and offering BioBank management advice to the organization over the past several years.

Amy Hess lives in Glen Ellyn, Illinois with her husband Steve and children Daniel (CFC-age 7), Sarah (age 5) and Laura (age 3). Daniel was unofficially diagnosed with CFC in 2001 at seven months of age, clinically diagnosed at the 2003 Rockville, Maryland CFC conference at age 2 and confirmed last year through genetic testing. The information and support the Hess family has received through CFC International has been invaluable to Daniel, family members and their doctors and therapists.


Amy Hess

Before Daniel was born Amy worked in television advertising sales for nine years at CBS, PAX and Discovery Networks. Amy has volunteered for the past 18 years with teenage mothers in Chicago and its suburbs. She also volunteers with parent support groups and special events at the local Easter Seals, where Daniel receives therapies.

Amy has been fortunate to attend the last three CFC conferences. She has enjoyed meeting the many wonderful families and doctors who have attended. She is grateful to belong to such a caring, involved, information-sharing network and looks forward to helping CFC International as a board member.

Resignation from Board of Directors Diana Zeunen

Dear CFC International Members,

Fourteen years ago, my son Ronnie was born with numerous medical and physical complications. As most of you know it is painstakingly hard to explore the possibilities of a diagnosis when you spend every moment of every day focused on your baby's health and well-being. It wasn't until Ronnie was three-years old that I found the strength to research and discover that he had the very rare genetic disorder called Cardio-Facio-Cutaneous Syndrome. Immediately I connected with, what was then called The CFC Family Support Network. Clearly it was a life altering experience due to the fact that priceless information was now at my fingertips and I no longer felt isolated from the world.

It wasn't long after I became a member of, what is now called CFC International that I was asked to serve on the board of directors and act as treasurer. Recently I resigned because of other obligations. However, I would like to take this opportunity to thank the members of CFC International for allowing me to serve as a member of the board of directors for the past 9 years. My work with CFC International has been rewarding to say the least. I have made wonderful friends, learned so much and feel I have gained a new family who understands the joys and difficulties of raising a child with special needs. Thank you for your continued support, ideas and faith in me as a board member. I hope to continue helping others who join our organization.

Sincerely, Diana Zeunen

Celebrating 10 YEARS of Family Support, Research and Education

~~ 1999 to 2009 ~~

2009 CFC International Conference
Berkeley, California
August 2-4th, 2009

Join us at the beautiful Doubletree Hotel and Executive Meeting Center at the Berkeley Marina as we host our 5th International CFC Syndrome Family Conference and Medical Clinic. This is a very special conference as we celebrate our 10th Anniversary as an incorporated non-profit organization.

We are grateful and excited to share our conference with researchers and scientists from around the world who will also be attending the National Institute of Health sponsored scientific meeting entitled, "Genetic Syndromes of the Ras/MAPK Pathway: From Bedside to Bench and Back".

You will also have the chance to meet families and individuals affected by Costello Syndrome and Noonan Syndrome whom we share the same genetic pathway with. They are our CFC cousins!

On the evening of August 1st, CFC, CS and NS are co-sponsoring a dessert social get together to show off our kids to the researchers and scientists who are working hard at treatments and therapies to help our children.

See you in California next August!

The Doubletree Hotel & Executive Meeting Center at the Berkeley Marina, a world-class full service meeting and conference center overlooking San Francisco Bay, providing superior service in a resort-like setting. Our exceptional hotel is only 20 minutes from the Oakland International Airport and convenient to all major highways.

Joint Symposium meeting CFC, Noonan and Costello Syndromes!

CFC International will hold its 5th International Conference and Medical Clinic program on August 2-4, 2008 in Berkley, California. In addition to this event there will be a joint Symposium meeting of the Genetic Syndromes of the RAS/MAP kinase Pathway on August 1-2. 2009 at the Berkley Double Tree Marina. The world's leading researchers and clinicians will give presentations on many topics ranging from advances in understanding the underlying biochemistry, to the latest findings utilizing various models systems and what future research can be expected on CFC, Costello, Noonan and Neurofibromatosis, which are all on this pathway. This exciting Scientific meeting will bring together CFC, Noonan and Costello Syndromes for the first time in the USA!

During the family conference we have the following tentative workshop program planned along with other exciting keynote speakers:

- Behavioral Issues
- Skin problems encountered with CFC
- Feeding Disorders & GI problems
- Endocrinology
- Wills & Estate Planning
- Transition Planning
- Orthopedic concerns
- Ophthalmology Research Findings
- Speech & Language Disorders
- Genetic Testing and What does it all mean?
- Cardiology Concerns with CFC Syndrome
- Working with your Primary Care Peditrician

For a conference registration form please check our website after January 1, 2009
www.cfcsyndrome.org

Caudwell Children Assists UK Families with Funding to Attend Conference 2009 in the United States

The Caudwell Children based charity in the United Kingdom has generously offered to assist UK families with CFC, Costello, or Noonan syndrome children to attend the 2009 conferences in California, USA. The whole family will be funded with flights, hotel and food as long as the affected individual is 18 years old or younger. The limitation on Caudwell Children being able to support a family is that the total family income must be less than 44,000 UK pounds per year.

Caudwell Children Contact: Lisa Bates Phone 0178 260 0608
C/O Caudwell Children http://www.caudwellchildren.com/
Minton Building
Shelton Road
Stoke ST4 7RY

183 Brown Road
Vestal, NY 13850

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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 Biobank, which is critical to future research.

Enclosed please find a check in the amount of $ _____

Gift in Honor of _____

Gift in Memory of _____

Please make checks payable to CFC International & mail to: 183 Brown Road
Vestal, NY 13850

Name: _____

Address: _____

City: _____ State: _____ Zip: _____

Phone: _____

Email: _____