



CFCinternational

Cardio-Facio-Cutaneous Syndrome

The CFC Chronicle

Caring, Facilitating & Connecting

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Featured Family: Zachiah's Journey

In the middle of our world famous scenery, snow capped mountains to dense native forest lay the plains of Blenheim, a small town commonly known for producing award winning wines. And this town, our home town is where we live, love and are blessed to raise our beautiful family consisting of myself Karrin, my husband Martin, Nathanael 10yrs, Danielle 8yrs and Zachiah 6 yrs.



Nathanael, Martin, Zachiah, Karin and Danielle

On the 10th of February, 2004 we welcomed Zachiah into the world. He was born three weeks early and the 2nd of our children to be born at home by choice. After a normal pregnancy he arrived fit and healthy. Within the first six weeks, although being breastfed in the same manner as his brother and sister his weight plateaued then began to fall off the growth chart to create a centile of his own. Tube feeding began, and then stopped when sufficient weight was put on and the stress of home tube feeding became all too much. Other than this he showed no other signs of ill health. Decisions were made not to insert a Gtube as his appetite increased unlike his weight and he began to eat as most normal children do. Doctors now waited for his growth graph to take off as it should have done, but today it still continues along its own path. Beautiful hair and a face that lit up the room when he wasn't crying masked the fragile thin child under his clothes.

Martin and I own and work for a small 3rd generation family Equestrian/canvas business - Brooks Saddlers, along with having both our families here we are blessed with Martin being able to step away from work to care for two other small children while I spent time in and out of the hospital with Zachiah. The hospital we are lucky to have is only a five minute drive away, one of the advantages of being in a small town.

Blood test after blood test was done. Fragile thin arms meant blood was drawn by pin pick from his heel. Each test returned negative with a sigh of relief and thanks to God yet a question still left as to what was happening in this beautiful little boy's body.

FAILURE TO THRIVE seemed to be the only certain thing doctors knew.

2006 saw our worst medical fear of seizures starting and although medicated for convulsive seizures now, he still often has absent ones.

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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

In this final newsletter of 2010 I wanted to send a thank you to our board, medical staff, and volunteers for their ongoing commitment to CFC International. 2010 has been a year of challenges, growth and accomplishments.

The end of next year brings about the final year of our first Strategic Plan (2005 – 2011) formulated by our board during our face-to-face meeting back in 2005 at the Orlando, FL conference. I am proud to say that we have accomplished all our goals that the board established! Our board will meet again at the Chicago event and jointly set up the next Strategic Plan for CFC International.

During this year we started work with The Noonan Syndrome Support Group and The US Costello Syndrome Family Network to establish a USA 'Rasopathies' (developmental syndromes of Ras/MAPK pathway dysregulation) joint organization. Each organization has a team of leaders who work closely with a chair to develop an accurate way to track all our data. This collaborated effort will set up a new Registry to track all the individuals born on this rare and complex pathway. In addition to the USA 'Rasopathies', our three organization leaders are spearheading a joint Conference for families and researchers this coming July in Chicago. The economic challenges in today's difficult times make joint conference planning a smart move. All our families have similar education, legal, social and health care planning issues so sharing the workshops speakers and professional childcare saves us considerable funds. We will still have separate meals and private meeting space planned as well. As usual, our traditional Mom and Dad's Nite out will be held at the conference.

The 2009 to 2010 years have brought about tremendous growth. We now have a Join button on our website and this data has clearly illuminated how many new babies are being tested for syndromes on the RAS-MAPK pathway. I believe that CFC Syndrome is not as extremely rare as originally thought back in the 1980's and 1990's. Our organization's data collection will be valuable for future research so we can keep families informed.

I wish you all a healthy and happy 2011!

Brenda Conger

E-mail: bconger@cfc syndrome.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.

Donations in honor of:

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Maureen Sidbury
Daniel Hess
Bobbie Olsen
Nola Iacobelli
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Clara Ruth Edwards
Richard Borkowski
Lindsay Levenhagen
Tom & Laurie Levenhagen
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In Memory of Nora

Our beautiful baby girl, Nora Jane Volker, filled our lives with love and laughter for exactly 14 months. She was born with an acute heart condition called DORV with a hypoplastic left ventricle. At her birth there was a lot of hope her heart would develop in a way that would only require one heart surgery. It was not meant to be that way though and she faced challenge after challenge including two open heart surgeries, a diaphragm placcation, anal rectal surgery, tracheotomy with ventilator, 24/7 nursing her entire life, and two lengthy hospitalizations: 10 weeks after birth and 6 weeks after second heart surgery until her death on August 29, 2010.

Despite these challenges Nora managed to teach the people that surrounded her lessons that last a lifetime. First and foremost she taught us how to love selflessly, to courageously face the future, to graciously tolerate discomfort, to greet everyone with a smile, and to move forward with willful determination. Precious daughter thank you for all you gave us in your short beautiful life.

We are forever changed and forever grateful. You are missed but we know you are finally free.

www.caringbridge.org/visit/norajane

Molly & Aaron Volker
St. Cloud, Minnesota



Racing for Christina

Let's start to begin on how I got started to run long distances. Around this time last year, a group of my co-workers talked about running a half-marathon. I piped up and said I would like to do that. They all laughed because I was not in the greatest shape. A half-marathon is a 21K race and that was a bit ambitious as I had run a maximum of 10K but that was in high school, which was over 20 years ago. Well once someone says I cannot do that; it motivates me to prove them wrong. I trained by myself and have been since day one. In Sept 2009 I ran the Scotia Bank half marathon and did it in a time of 2:29. I was very proud of myself.

My cousin Jeremy is also into running and thanks to him, he kept pushing me to train for that race. He then convinced me to run the Run Around the Bay in Hamilton this past March, which was 30K. It took me 3hr and 59 minutes. My cousin and I then committed to run the Scotia Bank full marathon on Sept 26 and the distance is 42K which is a distance I thought to myself was unattainable at one point in time.

As such I took my training to a more serious level. This year, I changed my eating habits by cutting out all the bad carbohydrates such as white bread, rice and pasta. I also gave up drinking beer during the summer (though I will have the odd one). This is a huge sacrifice for me because for those who know me know how much I love food and beer. I have lost 20 pounds and am down a weight that I have not been since high school. I am in the best shape in my life for some time (knock on wood) and it feels great. The biggest challenge I am facing these days is finding clothes that still fit me. I've had to tailor my pants 3 sizes down and have had to continually buy new clothes. Not cheap.

Aside from my running I have decided to try and raise money for a specific charity. This charity is near and dear to me because of one specific family and one little special girl. Her name is Christina as she is my dear cousin's (Monique) daughter. My cousin has been married to her husband for approx 19 years. Her daughter is 8 years of age and she also has 2 boys aged 6 and 17. So least to say it is a very busy household.

As for Christina, she is a special needs child who needs constant supervision 24/7. Christina has brought much love and compassion into their home along with that there has been a lot of emotional turmoil as well. It started from birth and failure to thrive and not placing a g-tube till she was 7 months where she had only gained two pounds. Once the g-

tube was placed things started looking good as she started to gain weight and look a lot more like a healthy baby that they hoped she would have been. For the next few years Christina was not meeting milestones so they had all the services put into place but still no major progress. She did sit up at 8 months but continued to take nothing orally and did not walk or was in any way mobile. At about 6 years of age with the help of her nanny and amazing teachers she started to use a walker, leg braces and seemed to be on her way to becoming mobile. Then tragedy hit in 2008 when she suffered a status seizure lasting at least 2 hours. As a result she suffered brain injury to her frontal lobe of her brain and had lost all her physical gains that she had made. She stayed at Sick Kids for 4 weeks then a rehab center for children for three. When she finished her stint there she was still not back to her baseline physically and still is not to this day. Getting there but still has a long way to go.



Christina and Andre

Since the major seizure, Christina has been in and out of the hospital for the past 2 years with her family always by her side. This has been very stressful for them but with the help and support

of friends and some family they were able to get by. Christina has brought much joy to their lives and they have said they would not change her for anything in the world but they do have goals for her. They want her to be as independent as she can. As of right now she is dependent for every aspect of her life. She does not talk or have any formal communication and her walking can be done with only assistance. She likes to hit her chin when she is angry or upset or requires attention.

The biggest challenge for my cousin is not knowing exactly what was wrong with her. At one point in Christina's life, they diagnosed her with severe autism and tried to provide treatment based on that. Only this past month were the specialists and doctors able to identify it as cardiofaciocutaneous syndrome, which is an extremely rare disease. There is no cure for this at the moment, only treatment. As such I have asked my friends and family to help by donating to CFC International.

Only this past year have I realized the sacrifices my cousin and her family have made for this girl and though my spare time is scarce to train for this race, this is nowhere nearly the sacrifice that my cousin and her family have done for Christina. I commend them for being so positive in such a stressful environment.



Andrea running the marathon

Sept 26 was race day! It was a great day weather wise to run. I knew that there was a good chance the course record would be broken (not by me of course) and sure enough it was. Anyways, it was definitely a tough go. I hit the brick wall a lot earlier than I anticipated as it hit me at the 18K mark. Thankfully I had my energy jubes and picked it up at the 30K mark to finish in an overall time of 4:54 and was extremely happy that

my family and my cousin's family were there to see me at the end. Though my goal was to finish it at 4:30, I will take it and be thankful. I had my first pitcher of beer after my race. Well deserved I say! This was truly a memorable day for me and I wish to thank everyone who supported me throughout this process.

On behalf of Christina, I want to thank you all for your support and love as we reached our goal of raising over \$1000 for CFC International. They will be ecstatic for the money we raised. If it were not for her, I would not have been able to spend the quality time with my family/friends this past summer nor would I have met some new friends along the way. Thank you Christina.

Health and happiness to you all!

André Wong
Canada

Tobby Family Establishes Research Fund

Well we did it. I kept my word. We had a very successful first Hawaii CFC fundraiser. It all started with me attending the American Society of Human Genetics conference here in Honolulu last year as a CFC parent advocate. It was there at Dr Kate Rauens session, yes our Dr. Kate Rauen who sits on our medical advisory board, the one who discovered the CFC genes causing this syndrome which resulted in a blood test so individuals can now be diagnosed...yeah that fabulous, extraordinary Dr. Kate Rauen.....shared information at that conference, that when examining a CFC developmental model using zebra fish, the effects of the CFC genetic change can be normalized. What was even more amazing was the team of researchers found a small window of treatment opportunity --- in that they applied the drug for only one hour and took it away, but the embryo continued to develop normally even with no more drug exposure!! Small fish but a BIG success! HUGE!!! So now



The Tobby Family

I want to wallpaper Amara's room with Zebra fish. Viva la zebra fish!!!

I was so impressed with that total experience, witnessing the dedication of researchers to our families that I left there feeling I

had to do something to help promote research. What could I bring to this table? I walked away with one thing in mind. I would host a fundraiser. I can do that.

Although I had never done any fundraising, I can throw a party. God knows how many of those I have done over the years. So I shared my plan with a friend who does sales representation like me, Marilyn Nagel, and she immediately volunteered to help. She had experience with fundraising for some local groups. Then our friend and president of our local organization here, Hawaii Sales Representative Assoc (HSRA) Ginny Wright graciously volunteered to help us too. With three hearts dedicated to a mission to help CFC

International establish a research fund, we had a date, a plan and mailed 175 invitations.



Hula dancers Tommi, her mom Jen and Tomas

Our event was held on Nov 7th at our local Elks Club in Waikiki. This is a beautiful location with stunning ocean views,

valet parking, buffet, pupu's and a no host bar. We collected great items for our silent auction from friends and local sales people. Everyone enjoyed the live music of Hawaiian piano stylings by our dear family friend Joe Mahoe along with Zumba by my new friends and teachers Jen and Tomas. Their lovely young daughter Tommi entertained all with a beautiful hula. All volunteered their time & talents from the heart. It was so amazing and beautiful. In attendance we had 48 friends join us. Jan Harada, Amara's EA volunteered to be our party photographer. She captured some great moments for us. Her special girlfriends she's known for



Savannah, Amara, Timmy & Alissa

years, Alissa and Savannah and boyfriend Timmy joined Amara. We are so blessed to have such wonderful people around us.

Everyone had a great time. We raised a grand total of \$5,200 dollars to establish a CFC International Research Fund. Yahoo! We hope to make it an annual event and raise millions, because our ultimate goal is to make CFC a thing of the past or as we say in the islands...all Pau!

Cruisin' Buddies Annual Memory Cruise



Cars on display at the Memory Cruise

On September 10, 2010 the Cruisin' Buddies Rod, Custom & Classic Car Club, Inc. from Binghamton, NY held their annual Memory Cruise. The parade of cars drove from

Endicott to Green field Ballpark in Johnson City, NY. CFC International was awarded a \$500 check from the Cruisin' Buddies. Clifford enjoyed a fun filled night of dancing, and eating his favorite meal of hotdogs with all his friends at Cruisin' Buddies.



Cruisin' Buddies Ron DeHaas and Clifford Conger

The evening continued on with another entertaining night of 50's musical skits with members dressing up and participating on stage. CFC International thanks Cruisin' Buddies for their generosity through the years!

Photo Gallery



Shayanna Smith, age 4,



Kate Brockwell, age 13



Daniel Hess, age 9



Avery Clark,
age 5

Dulce Flores,
Age 11



Leah Borian, 17 months



Francesco Serpe, age 5



McKenna Garcia, age 7



Amara Tobby, age 18



Logan Richardson, age 8



Josh Schrock, age 15



Clara Edwards,
age 2

Cameron, age 6
and Regan,
age 4 CFC



Brittany Bredikis, age 15



Emily Santa Cruz, age 13



Alaina Butcher, age 10 with
friend Joshy

Featured Family: Zachiah's Journey.....continued

Finally four years after consultation with a leading geneticist in New Zealand and waiting nine months for the blood test to be confirmed, Zachiah was diagnosed with CFC syndrome with a MEK1 mutation. As to date Zachiah may be one of only two in NZ to be diagnosed with this rare syndrome.

As per all other syndromes I had preread all I thought there was to know so I was not shocked by the diagnosis and was confident in sharing with our paediatrician things about CFC I had learnt and they were only just learning about.



Zachiah

Low in muscle tone we began therapy with our physio. Small town specialists are limited and consultation with them is not regular so we began our own in home therapy and

shared ideas with a friend who has a boy with cerebral palsy close in age to Zachiah. The two boys started hydrotherapy. With a love for water, bath times are by far his most relaxed and favourite part of the day.

Growth hormone treatment has just been stopped after three years of treatment with little significant growth. It amazes me that unlike his ability to grow at a normal rate his hair and nails need regular trims. Treatments, medications and equipment in New Zealand are all funded and so far we remain continually blessed.

Today Zachiah still does not walk. Sensory issues along with weight bearing plague him although we do have daily use of a kid walk walker to build up strength standing and support limited walking. Main mobility is done in a wheelchair. He has learnt and continues to build

up arm strength pushing the wheels to move himself. Cheekily he has learnt that he can do this in his walker by lifting his feet and moving the large wheels.

Although he does not walk he scoots around on his bottom and is starting to push through his feet to climb onto things. He has very limited speech but can make himself well known to those he is often around. His humour is unreal, quick and witty. Along with the many mysteries that are Zachiah is his unusual ability to clearly say a word and then never repeat it again. The only sign he has learnt is finished, bless him, as often he will tell us this as we start something he clearly does not want to do.

Unlike a lot of CFC kids Zachiah does not have a heart condition nor is G-tube fed. In the first year of his life Zachiah was a good eater but the introduction of new foods now days usually induces vomiting so with dietician's support his diet consists of his favourite "white chocolate buttons" which we buy by the 10kg box full and a concoction smoothie of pediasure with yoghurt, banana



Zachiah having fun with his dad on a self made slide

and weetbix.

Zachiah attends a mainstream primary school with his brother and sister. For the last year he has attended three hours a day supported by a teacher aide and is due to start full days as our last term for the year begins. Late last year Danielle was diagnosed with autism. And although she shows little interest in Zachiah small comments I overhear melt my heart. This week it was, with a whisper – Zachiah, you me and Jesus, we are all friends. Nathanael continues to show us his caring nature as big brother. He loves animals and is like a lioness with her

cub. When on the floor together he lets Zachiah crawl/climb all over him.

As a family we like to holiday around our amazing country so have bought a motor home which has made holiday with Zachiah easier and more manageable than the tent. In NZ we have a government paid respite care system and we pick and choose holiday destinations to take him on or leave him behind in the care of loving extend family.

Zachiah is Hebrew for Zachariah - meaning God has remembered. And that he has. Given into our loving care we consider ourselves truly blessed and believe God has lessons to teach us through him. Challenges include irritability, from the brightness of sunlight to self abusive behaviour of banging his head with his hand or on the floor and communication barriers.

For every tear there is laughter and his laughter like his smile is contagious. These moments make the bad ones bearable. God knows the beginning and the end and we put our trust in him for the in-between.

Karrin Brooks
Blenheim, New Zealand

First COSTELLO/CFC Conference, Thistel Hotel, London Heathrow, England

September 8 -11, 2010

*Written by Claire Griffiths, Mummy to Esther Robinson
Aged 7½ CFC Confirmed, England*

Firstly can I say a huge thank you to Colin, Cath and Helaina Stone, as without your vision, passion and caring attitude to our angels, none of this would have happened. Your sheer love for your daughter and other children has made Costello Kids and the incorporation of CFC UK and the RASopathies Network possible and truly unique.

Also a special thank you to Trisha Hannan, (mum to Emily) for all the hard work in co-organising such a wonderful event.

I'm very proud to say as Mummy to Esther the first UK Costello and CFC Conference was held at Thistle Hotel, London Heathrow from Wednesday 8th to Saturday 11th September 2010, and a wonderful time was had by all who attended.



Esther, Claire, Michael, and Jasmine Robinson

We were blessed by following:

- 8 Families from the UK (there were at least 3 more that showed interested but couldn't make the dates)
- 2 Families from Australia
- 1 Family from France
- 1 Family from Heidelberg
- 14 Families from the Costello Kids, some of whom were international guests
- Thursday full conference and dinner gave us 150 guests.

Wednesday started with registration at 4.00pm and then an informal dinner where everyone enjoyed catching up with old friends and making new ones. Thursday's breakfast was traditionally English which filled us for the day ahead, the children went to day care and the conference started at 9.30 prompt with an opening speech from Dr Bronwyn Kerr. We also had the following speakers:-

- The disorders of the RAS/MAPK pathway, an overview.
By Dr Emma Burkitt-Wright
- Management of severe speech and feeding difficulty.
By Rebecca Howarth
- Understanding differences about growth.
By Professor Peter Clayton
- The Skeleton in CS and CFC; current knowledge and research.
By Dr David Stevenson
- The Transition to Adult Services.
By Francis Binns and Kim Laurie

Break for Lunch

- Clinics for those who wish for an individual consultation
- Cancer risks in the RAS/MAPK disorders.
By Dr Bronwyn Kerr
- Panel Discussion

Evening Gala Dinner from 7.00pm was in a beautiful room all decorated with balloons, goodie bags for the children, 3 course hot dinner, raffle, auction and games followed by a singer and disco. A fun night filled with laughter and smiles.....a perfect end to a most informative but hectic day!!



Christie Dodd, Trinity Brown, Esther Robinson, Emily Hannon, & Nancy Newton

On Friday after an early breakfast, two coaches left the hotel bound for the House of Lords at 9.00am prompt. Travelling through London along an area called Embankment, encompassing some of the most unique English landmarks ie: Houses of Parliament, London Eye (Millennium Wheel), Tower Bridge, London Bridge, Canary Wharf Business Tower, it gives you a true idea of how beautiful our capital city is and how steeped in history and traditions these buildings are. Everywhere exudes character and charm, and as we pull up outside the House of Lords it's clear to see the true emotion and of what today has installed. When asked by one of our international families what was the function of The House of Lords, we could only compare it to be the same as the American Senate.

As an English family it was such a privilege and honour to have been invited to such a beautiful and fascinating day. After entering through Black Rod's Garden Entrance and going through strict security, we then entered into a courtyard which took us directly into the Westminster Hall which is where all Monarchs are

laid in State after their death, before their burial. This is where the Queen Mother was laid to rest after her Death in March 2002, and where the famous photo of the four Princes at each end of her coffin was taken.

We were then taken on a guided tour of the House of Lords, taking in the House of Lord's chamber, House of Commons (where the Prime Minister's Question Time – which is a weekly debate with the Government opposition), the many grande hallways and traditional rooms associated with these historic buildings.

We then had private invitations from The Lord Hameed of Hampstead and Aaron Kenneth Ward-Atherton - The Lord of Witley & Hurcott to attend a reception in the Cholmondeley Room, House of Lords on behalf of:

“The RASopathies Network UK”

This is the day that the RASopathies Network is launched supported by the Genetic Alliance UK. There is an official House of Lords Programme for this special event which you can download separately to view. This explains all about the RASopathies Network, the hosts

and the doctors involved.



Brandon Brown, Nancy Newton, Christie Dodd, Trinity Brown & Kieron Griffin

We had traditional afternoon English Tea, followed by various speeches and official photo shoot (to be commissioned). This was truly a wonderful time for our groups and a very emotional day, which can never be replaced.

On Saturday at 9.15am two coaches left for the London Zoo, the weather was fairly kind to us all and we had a fantastic day out before heading back to the hotel, and then our onward travels home.

President Signs Rosa's Law!

President Obama signed S. 2781, Rosa's Law on October 5, 2010. Although this new law does not change any services for people with disabilities, it still represents a historic change for national disability policy.

From now on, the phrase "mental retardation" will no longer be a part of any federal rule or law, whether it is related to education, health or labor. It will be replaced with the phrase "intellectual disabilities" everywhere it currently appears.

It is named for Rosa Marcellino, a 9-year-old Maryland girl from Edgewater who has been diagnosed with Down syndrome. Rosa worked with Maryland state legislator to pass legislation in the state last year.

Nina Marcellino, Rosa's mother, said she was thrilled to see the measure advance in Washington. She said the legislation will move the nation closer to shedding a stigmatizing label attached to people with intellectual disabilities.

"This has always been about so much more than just changing words or political correctness," she said in a statement. "It's about marking a new era where the dignity of people with intellectual disabilities is respected and their value appreciated."

The measure was introduced by Sen. Barbara Mikulski, D-Md., who met Rosa's mother at a meeting on special education. Mikulski said that if Rosa's law passed in Maryland, she would introduce it in the Senate.

The bill does not affect services, rights or educational opportunities for people with intellectual disabilities. It makes language used in federal law consistent with language used by the Centers for Disease Control and Prevention, the World Health Organization, and the White House through the President's Committee for People with Intellectual Disabilities.

Conference Frequently Asked Questions

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

Every two years we meet in different cities to listen to speakers, conduct medical clinic exams; discuss concerns and problems with one another and to introduce our

families to each other. We learn from educators, researchers and medical professionals and mostly from each other. We strive to better understand our own children and what we can do to help them. We gain knowledge, perspective and hope. We also have a lot of fun meeting new friends and checking in with old ones from past conferences.

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

The conference is typically held in late July. This seems to be a good time to have it as many children from around the world are out of school at this time. We usually, though not always, kick-off the conference on a Thursday night with a mixer that normally includes desserts and beverages. This gives us a chance to say hello, meet new and old friends, and get excited and ready for the next two busy days.



Q. Who may attend the conference?

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

Q. Will childcare be provided?

Yes, we've always provided professional childcare. This is a considerable cost (over \$5,000 for 30 slots) but the staff are insured, dependable and they bring in all the toys and activities. We anticipate a program that the children will enjoy with many activities to keep them busy and happy. We know personally that if parents know their children are happy, they can get the most out of the meetings. We can only allow CFC children and their siblings into this program due to space and cost. Childcare will cost \$35. per child for two full days of childcare. This is a small donation toward the childcare. We have a family who is funding the majority of the bill so the \$35 per child fee is a small portion with the majority funded by a generous sponsor.

Q. How old are the children in childcare?

We generally have children of all ages - from toddlers to age 17. We have both typically developing children and our children who are affected with CFC syndrome in childcare. This year our childcare services will combine CFC, Noonan and Costello individuals and their siblings as well. Infants are usually with their parents or extended family members and the adults take turns attending the sessions. Many times families bring grandparents or an aunt or uncle to help out. Please note that we cannot and do not provide one-on-one childcare or nursing care.



Q. What is included in the price of registration?

Each family is responsible for their hotel room and parking; it is not a part of

registration. Registration fees have been set at \$110 per adult, \$50 for children ages 6-12, CFC children and all children under age 5 are free. Parking has been discounted from \$25 per day down to \$7 per day. The registration fee includes the group meals offered, the facility for our sessions, printed materials, and clinic staff and speaker cost. CFC International funds the opening reception. For many years, we have charged each attendee a very nominal registration fee for the conference. It is our desire to make it possible for as many families wishing to attend the conference to do so. We will continue to do our best to make it economically feasible for our members to attend. Due to the generosity of our members and their friends who organize fundraisers or donate during the year, we are able to host the conference & medical clinic program every two years (at a cost of approximately \$30,000 per conference).

Q. What is the Medical Clinic program and who can attend?

The Medical Clinic program is free of charge to all our registered CFC children. There is no fee to register your child. The application for the CFC Registry can be found on our website <http://www.cfcsyndrome.org/registry.shtml>. Clinic day is extremely busy for the medical team involved and we set up folders on each child to be seen at clinic. Some

researchers wish to review the clinic folders the night before. The genetics/cardiology teams meet with only the new children that have not been seen in this clinic before. We set up a clinic schedule for the conference a few months before and only have so many slots for that one day. Clinic spots are reserved on a first come, first serve basis according to the date we have received your paid in full conference registration form. We will not hold any clinic spots for families who have not sent in the CFC Registry and related medical reports on their child.

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

Our general sessions tend to give an overview of the syndrome and any new research updates. The smaller breakout sessions are joint workshops shared by CFC, Noonan and Costello family members. You can visit the website and get a feel for what the conference will look like this year. As we get closer to the date, the schedule and more information about the sessions will become available.

Q. How do I register for the conference?

You register for the conference by going to our website and finding the area for the Conference under “News & Events” and then “Conference Information”. The registration form should be printed out and mailed in with your payment in full. No partial payments will be accepted and no holds on clinic spots are granted until all paperwork with payment is received.

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

Being together at the same hotel is so important and lends itself to the very purpose of our conference. The opportunity to spend time with other families living the same life as you are is well worth the money saved, but time lost, in commuting to the hotel from other accommodations. Most people secure their hotel room first. We encourage you to do this early. We have secured a set block of rooms at a reduced rate of \$120 per night plus tax at the Westin O’Hare. Once the rooms are gone we may not be able to obtain more rooms. The Costello and Noonan group will also be at this conference as well as the Scientific meeting so there are very few extra rooms to be located. We encourage you to register for the conference at your earliest convenience. Although it has never happened to date, we cannot guarantee that if we

are inundated with attendees that we will not enforce a cut-off. This would mean we have maxed out on numbers that can be accommodated properly. So please register with the Westin Chicago O'Hare early: <http://www.starwoodmeeting.com/Book/cfg26a> or call 1-847-698-6000 to reserve your room in the CFC International block.



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

CFC International

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

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

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

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

Does she plan to join us for any meals? Yes, she pays.
Does she want to sit in on one session? Yes, she pays.
Will she join us for the social on Thursday evening? Yes, she pays.
Does she want to socialize with other families in the conference setting? Yes, she pays.

Not paying a registration fee for an individual present at the hotel and connected to a CFC International family would indicate that person is taking care of the children (or vacationing) in an atmosphere that is not in any way a part of the conference.

Q. I am a teacher/therapist/nurse/etc. for a very wonderful child with CFC syndrome, and I would love to attend the conference. What is my fee?

All professionals are expected to pay the full registration fee of \$110.

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

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

Q. Who do I contact with additional questions?

Please e-mail CFC International at info@cfcsyndrome.org

Quality of Life CFC Research Project

Study Title: Health-related quality of life in pediatric patients with cardio-facio-cutaneous syndrome

Project Director: Stephanie Sottile
Boswell Room A08
300 Pasteur Drive
Stanford, CA 94305
T: 650.724.7190 voicebox 5

Research Team: Dr. Katherine Rauen, MD, PhD
Pilar Magoulas, MS, CGC
Andrea Kwan, MS, CGC

The purpose of this study is to investigate the quality of life in children with cardio-facio-cutaneous syndrome. Parents/caregivers who are interested in participating in this study will be asked to complete a quality of life survey for their child. The survey is no more than 100 questions and will take less than 1 hour to complete. In order to be eligible to participate in this study parents/caregivers must: 1) have a child who has been diagnosed with CFC 2) the parent/caregiver must be greater than 18 years of age 3) the parent/caregiver must speak English.

If you are interested in participating in this study, please contact:

Molly Santa Cruz
CFC International
molly@cfcsyndrome.org
phone: 805-481-8334

~~ Save the Date ~~

In conjunction with Family Forums organized by:

- CFC International
- Costello Syndrome Family Network (CSFN)
- The Noonan Syndrome Support Group (TNSSG)
- NF Inc.
- Children's Tumor Foundation

**GENETIC SYNDROMES OF THE RAS/MAPK PATHWAY:
FINDING OUR WAY BACK TO THE BEDSIDE**

July 28-30, 2011

The Westin O'Hare ~ Chicago, Illinois

**YOUNG INVESTIGATOR COMPETITION PRIZE:
TRAVEL EXPENSES TO THE SYMPOSIUM**

Contacts:

Bruce Gelb, MD: carolyn.quinn@mssm.edu (Assistant)

Amy Roberts, MD: amy.roberts@cardio.chboston.org

Lisa Schoyer, MFA: taos@earthlink.net

If interested in attending the Family Forums, contact:

CSFN: Tammy Moore: costellosyndrome@gmail.com

CFC International: Brenda Conger: bconger@cfc syndrome.org

TNSSG: Jeff Jasper, PhD: jjasper00@hotmail.com

NF1: Debbie Bell, NF Inc., CA: dbell@nfcalfornia.org



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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 Tissue and 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 the address above.

Name: _____

Address: _____

City: _____ State: _____ Zip: _____

Phone: _____

Email: _____