



CFCinternational

Cardio-Facio-Cutaneous Syndrome

The CFC Chronicle

Caring, Facilitating & Connecting

Volume XIII Number I

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CFC International 2011 Conference

- Rosemont, Illinois, USA July 28 – 30, 2011 -

Connect with your CFC Family! Join us in the Chicago area this summer for three days of exciting talks on genetics, ophthalmology, endocrinology, future research, feeding disorders, wills & estate planning, and more! This is a wonderful opportunity to meet other families affected by Cardio-Facio-Cutaneous Syndrome. Come to learn, ask questions, share and connect face-to-face with our “CFC Family” who knows exactly what you are going through! Register after March 1, 2011 by downloading the registration form on our website:

www.cfcsyndrome.org



Book your hotel room at the Westin O’Hare now by calling 847-698-6000 and mention CFC International to get the special group rate of \$120 plus taxes or book online at:
<http://www.starwoodmeeting.com/Book/cfg26a>



In this Edition:

Message from the President	2
Donations	3
Our Little Teacher Sidney	5
Genetic Diseases of Children Conference	6
New website Helps People With Special Needs	6
Why Genetic Testing Is Important	7
Photo Gallery	9
Featured Family: Our Road to a Diagnosis	10
Understanding the Dental Characteristics of CFC	12
College Application Essay	13
Managing the Special Education Maze	14

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

These past few months have been very difficult for our organization. All of our families have been devastated by the recent deaths within our group. When any of the children are lost our whole CFC family organization grieves together with these families. When Linda Kohler called me about the death of her daughter Stephanie, the first thing she asked was what could the family do to help with research. These wonderful parents gave us all the ultimate gift and enrolled Stephanie's tissue and organs into our NDRI tissue bank program to further research on CFC syndrome.

While attending the Genetic Diseases of Children Conference in New York City this past month, I was so pleased to find out that our organization is ahead of many others by leaps and bounds. The latest talk had been about organizations pulling together to enroll into registries for further research on their syndromes. The importance of the registries for rare syndromes like ours is that researchers can find out how many individuals in the world are affected by a disorder, where in the world do all these people live, and what mutation of the genes do these individuals carry. The importance of all of this data collection is that when researchers set up clinical trials, there is a set known population of individuals who can apply to enroll. So where are we as an organization in all of this planning? First, we have had a CFC Registry for over 10 years and recently we have added a join button to our website so that family members can immediately join our private CFC listserv to remain up to date on all the latest news. We keep track of individuals from all over the world and are able to now track vital information on the children's ages upon diagnosis and the gene mutations. As I mentioned, we are really on top of what needs to be done to partner with researchers thanks to all the families who send in the CFC Registry. This summer at the Chicago conference, the researchers who are involved with the RAS-MAPK Pathway (CFC, Noonan, Costello, Neurofibromatosis, Leopard syndromes), will also convene to further their efforts to move research along. I know that our CFC Registry will serve as a model to the other organizations as we move into a partnership to come up with a joint Rasopathy Registry. Thank you to all our families and board members who work tirelessly on these projects.

Brenda Conger

E-mail: bconger@cfcysndrome.org

Thank you!

Donations

CFC International is grateful to its supporters for their generosity. We extend our deepest thanks to the contributors listed below for their kind donations.

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Angel on loan.....

*An angel was given to me on January 6, 1999.
We knew right away we would only have her a very short time.
As each day passed we watched her grow.
We all fell in love with her as our compassion would show.
We knew when the day would come, we would not want her to go.*



*We all wanted what
is best for our
Angel sent to us
from above.
Harley is my
daughter, my
teacher and my
best friend.
I know a day will
come when we will
meet again.*

*I know Harley is an Angel that was sent to me on loan.
I know Harley accomplished all that she was to do down here,
and God said "It's time Harley for you to come home".
With God's spoken words, Harley left us on March 12, 2009.*

*Our special Angel on loan was finally able to run, walk, talk,
crawl and play, because she is now in the arms of God and
that is how she will stay.
I love you my daughter, my teacher and my very best friend.*

*Love Always,
Mommy*

*Written June 25, 2009
In Loving Memory of Harley D'Nee' Melvin
Sincerely, Renee' Melvin (RIP Harley 01/06/99--03/12/09).*

Cookies to Help A Cousin

Mario Catania, an 8th grader at H.B. Whitehorne Middle School in Verona, New Jersey, has started a community service project to benefit a young cousin with a rare disease. It is called "Cookies for Cousin", and it is designed to aid research into Cardio-Facio-Cutaneous Syndrome. Catania's cousin Nola Rose Iacobelli was born with the condition.



Mario Catania

To help, Mario is making and selling bags of chocolate chip, peanut butter and sugar cookies. The cookies are \$7 a bag and 100% of the proceeds go to CFC International.

Soft Clothing for All Children

Many children are extra-sensitive to the texture and feel of clothing. Soft® is the first line of inclusive clothing designed with the needs of all children in mind, including those with Autism, Sensory Processing Disorder, ADHD, and tactile defensiveness/sensitivity. We use flat seaming and seamless construction for extra comfort, 100% of the softest combed cotton and our specially developed Soft Sensory Blends, wide collars, encased elastic waistbands, printed labels (tagless), custom fits, and much more.

Created by a Mom and Special Education teacher, our goal is to design clothing that can enhance social skills, sensory organization, concentration, by addressing unique sensory needs, but without sacrificing personal style and self expression.

More than 1 in 150 children have sensory needs that are largely unmet by the children's apparel market today. Soft is about to revolutionize children's apparel. We are the first inclusive and universally designed line of clothing geared toward children with sensory or tactile sensitivity, a common symptom of Autism, Aspergers, and Sensory Processing Disorder. Soft is designed and constructed considering the needs of all children, particularly, sensory sensitive dressers.

Soft provides comfort and style for all children. After conducting over two years of in-depth market research, we learned a lot about what parents want and what children like.

We have incorporated that feedback to create stylish and comfortable clothes for the people who need it most. We use:

- Flat seaming totally seamless construction throughout (for extra comfort)
- Soft high quality cotton (combed, bio-washed and pre-washed for extra soft and smoothness)
- Wide collars (for a roomy fit)
- Encased elastic (waistbands that don't pinch)
- Printed labels throughout -- printed with water based ink (for a smooth non-plasticity feel that won't itch)
- Vegetable dyes, natural enzyme washes, and water based digital prints (to protect against allergies)

<http://www.softclothing.net>

Grandmother Partners with her Eastern Star Chapter & Applebee's to Raise Funds

Applebee's offers community organizations an outstanding fundraising opportunity with the Flapjack Fundraiser Program. Grandmother Melanie Trimm thought it would be an awesome idea to take this opportunity to try to raise money for CFC International. Her Eastern Star chapter, Rebekah #809, is based in Chicago IL, but with several members located here in Springfield, agreed to host the fundraiser.



Becky and Jackson Wadell, Grandma Tina Mitchell with Shayanna

Applebee's offered their restaurant during non-business hours on Saturday, November 13, 2010 from 8:00am-10:00am and provided a template to sell tickets at \$5 each. The hosting organization keeps all of the ticket sale revenue, minus \$1 per attendee which is paid to Applebee's. All the organization had to do was provide volunteers to work that

morning to host, seat, serve, and bus. Breakfast included 3 pancakes, 2 slices of bacon, and unlimited beverages. We sold tickets to family, friends, and co-workers in advance of the breakfast and planned to sell the remaining tickets at the restaurant on the day of the event. Unfortunately, we were hit with a cold and windy Saturday and attendance was not what we had hoped for. Despite the weather, we were able to raise a total of \$475.50 for CFC International. I wish we could have raised more and hopefully we can do it again in the near future. The best part of the day was having an opportunity to meet the Waddell family from Decatur, IL. Although we were busy serving customers, it was fun to take time to have Shyanna and Jack finally meet!

Our Little Teacher Sidney

On January 19, 2011, we got the news that we had been waiting 2 months to hear. Our baby girl Sidney had CFC syndrome. We were happy to finally know what she had but sad because we would never get the chance to see her through it. Sidney passed away on December 7, 2010, before we could fully understand CFC and take this challenge on. It was her 3 month birthday.



Sidney

Sidney Jean Blackwood Jones was born on September 7, 2010. I measured at 53 weeks when I delivered her and I was only 35 weeks along. We knew during my pregnancy that something was wrong. We weren't fully aware of the extent of her problems though. They couldn't tell us much when she was in utero because there was so much fluid.

But we held on to the hope

that I would deliver a healthy baby girl.

She was born 8lbs 4oz and was a beautiful angel with curly hair. She had feeding problems right from the beginning and we were told she suffered a brain injury in utero which contributed to her seizures. Now we know that was not the case. Her brain being underdeveloped was a part of her CFC.

Sidney stayed in the hospital for over 3 weeks. We brought her home ready to make sure she would thrive. Noah

and I learned how to feed her thru her NG tube, gave her seizure meds, took her to her appointments, held and loved her. Our other children, Samantha (8) and Grant (5) loved being her big sister and brother. They couldn't wait to help us give her a bath, read to her and have her play with them in their "fort." We were ready to take on her world but she was only home for 3 weeks when we noticed that she wasn't looking good.



Samantha, Grant and Sidney

When we had brought her home the first time, they told us she had a heart murmur but it would be okay. When we took her into a hospital this time, they noticed that it was more than that. She had pulmonary stenosis and they would have to do a balloon procedure to open up a heart valve. Her little

body couldn't survive and she spent the next 6 weeks bloated, hooked up to tubes with meds coming and fluids exiting her body and her lungs and heart were fighting against each other. She also had several blood transfusions, struggled fighting off infections and we weren't able to hold her.

We spent the days rubbing her toes, kissing her forehead and staring at her. She didn't tolerate us touching her much and the last time we really truly held her was the night before her procedure. The very last time would be when we said goodbye.

Even though she was only with us for 3 short months, she taught Noah, the kids and I more than we could ever teach her. She taught us to be strong, take care of each other and most of all cherish each moment we have because you never know what is in store for you. She also brought out the best in people. We were very lucky to have such an amazing family and community where everyone helped us out.

We have spent the last few months trying to figure out our role as parents of two again, how to survive each day without her and how we will make sure she is remembered. She and I will be studied to hopefully help doctors see the signs of CFC during pregnancy and we hope she will be able to help others with what they have learned from her.

When Sidney was first born, I opened one of Samantha's prayer books and read the following verse: "Before I made you in your mother's womb, I chose you. Before you were born, I set you apart for a special work." Jeremiah 1:5. This is our motivation and it gets us thru each day. We love and miss her more than anything but we know she will live on thru us. Until we meet again, Angel Baby.

Ashley Jones
Los Banos, California

Genetic Diseases of Children Conference

Advancing Research & Care

The Genetic Diseases of Children Conference was held March 8-9th at the Sheraton New York Hotel & Towers. Three members from CFC International attended this national event along with 500 other attendees. Brenda Conger, Jenny Iacobelli, and Laura Mitchell were all in attendance. Brenda presented at three sessions in the family track of "Sharing Family Experiences".



Brenda Conger, Jenny Iacobelli, Laura Mitchell

This conference was an exceptional opportunity to direct collective focus towards creating opportunities to advance research

and improve the delivery of health care for children living with genetic disorders. The goal of the conference was to develop recommendations for establishing a comprehensive, state-based model that will effectively: facilitate early diagnosis; improve access and quality of care; optimize the coordination of provider services; and foster high impact clinical research to expedite improved medical treatments.

The conference offered a comprehensive agenda presented through four plenary sessions and over 35 panel discussions covering five independent themes:

- Reaching An Early Diagnosis

- Sharing Family Experiences
- Optimizing Care And Coordination Of Services
- Children Living With A Genetic Disorder: Meeting Their Needs
- Accelerating Research

The Genetics Diseases of Children Conference was presented by the New York State Department of Health's Wadsworth Center in conjunction with the National Institutes of Health's Office of Rare Diseases Research, the National Institutes of Health's Office of Dietary Supplements, Genetic Alliance, the National Organization for Rare Disorders (NORD) and the Jeffrey Modell Foundation.

New website Helps People With Special Needs

Navigate Health Care Reform

The landmark health care reform legislation enacted this past spring includes new programs and services designed to help people with special needs and also alters the way many existing government benefit programs work. Several months later, the federal government has designed a new website, HealthCare.gov that helps people, including those with special needs, navigate the new law.

The new law has many moving parts that will affect every one differently. The new website aims to provide consumers with some answers to their health care questions in easily understood language. Since most of the changes to the health care system will be phased in over time, the site also features a helpful timeline that illustrates exactly what benefits will begin and who will receive them.

The HealthCare.gov site includes a section devoted entirely to helping people with special needs. This section outlines changes in the health care legislation that may affect people with special needs, and focuses on the portion of the new legislation that prevents insurers from denying coverage for patients with pre-existing conditions -- something that advocates for people with special needs have long sought. The website also provides tools for people looking for health insurance, including descriptions of both public and private programs that may be of use to people with special needs.

Why Genetic Testing Is Important

by Lisa Schoyer (Quin's Mom, and Chair of the Costello Syndrome Research Advisory Group)

Perhaps chasing a gene test is one of those efforts that don't make it on the Do List because of all the other Must Do's, day in and day out. And chasing authorization to get the gene test done seems futile or requires energy that could be used elsewhere, including taking care of yourself (crucial to being a good caregiver). For those of you whose routine for your children is manageable, we know how easy the boat can be rocked. But, if you do find time or the opportunity (like the next time your child is in the hospital), here are some things to consider:

1. Testing helps you tap into what's known (so far) about each of the sub-mutations.

(For Costello syndrome) While children with the G12S mutation (about 80% of our children) show a broad spectrum of issues, a few of the rarer mutations appear to have more or fewer issues. Because the numbers are so small, like fewer than 10 children with G13C, it's really hard to tell if it's the family genes or the Costello mutation causing it. For those rarer mutations, the information we do have provides some ideas to start with – and can only grow as more families get their children tested and share their children's information.

For CFC syndrome, there are four known mutations identified so far: BRAF, KRAS, MAP2K1 and MAP2K2. In addition to continuing to search for more mutations responsible for causing CFC syndrome, the only way researchers will be able to get a better understanding of what each mutation looks like is to know what mutation the child has.

2. Testing to share with researchers is a gift to future families because it adds to what they know so far.

- Because our children's syndrome is so rare, every little bit of information helps to clarify the difference between what's Costello or CFC and what's inherited from the parents.
- The way doctors identify best practice is to look for patterns. Patterns only come from individual families sharing.
- If the child tests negative for all known RASopathy mutations, the child's information is put in a pool of "one of the RASopathies - clinically, but with an unknown molecular mutation" from which patterns may emerge.

An Important Story:

Two adult sisters in Norway who were clinically diagnosed with Costello syndrome decades ago were tested in 2007, and it turned out that one has the G12A sub-mutation of HRAS (so: definitely Costello) and the other, who was having seizures, actually has a KRAS mutation. Her diagnosis was changed to CFC syndrome. These two sisters had contributed to early thoughts of Costello syndrome as an autosomal recessive syndrome, so the results of the testing are even more important, because they corrected an early scientific assumption. Though our researchers were pretty confident that CS isn't autosomal recessive by the time the gene causing CS was identified, having these two women's test results led to:

- the benefit of confirming a clinical diagnosis with a gene test for this family: it confirmed one, and helped make a more accurate diagnosis for the other, and
- being able to add information to what researchers and doctors know about G12A and KRAS – it explained why the sister with the KRAS gene had seizures while the other did not, building a more tailored approach for future children.

Where to test:

First choice: Ask your private insurance or government insurance to pay for the test. If they say no, then try to appeal. The case of the Norwegian sisters shows how knowing in advance can help your child's doctor develop a medical plan for your child. Risk of cancer is much higher for children with any of the HRAS mutations in comparison to the other syndromes - another reason to advocate for a gene test.

Second choice: Find a reputable researcher accepting research participants (we can help). While this is a free option, the reason it's not first choice is because the researcher's "client" is the organization that provided the research grant, not the individual family. They look at all participants as a group affected by the syndrome in their research study. What they share with the family isn't required, so if they do, it's a courtesy. A CLIA-approved laboratory (in the US) looks to the individual as the client, so the lab is better prepared to provide information to the family (through the doctor who ordered the test).

Here's a weblink to a really great one-stop site for labs that test for Ras syndromes to show your child's doctor. It's a list of labs that have informed the GeneTests.org folks that they do the test, so it's not a complete list, but it's a good place to start:

http://www.ncbi.nlm.nih.gov/sites/GeneTests/lab/clinical_disease_id/240294?db=genetests/

If you have any questions, please don't hesitate to contact me at http://www.costellokids.com/contact/contact_Lisa_Schoyer.php

Stephanie Kohler 12/2/86 – 3/28/11

Imperfection?

Our precious girl was born, with too many challenges for one girl
But she never thought she had them, with her cute smile and wavy curls

What is perfection? Is it the beauty of a model or glitz of a star?
Or is it her capacity to love every human near and far.

Is it the perfect piece of art, the writing of a book?
Or the smile when you are hurting, because of mommy's worried look.

Is it the strength of the power lifter, the boxer's crushing blow?
Or the loving caress to daddy's face, when he is feeling low.

Is it the Nobel prize, Oscar or the Mensas oh so bright?
Or the model for your brothers to know wrong from right



Steph was an angel from the start, an angel without sin
An angel that without fail could make the world grin

She was so strong, so tough, she never showed her pain,
She got close to heaven, when she stroked the horse's mane

Our precious girl's body was frail, but her spirit was so strong,
God needed her back in heaven, he only loaned her for so long.

Imperfection is most likely what some might see at first
An angel is what we saw, and now our hearts have burst.

"Imperfection" is not the word for her, but an apostrophe and space help change and link it
"I'm perfection" is so much better, but of course our Stephanie would never think it.

Because being perfect is being humble, putting others ahead of you.
Being perfect is reserved by God for the paradoxically imperfect few.

By: Jeff Kohler, Father

Photo Gallery



Nola Iacobelli celebrates her 3rd birthday



Daniel Hess enjoy a winter snow fort



Maddie Mitchell as the Northern Star in a Christmas play at school



Ashton Nagy celebrates his 7th birthday



Clifford Conger, age 17 competing in the Special Olympics Winter Games



Graham Randall, age 3, on the move!



Meg Young enjoying the winter snow

Featured Family: Our Road to a Diagnosis

Greetings from sunny (and flooding) South Africa!

It's been fifteen years of doctors, hospitals, pain, tears and doubt, but finally a diagnosis!

After an eventful pregnancy where I had slipped discs in my back, hip displacement, and a hormonal rash, Brittany was born three and a half weeks premature on 30th October 1995, weighing in at a hefty 3.9kg. She was admitted to the neo-natal ward due to excessive sweating and breathing irregularities.

By the time she reached three months old, I was concerned about her delayed milestones, such as not being able to hold her head up and so started months and years of tests as well as numerous operations.

It started when she had a bilateral myringotomy, followed by a skull ultrasound which showed her head growth to be too rapid. Grommets and a tonsillectomy & adenoidectomy were performed at the age of five months. Thereafter it was a kidney and brain sonar, CT brain scan, sonar of the abdomen (and in between this many, many blood tests and for a mom who was petrified of needles, this was very difficult for me to watch), and a skeletal survey, all of which could give us no answers.

Then it was off to Potchefstroom University for metabolic testing – still no answers!

I started her at occupational therapy at the age of ten months and at this stage her weight was 6.6kg, she was unable to hold a bottle, still unable to hold her head up unaided, still not able to eat solids, sensitive to loud noises, kicked with one leg only and had extra sensory defensiveness – so we started the “brushing” process. Her developmental age was put at three months.

More blood tests, a CAT scan of the brain, X-rays of the left wrist to check her bone age, were followed by an operation in which they had to remove a vein under her eye which kept “popping” and bleeding - at one stage, there was so much blood I thought a wild animal had got hold of her during the night – after all this is Africa!!

Right before her first birthday, we took her for genetic counselling. At this stage her weight was 7.4kg, she had a shortish neck, fetal pads on her fingers, bilateral epicanthic folds, developmental delay and presented with a hepatosplenomegaly. She was referred immediately to the oncology unit for a bone marrow biopsy, liver biopsy, lumbar punches and sweat tests (these were horribly painful). Their suggestion was Liver Storage Disease or Haemo-phago-cytic-lympho-hystio-cytosis, but neither was ever confirmed.



The family Atherstone

During the following year she was hospitalised numerous times with urinary tract infections, bronchial pneumonia, failure to thrive, chest infections, gastro and an “eating disorder” and was fed intravenously.

At the age of three, we stopped all testing – as parents we could not deal with the pain our little one was going through.

At age five, she had to have all her baby teeth removed as they had come in with no enamel on them. Then just to make matters worse, or perhaps she was just being a little girl, she broke her leg after falling off the see-saw.

Two years later, she developed multiple mollusca and had 59 lesions removed from her chest, neck and groin area.

A year went by and we felt that we were ready to face more tests to get answers after numerous tests and X-rays which showed severe stunted growth, it was suggested that she had Turner Syndrome – once again, this was not confirmed.

During the month of March 2006, I felt as though I was in a movie – everything happened so fast blood tests with glucose injections, liver function test showed that her enzymes were elevated and we were referred to an endocrinologist who then referred us to genetics. Our geneticist was concerned about the wrinkly skin under her feet, hands and on her neck and our gastroenterologist did more tests (and eventually had to do a gastroscopy and bowel biopsy which showed an infection in the stomach lining and detected an abnormality in the bowel) and so, her DNA took to the air and was flown to London to undergo clinical trials for a syndrome – it would be seven months before we had some answers!

During this time, Brittany had been attending school – a small, farm school with much individual attention, however they did suggest that I put her in a “special school”. I refused and took her off for psychological testing – they found that she had obsessive compulsive disorder, tactile defensiveness, separation anxiety, a damaged neuron in the brain which affects spatial development, behavioural problems, educational problems and psychological problems – but hey, nothing we could not deal with – and really not as bad as it seemed at the time.



Brittany and Karen Atherstone

In November of 2006, we had a clinical diagnosis of CFC Syndrome. We were happy that we finally had an answer, accepted it and moved on!

Four years went by, and on the 21st September 2010, we got a call from genetics to say that they had withdrawn the previous clinical diagnosis of CFC and after conducting more genetic tests on her DNA, it showed that she has the SHOC2 4A>G gene and was given a final diagnosis of Noonan-like Syndrome with Loose Anagen Hair.

By the way, this comes with a wonderfully tanned looking skin (without having to use a sunbed). Yes, her hair is sparse, but we have clip on pony tails and in time we will look at hair extensions once she can look after them herself, she has a slight heart murmur and will have to go for annual cardiac examinations. She has also started wearing glasses for watching TV. She still has major separation anxiety and will not sleep out at friends, but if that is all I have got to be anxious about, well hehllllloooooo!!!

We are proud to say that Brittany passed her primary school (Grade 7) with pretty good results, given the problems she has faced, and even won the Principal’s Trophy last year for her perseverance despite her setbacks in life. However, I have taken the decision to home school her for high school and will be following thematic unit studies (thanks to the internet) and doing more life skills training such as beading, cooking, sewing etc, and she will be taking up pony club, dancing and other activities in our area.

All is not lost – we lived through fifteen years of dealing with the symptoms as they appeared not knowing any different and nothing has changed! We have a wonderful daughter who is turning 16 this year!

Now a little more about the rest of our family. My hubby Dave is General Manager of Operations for Bill Harrop’s Original Balloon Safaris. He joined the company after 33 years in the hotel industry. We live on a farm out of Johannesburg with the most beautiful mountain view. (He is also in the process of obtaining his pilots license!!)

In fact, at the moment we have 3 dogs, a tortoise, a family of mongoose, rabbits, whistling ducks, Egyptian geese and a family of porcupine on the property as well as

..... wait for it a crocodile (approx. 1m baby) in our front dam – of the view from our verandah. We caught a 2.1 m croc weighing in at 36kg two weeks ago which has gone off to the Reptile Park. These were not intended, but with the recent floods a lot of crocs have come up from the river to escape the floods!!

I am in the wedding industry and work as the Marketing Manager for Oakfield Farm Wedding Venue. I will carry on working for them from home as of February. I was also in the tourism & hotel industry for a long time prior to this.

Ashleigh, our eldest daughter is turning 20 in two weeks time and is studying part time to be a pre-school teacher and wants to specialize in mentally and physically challenged children. Whilst studying she has a wonderful job at a pre-school 6km from our home – also on a farm with horses and a spectacular view of the mountains.

Karen Atherstone
Mom to Brittany (15)
E-Mail: dave@balloon.co.za

Understanding the Dental Characteristics of CFC

Although the Ras/MAPK pathway that is affected in CFC syndrome is known to play an important role in tooth development, the teeth of individuals with CFC had not been well characterized. Thus, our team, including Ophir Klein (geneticist), Kate Rauen (geneticist), Sneha Oberoi (orthodontist), Cecilia Fairley (genetic counselor), Cyril Charles (evolutionary biologist), Jessica Groth (pediatric dentist) and Alice Goodwin (dentist) from the University of California, San Francisco (UCSF), examined the teeth of patients at the CFC Family Network Conference in Berkeley, CA in August 2009. At the conference, we reviewed dental histories and x-rays, performed intra- and extra-oral exams, and took pictures of the face and inside of the mouth. In total, we examined 25 patients. We were able to confirm many of the previously described craniofacial characteristics of CFC in a large group of individuals, including macrocephaly (proportionally large head), bitemporal narrowing (narrowing at the temples), sparse, curly hair, hypertelorism (widened space between the eyes), ptosis

(drooping eye lids), and small chin. In addition, we characterized the teeth of patients with CFC, which had not yet been done, and found several dental characteristics.

These characteristics are found in the general population as well, but they appear more frequently in CFC patients. We found that most patients (80%) had a convex facial profile. We also found malocclusion, most commonly, anterior open bite (40%) (with back teeth touching, front teeth are open) and narrow maxilla with posterior crossbite (28%) (lower teeth are outside of the upper teeth). In addition, most patients had a high arched, narrow palate (81%). We also found several characteristic habits, including open mouth posture (32%) and tongue thrusting (28%). Otherwise, we found that the teeth of individuals with CFC were normal in size, shape and number.

We plan to attend the Family Conference in Chicago, IL in July 2011 to perform dental exams, review x-rays, and collect lost teeth that families are willing to donate with a focus on characterizing the enamel structure of CFC teeth. With this project, we hope to further our understanding of craniofacial and dental development in the RASopathies and gain a greater understanding of enamel formation in general.



Ophir Klein, Sneha Oberoi, Cecilia Fairley, Cyril Charles, Jessica Groth and Alice Goodwin

Thank you again to all of the children and families who participated in the study in 2009. We look forward to attending the Family Conference in July 2011, and we hope to recognize faces from the previous conference and see new faces! Thank you again!

College Application Essay

By: *Rachel Del Ciello, Glen Ellyn, IL*

I did not *want* to know more about him. Simply put, I *had* to know more about that little boy. Many times I had watched him scoot down the driveway on his bottom out the back window of my house. I will admit it...I stared at him. I had a perfect view of the driveway that belonged to the neighbors that had recently moved in behind us. I used this view to watch the multitudes of people go in and out of the house each day, but more recently, I had started using it to watch that boy. He would laugh and play outside with the same joy that any other child does. But he was not like any other child. I did not know this little boy, with his brown curls springing off his head, scooting his way around outside of his house. Questions ran through my head as though they were on the marquee outside a movie theatre. Why does he scoot like that? Can he not walk? What is wrong with him? Why do all of those people keep going in and out of their house? All of the things I wondered, I knew were harmless, though they have poor connotations. I wanted to know more because I was interested in his differences. I did not think he was strange, or stare with disgusted wonder. I thought he was adorable and interesting, in all the ways a three-year-old was. And when I met him, I found out he was much more extraordinary that I had first thought.



Rachel and Daniel Hess

I did not formally meet that little boy until close to a year after he moved in. His name was Daniel. He was one of the happiest little boys I had ever met. And those people streaming in and out of his house were nurses and therapists that came to take care of him. My thoughts

regarding his special differences were immediately confirmed. I, then, discovered that he had a syndrome I had never heard of before. Anything and everything to do with special needs had always fascinated me. I was learning American Sign Language and wanted to be a special education teacher. From an early age, I had devoted myself to children with special needs. One thing I had always known was that there was so much more in the special needs category than autism, Down syndrome and cerebral palsy, that those were just some of the most common disabilities. What I did not know was the extent to which they were all different, yet so much the same. I quickly learned that Daniel has a rare syndrome most commonly known as CFC syndrome, the more condensed version of Cardio-Facio-Cutaneous Syndrome. This syndrome gives a child very distinguishing features, much like Down's syndrome does for a child. These features included the cute, tight, little curls that top Daniel's head and his small stature. It was also the reason for the GI tube Daniel happily shows off when asked to see his tummy.

Each time I came into contact with Daniel, I learned and observed something new about him that answered the questions that so frequently popped into my head. The reason he did not walk? He simply was not strong enough. The syndrome had not allowed for his muscles to grow at a rate that would allow him to hold up his own weight. What was wrong with him? Though it sounded like an awful way to put it, it was the syndrome. There were so many ways that Daniel is different than every other child, and so many ways that he is the same as them. I rapidly learned more about his syndrome and shared my knowledge about it with others. My passion for special needs children expanded so much farther after I met Daniel. I wanted to learn everything about him and how he functioned. All of my desires to know more were fulfilled in the many hours I have been at their house and therapy sessions I have observed. I was witness to speech and language therapy, occupational therapy and even horse therapy. Daniel has been a great learning tool for all the lives that he has touched. He helped my Girl Scout troop and I learn more about caring for children with special needs. He also helped me learn what I wanted to do with my life.

Without Daniel and my ability to observe him during some of his therapies, I never would have found my path in life: special education with focus in occupational therapy. There was one project for my Girl Scout interest project that I had to shadow a therapist. Since I knew how

many therapies Daniel attended on a regular basis, he was my go-to guy to find a therapist to shadow. It was the day that opened my eyes to the rest of my life. I accompanied Daniel and his nurse, along with my mom, to Easter Seals of DuPage to his weekly OT session. I was familiar with the concept of OT, helping people learn to do things to help them complete everyday tasks. When I entered the room following the confident little boy, I was surprised to see the room filled with mats and swings hanging from the ceiling. I did not really know what to expect, but it was not that. What I gathered from that day that I observed Daniel climbing around and playing “mail man” by throwing bean bags into a tube from lying atop a swing was that OT was essentially play time. This play time worked his large motor skills and also his confidence. Daniel climbed through an obstacle course and picked up puzzle pieces to focus on using his muscles and helped him complete the goal of putting the puzzle piece together. The end of this session focused on the fine motor skills of writing. Though this was a hard task for Daniel, much like parts of the obstacle course had been, he was able to effectively channel his focus to stay calm.

Occupational therapy opened my eyes to all the wonders it did for children, especially those with special needs. This therapy was not a torture for children, it was fun. It was playtime with a purpose, and I loved the idea of that. I learned that day, and from that sweet little seven-year-old, what I wanted to do with the rest of my life. I wanted to help special needs children be able to perform the everyday tasks that most people take for advantage. I learned from talking to Daniel’s therapist that occupational therapists assist children in learning everything from the basics of standing and walking to climbing, throwing and jumping. Daniel has had a profound influence on me in the short six years I have known him. Everything from his smile to the way he yells “RAAAACHELLLL” when he wants to get my attention thrills me. I do not think I would be as driven and value my life and the people around me as much if I had not met him. He has taught me, through all the pain and troubles he has been through, that good days are good days, and you move on from bad days and look to have a better day tomorrow.

Managing the Special Education Maze

Our experience with Jared's education has been a roller coaster ride. I hope that by sharing it, others can know what to expect and better navigate the maze of IEP's and 504's that make up public education for those with disabilities. Jared wasn't diagnosed with CFC Syndrome until he was 5. We knew something was wrong long before then, as he had 4 surgeries by the time he was 3 years old. He was delayed in meeting the usual milestones of crawling and walking, and his speech was unintelligible until he was about 4. Because of his developmental delay, Jared received in-home visits from the school district’s Early Intervention program. I believe there was some speech and occupational therapy back then. When he was 3, he started pre-school at an elementary school. A Special-Education bus would pick him up and drop him off at the door of our house. The highlight of his day was riding that bus. This time in Jared’s life began our first exposure to IEP's -- Individualized Educational Plans. After testing, an IEP must be completed for a student to enroll in Special Education services in the public education system. This was all very new to us and we still don't understand it all. Once a year we would sit down with district personnel and Jared's teachers and come up with specific goals for Jared to reach. We would also discuss the goals he had from last year and how he was performing.

It was around the time that Jared was 4 when we realized he was not making very good progress with the speech therapist from school. I don't remember where we found her, but we were put in touch with a speech therapist who worked from her home. Jared would go once a week for a 1/2 hour session. She knew just what sounds and letters and letter combinations to work on, and she had incredible motivational skills. We had to pay for these sessions out of our own pocket as our insurance denied our request for reimbursement. But in retrospect, it was well worth every penny. We credit Jared's very good speech skills to her help. We also learned this important lesson: if things aren't progressing through the free public educational services, seek help from the private sector. From his public preschool, Jared went to Kindergarten, where we found out that his teacher just let him play. In 1st grade, he started out with a very kind teacher in the Resource Room program. We knew this

wasn't intensive enough for him, so we moved him to a self-contained Learning Center. Jared received much more one-on-one attention there. For me, finding the right person has always been important. His Learning Center teacher was definitely the right one for Jared. Like the speech therapist, Mr. H knew how to motivate



Jared Stowell

Jared. He ended up staying there from 1st through 6th grade. He made excellent progress.

Jared's IEP team came up with a plan to transition him to Junior High. Included in the plan was moving from Special Education self contained class to Resource Room services for 3 periods, with 3 periods of mainstream classes. He would be able to be a peer tutor to the students in the severely disabled Special Education class. He would be escorted between classes by peer tutors and he would eat lunch early with the Special Ed class. During the summer between 6th and 7th grade, the school dropped the ball, and we were left scrambling to figure out what to do. Jared ended up back in the 6th grade, but he still made the transition to a mix of Resource Room services and mainstream class. This gave him one more year of physical growth and he got to know his peers, which has helped him feel more at home in Junior High this year.

At the end of Jared's 2nd year in 6th grade, we made the same plan as the year before for his transition. This time, with a little prodding from us, the school came through with most of the elements of the plan. The Resource Room teacher has really come through with Jared. She attends Jared's math class with him and assists him right in the classroom. Jared's weakest subject has

always been math. He couldn't add single digit numbers in the third grade. His end-of-level testing after two years of 6th grade placed him in the 54th percentile. He has gotten B's every term and is learning pre-algebra, fractions, and decimals. His progress has been remarkable. Most of the mainstream teachers have been great to work with, and he has received a 3.3 GPA in both terms so far, with this term looking even better. Socially, Jared doesn't always make good choices. Some of the kids push him to "scare" girls and he gets in kid's faces sometimes and makes strange noises. He has always had a difficult time with change and he does act out and tries to be the center of attention when he shouldn't. But these are issues everyone is aware of and working on. Overall Jared is progressing and we have reason to be optimistic about his future.

Kyle Stowell

Farmington, Utah



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