



CFC international

Cardio-Facio-Cutaneous Syndrome

The CFC Chronicle

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2007 CFC 4th International Conference Overview

(By Kyle Stowell)

The 2007 conference for CFC International started with registration at 4 PM on Thursday, July 12. Volunteers and board members helped to hand out conference bags and schedules. As families came to register, it was great to watch as they came and saw familiar faces. Some stayed to talk with old friends from past conferences.



Participants at the Fourth International CFC Conference, Orlando July 12-14 2007.

Starting at 6 PM, families gathered at the Welcome Reception. Acquaintances were renewed and families were able to see in person those who they only knew by name from the email listserv. One of the main purposes of the conference is to allow families to interact with each other. There is a bond between these people who have so much in common because of CFC Syndrome. The information gained by speaking with those who have had such similar experiences is invaluable. The soul is lifted by the acceptance and support that CFC families offer each other. This interaction was continued Friday night at Mom's night out and also Saturday with a Dad's night out.

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



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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.

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Message from the President

The 4th International CFC Family Conference and Clinic program was held at the Rosen Centre Hotel in Orlando, Florida from July 12-14. Twenty five states in the USA and 6 other countries outside of the USA brought in 175 attendees to this record breaking attendance event. So many new families with very young children who already had a confirmed molecular diagnosis of CFC Syndrome increased our attendance rate for this conference. What was also wonderful was meeting first time attendees who also had older individuals with CFC. No matter where families came from the similarities in behaviour, health concerns and eating habits were often the same. During this weekend event we were united as one big CFC family sharing experiences, support and information with each other.

A special part of our conference is the Saturday night showing of the DVD of children & family photos set to music. This year the Stowell family from Utah coordinated this project and as always, the production brought many a tear. We celebrate the accomplishments of each and every child while never forgetting the children who have passed away but are still so much a part of our lives.

So many behind the scene efforts go into making a successful conference. Many people came forward to pitch in and help this year. We had volunteers working all throughout the weekend, prior and even after the event to cover more T-shirt and DVD sales. Our annual silent auction with themed baskets brought in a total of \$4,133. Thank you to all the families who helped out even to those of you who could not attend yet shipping in a donation for the silent auction. Without your generosity we would not be able to host this crucial event for families every two years.

We look forward to an even bigger and better event in 2009 and hope that those of you who could not attend the 2007 conference will consider joining us in 2009.

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.

<i>Donations in Honor of:</i>	Madison Placido	<i>General Donations:</i>
Megan Ankeny Remos Killian Paige Conger Gary & Louise Childs Clifford Conger The Conger Family The Sperber Family Jack Doyle Carole Drake Melissa Smith Jacelyn Hickman Sean Crowshaw Camden Kline Kevin & Susanne Machlinski Luke Lydkisen Thomas & Michele Francione Joseph & Michele Grogan Todd & Nancy Anderson Jake Metcalf Kelly & Brenda Metcalf Aaron Rose Grace Yeager Emily Santa Cruz Judy Devine Susan Hermreck Sandra Knotts	Fernando & Imelda Placido Ashley Thompson Antonia Davis Gregory & Janet Seward Meg Young Terri & Shawn Briggs Wilburn & Jaunell Riddell Richard Soloman Jr. <i>In Memory of:</i> Isabella Hope Dahle Diane Dahle Mabel Dezzani Adam & Jeri Koehler Lynn Kipp Al & Gayle Kryger David Koehler Adam & Jeri Koehler Joseph Marano The Hoeck Family Elizabeth Paschall Robert & Rebecca Politzer Anthony Verrino Jr. Geraldine Flowers Joan Joseph Adam Pichler Susan Trevas	Anonymous Akron Children’s Hospital Employee Foundation Matthew Brockwell Estate of Ron Clark Jennifer Day Christopher Dietrick Kent Donald Edward Giegucz Maxine Granato Susan Hopkins William Lewis Kristine Lindsey Maribeth Linmore Edward Machir Kenneth Wade Martin Brian & Margery Mather John MacArthur & Renee Khatami Nicole McKean Wendy Morris David & Bobbi Olsen Carol Petras Mary Beth Strausbaugh Thompson Family & Friends Tyco Matching Gifts Program Lisa Vallequette Lawrence Williams Tamara Williams Tracy Bushman

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 the CFC International, you may usually "write us in" on the campaign giving form. If you are unsure whether your gift will be directed to us, speak with your local workplace campaign coordinator for assistance.



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

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

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

"A NIGHT OF FUN TO HONOR LUKE AND HIS CFC ANGEL FRIENDS"

Our family hosted an evening filled with good food, good drink, good friends and family all for a GREAT cause!!! On Saturday, March 24th, 2007 about 200 of our closest friends and family joined us at a local firehouse. The event was catered with food and donated beverages



Silent auction item

and a disc jockey who donated his time as well (he is a med student with a big heart)!

We had many great baskets for auction and other items as well and a 32" flat screen TV. for a special raffle. Our fundraiser was above and beyond our expectations raising \$22,384.00!



Lydiksen fundraiser hosts a full house

It was so nice to see many friends and family that you don't get to see that often and they were absolutely thrilled to see Luke and the

wonderful progress he has made over the years! The CFC Angels on the poster and the PowerPoint presentation were a powerful moment for all who viewed!

Angie, Erik, Erik (10 yrs.) and Luke (8 yrs. CFC) Lydiksen Seymour, Connecticut

Golf Event Funds CFC International

It was a beautiful day for golfing in Connecticut as the Hickman family prepared for their 1st Annual CFC International Golf Tournament in honor of Jacelyn Hickman. The event was held at the Keney Golf Course in Hartford, CT on May 12, 2007. Many new golfers as well as the experienced from all over Connecticut came out to support our wonderful cause. There was much interest in our organization and it was great to raise awareness.



Even the losing team takes a prize!

There were beautiful baskets and other prizes donated by many family members and friends. The local business community donated many items included in the baskets. There was also a 50/50 raffle that was generously donated back to CFC by the winner! The event was also made possible through wonderful donations and support



from all the brothers of Lambda Theta Phi Latin Fraternity, Inc especially Leo Espejo and Luis Rodriguez.

We would like to extend many thanks to the Zeunen family in Wilmington, NC who offered warm words of encouragement and unlimited knowledge in running a tournament. Also to the Conger family who traveled all the way out to CT in order to support our event. Cliffy was a great hit and he enjoyed riding the golf carts.

Thank you all for helping to make our 1st annual CFC International Golf Tournament in Connecticut such a big success. We raised over \$9,000. Hope to see everyone again next time.

Mildred & James Hickman
Vernon, Connecticut

5th Annual CFC International “Toast the Angels” Wine Tasting & Dinner

Hosted by Cliff & Brenda Conger

The 5th Annual CFC International “Toast the Angels Wine Tasting” was held at the Kalurah Shrine Center on Saturday, March 29th.



Clifford and Luke take a stroll

It was another great success. Chef Russ Rodrigez masterfully cooked up a fantastic 5 course meal and with his creative wit, talked about each course along the way. Sharon Wild, representing Château Lafayette Reneau Winery located in the Finger Lakes region of New York State, explained



Jacelyn picks the winning ticket

the wine we were about to taste with each course. As you’d expect, both the wine and food were delicious.

We had a record attendance of 162 people joining us this fifth year. We were blessed to have several CFC families who travelled from great distances to support our event. The CFC families that attended were Dana and Chuck Klein from Maryland, Mildred and James Hickman from Connecticut, and Angie and Eric Lydiksen from Connecticut. It was great to spend time with them.



Our faithful waitresses who pretend to be bartenders: Paige Conger, Audrey Ryan, and Brie Spak

The CFC Silent Auction was the largest so far with over 50 items donated for the event. There were a lot of interesting items donated for the auction. A few items were actively bid on and talked about throughout the evening. Susan Fedor, a local jewellery designer, displayed and sold her beautiful hand made designs of earrings, necklaces and bracelets. At the end of the evening Sue donated 20% of the sales to CFC.

We raised a total of \$6,041 for CFC International. The crowd that attended had a wonderful time. Brenda and I had a lot help from seniors attending our local high school, the Cruisin’ Buddies Car Club, the talents of Melissa and Chef Russ Rodrigez, Château Lafayette Reneau Winery, and Sue Fedor that made the evening a huge success. Thank you everyone for your help and dedication to our worthy cause.

Cliff Conger

Friendship is the only cement that will ever hold the world together.

Woodrow Wilson

2007 CFC 4th International Conference Overview (Continued)

Friday morning began with breakfast. As we finished eating, Dr. Giovanni Neri spoke to the group about his interaction with CFC Syndrome and the support group. Dr. Neri has been involved with CFC International since the beginning. He explained how he viewed CFC Syndrome as a separate and distinct syndrome from Noonan Syndrome. This view came to be accepted and eventually was proven correct through the identification of genes for both syndromes. We then heard a presentation from Dr. Katherine Rauen from UCSF. She led the group that identified mutations in all 23 sets of DNA that were obtained from the CFC International DNA BioBank. Dr. Judith Allanson then presented some interesting statistical findings from the CFC Registry. The Registry is the form that families fill out when they join the support group. Trends within CFC Syndrome have become apparent by analyzing the information from the Registry. Dr. Grace Yoon finished off the morning presentations by presenting facts from her neurology study. It was thought that seizures were not common in CFC Syndrome, but it is now estimated that some 40% of CFC kids have seizures.

Following lunch, there was a Q & A session with the medical advisory board. Pilar Magoulas, the Genetics Counselor on our Board of Directors, selected questions that were submitted with the conference registrations. It was apparent that the doctors attending the conference are caring and compassionate. They want our kids to have great care and to meet their potential.

The afternoon of Friday, and the morning and afternoon on Saturday were spent in workshop sessions. Most of these were taught on various medical issues that are faced by those with CFC. Dr. Kavamura spoke on skin issues. Dr. Noonan presented on heart problems. Dr. Maerz, Cliffy Conger's pediatrician, spoke about dealing with primary care physicians. Dr. Kent Reinker spoke on orthopedic issues and then spent Saturday afternoon offering evaluations of our kids. Linda Bonnar spoke about some of the technology that is available to help our children communicate. Oral and motor issues were discussed by Charlene Larsen. Members of our

Board of Directors spoke on fund-raising. Without regular fund-raising by you, our members, research into CFC would virtually come to a halt. Dr. Eric Johnson explained to us the basics of genetics. Rob Davies, who has given wonderful presentations about planning for the needs of our disabled kids was not able to attend this conference. Mike and Marcie Scholl filled in nicely for him in teaching us about trusts and wills, and also transition planning.

At 6 PM on Saturday, all of the CFC kids were lined up for a group photo. Many of the doctors pitched in, holding CFC infants and toddlers. Many more individual and small group photos were also taken. There is nothing like seeing 5 little curly heads together at once!

The evening was completed with the dinner, DVD slide show, and silent auction that has become a tradition of CFC conferences. It is fun to see the family photos on the DVD as well as the cute and funny photos of the CFC kids. The Memorial section was also very moving. The silent auction was great as there were so many different baskets and items to buy. Some of the most popular items this year were the Disney baskets. The auction raised a record \$5,050. This was matched by a \$10,000 donation from the generous Olsen's, Daniel Hess' grandparents.

That evening and the next day, good byes were said to friends, old and new, as families headed home. We are looking forward to the next conference to renew these friendships again, increase knowledge of CFC Syndrome, and further research into CFC.



If you have a loved one who is hospitalized or in care outside of the home you might want to consider setting up a private website on CarePages to keep family and friends up to date on their progress. CarePages is a free service and can be set up in 7 minutes or less! Upload photos and post updates as desired. www.carepages.com/familyrn

What is Cardiofaciocutaneous (CFC) syndrome?

CFC syndrome is a rare genetic condition caused by mutations (changes) in one of three genes (BRAF, MEK1, and MEK2). While there is a wide spectrum of severity in CFC syndrome, most individuals will have some degree of heart (cardio-) involvement, facial (facio-) characteristics, and skin (cutaneous) abnormalities. Below is a summary of common findings in CFC syndrome:

Heart: Pulmonic stenosis (narrowing of the artery going from the heart to the lungs), atrial septal defects (holes in the upper chambers of the heart), ventricular septal defects (holes in the lower chambers of the heart), hypertrophic cardiomyopathy (enlarged heart muscle).

Facial features: Large forehead, relative macrocephaly (large head), narrowing at the sides of the forehead, down-slanting eyes, ptosis (droopy eyelid), depressed nasal bridge, rotated ears.

Skin and hair: Dry, thickened (hyperkeratotic), or scaly (ichthyotic), eczema (extreme dryness of skin and itchiness); sparse, curly, wooly or brittle hair; eyelashes and eyebrows may be absent or sparse.

Other: Decreased vision and acuity, feeding difficulty, failure to thrive, reflux; growth delay, short stature; hypotonia (low muscle tone), seizures, cognitive impairment and developmental delay (ranging from mild to severe).

Early diagnosis is essential

Historically, the diagnosis of CFC was based on the clinical features, medical, and developmental history of the child. However, there is now genetic testing for any individual suspected of having the diagnosis. Currently, there is no cure to treat all of the symptoms of CFC syndrome, however, with proper management, referral to appropriate specialists, and early intervention, much can be done to improve the health and quality of life of individuals with CFC syndrome.



Jack & Judy
Ohio



Gary & Marcus
England



Sarah & Maddie
Wisconsin & Ohio



Sara & Regan
Massachusetts



Thea & Fleur
The Netherlands



Jennifer & Lauren
Texas

Photo Gallery - Memories from Orlando - July 2007



**Nancy, Australia, Jared, Utah, Clifford, New York
Jack, Ohio, and Brennan, Alabama**



Aunt Peggy & Nick, Missouri



Louise, The Netherlands



Jennifer & Alaina, Minnesota



Andrew & Julia, Michigan



Camden, Maryland



Amanda, Oklahoma



James & Jazzy Hickman, Connecticut



Esther, England



Ernie, Emily & Molly, California



Mary & Rachel, Texas



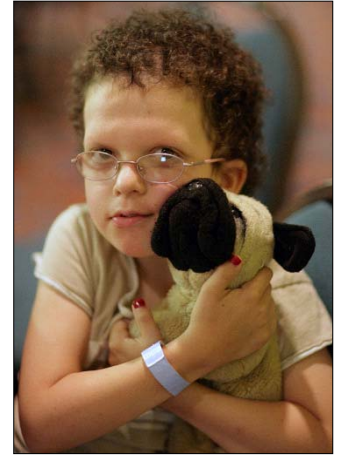
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Dr. Kavamura & Avery Clark



Flores family, Mexico



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Tanda & Marcus, Denmark



Palombo family, Pennsylvania



Theo, Maryland, Jason, Utah & Nick, Denmark



Emily Hannan, England, swimming with the dolphins

McCann Family

Given that one of the signature traits of CFC is “sparse, curly, friable hair”, we hardly expected that at the CFC International Conference we’d be able to identify Melissa as ‘the bald one’ when introducing ourselves as her parents.

It was one of a number of surprises we came across in Orlando. We arrived not quite knowing what to expect. Our decision to attend was made fairly spontaneously at the end of May. After 21 years of having Melissa being unique in terms of diagnosis in the disability circles we participate in at home (Vancouver, BC), we were entering a realm where everyone had the same diagnosis.

As a parent active on boards and advocacy groups at both the civic and provincial (state) levels I believe that families are better off concentrating on the sameness of the challenges faced by persons with disabilities rather than the needs of specific diagnosis. Too often the creation of label specific silo’s by disability groups allows government to play off internal politics instead of addressing issues that are faced by all. In reality all of us in the disability community have a common goal; for the people we love and support to have a good life.

Hanging around the registration desk, and spending a few hours at the Thursday evening social it became clear that while their abilities ran the gamut, the characters of the CFC children and young adults held many commonalities. And once you started talking to other parents, more similarities emerged and in one particular case we seem to have found Melissa’s doppelganger (hello the Scholls).

Over the course of the three days we met many people, connected strongly with several families and took advantage of the medical teams’ expertise. As at many conferences parents tend to gravitate to other parents by age of the child or issues being faced. We are all involved with different challenges at any given time and that sometimes predetermines with whom we bond.

At check-in, Brenda put the long arm of Conference organizer on us, asking Bernice and I to participate on a panel discussion revolving around transition. Of particular interest for her was our recent purchase of a home for Melissa and the living model we are hoping to create there.

Several years ago we created a document we call “A Vision for Melissa”. It is an outline of who Melissa is, what our dreams for her are, and what kind of life we desire her to live. In that document we wrote:

“People around the world recognize home as a haven. Home is a caring, accepting space where Melissa and her supports (paid and natural) can fulfill their potential, both individually and collectively. A place where they all can participate in quality life experiences, recognizing and celebrating diversity and accomplishments. Home is a place where we will support Melissa’s quest to have control over her world and environment. It will be a space that allows Melissa as much freedom as possible, while at the same time ensuring her safety and security. Home will be a place where the expectation is respect and care, not rules and regulations. It will allow Melissa to choose where she lives, with whom and who will support her. “

In January, 2006 we took part in a focus group helping a family directed organization called Planned Lifetime Advocacy Network (www.plan.ca) develop a package of proposals to advocate for at the Federal Government level. The two primary planks being a Registered Disability Savings Plan that would allow families to set aside money in a plan that would grow tax free until withdrawal and a home ownership strategy for people with disabilities. At that meeting we asked the question of other parents, “isn’t looking after one house more than enough work?”

Three months later we were touring homes with a real estate agent.

What happened in the meantime? Government funding. In British Columbia support funding for people with disabilities has always been administered by a provincial government department, but as waitlists have grown and the lack of services have become a political liability, a new administrative body known as Community Living BC was created and a wholesale transfer of responsibility to this now arms length body has taken place. In the early days (Jan/Feb 06) of transition, government workers being forcibly transferred to the new organization loosened the purse strings and we secured funding to support Melissa living out of our family home beginning in April 2006. The funding is approximately one-third of what a group home placement would cost the agency so it requires us to be creative in using this money. The money is for direct support only, and not for the purchase of property. It also arrived on our doorstep three to four years earlier than we intended to have Melissa moving out, but like any

government support you take it when it comes because you never know when it might be available again.

With the funding for support services approved we needed to accelerate our quest for an independent living situation for Melissa. We knew that group homes with their rigid schedules and staff focus were not what we wanted for Melissa. And I say that as the past-president of the Burnaby Association for Community Living (www.gobaci.com) which operates 14 group homes, sheltered workshops and many other supports. We looked at Pri-Care, where Melissa would live with another family who would support her, for payment, but felt that Melissa already had a family and did not need another. Just as importantly we did not feel we or Melissa's extended family would feel comfortable stopping by for a visit.



Melissa McCann

That left the option of us buying a home for Melissa, because we knew that we did not want Melissa to be unsettled by the vagaries to landlord/tenant issues.

Our initial plan was that Melissa would live in a house with two or three friends who have

disabilities but are able to live semi-independently. The young women would share in the rent and living expenses and benefit from the support of Melissa's paid staff. As the individuals we had in mind do not receive any additional support dollars beyond their monthly disability benefits payment we thought we were making a great offer. We proceeded down that path, encouraged by previous conversations with these young women as they hung out with Melissa about their desire to move out, and reality hit. We realized while their parents may support the idea of their children living out of the home they weren't ready to share the responsibility so this situation would see us looking after household shopping, cleaning and administration on our own. As an added bonus these young women have boyfriends, so the picture suddenly become much more than we felt we wanted to cope with or be responsible for.

Time to refocus and continue the arduous process of looking at homes that had to meet our criteria: reasonably close to our home; have good transit access; and not need a lot of renovation work because we just don't have that amount of time and energy. We also reevaluated our goal of who would live with Melissa and decided that we

would like to create a community of young women living with Melissa who would share their lives with Melissa, natural support, along with some paid support. Here is what that vision has become in black & white:

Our vision is a home where four young women share this time in their lives. It is a safe, caring, respectful environment where each housemate aspires to reach their goals, dreams and potential; both individually and collectively. A home, where family and friends feel welcomed.

The home is located in Burnaby's Garden Village neighbourhood, north of Central Park and near major transit routes. It is a four bedroom newly renovated home with a spacious backyard and attractive low maintenance garden. This is a Pet Free & Smoke Free home.

Melissa wants to live in a home where she is safe, where she is free to be herself. Family and friends are critical to her well-being and to the richness of her life. She wants to make choices about matters big and small. She wants to pursue her interests and passions. Melissa wants to feel welcomed and valued in her home, neighbourhood and communities. She wants to contribute and be recognized for her contributions. All people have the right to say I want to live my life my way; these are my hopes and dreams to achieve self-actualization. Melissa wants a "life"; to be accepted, to have friends, to have fun, to be respected, valued and loved.

Melissa's wish: Accept me as I am. Respect, value and appreciate me, affirming my positive self-outlook. Support me with encouragement, guiding me to make positive life choices. Do not see me as needy or as a client but as your housemate and in time, I hope, as your friend.

Melissa spends her days in social and volunteer roles that revolve around her passions: people, babies, and dogs. She is supported on an individual basis by the Burnaby Association for Community Inclusion, Mondays through Fridays (8 am -3:30 pm). Melissa's Microboard, Melissa's Life With Friends Society, fills in for all other hours of support needed. At this time Melissa will be out of her home each weekend from Friday at 9:30 am until Sunday at 7:30 pm.

Melissa gives us the privilege to experience just how simply living and loving is done. Melissa is about unconditional love. Her life brings us joy and sorrow, in essence affirming the experience and purpose of life itself. Melissa daily faces the challenges that come from living with no speech, functional hearing, vision, coordination



and balance, decreased organ function and appearances that differ significantly from her peers. Yet, were you to ask anyone who knows Melissa what her most distinguishing characteristic is, they would say “her incredible smile”. Melissa’s smile gives comfort and hope. Melissa teaches us about happiness, contentment, insight, inner peace and a love for the smallest things.

Melissa uses a wide range of communication modes (facial expression, body language, gestures, modified sign language, vocalization, and physically leading). Melissa understands verbal speech but she also closely monitors your use of communication modes to gain a fuller understanding of what you are saying.

In July of 2006 we purchased a 4 bedroom home about 1.5 miles from ours and began the quest for peers to become her housemates. The house is in our name and we are now landlords, as Melissa cannot legally enter into contracts without our going to court for an expensive legal process called committeeship.

In terms of accessing the funding provided, it was originally contracted with the agency that provides Melissa’s day supports and we had the option of having the contract be made directly with us or we could create a Microboard to administer the support dollars. We have chosen to create Melissa’s Life With Friends Society, a registered organization whose sole purpose is to support Melissa in her independent living endeavors. The five person board includes myself, two of Melissa’s aunts and two family friends. Bernice and Melissa’s two brothers are not on the Board so we have a broader range of people familiar with what is going on should something happen to us. The Microboard also creates a liability shield for our family assets should a staff person or roommate seek to take legal action against the Society. More information about Microboards can be found at www.microboard.org.

In the past several months we have spent a lot of time living between two houses. We have gone through two housemates and learned that Melissa’s peers at 21 are not ready to run a household. Our hope for the next housemates is that they will be a little older and more prepared for the responsibilities of a house such as; getting the garbage to the curb with some regularity, acknowledging that toilets may need to be cleaned occasionally, and that food shouldn’t be composting in the refrigerator.

It has been an intense learning curve, but one that we feel is taking us and Melissa in the right direction. She deserves to have the same opportunities and standard of

living as her siblings. How we get to that point is just a different path.

Just like the families at the CFC International Conference, we all have the same diagnosis but have all arrived in Orlando by different methods of transportation.

Journey from Costello to CFC

Amara Marie Tobby was born on April 1st, 1992 in Greenbrae, California, just a few minutes north of San Francisco. She arrived into this world as a healthy baby girl. We left the hospital the following day with our new bundle of joy. We were so happy to have this precious little girl in our lives. We had no idea the journey she had begun for us would take us up the road less traveled.

At the time my father was ailing and 3 months after Amara’s birth he left his body. The loss of my father was a very difficult experience for me because I found myself caught between deep sorrow and great joy. It was very strange place to be. I did my best to handle it and often felt guilty that my emotions played a role with Amara’s developing health condition.

Two weeks after we buried my father we found ourselves insisting that our pediatrician, Dr. Maisel, run some tests on Amara since she was now consistently projectile vomiting and not gaining weight. After my husband became involved the doctor finally agreed to run some tests which resulted in the awareness that Amara had elevated calcium in her urine. Since the doctor wasn’t clear what this may mean she sent us to the University of California, San Francisco for further testing. There Amara was placed in the ICU and an NG tube was inserted. We spent the next ten days being evaluated by many different specialists that included neurology, GI, genetics, endocrinology, and ophthalmology. There were so many specialists all trying to find an answer. It appeared Amara was such a mystery and turned out she was. After all the testing, poking and prodding of this little baby we finally were sent home with the diagnosis, “failure to thrive”. We left UCSF with the hope Amara would thrive and grow out of whatever it was she had.

The months went by with frequent visits to the pediatrician and follow-ups with the UCSF specialist. She began to gain weight slowly and development remained delayed but she was stabilized and happy. I fortunately was able to be home with her and our son Teddy, who was four years old at the time. I made the

best of a situation that was filled with unknowns that caused much fear in me. I prayed a lot for Amara; mostly that she would know joy.

I was frustrated that no one could define clearly what was really going on with Amara. I finally got the courage to go for a second opinion to Stanford's Lucille Packard Children's Hospital for GI and the visit with that team of specialists brought two new suggestions. One, change the formula because due to Amara's distended tummy they felt she was allergic to Similac and suggested we use Isomil, a soy based formula. Two, to have a neurological evaluation because they thought the way her eyes stared could mean she was having petit-mal seizures. I was so surprised to find this a possibility.

About a month and a half later she had her first granmal seizure that lasted 45 minutes. We thought we were losing our baby girl. But by the grace of God this little angel pulled through. She was 18 months at the time and so began the journey of a million meds. Trying to find what would control this; what later become known as epilepsy. Of course we were also taking her for her regular immunizations. After she presented with the seizure disorder the new pediatrician I chose, Dr. Ernster, suggested that we no longer give her the pertussis shot. For those who don't know, that's the "P" in the "DPT" shot. Pertussis immunization is known to cause seizures in some children.

Either shortly before her first seizure or right after, Dr. Mahin Golabi, who was the pediatric geneticist we had seen at UCSF, gave us the diagnosis of Costello Syndrome. When she showed me some pictures from a medical journal for a moment I thought it was Amara. It was scary to see the similarity. She told me since it was a clinical diagnosis and there is no test to prove this I could dismiss it and hope for the best. I was free to move forward with an open mind and see what transpires. It was so heartbreaking but it did help explain some things such as the feeding issues, difficulty with weight gain, strabismus, but it didn't explain the seizures. There would be many years of explorations, alternative doctors, and therapists that would try to assist me in improving Amara's quality of life.

In 1996 we decided since we had exhausted all of the best specialists in the area, and possibly the country, we would move to the Hawaiian Islands since this is where my parents were originally from. We thought going back to our roots would be good for all of us spiritually and it would be a beautiful place to raise Amara and Teddy. My husband Ted was presented with an offer we

couldn't refuse so off we flew to Hawaii to create a new life for our family. We left California after it being our home since birth with the blessing of our mom's, relatives and all our friends. Moving to Hawaii turned out to be the best decision we ever made. The island lifestyle and community truly embraced us with the Aloha spirit. It was a challenge at first to find the team of doctors, specialists, therapists and educators we would need for Amara's care but eventually we were successful.

I believe after we were here a year we were able to get our first computer for our family through a special program because of Amara's mental disability. That computer would open my world to the amazing internet and give me access to the "information highway" that would eventually change our lives. Because I had the uncertain diagnosis of Costello Syndrome, I started searching online and came across Colin & Cath Stone of Manchester, England. They had put up a website and were just beginning to link up the families and children who had been diagnosed with Costello Syndrome. I was able to phone them and we shared our experiences. They were the only ones I knew who were dealing with a similar situation. As time unfolded I would visit the site and view more pictures of the children and began to feel that Amara was not a Costello kid.



Toby Family at Disney

In 2001 we planned a trip back to the Bay Area to visit family and friends; Amara was now ten years old. I thought since we were going to be in San Francisco I

might as well see if we could get an appointment with the Genetics clinic and hopefully get to see Dr. Golabi again. When I telephoned for an appointment I was connected to Vicki Cox, the genetic counselor who happened to be the same counselor Amara had on the first visit back in 1992. I was so happy to speak with someone who knew Amara and was familiar with us. She suggested that since Dr. Golabi had moved on we must see Dr. Kate Rauen. I was thrilled to get the appointment. When we met Dr. Kate we were so impressed with her true concern, her compassion and kindness. It was at this



first meeting that she shared with us that they were actively looking for the Costello gene and wondered if we would be interested in donating Amara's blood. We were more than willing to participate. I felt anything we could do to help, if not for our daughter, certainly for the next generation. Dr. Rauen also asked if I could provide some pictures of Amara. We gladly obliged since by now I was getting technologically savvy and happened to bring my Sony floppy disc digital camera. Right there in that clinic we took many pictures of Amara and gave the floppy disc to Dr. Rauen to keep for use as she wished. Later I would learn that it was something in some of those pictures that triggered a thought within her where she should look for the CFC gene. What a miracle!

In November of 2005, Dr. Kate called me to inform us that the gene for Costello had indeed been discovered and that it was called HRAS. She told us that she wanted to run Amara's cell line again because Amara didn't show the mutation and she would call us back in a few months.

As promised, on January 25, 2006, the day before the announcement of the CFC gene discovery, I received a call from Dr. Kate Rauen. I was so happy and surprised to hear from her. The conversation began with an unnecessary apology that she mistakenly thought Amara had Costello Syndrome. By this time I intuitively doubted that Amara had Costello mainly because of all the seizures. Then she told me the good news was she did know what Amara had because she had discovered the genes for CFC and the gene discovery was about to be published in Science Medical Journal. I was shocked, but very excited to finally have an answer after waiting 14 years. She told me she would call back in a couple of weeks because there would be an actual blood test we could take to confirm this for ourselves. It was on this day that I was given the confirmed diagnosis of CFC Syndrome which was later reconfirmed by the new blood test. I was so joyful and happy and still am!

I was told to contact Brenda Conger to ask to be connected to the CFC list serve and to visit the CFC website. This was so amazing to me to find so many families and children dealing with the same challenges after really feeling alone for 14 years. But, the most amazing thing that came from this awareness, this new knowledge, was that it freed my mind of all worry, any guilt. I knew we would be ok; it was so inspiring to know I was not alone. I was so happy to find CFC International and knew I finally had arrived home. I was impressed how organized and professional the CFC International

organization is, but mostly how much love I could feel in the many emails I would view.

When I heard there would be a conference in Orlando, Florida the following summer, I decided in that moment we would be there. If I had to paddle a canoe and swim; there would be nothing that would stop us from going. To be able to make it to our first medical conference ever was so exciting and uplifting. To meet all the beautiful children and their dynamic families; and to finally have had the opportunity to meet everyone this summer has truly been God's grace. The many workshops were very informative and the opportunity to meet the doctors on the CFC Medical Advisory Board was amazing. They are all so brilliant and passionate about our cause. In hindsight, I wish I would have asked more questions, but I can always email.

I know deep within my heart that great things are going to happen for all of our families. For me, "the future looks so bright it burns my eyes"! I have great gratitude and admiration to Dr. Kate Rauen for the gene discovery and for introducing me to this incredible organization called CFC International.

We have learned so much from having our angel Amara in our lives. She has opened our hearts to know unconditional love; she has taught us great patience and the meaning of hope and faith. She has shown us to walk with a grateful heart and to never ever give up. She has taught us to look within to self examine our lives and change. There are many gifts she has given that have made us better people and a stronger, more courageous family. Thank you, thank you, to all of you who share the journey with so much love, courage and a wonderful positive attitude. We look forward to hopefully seeing you all again at the next fabulous CFC International conference in San Francisco (wishful thinking).

Until then our fond Aloha!

The Tobby Family

Flora, Ted, Teddy (19 yrs the Best Big Brother) and Amara Marie (15yrs our angel)

The Weston Family Story

Isn't it funny, the most complicated of puzzles always seems so obvious...once you've discovered the solution? This was definitely the case with Marcus.

On the 17th October 2004, my friends and their children had come round to our house to play. I had

finished work just three days before and was already feeling quite strong twinges but I still had almost four weeks to go before the baby was due. I was huge! That afternoon, my friends joked that I seemed preoccupied; and I was ... I was wondering whether the baby was coming! My bump was so big, there was no way it would stay in there for another 4 weeks; there wasn't any room left!

Three days later, I was in bed cuddling my husband and my newborn baby boy. We were ecstatic. He was huge: 7lb 15oz and three weeks early. The midwife commented that he appeared more mature and we noticed his face was pretty puffy and bruised. He also looked as though he hadn't quite grown into his skin. We put this down to the fact he was early and was stuck in my pelvis for three days. That first night, I noticed he didn't feed well, but thought little of it. The next day, Marcus was checked over by the Paediatrician who noted his droopy eyelid before allowing us to take our new bundle home.

After just three days at home, we had to take Marcus back to the hospital. He had quite a nasty eye infection and his droopy eyelid was really swollen. We were pretty concerned about his eyes; they looked really sore, very swollen and a bit wonky. Over the following two weeks, his eyes improved but his feeding was becoming problematic. I was breast feeding but knew he was not feeding for long enough or often enough. He was also vomiting pretty regularly. However, I wasn't overly concerned until he was 2 weeks old and had still not regained his birth-weight.

At three weeks old, Marcus contracted RSV virus and refused to feed altogether and lost another 8oz - he was now four weeks old and only 6lb 15 oz, a whole pound lighter than when he was born and he looked terrible. We were desperately worried about Marcus, he'd been prescribed reflux medicine to control the vomiting. It didn't make any difference and I began bottle feeding so I could be sure about how much milk he was receiving. Life was becoming increasingly difficult by this stage. We had to take towels with us if we went out because he was vomiting so much. I was also spending almost all day feeding Marcus, drip by drip. He would arch his back and scream every time the milk touched his tongue. It was as if he knew it would cause him pain later down the line. He started to scream when you brought the bottle near him. Despite spending every waking minute trying to feed him, he still wasn't taking enough to grow. Marcus also slept through at night, I'd have set the alarm clock to wake me up so I could force feed Marcus - it was such a dreadful time. I'd sit in bed sobbing, why won't my baby eat?

At this stage, Marcus was admitted to hospital where they discovered his heart problems - which are quite mild fortunately. The doctors also changed his milk, added some more reflux drugs and started naso-gastric feeding. We went home for another couple of weeks but Marcus still didn't gain weight. The vomiting worsened as he couldn't cope with the increased volumes we were forcing on him with the NG feeds. Going out was impossible with the vomiting and every time someone asked about his NG tube, I cried. I didn't know why he had it, why he wasn't growing or what was wrong with my baby.

Around this time, our GP commented that Marcus' ears were rather low-set. I was furious, what did his ears have to do with anything? I was more concerned about his feeding, that's why I went to see him. He said, "You can't make a baby eat if they're not hungry." Great! Really helpful....NOT! I got straight on-line and Googled "Low-set ears." I was gob-smacked by what came up on screen. Noonan Syndrome. This was the first time I'd really considered that there may be something serious and long-term wrong with Marcus. I read the symptoms: low set ears, wide-spaced eyes, loose skin, pigeon chest, heart-defects, and developmental delay. Marcus was lying across my knee. I looked at him and then at the screen...and back to him. I knew in my stomach, this was it, The GP was right.

Eventually, Marcus was readmitted and remained in



The Weston family

hospital for another 7 weeks while the doctors tried bolus feeds, continuous feeds, experimented with milk and reflux drugs. He had all sorts of scans and x-rays to try and determine his problems but to no avail. I asked the doctors there about Noonan Syndrome - they didn't think so, his feeding problems were too severe, they didn't fit.

At 15 weeks old, we said goodbye to the brilliant staff at Northallerton and Marcus was admitted to the Royal Victoria Infirmary in Newcastle, 50 miles away from home. He had just regained his birth weight and the doctors there decided to perform biopsies of his gut and insert a central line so they could get some nutrition into him. His gut biopsies were normal, as was the barium study, MRI scan, chromosome studies and abdominal scans. Marcus was fed via parenteral nutrition and NG tube for the following few weeks.

Meanwhile, Joely was almost 2 and her life had been turned upside down with Marcus' arrival. We were all missing each other but there was no easy way to manage the situation. When Marcus was transferred to Newcastle, family life became easier in some ways as we were all able to stay in Crawford House, accommodation provided for the families of poorly children by The Sick Children's Trust. We ended up living there for 3 months.

At first, Marcus seemed to make little progress and the doctors were no nearer to determining why he couldn't feed. We were visited by the genetics team who felt there might be a genetic reason for his problems. Again, I asked about Noonan syndrome, and they said they didn't think his problems fit that pattern.

After 4 weeks at the RVI, Dr Bunn struck gold. She thought it might be worth feeding Marcus into the top of the bowel, into his jejunum. It worked, Marcus gained weight, the vomiting improved and he slowly came off the parenteral nutrition. We decided it would be best for Marcus if he had a surgical jejunostomy inserted so we could take him home and feed him that way. We were so excited, we had a solution.

Our excitement was very short lived. At 5 months old, he went to have the surgery to insert the feeding tube. It all appeared to go well but later that evening; it became clear that Marcus was very unwell. His heart was racing, blood pressure was high and his breathing was much more rapid than usual. He was writhing in pain. The surgeon sat with us all night, then Marcus' nappy filled with blood; rich, dark and red. He had a couple of blood transfusions before being taken back to theatre in the morning.

Then we waited...and waited... After about four hours, the surgeon came and explained that Marcus had malrotation and because he'd inserted the J-tube using key hole methods, the gut had knotted in two places and caused a volvulus. He felt the surgery had gone well; he'd re-situated the j-tube on the other side of the abdomen and

performed a Ladds procedure to correct the malrotation. He said we'd be able to see him soon...

4 hours later, they still wouldn't let us see him and no-one could tell us anything. Gary and I were going out of our minds. Eventually, it all got too much and I threatened to go and find Marcus myself. The Sister took us to PICU and sat with us in an office, the surgeon, who told us the surgery had gone well, entered. I knew it was bad. I was terrified. Why had he brought us into his office? Had Marcus died? I waited for him to speak; I didn't trust my own voice. His words are a blur to me now but he explained that Marcus had taken a turn for the worse while in recovery. He had sepsis and he was on a ventilator. "Can we see him?" I heard myself ask. I held Gary's hand very tightly as he led me into a cubicle. There was a baby on the bed, intubated, sedated, swollen and very bruised. He didn't look at all like he did this morning. I had to look closely at his face just to make sure they hadn't made a mistake. "Is he going to die?" I asked suddenly.

The doctor replied in a soft Scottish accent, "I suppose the honest answer is yes, he could ... but I don't think he will." That was enough for me; I focused on the surgeon's faith or belief in Marcus. I didn't think he would die either.

My Dad drove up to Newcastle that night and the three of us sat in vigil around Marcus' bed all night. We stared at the monitors; praying, willing the numbers to reverse - willing his temperature and heart rate down. Marcus little body lay still on the bed for most of the evening, only moving when the sedation wore off. We sat watching the rise and fall of his little chest, inflated with every compression of the concertina within the ventilator chamber. His tiny body was covered in wires, tubes, canulas, blood and adhesive plaster. By morning, the worst was over, his temperature was down, his heart rate had stabilised. Our prayers had been answered.

After a week, Marcus made his way back to ward 7 and we were able to start using the jejunostomy to feed him. We did have a few teething problems but by the time Marcus was six months old, we were all back at home in Richmond.

This marked a new phase in our lives. It was very frightening at first, having a small baby fed permanently, twenty hours a day via a pump. He was attached to a drip stand at home and we had a little ruck-sack to put the pump in when we went out. Gradually we got used to it and all the people coming to our house to attend to Marcus' needs - we now fondly refer to them all as 'his



entourage'. They've become friends, the faces change sometimes but their presence is ever felt. They come in the shape of the community nurse, health visitor, dietician, physiotherapist, speech therapist ...

Marcus still didn't grow too well, and the next year was a struggle. We tried many different types of milks and regimes. Eventually, we found that Paediasure worked and his weight improved dramatically but his vomiting gradually got worse again. Sometimes he'd vomit green bile up to 10 times a day, writhing and screaming in agony. We went back and forth to see the doctors but they couldn't find a reason why he was so sick.

Meanwhile, we were trying desperately to get him to feed orally. I managed to get him to drink water and eventually, after one particularly sick weekend, I stopped his feeds and waited. I offered him water which he took readily and then I tried offering some milk - he took it!

He has been taking his milk orally for a year now. At first he lost a lot of weight but he seems to have found his own line at the bottom of the centile chart now. I believe Marcus cannot cope with huge volumes of feed, he like little and often. He vomits very rarely now, only when he's drunk too much or is poorly. He doesn't eat solids yet but we're working very hard on that at the moment.

Marcus was diagnosed with CFC last November, just after his 2nd birthday. It came as a shock at first but

within a few days we realised how lucky we were to know for definite, what was wrong. Knowing is far, far better than not knowing. And all the pieces of the puzzle fit together and seem so obvious now. Marcus has always itched and sweated. He has sparse curly hair, low muscle tone, ptosis... you know all the rest ...

Marcus is doing really well. He's still fairly delayed but that's to be expected given that he spent the first year of his life completely malnourished. He will be three in October and he can bottom shuffle, stand supported, sit, roll and converse very effectively using a combination of Makaton, words and sounds. He's a complete joy to us and we appreciate each and every milestone all the more because we know how much effort and determination it's taken him to reach it.

At the conference, in July this year, we met our CFC family. We all share different versions of the same story. Thanks to the doctors who figured out the puzzle, we now know why and how Marcus struggled so much at the beginning and why he has so many complex problems, and it all seems so obvious now!

Gary, Sam, Joely and Marcus Weston (CFC age 2 ½)
North Yorkshire, England

Board of Directors announces new addition!



Stowell family

The newest member of the board of directors was appointed during the CFC conference. Kyle Stowell is from Farmington Utah, which is about 15 miles north of Salt Lake City. He is married to Jennifer and they have three boys -- Jason is 12, Jared (CFC) is 10, and Jordan is 2. Kyle grew up on a farm in a small town, Parowan, near Zion National Park in southern Utah. From 1989 to 1991 he lived in Perth Australia as an LDS missionary. He graduated from Brigham Young University with a degree in Electrical and Computer Engineering. He is the IT Manager at the Flying J Oil Refinery in North Salt Lake. He and Jennifer are active in their church, teaching Sunday School to the 14-15 year olds. Kyle hopes to aid CFC International with their technology needs. He has helped with the conversion of the CFC Registry into a web-based format, and designed the 2007 DVD slide show. He was a pre-med major at one point, and has some understanding of Mendelian Genetics and DNA. He also enjoys writing and is willing to contribute and help where needed to make progress on treating our CFC kids.

CFC Conference

CFC Orlando Conference Fundraising Workshop

As parent, board member and an extremely successful fundraiser, Judy Doyle presented a very informative workshop on fundraising.

The topic included event based fundraising - Fundraising through organizing events like chili cook-offs, golf tournaments, bowl-a-thons etc.

The subject included how you can make a difference by raising funds to help support CFC International and all that it provides to families.

Information was provided on the basic steps to a successful fundraiser, how to improve your fundraising strategy, understanding the basic elements of asking for money from individuals and businesses in your community as well as addressing public awareness.

Judy encouraged audience participation so that listeners could hear directly from families who have created successful fundraisers. Ideas were given and exchanged from simple jewellery sales to golf tournaments. Choose an event that fits the personality of your family and your target audience. For example if your family is into swimming, a swim-a-thon may be perfect, but a golf outing might not go over so well. Be creative and have fun with the playing.

The presentation showed that fundraisers can increase awareness of CFC syndrome and be fun for the family involved and their community. CFC International will provide brochures, a banner, a PowerPoint presentation on CFC Syndrome, photo collage poster and even write your thank you notes to donors. Information and support throughout your event planning are only a phone call away. Members of CFC International will advise you along the way.

Overall it was a very educational workshop for anyone planning to organize a fundraiser. Thank You Judy.

Working with Your Primary Care Pediatrician Workshop

By Kyle Stowell

Some of the things Dr. Maerz recommended in order to get your Pediatrician's undivided attention were:

1. Ask for a return appointment. A doctor is very busy and usually has a full schedule. If what you need to discuss can't be covered quickly, it would be helpful to

gain the doctors undivided attention by coming back another time, and/or scheduling a longer appointment.

2. Be specific. Sometimes parents are not very detailed or specific in their descriptions of what is wrong.

3. When setting up an appointment, request the last apt of the day. Usually doctors are behind schedule and always trying to get out of the room with you in order to catch up. If you have the last appointment of the day, the doctor is not rushed to get to the next room and can usually spend more time with you.

4. Get to know the staff at the doctor's office. Dr. Maerz suggested baking cookies or bringing in food, or even flowers, if you really want to get the staff's attention. Having a personal relationship with the doctor's staff can help facilitate appointments and care.

5. Give the doctor reminders -- jog his memory with statements like "you may not remember me, but my son has CFC Syndrome." This is a very, very rare syndrome. It is unlikely that a doctor has even heard of CFC, let alone knows much about it.

Dr. Maerz was really a character. He's a remarkable person and the Conger's are lucky to have him as a doctor. One question was asked about getting referrals to other doctors, specifically to a Gastroenterologist to discuss vomiting and feeding issues. They had repeatedly asked for the referral, but were repeatedly turned down. His answer was to stuff the kid with formula right before the appointment, and then pinch him hard when they got in



Dr. John Maerz with Brenda, Clifford & Cliff

with the doctor so that the baby would demonstrate to the doctor why there was a problem. He also suggested that crying (mom or dad) was something that always tugged at his strings and motivated him to take action for a patient. The suggestion was also made by audience members that if your insurance does not require primary care physicians to make referrals, to refer yourself to the necessary specialist. But care should be taken because insurance loves to deny coverage if correct procedures are not followed, including getting referrals where necessary.

There are some other resources he suggested for parents including Parent To Parent, and the Exceptional Parent Resource guide.

Orthopedics

By Kent Reinker, MD

It's clear from the children that I saw at the Orlando conference that a crouch gait is fairly common, and that tightness of the gastrocnemus causes tiptoeing and knee flexion in a number of patients. I wasn't able to tell for sure whether the crouching was due entirely to gastrocnemus tightness, hip flexor tightness, or balance issues, or all three. I suspect it's a complex combination of factors. I saw some children who are unable to bend backward and have hips that can't fully extend. These children walk tilted forward. This is quite unusual in other populations; I'd love to see some radiographs of the spine on these kids. Flat feet are common, but are common in the general population as well – this isn't limited to the CFC patients. I saw a couple

children, however, who have an unusual form of flat foot, what we would call a pes valgus. This may be something that is much more common in CFC. Finally, a number of children had spinal problems,



Dr. Reinker meeting with children at the Orlando clinic

though scoliosis was less frequent than I would have suspected. It was a pleasure to attend the meeting; thank you for inviting me.

Orange County Sheriff's Office Reaches Out to Help CFC Family

During the past two Orlando conferences for CFC International, the Orange County Sheriff's Office (OCISO) has stepped forward to assist with conference planning, volunteers, receiving shipments of boxes and donations for the silent auction. This year the friends from the Conger family's extended family (OCISO) went above and beyond the call of duty. During the Saturday night banquet at the Rosen Centre, 8 1/2 year old Harley Melvin became ill. Luckily Dr. Ines Kavamura was seated at the same table with the Melvin family and commented that Harley's coloring was very grey and she did not look good. Mom Renee had mentioned that Harley had been having a seizure which is quite common for her but the coloring change was not usual. Dr. Kavamura felt Harley should get to the hospital and the Rosen staff immediately called an ambulance. Harley was taken to the closest hospital but later transferred to Arnold Palmer Hospital where she was

treated for heart failure and later kidney problems. Over a liter of fluid was drained from Harley's heart.

The next day Brenda Conger alerted Tracie Esagro, Administrative Assistant to Captain Patty Wells about this emergency. From her home, Tracie immediately started networking within the Sheriff's Office and made connections for the Melvin family to receive a complementary room located closer to the hospital for the next three days. In addition to the room, the Sheriff's Office contacted Shannon Clayton from Pointe Orlando and Shannon arranged for food to be brought in. Two



Harley

dinner donations for the entire family were delivered by officers to the Melvin family at the hotel. In addition, Tracie made arrangements for the family to get tickets to Wet n' Wild since there was another child who had saved up his own money to go to this water park but the money was needed when Harley became ill.

The outreach to visitors has been a tradition to Captain Patricia Wells and her Administrative Assistant. Extending a helping hand is nothing new to these remarkable two women and the staff at Sector V. Orlando has long been perceived as the place where dreams come true! But kids aren't the only ones with dreams! If asked, "What did you want to be when you were a child?" the overwhelming response is a police officer. Well the Orange County Sheriff's Office implemented an innovative program where community members can actually fulfill that dream – sort of!

The T.I.P.S. Program (Tourist Information Patrol Specialist) is the brainchild of Sector 5 Patrol Captain Patricia Wells. Her vision was to establish a unit where uniform volunteers would drive specially marked police cars and offer assistance to tourists visiting the International Drive area. The assistance offered would range from providing area driving directions to directing traffic, assisting with a disabled vehicle, offering crime prevention tips and reducing potential criminal activity through high visibility patrol in hotel and business parking lots. The assistance to the Melvin family went above and beyond the normal situation for extending help to tourists.

Harley was eventually returned home to Louisiana by an Angel Flight. Her condition remains serious after a bout of pneumonia and further cardiology and respiratory problems. As of publication Harley has been sent home from Our Lady of The Lake Children's Hospital where hospice services have been sent into the home. We are keeping the Melvin family in our prayers and thank God for our friends at the Orange County Sheriff's Office who stepped in to help this family who travelled to the land of magic to find that the magic of friendship is far more powerful than anything found at Disney.



CFC international

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Participate in an IRB-approved study of language and learning in CFC syndrome!

Families of individuals with CFC syndrome ages 1-25 are invited to participate in a research study to investigate communication and literacy in children and adolescents with CFC. This research is being conducted by Rene Pierpont, M.S., as part of a doctoral dissertation at the University of Wisconsin. The study has been approved by the CFC International IRB as well as the University of Wisconsin IRB. A large number of wonderful families participated in this study at the CFC International Meeting in Orlando, but we are still looking for more participants!!

Parents of infants, children and adolescents with CFC can participate in this research by filling out a set of questionnaires about their child's language development and learning. These questionnaires take about 45 min. to fill out and can be completed through the mail. Families will receive \$15 upon completion of the questionnaires.

Families should contact the researcher directly to make arrangements to participate or to request further information about the study:

Rene Pierpont
Department of Psychology
University of Wisconsin - Madison
eipierpont@wisc.edu
608-772-1980

We look forward to your participation and to learning more about CFC syndrome!!!