CFC international
Cardio-Facio-Cutaneous Syndrome

The CFC Chronicle

Caring, Facilitating & Connecting

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Dolphin Therapy to Genetics Convention and beyond….

By Flora Tobby

When my daughter Amara was born on April 1, 1992, never in my wildest dreams did I imagine the great significance she would bring into our lives. That first year was filled with the big unknown, what did our daughter have? No one at the time could give us a real diagnosis. By the time she was 18 months old she had been diagnosed with "failure to thrive" which was the most horrifying term to me, then to "cocktail syndrome" that sounded better, then to Noonan, then to Costello where we finally settled. Of course back then, which seems like ancient times now since it was pre-internet I felt very isolated. Especially since they had told me she was 1 of 300 in the world. I thought what are the odds of me, of us, getting the 1 of 300 that didn't like to eat food? After all, her dad is in the business of fine dining, who doesn't like food?

Flora & Amara Tobby along with other Genetic Alliance advocates who attended the ASHG meeting

So in 1996 since there was not much for treatment they could do for us in the Bay Area, we decided to move to paradise, yes Hawaii, and begin a new life. My grandparents and parents had grown up here and we had heard that medical care was better and that hospitals were incorporating alternative/eastern treatment with western medicine. I thought that was a novel, contemporary, great idea. However the medical care turned out to be not as expected but certainly compassionate once I found my way. The problem, small demographic, fewer choices in specialists. But maybe looking back that was a good thing because it brought me back to the University of California San Francisco Medical Center years later where we would have the opportunity of meeting the extraordinary Dr. Kate Rauen who of course would bring us to learn about CFC. The decision to move my then young family to the islands turned out to be one of the best decisions of my life.

Here I was able to reach out through Amara's elementary school to other parents with children of special needs through a family support group I initiated in order to find my way to familiarize myself with the local system. That was truly so helpful because I connected with many other moms trying to navigate their way through education and the medical services system. We would meet once a month on the campus and a small group of committed moms participated.

There were no criteria other than be a mom of a child with a medical challenge. There I met my best friend Young Sun who had a boy named Timmy who to this day is still Amara's boyfriend. It was she who first planted the idea of dolphin therapy. We thought since we are in Hawaii why not get the kids in the water with the dolphins? And you know we made it happen. It was one of the most memorable and remarkable experiences with Amara.

Dolphin Therapy …. continued on page 7.

In this Edition:

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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

2010 is off to a wonderful start. The transfer of our stored DNA from our self funded BioBank program has been successful. The DNA left the lab of Gene logic in Maryland and is now stored through the National Disease Research Interchange (NDRI) program in Pennsylvania. We now have an agreement with NDRI to be members of the tissue bank program along with the storage of our DNA. Tissue can be obtained in two different ways. One way to obtain tissue would be if a child is going in for surgery. The parents would alert the tissue bank and NDRI would take care of all the arrangements prior. The other way is if a child passes away and the parents wish to donate tissue for further research. Those arrangements would also be taken care of totally by NDRI. Either way, parents should first register with the tissue bank ahead of time at www.ndriresource.org. There is no charge to families to participate in this valuable program that will help fuel more research.

Conference planning for 2011 has already begun. We are searching potential sites in the Chicago area with a professional conference planner. The international conference event will take place during the summer of 2011. The conference will also be at the same hotel with the Noonan and Costello groups as well. We are hoping to share some of the workshop speakers within the three groups since many of the concerns are similar to all of us. Further updates will be posted on our website and private family list serve.

It can be said that we are all teachers and students during our life times. I have always viewed my role with our two children to be one of teacher and role model. This past week's ski accident left me with a torn up knee. Our son Clifford who was born with CFC is now my teacher. As I learned to maneuver with crutches and a leg brace he saw that I was afraid to come down the stairs in our house. He looked at me and said, "mom, why don't you come down the way I do when I have to carry heavy things? I just scoot down on my bottom." Hey, it now works for me and our son is now the teacher! He asked me yesterday if I planned to ski this weekend since Sunday is the day we normally go up to Greek Peak. I told him no and that I was done for the season. He said, "me too." He definitely does not have his dad's love for the slopes and is anxious for spring when he can get his John Deere tractor out of the storage shed.

Brenda Conger
E-mail: bconger@cfcsyndrome.org

Donations

Thank you!

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

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Explaining Special Ed

If your child is eligible (now or in the future) for Special Services through your school system, there are many CFC International parents who have shared their hard won knowledge of how the "system" works. Although this information applies to US families, other countries may have similar administrative procedures.

The Special Education system is devised to provide a seamless process for children with special needs. Babies from birth to age 3 receive Early Intervention services, 3 to 5 year olds receive special pre school and then school age children are in the special education category. Each state in the US administers the programs and the funds according to their own specifications (and has differing terminology).

The important thing to remember is that these services are your right. You, and more importantly, your child, are entitled to them.

Every state has an office for Children With Special Needs, usually managed day to day on a county wide basis. Your local school district is the agency in charge of delivering the services through its Office of Special Services (usually social workers, psychologists and therapists).

So get used to alphabet soup as each agency has its own pet name for itself and others. Learn the jargon. Declare yourself to your local school district and work through them.

This is especially important because CFC syndrome is not a "reportable" condition in the newborn nursery the way Down Syndrome or spina bifida are, because the hospital needs to report to the state as these children will definitely need services. You have to contact your state agency and apply. They won't come after you.

Don't forget that you and your child have a right to these services. Don't be afraid of labels that will secure for your child the services they need. The labels go away once the child ages out of the system. Get to know advocacy organizations in your area.

You will become an expert. And perhaps a mentor to the next person in line.

Vestal Backyard Friends Meet Up Again Along the Ras/MAPK Pathway

Brenda Shaffer and Nancy Haddad grew up in the 1960's as childhood friends who hung out in the swamps and hills behind Harpur College (currently known as Binghamton University). They explored the then largely family populated neighborhood with other nearby neighborhood girls. Back in the 60's there were no computers, IPod's, home movies or extensive competitive girl's sports programs at the schools. What's a girl to do other than set up playhouses, explore the woods and get into dress up clothes? These were the best of times! We all grew up in families where our mothers stayed home, baked great treats and the fathers went to work daily. IBM ANSCO, Singer Link and GE were the predominant employers in the area. The black and white television sets had only 3 channels and family night was Sunday night watching Disney.

As Brenda and Nancy and the rest of the neighborhood gang grew older they developed their own cluster of friends in middle and high school. College took the girls to different areas and teaching careers. Brenda and Nancy eventually took a road trip right after college to interview with school districts further south. Nancy landed an elementary teaching job in Virginia and Brenda worked a couple of months in Maryland before moving back to the Binghamton, NY area to open up the first resource room in the middle school teaching special education for the Binghamton City School District.

Nancy went on to marry Jim Kadlecik whom she met in college. They had their first baby, a healthy girl, born in 1982. Brenda was married later in 1985 to Cliff Conger and they had their first baby, also a girl, in 1989. Nancy and Brenda both had miscarriages between their first and second babies. Nancy then delivered Jena in 1987. Jena was born deaf and had hypertrophic cardiomyopathy along with a deformed pulmonary valve. She underwent surgery in 1988 for the valve. Jena was tested for Turner's syndrome and seen by a variety of doctors up in the Syracuse, NY area after the family moved from Virginia. Jena was not expected to live long. Noonan Syndrome was eventually brought up to the family prior to the first Noonan gene discovery.

In the years following the birth of Conger baby number two (Clifford), Brenda's life was busy with visits to many medical specialists at Children's Hospital of Philadelphia, keeping up with daughter Paige and work. There was no time for connecting with friends or former neighbors.

Fate eventually reconnected Brenda and Nancy when Nancy came down from Syracuse to visit her parents. One time after a lengthy discussion on Noonan and CFC, Brenda convinced Nancy to travel to Maryland with her to the Noonan Syndrome national conference. Jena's diagnosis still was not clear even to the medical specialists attending the conference. Life went on and Nancy eventually balanced life as a teacher and a married woman with three daughters.

Clifford, Brenda, Nancy and Jena

As Brenda continued to learn more and more about the genetic testing for CFC and Noonan Syndromes she gently encouraged Nancy to consider genetic testing while Jena remained on the family medical insurance. At this same time, older sister Deidre who had recently married started to think about family planning. The importance of uncovering Jena's genetic condition took on more of a focus for everyone. Over the summer of 2009 Brenda and Nancy phone conferenced about labs that tested for Noonan Syndrome and the specific genes that were now published for testing. At first Nancy's insurance carrier would not allow the testing since she had not been seen in genetics in so many years. Finally after Nancy worked through the insurance system, permission was granted and in October of 2009 Jena's blood was drawn and shipped to a lab for testing. In November Nancy and her family finally had answers after 22 long years. Jena tested positive for Noonan Syndrome. More amazing is how two close childhood friends who lived two houses away could end up with children born on the same MAPK Pathway. Now what are those strange odds?????

A Tale of Two Hands

My name is Kenneth Ankeny. I am the proud father of CFC Angel Miss Megan Ankeny. I have an ironic story to tell that dates back to about 1970. Keep in mind this was about 40 years ago so some of the language may not be politically correct.

I was born and raised in Albuquerque, New Mexico where I attended a small private school for boys from 5th grade to 12th grade. Everyone in the school knew each other because the classes were so small. I believe I was in 8th or 9th grade and about 14 years old at the time.

One of my classes was biology in which I had about 20 classmates. We were studying genetics at the time, and one day our professor had us come in, sit at our desk, and open our hands palms up. He advised everyone to observe the lines in their hands, and if anyone had a single crease running all the way across his palm they would be retarded. At this age, I had never heard any such nonsense, nor I had I ever paid any attention to my so called life lines, but when I looked down at my hands, I was astonished to see I had a single crease on my right hand.

Maria, Ken and Megan Ankeny

Well I knew I wasn't retarded and hoped my professor knew as well, but he was in shock that I had a single crease. As long as he had been teaching, he had never seen one other than in biology textbooks.

Needless to say word spread through the whole school quickly, and I was the butt of many jokes for the next several weeks. Summer break could not come soon enough that year!!!

For the next several years I didn't think much about my unique situation, however I would take notice of other peoples hands when I shook them or saw them open, just to see if anyone else I knew or met had that single crease.

The ironic part of the story comes 23 years later when Megan was born in 1993. Because of my wife's abnormal pregnancy and the fact that Megan would probably have severe medical problems at birth, we knew she would be in the Newborn Intensive Care Unit for some time. After 3 or 4 days of getting Megan stabilized, a genetic specialist visited our room to observe Megan and the first thing she did was open her hands, look at her crease lines come straight to me, open my hands, and look at my crease lines. She told us Megan had the exact same single crease on her right hand as I did and it was a common characteristic in people with mental disabilities or syndromes. Of course the first thought that came to my mind was that day in class 23 years ago.

I have since been told by this specialist that about 5% of the population has a single crease life line, however other than myself, Megan and an uncle who has a single crease on both hands (1% of population) I have never met anyone else with this uniqueness.

I would be curious to know how many of the other CFC kids have this characteristic and if any CFC parents share it with them. I hope other CFC families can relate or share a similar story to my experience. The Ankeny Family hopes everyone had a Happy Holiday Season, we look forward to seeing everyone at the next conference.

Ken Ankeny

Save the date!

8th Annual Toast the Angels Wine Tasting & Dinner

April 17, 2010 5:30 P.M
Kaluarah Shrine Center, Endicott, NY

$55 per person
Call 607-206-4375 for reservations

CFC Family & Board Member Participate in Genetics Seminar

Dr. John Lynn Jeffries, Pilar Magoulas and Jeremy Clark

On September 22, 2009, Pilar Magoulas, one of our board members and a genetic counselor at Texas Children's Hospital in Houston, Texas, Jeremy Clark (father of Avery Clark, CFC), and Dr. John Lynn Jeffries, a cardiologist at Texas Children's Hospital, gave a joint presentation entitled: "Understanding the variability in Noonan, Cardio-Facio-Cutaneous and Costello Syndromes."

This presentation was part of a monthly "Evenings with Genetics" seminar series that is sponsored by the Department of Medical and Human Genetics at Baylor College of Medicine and Texas Children's Hospital. This seminar series offers the most current information on care and research for many genetic conditions and is open to the public. The goal of these seminars is to highlight the advances in genetic research as well as encourage networking within our community.

Jeremy, Brandy, and Avery Clark, Pilar Magoulas, Theresa Aguilar, & Dr. John Lynn Jeffries

Pilar Magoulas gave a brief description of the clinical features, the genetic basis, and similarities and differences between the three syndromes, while Dr. Jeffries focused on the cardiac manifestations and management in children and adults with these conditions. Jeremy Clark closed the presentation by giving a wonderful portrayal of the journey that he and his family have encountered, beginning with the uncertainty and waiting period before a diagnosis was made to the acceptance of the diagnosis and appreciation of support that they have received from other CFC families.

This seminar was very well attended by parents, family members, and caregivers of children with either confirmed or suspected CFC, Noonan, and Costello syndromes, as well as health care providers from the Houston area.

CFC International Joins NDRI Rare Disease Partnership

A larger collection of biomaterials, and their related medical records, is essential for research into CFC Syndrome. Our new partnership with the National Disease Research Interchange (NDRI) will aid research to better understand and treat CFC Syndrome. NDRI (www.ndriresource.org) is a non-profit organization with nearly 30 years experience in obtaining, banking and distributing human cells, tissues and organs to researchers. NDRI receives funding for their Rare Disease program from the National Institutes of Health and support from the NIH Office of Rare Diseases Research.

Through the partnership between NDRI and CFC International, you have the opportunity to become part of the NDRI Private Donor Registry and consent to donate tissues for research. Donating tissues has been designed to be as simple and sensitive to you and your families' needs as possible. You will receive a packet of information describing the donation process, including consent and medical history forms. A Rare Disease Private Donor Coordinator is available to answer any questions that you may have. There is no cost to you for donating tissue.

NDRI
National Disease Research Interchange
The National Resource Center

NDRI follows strict governmental regulations and guidelines regarding donor consent and confidentiality; tissue samples are only provided to approved biomedical researchers. NDRI has a database of researchers who are seeking human tissues, matches donors with appropriate requests, then sends tissues directly to researchers. Your personal details remain strictly confidential and no donor information is given to the scientists.

To receive more information on tissue donation and how to join the the NDRI Private Donor Registry, please contact a Rare Disease Coordinator at raredisease@ndriresource.org, or by phone at 800-222-6374.

For further information about NDRI, visit www.ndriresource.org.

Dolphin Therapy to Genetics Convention and beyond….continued.

We were able to do this through a grant that one of the staff members, Lisa Maetani, from the Dept. of Health had heard of our small group and came to observe. It was she who wrote the grant to get the funding. I don't believe I knew want a grant was back then and certainly not how to write for one, but looking back this was my first step into becoming part of a bigger circle and realizing that with a little effort you can bring people together and accomplish your dream.

So after attending my second CFC International Family Conference in Berkeley, California this past summer I was asked by our CFC President Brenda Conger if I would be willing to attend the ASHG here in Honolulu. I of course was more than willing to represent our group but first had no idea what ASHG was. It's the American Society of Human Genetics and they have been around since the 1947. WOW!! The scientists, geneticists, clinicians, and industry all converge once a year in some city in the world to discuss everything to do with genetics and the human genome. We'll talk about how way over my head it was and that I agreed to go where this mom had not gone before. Once I sent in the application I received a catalog of events that would take place at the conference that was an inch thick. I had been to numerous fashion apparel trade shows but this was quite different. It was me walking into another world that Amara once again led me to. I was told my only requirement was to write an article after the conference to share my experience. So here I am trying to do that. I would have the privilege of being joined with another mom, Lisa Schoyer, who is President of the Costello Syndrome Group in California.

On October 20th I, yes me, Flora Tobby, attended my first Scientific Conference known as ASHG, or the American Society of Human Genetics. A mouth full that at first was hard to swallow or believe that you would find me in the company of brilliant scientists, geneticists, physicians etc. After all, I'm just a MOM!

The first meeting was with the Genetic Alliance who is responsible for the BioBank that houses all the DNA of our children. CFC International has worked hard and encouraged all of us to share our medical information of our children so that research is possible. Hence the discovery of the CFC genes and a blood test that would bring the gift of early diagnosis so parents would no longer be in the dark like I was for 14 years. They were responsible for providing funding so a parent advocate for CFC International could attend. There Lisa Schoyer and I met another mom, Geraldine Bliss from a group called Phelan McDermound Syndrome who was attending from Texas in hopes of being able to find doctors to create a research board for their family support organization. She was so impressed with the CFC and Costello syndrome support groups since we have made such great progress.

Our first session we attended was titled The Rasopathies:Germline Dysregulation of Ras/MAPK Signal Transduction, how's that for another mind full? Of course Dr. Rauen was a speaker and shared that in their research with embryos of zebra fish, a small window of opportunity exists where they have been able to correct the outcome of CFC. Now keep in mind this is the tiny embryo of a zebra fish however it's a huge accomplishment since it represents great potential for the future of individuals with CFC Syndrome. This was one of many, many amazing things that were shared at ASHG. We were told in one of many meetings I participated in over the course of four intense days that a "tsunami of information" is coming our way. We are on the doorstep of the future where things men have never dreamed possible will be possible. There are many things for all to consider as these events will unfold. I left this experience so inspired and enlightened about what the world of genetics has to offer humanity. I actually had the opportunity to sit with the Dr. and his wife of UC Santa Cruz who is the one responsible for mapping the human genome onto the internet. AMAZING!

I wish to encourage all of you who have been on this journey and those who have just began how vitally important it is for all of us to send in and keep sending in the medical information to CFC International since we have the most wonderful group of doctors, geneticists and researchers interested, willing and able to help move us forward into clinical trials. The question now is what do we really want for our CFC children? Think about it. I for myself am so grateful to Brenda Conger and Dr. Kate Rauen who have laid the foundation to bring me, us, from isolation into infinite possibility. If not for my daughter Amara then for yours, I am now willing to bring to the table the intention to assist them in raising funds for CFC International. We plan to organize our first fundraiser here in Honolulu, HI this year so that one day CFC will become a syndrome of our past. May we all this year find the clarity on how we want to move forward for our children, ourselves and humanity. I leave you with this…"the future looks so bright, it burns my eyes."

Happy New Year to you and yours!!! Aloha,

Flora, Ted, and Amara (my angel) Tobby.

Photo Gallery

Twins Luke & Brett from Illinois waiting for their clinic appointments

Clifford Conger entertaining the crowd at the Lion's Club Christmas

Kolosia and Vaasi from California

Nola Rose Iacobelli all ready for Christmas

Jared Stowell, Utah & Brett Hanson, Ilinois

Megan Ankeny all decked out in holiday red

Siblings of CFC & Noonan children have their special time at the pool

Dental team from University of California, San Francisco

CFC International board meeting with NORD facilitator Jean Campbell

Jaime Phillips from Idaho, handing out water to the medical clinic team

Dr. McLaughlin-Beltz reviews Dr. Guntram Krzok's son's photos

Jean Campbell from National Organization for Rare Disorders and Rosemarie Pavilonis, Illinois man the registration table

Dr. Alicia Romano from Children's Physicians of Westchester, NY presenting a seminar on endocrinology

Brenda Conger & Molly Santa Cruz getting signs ready for the conference

Dr Eric Johnson makes a point during his Genetic Testing workshop

Cliff Conger & Dr. Noonan

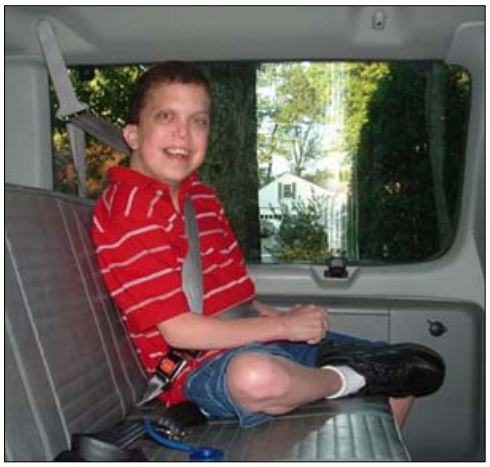
Dr. Allanson from Children's Hospital of Eastern Ontario chats with a parent

Featured Family: Stephen Patsos

Our son, Stephen, was born on January 14, 1991. He arrived 2 1/2 months early. I was diagnosed with polyhydramnios, too much amniotic fluid, which probably added to his prematurity.

Stephen remained in the hospital for 2 months. As we look back, the only issue brought to our attention were his low set ears. They performed tests and found his chromosomes to be normal. They sent us on our way saying that he should be fine.

Two months at home and Stephen was failing to thrive. He had difficulty with sucking and swallowing, not being able to breathe through his nose during either action. Upper airway noises were not normal. He was then admitted to Mass. General Hospital with failure to thrive and upper airway narrowing. Seeing that he could not feed and breathe through his nose, an ENT surgeon decided that he simply would not be able to eat without a tracheotomy. This would be done and kept in until his nasal passages were able to enlarge with time. The tracheotomy was done at five months of age. Literally, the next day, he simply refused anything in his mouth. He then required a gastrostomy tube, which he has had ever since. After 2 more months in the hospital, he finally came home.



Stephen Patsos

Stephen began to grow well. His health was good, but he did not appear to be reaching his milestones. He did not look like other children. In the old days, he would be categorized as the FLK Syndrome, that last chapter in the books.....Funny Looking Kid Syndrome, which simply, in retrospect, must have been many children without a diagnosis.

His head was larger than normal, his eyes were proptotic, and his ears were set back and quite low. He was sent to a geneticist around 18 months of age and also had his trach removed. His upper airway had grown enough, he was breathing well, but his Gtube remains to this day.

We spent many years visiting the genetics floor at Children's Hospital in Boston. Stephen was either undiagnosed or misdiagnosed for about 14 years. One of his diagnoses was mitochondrial disease due to a muscle biopsy. We never felt comfortable with that because he never shared any of the characteristics with the other children similarly diagnosed. As it turns out, a few years later we were told the muscle biopsies were coming back as false positive because of the frozen fixation prior to being tested.

The Patsos Family

We were now back to square one with no diagnosis. Our geneticist moved and we were given a new one named Dr. Gerald Cox. He seemed to know what Stephen had the moment he walked through the door. It seemed like a miracle with CFC being so rare.

We went to the CFC conference in Maryland in 2003. Doctors Neri and Opitz examined Stephen and felt, because of his lack of skin abnormalities, that he did not have CFC. However, he looked exactly like many of the children at the conference. Also, the next morning was the last slide show we saw, which stated that 20% of them have normal skin!

We went back to Boston without a diagnosis, but our doctor there stood firm on his diagnosis. We were left somewhat confused, but without a specific treatment anyway, we went on.

Our next visit with Dr. Cox brought hope because they had developed a blood test to diagnose CFC. Stephen was tested and it was confirmed that he had CFC Syndrome. We were excited, relieved, and yet fearful at the same time.

Stephen has remained relatively healthy. His cardiac system has very mild abnormalities and is stable, never needing any cardiac medication. Oral feeding has continued to improve, albeit slowly. He now can eat 3 meals a day of pureed food and receives 4 cans of Pediasure a day via his Gtube.

Stephen lives at home with his mom, dad, and 2 brothers. I am a registered nurse and his dad is a general surgeon in the community. This has certainly helped us through our journey. Rick, his older brother, is 21 and Johnny is 16. Stephen loves them both and has fun with them, in his own way.

Stephen turned 19 on January 14, 2010. He has blessed our lives in more ways than we could ever imagine. We used to quietly, in our minds, plan his funeral. Today, we cherish every day we have with him as well as our other 2 sons. We have realized what a true gift from God he really is and how lucky, not unlucky, we are to have him in our lives. He was given to us, like any other child, no matter how long he lives, for one reason: to love him.

Skin Findings in CFC

By: Dawn Siegel, MD Oregon Health & Science University, Portland, Oregon

The skin has always provided useful clues in helping to make the diagnosis of CFC. Now with gene testing available, we are beginning to link the changes in the skin with the changes in the gene. For the past couple of years, Dr. Siegel, Dr. Rauen and their colleagues have been working to characterize the skin problems in CFC and have come up with some interesting findings. Over sixty families filled out our surveys and all of the information and comments were very helpful. Thank you to everyone who participated!

One of the most striking skin findings was the large number of moles (nevi) that can develop during the teenage years. Moles are a type of skin lesion made up of the pigment cells called melanocytes. Many of the individuals in the study had over 50 moles, and a handful of the individuals had over 100 moles. These moles begin to appear in early childhood even in the general population, but in CFC syndrome they seem to increase to larger numbers than the general population during puberty and the teenage years. Despite the large number of moles, there have not been any reports of skin cancers or melanoma.

Rough, bumpy skin on the face, arms and legs (keratosis pilaris) was present in a majority of the individuals in the study. This condition (keratosis pilaris) can also occur in the general population, but usually only in about 1/3 of people. Some of the parents reported that it improved with age. Sparse or absent eyebrow hair, with redness of the skin under the eyebrow was reported in most of the individuals. This is uncommon in the general population and is one of the characteristic features in CFC.

Strawberry birthmarks (infantile hemangiomas) occur in about 5% of newborns in the general population. In this study, the number of children with CFC who had a hemangioma was higher than in the general population. This type of birthmark is made up of small blood vessels. Hemangiomas are usually very faint and flat at birth then grow over the first couple months of life. These birthmarks then stop growing and gradually fade away over several years. Sometimes the skin looks pink or stretched after the birthmark has resolved.

Dr. Dawn Siegel

This study was also helpful because we were able to identify some skin findings that are not common in CFC, but are present in other similar conditions. For example, there were no individuals with over five café au lait spots, helping to distinguish CFC from neurofibromatosis.

We also learned that the CFC families are doing a fantastic job with sun protection. Most of the families reported using sunscreen and hats, although a majority reported that their child does not like to be out in the sun and does not tolerate the heat. In addition, the development of body odor in early childhood occurred frequently. Fast growing fingernails were reported in a majority of the surveys and some parents reported cutting their child's nails a couple times per week.

Thank you so much to everyone who participated in the study- you are all helping us learn more about CFC and how to take care of the skin issues that arise. We are grateful to CFC International, Molly Santa Cruz and Brenda Conger for helping us collect all of the surveys!

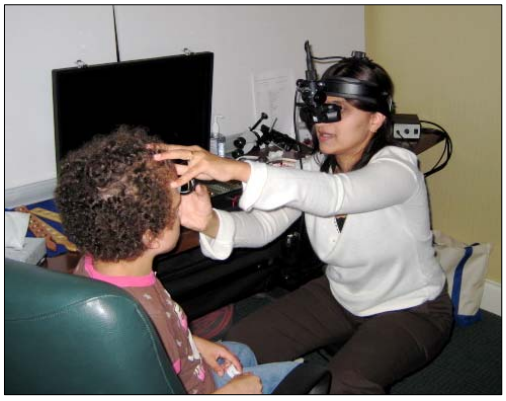
The Berkeley Conference was an Eye Opener

The logo for CFC has a child wearing spectacles– this shows how CFC kids commonly have eye involvement!! Our vision team consisted of Drs. Suma Shankar and Dr. Kate Rauen from University of California San Francisco, Dr. Deborah Orel-Bixler and her team of optometrists from

University of California Berkeley and Dr. Terri Young from Duke University. We also want to thank all the parent organization team leaders and volunteers who helped in this successful undertaking. We had the unique privilege to meet with many CFC families along with Noonan and Costello syndrome families to perform ocular examinations during the bi-annual symposium in Berkeley.

Clifford Conger gets his eyes tested

The importance of the role of the Ras/MAPK cascade in the development of the eye and visual pathway (includes the optic nerve and tracts responsible for transmitting the nerve impulses from the eye to the visual cortex of the brain) has been reported. Given that genetic alterations in the genes causing CFC syndrome cause disruption of Ras/MAPK signaling, we sought to evaluate the extent of eye development and visual issues in individuals with the Ras/MAPK genetic syndromes.



Dr. Shankar performing an eye examination

We began by obtaining previous medical records of eye examinations from members of CFC International. We received medical records from many CFC families---**And we thank all those that participated!** We performed eye examinations in 58 individuals including 28 with CFC, 16 with Costello syndrome and 14 with Noonan syndrome during the conference using UC Berkeley's state-of the-art ophthalmic equipment.

Not surprisingly, visual problems were reported in almost all individuals with all three syndromes. The most common reported eye findings included strabismus– exotropia (an eye or eyes turning outward), esotropia (inward crossing of the eyes); refractive errors including myopia (near-sightedness), hyperopia (far-sightedness) and astigmatism. Other issues included nystagmus (rhythmic shaking of the eyes); ptosis (droopy eye-lids) and optic nerve anomalies. Outcomes from these structural issues were problems with depth perception, abnormal head posturing and amblyopia (diminished visual acuity due to unequal eye use). Functional studies showed that most individuals had reasonably good vision in one or both eyes, but lacked depth perception.

Dr. Suma Shankar & Dr. Terri Young

In summary, although the majority of CFC participants have ocular issues, most are manageable similar to those in the general population. If the ptosis is significant enough to cover the pupil causing visual deprivation , early surgical correction is recommended to prevent amblyopia. Similarly, early correction of myopia or hyperopia with glasses projects a clear image to the eye (and brain) to help the child in achieving their full visual potential. We also recommend surgical correction of strabismus when indicated. Some individuals may have poor vision that is due to optic nerve problems and sometimes brain structure issues can cause visual problems. Appropriate management in these individuals necessitates consultation with neurologists and neurosurgeons. Based on the eye findings that can be seen in CFC syndrome, it is very important that early eye examinations be performed (scheduling a first eye examination by 3-6 months of age) if a child has been diagnosed at this early age. Then, it is strongly recommended that routine eye examinations be performed every 6-12 months during childhood. After childhood, annual exams should be performed, or sooner if any issues arise in the interim.

We again want to thank all the families that participated ---- you helped make this event a huge success.

Advancing Rare Disease Research: The Intersection of Patient Registries, Biospecimen Repositories, and Clinical Data

CFC International board members Brenda Conger and Luba Djurdjinovic attended the workshop in Bethesda, Maryland on January 11-12th. Wanda Robinson from The Noonan Syndrome Support Group and Lisa Schoyer representing the Costello Syndrome Family Network were also in attendance.

The objective of the 150 member attendees was to discuss the development of an infrastructure for an internet-based platform with common data elements utilizing a federated rare disease registry able to incorporate existing rare disease registries; patient organizations with no registry looking to establish one; and patients with no affiliation with a support group looking to belong to a registry. The expected outcome of the workshop was to gain acceptance of the concept of a federated rare disease patient registry and participation in creating this patient registry from as many curators of patient registries and other stakeholders as possible. Participating stakeholders attended to standardize common data elements, vocabulary, and open source software to enable the exchange of data and information to facilitate research collaborations.

Wanda, Luba, Brenda & Lisa

Housing and Transitional Service Questions from our Families

Answered in bold by Rob Davies, retired Director, Office of Housing Initiatives, New York State Office of Mental Retardation and Developmental Disabilities

Q. Although our CFC son won't need residential services for many years yet, I am trying to better understand the process. Both my husband and I are older parents and we have no extended family in state. I don't want our son to end up in a place we would not have chosen for him, if something were to happen to us. And I wonder if I could be using my energy now (volunteering or serving on a board) to develop a relationship with an agency or community that I do like in order to learn more or help our son transition eventually.

A. I would definitely say yes to this question. You need to get to know the services agencies in your community, learn about their philosophy, management style, staff turnover rates and fiscal record. Look at copies of their state inspection reports. Getting on committees and boards will help you in positioning yourself and your son for services earlier in his life. You will also be involved in decision making to move the agency in directions you feel are important.

Q. Right now our son is on our state's Katie Beckett waiver and we get some in home nursing support and Medicaid. He also has a special needs trust which appoints my (older) sister as his guardian-- but she lives out of state. If both my husband and I were to die and my sister had custody of our son, she could not take him out of state to live because he'd lose his waiver slot, right? And the waiting list is too long everywhere...and she could not manage to take care of him without the waiver support. In such a case what are the possible options? Would our son lose his Medicaid waiver slot on our death anyway (if he were under age)?

A. Having the special needs trust set up is good. You need back-up trustees if your sister dies. If you have no other family, look for nonprofit administrators of Special Needs Trusts in your state, at your bank or at a lawyer's office. You can have co-

trustees also so decision making is shared. For example, a bank trust officer and a nonprofit case management agency can act as co-trustees.

There is something called the interstate compact which allows transfer of persons with disabilities to other states. You should call your state developmental disabilities services agency and talk to the person in charge of interstate transfers for more details on how it works in your home state.

Your child will not lose his Medicaid funding upon your death as long as he needs the services. He may lose services due to state and federal budget cuts, but not because of parental death. It is more likely that he would more easily receive more services upon your death because he is already known by the system.

Q. Likewise, if we (his parents) are too old or deceased to be his appointed guardians sometime after he turns 18, and he had an appointed guardian out of state, would he have to receive services in the state his guardian lives in?

A. His guardian can live anywhere and he can stay where he is living.

Q. Another question has to do with how residential placement is chosen for an adult through the Developmental Disabilities Administration (assuming he has a DDA slot by then). At that point is he eligible for a group home or residential community in ONLY the area that we live, or the state we live? If we preferred a group home in another state or neighborhood, could we get him there by moving? Would we have to move temporarily or permanently?

A. As you know there are long waiting lists in all states and some states have better or more available services. Blue states vs. red states...more taxes = more services. In New York State people with highest needs are supposed to be placed first. However, squeaky wheels, politically powerful or knowing someone powerful or being an actively involved parent with a local agency all help move one higher up on the waiting list. You need to check with DDA on their policy on placement by region. Moving to another state and doing all the things I discussed will also work, but you must know the system and agencies in the area you live.

The Camp Hill organization is one I would recommend if you like what they have to offer. See http://www.camphill.org/ for more information.

Q. Rob, who would be best to talk to locally about these questions...what kind of person? (ie, lawyer, social worker, somebody at ARC?) Who is expert in this kind of knowledge?

A. I would try to find someone that has been in the system for a number of years and is a speaker at local/statewide conferences. Regional state staff, state ARC staff, or a case manager/social worker who is older, more experienced and still excited about their job. Creativity and a commitment to individualized services will help.

We could always have a conference call together with this type of person if you want.

Fair winds,
Rob

CFC International would like to thank Rob Davies for his answers to questions from our families. If you have further questions on this topic please feel free to email Rob Davies at rdmandate@yahoo.com .

"You will find as you look back upon your life that the moments when you have truly lived are the moments when you have done things in the spirit of love"

Henry Drummond

'He was trying to find his voice'

Ian Brown wins Charles Taylor Prize for memoir about his disabled son

Melissa Leong, National Post, Canada
Published: Tuesday, February 09, 2010

Ian Brown would not speculate whether his 13-year-old disabled son, Walker, would appreciate him winning the Charles Taylor Prize for Literary Non-Fiction.

But at the awards ceremony yesterday in Toronto, he told the audience that if Walker were there, he would be playing with jewellery, knocking over statues and speakers and he would be happy.

"So for the possibility that he might be grateful, I am, to you, most grateful," Brown told the 200 guests, including Giller Prize founder Jack Rabinovitch, Senator Linda Frum and former Governor General Adrienne Clarkson.

Brown, a broadcaster and writer for The Globe and Mail, won the $25,000 prize for his book, The Boy in the Moon: A Father's Search For His Disabled Son, published by Random House Canada. It's a memoir about his life with Walker, who is afflicted with the extremely rare cardiofaciocutaneous syndrome (CFC), a genetic mutation that affects about 150 people worldwide.

"For many years, I watched him go about the house, stand in a corner, playing with something or looking behind a blind or picking something up with his hands. I never knew what he was doing. Finally, it began to occur to me that what he was doing was trying to do was find his voice," Brown said.

"I wanted to find his voice."

Walker cannot speak or eat solid foods. He is fed through a tube in his stomach and wears a helmet to prevent him from hurting himself.

"The struggle was to accept him not as I wanted him to be but as he is, as an individual who has a contribution to make, no matter how slim that contribution might be and how resistant I am, because it means redefining what a successful life is," Brown said in an interview after the gala luncheon.

"It's really the struggle of the individual against generalities, and that's something that everyone is connected to."

The Boy in the Moon has also won British Columbia's National Award for Canadian Non-Fiction, which comes with a $40,000 prize.

The three-member jury for the 2010 Charles Taylor Prize--author Andrew Cohen, translator Sheila Fischman and historian Tim Cook, who won last year's prize for his book Shock Troops: Canadians Fighting the Great War, 1917-1918, Volume Two-- selected four nominees from among 125 books submitted by 34 publishers.

"We had books about a daughter travelling ... with her incontinent mother and a daughter travelling with her inebriated father," Cohen said. "We had books on saving Vancouver, saving Sudan and saving the delicatessen."

This year's three other nominees were University of Waterloo history professor John English's Just Watch Me: The Life of Pierre Elliott Trudeau, 1968-2000 (Knopf Canada); Montreal author Daniel Poliquin's Rene Levesque (Penguin Canada), a portrait of the founder of the Parti Quebecois; and Maclean's publisher and editor-in-chief Kenneth Whyte's The Uncrowned King: The Sensational Rise of William Randolph Hearst (Random House Canada), which revisits the life of the famed media baron. They each received a $2,000 prize.

"Narrative non-fiction is being ignored these days in favour of faster, more frequent, shorter blurts," Brown said. "I think narrative nonfiction will come back. This prize keeps it there."

The Charles Taylor Prize for Literary Non-Fiction was named after the late journalist and author Charles Taylor. It was created in 1998 and has been awarded to prestigious writers such as Carol Shields, Rudy Wiebe and Richard Gwyn.

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Would you like to contribute to CFC International?

Your donation and support will help:

- families to receive information about CFC syndrome;
- publish our newsletter;
- fund the biannual International CFC Family Conference & Clinic Program;
- maintain the CFC Tissue and Biobank, which is critical to future research.

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