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Cardio-Facio-Cutaneous Syndrome

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Caring, Facilitating & Connecting

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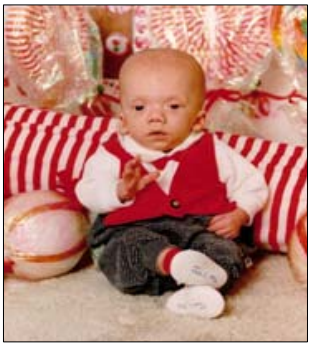
Michael's Journey from Noonan Syndrome to CFC

By Rosemarie Pavilonis



Michael Pavilonis

My husband Stan and I were blessed to have our first child, Michael on June 11, 1992. It was the best day of our lives, we gave birth to a healthy baby boy and we were so happy and thankful. The next morning our family doctor came in to visit me after he examined Michael and told me he thought Michael had Down syndrome. He heard a heart murmur and immediately ordered an echocardiogram. Our excitement and joy of being a parent suddenly turned into fear and worry; how would we know how to care for such an ill child. I shared a hospital room with two other mothers that gave birth to healthy babies and it was difficult for me to listen to their joy and excitement. Michael was diagnosed the next day with Noonan Syndrome by a geneticist. Genetic testing was not available in 1992 for Noonan Syndrome and a clinical diagnosis was the only option available, however every geneticist and physician that met him said he was a classic Noonan Syndrome child. Michael was born with pulmonary stenosis, atrial septal defect (ASD), ventricular septal defect (VSD), a webbed neck, droopy eyes, low set ears, large head, heart shaped lips, partial circumcision, undescended testicle, failure to suck, wide set eyes and nipples. At three weeks old he had another echocardiogram, was diagnosed with severe hypertrophic cardiomyopathy and we were told he may only survive a couple of weeks. The cardiologist recommended heart catheterization and surgery to reconstruct Michael's heart. We were frightened and wanted to help our precious little angel beat the odds, so we met with our pastor and our families and Michael was christened the next day. Our family doctor secured an appointment for us at Children's Memorial Hospital for a second opinion the very next day. A team of doctors was very interested in Michael's case and collectively recommended that Michael be put on a beta blocker called Verapamil to reduce the pressures and muscle blockage in Michael's heart. Verapamil came with possible life threatening side effects; however our only other option was a very risky surgery that he may not survive. Michael responded remarkably well to Verapamil and remained on it until he had his open heart surgery at 18 months old in January, 1994.

Michael had a successful multi surgical heart repair at Children's Memorial Hospital in Chicago. The surgeons cut out the bundles of muscle in Michael's ventricles, cut open his right ventricle to expand it and attached a Dacron patch to the opening so the blood could flow more freely in/out of the ventricle. They reconstructed his pulmonary artery above the valve and repaired the valve so blood would no longer be restricted. His ASD and VSD were repaired and his aortic valve was also repaired. The doctor was very aggressive in repairing both sides of Michael's heart in one surgery, as these are typically performed in two separate surgeries. Michael went into third degree heart block after surgery, which meant he did not have sinus rhythm because his heart's conductive system was damaged and his heart needed assistance from an external pacemaker. Michael caught a virus right after his surgery, therefore his pacemaker surgery was postponed. Three weeks later, his heart's sinus rhythm returned, he was no longer in heart block and he was immediately discharged from the hospital without a pacemaker.

Michael's Journey Continued on page 6.

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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.

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.

Medical Advisory Board

Message from the President

Recent tragic events in our hometown area of Binghamton, NY have made so many of us think about the future and how quickly life can change. Most of us raising special needs children feel as though we are the conductors of a very large orchestra. When one person is missing or sick in our orchestra we struggle to keep the melody flowing. But what happens if the conductor is gone? Dying and leaving behind a special needs child is often a topic that we often fail to discuss or plan for. I remember when our own son was so little and just hanging onto life. I kept mentally preparing for his death. With his recent 16th birthday and my near future retirement from my teaching job at Binghamton High School, I am forced to look at the next phase of life that entails planning for his future.

Future needs planning within our Conger family began last summer when we located a local lawyer who was experienced in guardianship and estate planning. He too has an older son with disabilities so I knew that he had walked before us with this exact struggle. He set up guardianship through our local courts and then counseled us on how to revise our will. Clifford's older sister Paige also attended this legal meeting. Our next hurdle will be to sit down as parents and work on our Letter of Intent. The Letter of Intent permits parents to communicate vital information about their son or daughter to future caregivers. I did locate an excellent resource for our Letter of Intent: www.specialneedslegalplanning.com

If you plan to attend the upcoming CFC conference in Berkeley, CA this summer you might also wish to attend the will and estate planning workshop as well. This just might be the motivation to get your family started on this very important task!

We have been extremely busy with lining up our many conference workshop speakers and medical teams to participate in this large event. We will be sharing our family conference days and some workshops and medical exams with the Noonan Syndrome Support Group. There is also private time for just the CFC International families to connect as well.

I look forward to reconnecting with old friends and meeting new friends in California this 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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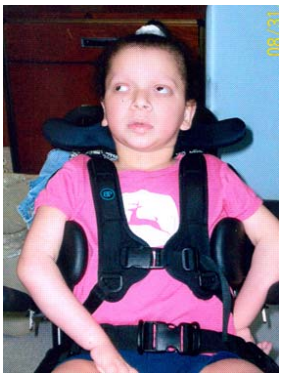
STAR UP ABOVE

Sweet Harley Melvin was sent from above,
our special girl who bought us much love,
her abilities were small but her attitude shone through,
she could light up a room with that cheeky smile too,
coz that was Harley's style you know,
her laughing eyes did really glow,
she never complained with all of her pain,
our darling angel she fought on in vain,
her life was planned; we can't change a thing,
the memories live on that her life did bring,
now the times we shared will stay in our hearts,
now God has chosen to give her a new start,
HER PAIN HAS GONE AS YOU CAN SEE,
BUT HER SPIRIT STAYS WITH OUR FAMILY,



Harley Melvin
1/6/99 - 3/11/09

so take your wings and fly high in the heavens my love,
our sweet darling Harley borrowed from above,
our shining star sent to us with care,
our love for you we did not spare,
you changed our lives, that is for sure,
but you made us open many a new door,
our special angel sent from above,
our CFC angel sent with much love.

DEDICATED TO THE MELVIN FAMILY
Written by Paula Newton, Australia

Doyle Chili Fundraiser 2009

We had our annual CFC Chili Bowl on Saturday, February 21, 2009. Weather here in Medina Ohio was pretty decent for us, the end of February. A bit of snow hit, just before the guests started to arrive. Luckily the weather or the economy didn't keep people away. About 190 family, friends and friends of friends made their way out to taste what our chili chefs had to offer. Once again, the Medina Eagle's donated their banquet room. Giant Eagle was a sponsor again this year donating most of the extra food and beverages. John and Cindy Boosinger and Maryann Bolton from SimplexGrinnell brought the hot dogs and helped man the food tables and Jim Shields covered the evening's beer tab. Pete Effinger supplied enough cheese to feed everyone and then some. Kristine Effinger, Lisa Kellogg and Carol Ryland baked up a storm. Our family covered the rest of the incidental expenses so the total amount from sponsors, donations, admission, chili entries, big screen TV raffle, raffle prizes, and silent auction items went directly to CFC International. This year's total amount was just under $13,000!

Chili Bowl 2009 Winners 1st Place Lisa Kellogg, tied for 2nd Place Cathy Bryenton and Stacy Bonitz

The chili competition was as hot as ever. 14 cooks entered for a chance to win the Chili Bowl's top spot. For the third year in a row, taking 1st and her fourth year placing, Lisa Kellogg won with her infamous traditional Texas style chili. Lisa will add her third Chili Bowl trophy and first place ribbon to her kitchen wall, #4 magnet to the frig and the apron to her collection. She graciously donated back her $100 in gold coins. For the second time Stacy Bonitz took the 2nd place ribbon, this year she shared her second place spot with newcomer, Cathy Bryenton. Believe it or not, Judy placed 3rd with a first time recipe she got from the Food Show. Congratulations to all of this year's winners!

We appreciate everyone who donated raffle prizes and silent auction items. We had a great selection again this year. A special thanks goes to Sam Meuler who helped out getting donations for the silent auction. The 52" LCD TV raffle was big hit again this year. Barb and Jim Berger were the lucky winners. Jim even followed through on his promise to dance on the table if he won the TV.

Jack had a great night. He made it all the way to 11 pm. He was busy passing out his artwork all evening. We were very fortunate to have two special guests with us. Kaci Smith and her parents Ken and Michelle Smith came again from Massillon. Isabella Misita from Chillicothe came up with her parents Mark and Peggy and her little brother. It was really cool to have three children with such a rare syndrome together at our event. We hope you had a chance to check out the CFC display table and see all of our gorgeous children.

Next years event is already on the calendar, Saturday, February 27, 2010. Planning a fund raiser is a lot of work, but for our family it has become something we really look forward to. It is a little like having a wedding every year but the work is well worth it. The amount of money our little Chili Bowl has raised for CFC is something that makes us very proud. As a parent of a child with CFC, I can see and experience the direct results of my efforts. If any of you are interested in planning a fund raising event for CFC, I would be happy to help in any way that I can. We welcome any of our CFC family to attend next year!

Conference Gift Baskets Support Event

Every year we host a conference, we have an "international" tradition. Families from all over take a great deal of pride in creating or obtaining gift baskets that feature favorite food, drinks and items from their home states and countries. The tables are set up for this silent auction on the night we have our final banquet dinner. The competition can become fierce as families bid against each other to take home a piece of culture from other regions.

At the 2007 Orlando conference, gift baskets raised $4,000 to help offset conference expenses. Even our hotel conference manager at the Rosen Centre in Orlando was so touched by our children that he ran home one night and grabbed his special golf visor hat that was a prized item from a prestigious tournament in Orlando, FL just a few months prior. He bought some golf balls to add to this "golf theme".

We will have another silent auction at the 2009 family conference in Berkeley, California this coming August. We encourage families to participate again. You can be as creative as you like and even ask businesses in your community to donate items for your baskets.

Basket donated by the Clark family from their home state of Texas from 2007 conference

Judy Doyle is coordinating the gift baskets so please contact her at jadoyle@zoominternet.net if you wish to participate. Even if you are not attending the conference but have an item that you would be willing to donate, it would be most welcome. Your donation does not need to be a whole basket but something valued at least $20.

A Make-A-Wish Happy Birthday

About two months ago, I contacted the Make-A-Wish Foundation. I filled out an application for my daughter McKenna to get a wish. About a month later, I was contacted by a representative. I was hoping to maybe get a swing for McKenna. I had no idea how wonderful making this wish was going to be.

Our family consists of my husband, Vince, myself, and our three daughters; Mikayla (13), Mariah (10 ½) , and McKenna (6). McKenna has a confirmed CFC diagnosis. She is also diagnosed PDD-NOS (early childhood autism). We live in Corsicana, Texas which is 50 miles south of Dallas.

I really didn't know what to say when requesting a wish. McKenna can't make a wish for herself. Some wishes, like trips to Disney, a play set, or meeting a famous person just didn't apply to her. She doesn't like to be outside or travel. She doesn't walk or talk, so other wishes just didn't apply either. After I talked to the representative and told her about McKenna, she mentioned a sensory room. I wasn't exactly sure what a sensory room was, but having a child with severe sensory issues, it sounded awesome!

I decided that a sensory room was going to be her wish. The local volunteer wish granter then contacted me. She brought catalogs for children with special needs and told me to pick out things that I thought McKenna could use. I thought - wait a minute. You're giving me the freedom to choose just about anything that I think my daughter could use? Something that I can't get on my own that she could benefit from? WOW! I was so excited for McKenna.

I met with her PT and OT and we started looking through catalogs. Abilitations is a great special needs catalog, and is where most of the things we picked out came from. We tried to address all of her needs. She is very tactile defensive. She likes to look at lights and colors. She loves to swing. All of these things helped us when choosing items.

McKenna's brand new sensory room contains these items: therapy net, tadpole swing, biggie adapted swing, two therapy balls, rocker balance board, mat, game terrain mat, speech mirror, Rifton toddler chair with tray, weighted blankets, weighted lap pad, octopad, snuggle wrap, morph nightlight, lighted fan with switch, vibrating blanket, vibrating tube, arm & leg weights, core disc, gel-filled wheelchair tray, exploring tunnel, vibrating lighted hand wands, sound activated lighted disk, tumble form roll, and various textured balls.

This is a dream come true. This equipment is helping in her therapy already. We celebrate what McKenna can do, not what she can't. Plus, SHE LOVES TO SWING!! That is definitely her favorite thing. She is so relaxed when she is swinging. Our family has been extremely blessed with this wish. McKenna's sixth birthday will always be extra special because it was also the day that a wish came true.

McKenna Garcia playing on her game terrain mat

Sheila Garcia

Michael's Journey from Noonan Syndrome to CFC (Continued)

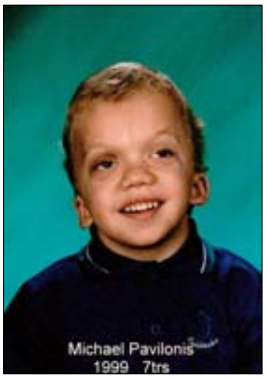
The first two years of Michael's life were frightening, educational, exhausting and full of love and support from our wonderful friends and family. Much of our time was spent trying to keep Michael alive and healthy, but we also had several other issues with Michael that we were dealing with and began to focus on them more after his heart surgery.

Michael received special services from Illinois' Early Intervention program at 6 months old and received special education, physical, occupational and speech therapy. He was unable to suck, we spent hours each day trying to feed him and he would usually vomit most of what he had eaten. Michael had significant speech/language and fine motor delays. He did not walk until he was 20 months old and had difficulty transitioning to different surfaces. He had constant ear infections, did not sleep through the night until he was 4 years old and was not completely potty trained until he was 6 years old. One of our biggest challenges was his irritability, anxiety and his inability to deal with change or disappointment. He had self abusive behavior such as biting his arm, banging his head on the floor or with his hand, biting and ripping his shirts and things throughout our home.

When Michael was 3 years old our options for Early Childhood education were very limited in Illinois and it became very confusing and stressful. We eventually got him into an appropriate educational program, however his progress at school was very slow and each year it seemed that he had forgotten what he had already learned. He had been diagnosed as mild/moderate retardation with a composite IQ of 62.

He was diagnosed with Autistic Spectrum disorder and ADHD when he was 6 years old and his educational needs were becoming much more complex and his self abusive behavior continued. We started him on Ritalin and that seemed to help him stay focused at school and reduce his anxiety. Michael's challenges are with speech/language, cognitive development, visual attention, sensory motor, visual motor and anxiety with self-injurious behavior. Michael has issues with visual tracking and fixating accurately on objects. His visual impairments affect his reading skills and daily functioning.

In 1999, when Michael was 7 years old he was accepted into a special Christian day school after a stressful challenge from our school district. It was a wonderful feeling knowing that Michael's educational needs would be met in a safe environment where he could gain some independence and reach his God given potential.



Michael at age 7

When Michael was born in 1992 we gathered as much information that was available about Noonan Syndrome and traveled to San Jose, California in 1993 to our first Noonan Syndrome conference. There were 8 families in attendance that traveled from Florida, New York, Chicago, California and Canada. This support group dissolved by the following year and a new Noonan Syndrome support group was founded by Wanda Robinson in 1996. We were involved with the Noonan Syndrome support group since its inception and traveled across the country to every conference annually for 11 years.

For years we were confused that Michael's cognitive development, motor skills and heart disease were unlike many of the other Noonan Syndrome children that we met at the conferences. One year I spent time at the Minnesota Noonan Syndrome Conference with Brenda Conger, President, and Founder of CFC International. Brenda and I decided to go shopping and we shared characteristics of her son Clifford (CFC) and Michael (unconfirmed Noonan Syndrome). Their similarities were remarkable and were more similar than any of the Noonan Syndrome children I had met. I attended the scientific symposium in Baltimore where they discussed research on Noonan, Costello and CFC syndromes and I started to look more closely at CFC. I continued to meet with the doctors and specialists at the Noonan Syndrome conferences and they were confident that Michael had Noonan Syndrome.


Michael at age 15

In 2007, at the Noonan Syndrome Conference there was new information about Noonan Syndrome and CFC genes. I met with Dr. Amy Roberts and agreed to have Michael participate in a Noonan Syndrome study. We met with Michael's geneticist to discuss the information that I learned regarding both syndromes and she was certain Michael had Noonan Syndrome and did not think it was necessary to test him for CFC but I asked her to test him anyway. In October, 2007, the test results were negative to all four Noonan Syndrome genes and positive to the CFC BRAF gene. When I first received the news from the genetic counselor she said 'it was mother's

intuition', I was overwhelmed with the news and relieved at the same time. We spent 15 years focusing on Noonan Syndrome, educating Michael's doctors, specialists, teachers, family and friends on what it is and how it has affected him. However, it does not matter what syndrome Michael has, we prioritize his special medical, education and social needs, provide support and develop opportunities for him to overcome his challenges.

Today, Michael is 16 years old and recently had pacemaker surgery to help him with his heart block issue which returned in 2007. He will remain in his special day school where he continues to receive special education and services until he is 22 years old. They are helping us plan for his transition to adult life.

I recently read an email message from a mother on the CFC list serve that stated 'I am at wits end and feel like I'm on my own' and I thought that's often how I feel. Several families responded just as I had and provided wonderful words of encouragement and support for the mother. Our journey to CFC has been a little bumpy, but it is a much easier ride when supported by CFC families that understand exactly what you are going through.

Effects of Germline Mutations Within the Ras Pathway on Bone Remodeling

Dr. Reinker will again be performing orthopedic exams on individuals with CFC syndrome at the 2009 conference, and Dr. Stevenson will be joining him during these examinations. Dr. Reinker has had a long-standing interest in the orthopedic problems associated with CFC syndrome.

Dr. Stevenson and his research team have been investigating the skeletal problems of individuals with syndromes of the Ras-MAPK pathway such as Cardio-facio-cutaneous (CFC) syndrome, Neurofibromatois type 1 (NF1), Noonan syndrome, and Costello syndrome. Although each of these syndromes are distinct genetic conditions, they have some clinical similarities including skeletal abnormalities.

Dr. Stevenson's research goal is to investigate the overlapping skeletal problems in syndromes of the Ras-MAPK pathway by evaluating the function of cells that remodel bone. Participation in research is entirely voluntary and one can elect to participate in some or all of the aspects of Dr. Stevenson's research protocol. A brief overview of his research investigating bone concerns in CFC syndrome is as follows:

1. They hypothesize that the resorption of bone is increased in CFC syndrome. Some of the breakdown products of bone are excreted in the urine. Therefore, two urine samples will be collected for analysis of an increase in bone resorption markers. The type of mutation will be correlated with the bone resorption values.
2. They will also look at the activity of cells that break down bone called "osteoclasts". The osteoclasts are cultured from cells that come from a peripheral blood sample. The osteoclasts do not survive long and need to be processed quickly and hence fresh blood samples would be needed. The function of the osteoclasts will be measured looking for increased activity of osteoclasts.
3. The last part of the study is to look at the microarchitecture and histomorphometry of bone samples in individuals with CFC syndrome. This would only be done if for some reason and individual with CFC syndrome were to undergo an orthopedic surgical procedure in which bone samples were to be removed and thrown away. In the future if this were to occur the investigators would ask that the bone samples be sent to them for analysis. This would be performed to look for evidence of decreased mineralization and abnormal bone structure.

The musculoskeletal findings in CFC syndrome are not well delineated. Individuals with NF1 have significant scoliosis and fractures with non-union and individuals with Costello syndrome and NF1 have been reported to have osteoporosis. Since all of these syndromes are due to mutations in genes that are closely related within a similar molecular pathway, a better understanding of the similarities and differences between these syndromes will likely lead to better management and treatment protocols. The success of treating skeletal abnormalities in one condition may help in the treatment of other related conditions.

California Conference Event Eye Exams

We are excited to announce that we have arranged eye exams for children with CFC, Costello and Noonan syndromes in the upcoming Ras/MAPK pathway family conferences to be held this summer. For children with CFC, exams will be held on Sunday August 2, 2009.

In a collaborative effort, three groups: Duke University, UC Berkeley, and UC San Francisco will be participating in this event: Terri L. Young, MD., Professor of Ophthalmology and Pediatrics, Duke University, Deborah Orel-Bixler, Professor of Clinical Optometry, PhD., OD., FAAO., and Dr. Tonya Watson, OD., PhD., a post-doctorate, University of California, Berkeley, Suma Shankar MD., PhD. and Katayoon Ebrahimi, MD., clinical fellows, University of California, San Francisco; in addition, parent and student

volunteers will be helping in coordinating the children and eye exams. All clinicians have expertise in eye exams of children and have special interests in Ras/MAPK pathway syndromes.

The exams are going to be performed in two of the conference rooms at the hotel and also in the traveling vision van from UC Berkeley. The van is equipped for performing 4 exams at once.

The exam itself will be similar to visiting you local eye doctor. It is anticipated that it may take about 1 to 1.5 hours. A questionnaire and consent form will have to be filled out before the exams. Eye drops will be used to dilate the eyes.

Please visit the CFC website at www.cfcsyndrome.org to locate the conference registration form and check yes for vision exam if you are interested in participating.

The eye exams will contribute towards putting together the ocular manifestations in children with the Ras/MAPK pathway syndromes and will also help in understanding the role of Ras/MAPK pathway in normal eye development.

Berkeley Conference Dental Exams

The genetic basis of CFC was recently identified. Knowing the causative gene for CFC now allows us to reevaluate the clinical characteristics of this rare syndrome. Dental findings in these patients have not been well characterized, although these individuals are thought to have dysplastic (abnormal) teeth. Now that the causative genes for CFC have been identified, we are attempting to gain a better understanding of the spectrum of tooth findings. In particular, we are interested in the percentage of mutation positive individuals with tooth findings and exactly defining what the findings are, such as missing teeth, too many teeth, abnormally shaped teeth, and poorly functioning teeth. We hope this study will help us understand the teeth of CFC syndrome individuals so that physicians and dentists may provide better anticipatory guidance and care for their patients. It will also assist in understanding the pathophysiology of the pathway affected in CFC in human development.

Dental exams will be held at the CFC International family conference on Sunday August 2nd. The members of the dental team for the CFC exams are all UCSF faculty or trainees and include:

Ophir Klein, MD, PhD - genetics and developmental biology of tooth anomalies.
Kate Rauen, MD, PhD - medical geneticist.
Sneha Oberoi, DDS - orthodontist that specializes in craniofacial anomalies and complex conditions.
Jessica Groth, DDS - pediatric dentistry.
Alice Goodwin, candidate for DDS/PhD – will assist the team with dental exams.

CFC Index Review Research Project

CFC Researchers Dr. Maria Ines Kavamura, Federal University of Sao Paulo, Brazil and Dr. Giovanni Neri, Universita Cattolica Del Sacro Cuore, Rome, Italy have a project underway to revise the CFC Index that was originally published in 2002.

Enormous progress in the knowledge of CFC syndrome has happened in the last couple of years. The finding of the mutated genes is a very important step towards the identification of CFC patients, the understanding of the mechanisms involved in the genesis of the disorder and, potentially, in the discovery of treatments that can be offered in the future. The fact that we now have a number of patients with proven CFC syndrome (carriers of BRAF, MEK1 or MEK2 mutations) allows us to define CFC clinically. Our proposal is to analyze CFC International BioBank clinical data from molecularly proven CFC patients and update the CFC Index, which was created before molecular tests were available and based on data from patients that presumably had CFC. The development of a clinical tool to diagnose CFC is of great importance for the clinicians and this study may also help to define the boundaries with phenotypic overlapping disorders, such as Noonan and Costello syndromes.

A glimpse is not a vision. But to a man on a mountain road at night, a glimpse of the next three feet of road may matter more than a vision of the horizon.

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CFC International Annual Report 2008

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.

To Our Families, Friends and Donors:
Every year at this time, I look back at our past year and summarize our organization's accomplishments. Although 2008 was the beginning of a very bleak time in economic history, we again saw a year when our dedicated volunteers, faithful contributors, caring physicians and brilliant researchers have created and accomplished so much. This annual report reflects our triumph of progress and hope.

Most of the people on the front lines of these efforts are parents who have a very personal reason for volunteering. But in each year of growth we meet more and more friends who walk with us to support us on this challenging journey in CFC land.

Cardio-Facio-Cutaneous (CFC) International is now ten years old and no longer a novice charity. We have accomplished a great deal in such a short time. But at our core, all of the people who comprise the CFC syndrome organization share a simple value. We are a family. We care deeply about each other. We give each other the strength, courage and hope to live, to laugh and to face whatever may come each day, knowing that we are not alone. We embrace each other and send messages of hope and prayers when a child is ill or a family is dealing with a death. Our mission is … "Forging a path to improve lives through family support, research and education". I know that we have made a huge difference in the world through our services to families and by promoting extraordinary new research on this rare condition. I hope all our families feel as proud as I do of our accomplishments. We could not have made such unbelievable progress without all of you. Thank you to each and every person!

Brenda Conger
President & Executive Director

CFC International 2008 Accomplishments

Family Services *(Led by Molly Santa Cruz and Judy Doyle)*

Family Service representatives contact and care for our families, giving them the strength, knowledge and tools to care for their own. The latest census shows that CFC International has grown to a total of 156 registered families from around the world. When new families reach out to CFC International, Molly Santa Cruz and Judy Doyle are there to welcome and support them. Many of our newer families tell us that they were anxious about contacting us for the first time, not knowing what they would find. Those who do find a warm voice and a gentle introduction to a new family. Judy Doyle connects them with the private family listserv—our global lifeline on the internet and the place where no question is too small, answers come quickly and everyone understands. Molly ships them out the new parent package with a note to contact her with any questions.

CFC International produces a fact sheet, Parent's Guide, brochure, and private family address directory. We ship new family packages out to all regions of the world. In 2008 20 packages were shipped out to new families who contacted us.

Individuals Registered With CFC International

Australia 4	France 1	Ireland 2	Netherlands 3	Switzerland 1
Canada 8	Germany 1	Israel 2	Samoa 1	USA 112
Denmark 2	Greece 1	Malaysia 1	Scotland 2	**Total 156**
England 12	Hong Kong 1	Mexico 1	South Africa 1	

As much as CFC International is an internet community, there is nothing like the opportunity to spend time together. Our families can't wait to attend a conference and meet others they have chatted on line with.

CFC Conference *(Led by Brenda Conger and Molly Santa Cruz)*

Planning for the 2009 Scientific, Medical and Family Conference began as the 2007 Conference was coming to a close. Molly Santa Cruz led the search, site visits, and negotiations for the location for this year's conference in Berkeley, California. Amy Hess researched professional child care companies for not only CFC International but for Noonan and Costello syndrome organizations. Pilar Magoulas has coordinated the Stanford University genetic counseling students to assist with the dental, ophthalmology, genetic, and cardiology appointments. Brenda Conger contacted workshop speakers and doctors to set up the specific talks and agenda and worked hand in hand with Wanda Robinson from the Noonan Syndrome Family Support group to partner with a number of shared workshops. Our conference closing ceremony DVD production is again created by board member and dad Kyle Stowell.

Science and Medicine

BioBank & Clinical Registry program *(Led by Brenda Conger, Molly Santa Cruz and Amy Hess)*

Our original BioBank program was focused on DNA collection and storage. Today we maintain the DNA storage as a partner project with the Genetic Alliance BioBank and other syndrome organizations. In 2008 we saw an increase in research on our genetically confirmed individuals with CFC thanks to the efforts made by the families who enrolled in the BioBank and the previous gene discoveries. Brenda receives all the shipments of clinical data and has student volunteers who blind the data to remove all confidential identifiers. After the data is blinded and sorted into the correct medical categories, the files are scanned into PDF files by a staff member at the Ferre Institute in Binghamton, NY. Amy Hess then is sent a list of what clinical data has been scanned in and enters this information on a confidential spreadsheet linked to the individual's BioBank number. Researchers submit study proposals and upon approval from our Scientific Advisory Board, they are shipped out specific data that involves their study on a CD file. Molly Santa Cruz works with researchers who request that parents fill out specific surveys. She sends the surveys out to parents and continues to follow up and keep track of the finished paperwork. A very nice team approach with detailed service and record keeping provided to our dedicated researchers saving them a great deal of time. All projects are done with use of the confidential BioBank numbers to protect confidentially of our families.

Research Projects

Collecting medical reports (clinical data) on CFC individuals since 2002 has really paid off! Years ago we asked parents to only send in their child's genetics reports along with a Medical Advisory Board approved Registry. As time went on we gradually asked for all medical reports that families could get their hands on. The reports encompass cardiology, genetics, ENT, ophthalmology, orthopedic, endocrinology, neurology, and any other specialist the CFC person met with. Today, the years and years of extreme data collection have brought about extensive research projects in 2008:

1. Ophthalmic and Hearing Manifestations in CFC – Duke University Eye Clinic and UCSF
2. 3D Digital Surface Photogrametry to capture, visualize and analyze Dysmorphic Facial Features – UCL Institute of Child Health, London
3. SOS1 Gene Mutation Studies on individuals found negative for CFC gene testing – Harvard Medical School
4. Communication & Adaptive Behavior in Individuals with CFC syndrome – University of Wisconsin - Madison
5. Prenatal Manifestations in CFC syndrome – UCSF
6. The Spectrum of Dermatologic Findings in 55 Mutation Positive Individuals with CFC syndrome – Oregon Health and Science University and UCSF
7. CFC Index Review – Universita Cattolica Del Sacro Cuore, Italy and Federal University of Sao Paulo, BraziL

None of the above projects would have materialized if it were not for the endless hours our college intern Amber Chavalier contributed to CFC International. Her scanning of volumes of medical records has enabled us to now ship large quantities of clinical data to researchers on CD's.

Scientific and Medical Meetings

2006 was the first year of Scientific meetings on CFC and Noonan syndrome. Thanks to Dr. Amy Roberts for coordinating this first ever meeting of the great minds from both organizations. Speakers from all over the world were in attendance as well as board members from CFC International. In 2007 a European Scientific Meeting was held in Barcelona, Spain with researchers from the European region. Brenda Conger represented the CFC International families at this event and shared literature and information on our parent driven organization. In early 2007 Dr. Kate Rauen from UCSF stepped up to the plate to represent all the disorders on the RAS/MAPK pathway and embarked on non-stop grant writing to pull together a first time USA Scientific meeting for CFC, Costello, Noonan and Neurofibromatosis. Support groups leaders in turn have been working on coordination of family conferences for this August 2009 event all at the same hotel in Berkeley, CA. A huge amount of coordination and work on everyone's part!

CFC International Annual Report 2008

Caring, Facilitating & Connecting

Contributions

CFC International raised $67,573 from contributors including our fundraisers in 2008. These financial supporters are so vital to the continued survival and progress of CFC International. Without your contributions, none of the important programs described above would be possible. Your donations fund advances in research. You fund physician awareness and the cost of operating and maintaining our Biobank, website and list serve—such vital links for our families and physicians. Our upcoming CFC International Conference at the Doubletree Marina Hotel in Berkeley, CA will cost over $30,000 to host and is a great investment.

Equally critical are the families and friends who led the fund raising efforts that raised this money. People don't give to institutions or charities; they give to a cause they have come to believe in. A rare and obscure disorder like CFC syndrome is not a high profile cause. There are no government or corporate grants or celebrities endorsing CFC syndrome, and most people have never heard of it let alone known someone who is affected. Except our donors.

Every one of them knows someone with CFC syndrome personally or has been told the story by a good friend who does. Every donor was asked to donate to CFC International by someone. These are our heroes. The ones who ask. The ones who tell their friends and family of the challenge of CFC syndrome and the hope of CFC International. The ones who understand that without financial support, progress stops. So our thanks go to our donors and we ask you to please keep CFC International at the forefront of your generosity, even in these difficult economic times. But we reserve our highest praise and deepest appreciation for our fund raisers, for they keep the lights shining… literally!

- Judy and Tim Doyle's annual chili cook off held in Ohio — Each year the Doyle family bring their community together to sample the best chili in their region. The silent auction, donations, TV raffle and chili samples raise money for CFC International at this very popular event.
- Meg Young loves to dance! Each year Meg's parents, Sherri and Scott, invite their friends and family to attend a dance event with proceeds to benefit CFC International.
- Cash in your Gold – Clifford Conger's Godmother, Beth Kaufman invited neighborhood friends over to cash in their old and broken gold jewelry. Beth donated her proceeds from hosting this party, along with some other friend's checks to CFC International and brought fundraising to a sparkling level.
- Each year Matt Brockwell has sent a letter with CFC syndrome information and updates on his daughter Kate's progress to a list of coworkers at PricewaterhouseCoopers to appeal for consideration of donations earmarked to CFC International during the United Way Campaign. This appeal continues to raise thousands of dollars each and every year. A simple letter from the heart is making a huge difference!
- Car club president, Ron DeHaus and the Binghamton, NY Cruisin' Buddies Rod and Custom car club love to get out and raise money for local charities. Each year the club shows off their cars at an annual Memory Cruise. Again they chose CFC International as their charity to support for their annual fall cruise.
- 6th Annual "Toast the Angels" – Brenda and Cliff Conger host this fabulous wine tasting and dinner year after year to honor all CFC children here and those who have passed. Friends continue to support this huge event and start asking about the next date right after the first of each year since it is a sure hit and reserved on many a calendar.

CFC International is a member of the Genetic Alliance and an Associate member of the National Organization for Rare Disorders

CFC International Income and Expenses 2008:

INCOME 2008		**EXPENSES 2008**	
Contributions	$27,564	Program Services	$22,139
Fundraisers	$40,009	Management & General	$18,539
Other	$6,402	Fundraising	$9,493
Total Support & Revenue	**$73,975**	**Total Expenses**	**$50,171**

CFC international

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Vestal, NY 13850

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RETURN SERVICE REQUESTED

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: _____