An International Group for Support and Information for ALL Chromosome 22 Disorders

CONNECTIONS

Issue 51 March 2012

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March is Trisomy Awareness Month and so it is a great time to also celebrate

Cat Eye Syndrome Awareness Day, March 22, 2012!

Visit https://www.facebook.com/CatEyeSyndromeAwarenessDay

Our cover girl is Miss Miranda, a sweet little mischief-maker who happened to have CES, and who we unfortunately lost in 2004. CES Awareness Day seemed appropriate to remember her. Her mom has the best photographs of her...and I'm grateful to Miranda for bringing her mom Laurie into my life :)

Thank you to all the families in the CES Facebook group who shared their stories and photos for this issue. I hope it will help bring awareness to this condition.

-Stephanie

Celebrating Our Beautiful Kids!

JORDAN

My daughter Jordan was born with CES, July 13th, 1993. She was a full turn (late actually) pregnancy and normal delivery. She was however held in NICU overnight because she had "swallowed amniotic fluid" and was being observed. At her first check-up (1 week) I asked the Dr. why her pupils were shaped the way they were and she was amazed. She hadn't notice and she was also the Dr. who checked her out upon delivery. This led to the LONG road of Dr.'s, specialists and so on and so forth. Bouncing around from place to place to put names and diagnosis on my sweet baby girl. Thankfully, she was reported to be in great health! She has bilateral colobomas in both eyes, with the right eye extending all the way to the optic nerve, but see's well enough. She was born with preauricular pits on both sides above her ears, and palpebral fissures or slightly slanted eye folds.

She grew and developed normally and at age 8 had to have tonsils and adenoids removed and tubes in her ears because of multiple infections. After the surgery she didn't seem to recover her voice and had severe nasal aspirations upon speaking. Which led us to speech therapy with a wonderful teacher that sent us to the RIGHT Cranial Facial Dr. at Scottish Rite in Atlanta, GA. He told us the problem and solution and turns out...another characteristic of CES that was uncovered because of a routine T&A. After all of this, now we understood why our baby girl would choke as an infant when she spit up. It would always go out her nose and we had to suction her daily! If only the answers were easier to find!!

Fast forward to today...our beautiful Jordan is almost 19! She is a college freshman at USC and aside from her academic struggles, she is pushing on. She will be having a surgical procedure done on Tuesday to remove one of the preauricular pits that has become active in the last year and is causing infection and terrible drainage. Fingers crossed that it is successful!

Aside from the occasional teasing during school and the reactions of fascination from strangers...my Jordan is a sweet, loving, responsible, beautiful young lady and I admire her so much for being the bravest person I know! I am so happy that we have this awareness and only wish it had been available all those years ago.

Thanks for letting me share my story and my daughter with you!

-Stacy

GEORGE

Our son, George, was born on November 29th, 2007. He was immediately lifeflighted to Nationwide Children's Hospital in Columbus, Ohio for several problems. He needed to have a colostomy and he also needed open-heart surgery. His colostomy surgery was performed when he was 1 day old, his open-heart surgery was performed when he was 7 weeks old. He had his colostomy reversed at 6 and 10 months of age in 2 surgeries.

He has now had a total of 7 surgeries, but is a fire-ball! Has lots of energy and loves to play outside and take four-wheeler rides. His sister is 50 weeks older than he is, so they are almost like twins.

-Cathy C.

Ryan, age 9, CES

Celebrating Our Beautiful Kids!

AYDIN

Our story started when I was 17 weeks pregnant. We had our 1st ultrasound and found out we were having a little boy. At this same visit the doctors came in and told us that I had a one artery umbilical cord and said with my age that our baby would have a syndrome called Edwards syndrome. They told us the baby would not survive. They asked if we would like to consider abortion. We told them no and that if God gave us this baby for 2 hrs 2 days or for 2 mins, there's a reason. We continued with the pregnancy and at 25 weeks I went in pre term labor. They said I was measuring 42 weeks and you could tell it. Sad thing is I only had barely a one pound baby in that big stomach so they drained amniotic fluid and sent it for testing at that time. I laid in the hospital for weeks. Then was sent home on strict bed rest I was able to keep the baby in till 35 weeks. At that point I went back in to labor and doctors said they need to deliver by c-section. I was measuring 42 weeks again and it was too dangerous to drain it this time so it was time to take baby. When he was born they sent his blood to genetics at Ft Lewis, who told us he was 99% sure he had cat eye syndrome. We were like shocked. We had never heard of it. We named our little 4 pound 17 in baby boy Aydin.

He was born with an imperforated anus. He also had TAPVR heart condition. He had an ostomy placed at 3 days. At 2 months his heart was repaired. He had the anus reconstructed at 6 months, the ostomy taken down at 9 months. He has now had a total of 9 intestinal surgeries; 2 liver surgeries, 1 cyst from the side of his head, a g tube placed, and a port placed. In total, 15 surgeries. He does have bilateral colobomas. With this he only had one small clip of vision in his lower left eye. He also wears hearing aids on both ears. We were told to teach him sign language for he would not talk. He learned the signing but he now has 300 words and says small sentences like 3 and 4 words. They said he might not walk but at 2 he began and now I am chasing him to keep up. He has had a development specialist test him and cognitively nothing is wrong. He is very smart. I am so glad that my Husband and I have a strong faith in God for we would not have gotten through this and there is still a lot to come. Aydin is suffering with the shut down of his GI tract every other week right now and is possibly going back to an ostomy. When this happens he goes in hospital to get TPN. He is not allowed to have any food or g tube feeds in his gut. Aydin did not have much that grew to begin with so they think that's his problem but even at this he is still going and learning new things and will start school on his 3rd birthday (which is March 13, 2012). We were so blessed to have a doctor that had seen this syndrome was able to test and confirm his suspicion and knew of the support group for this. It has been a true blessing to have an extended family thru the group. When we first got on the site we asked if any one was in the Tacoma, WA area and was able to meet little Joey W. This was when Aydin still had lots of surgeries and uncertainty; still a lot to overcome. Meeting Joey gave us hope to what the doctors had first said. Our little boy won't make it 2 days and look at our miracle now. He has overcome so much and is still going. He is one of the sweetest kids I know; so easy going. Aydin is one of five kids and our other kids have learned so much from him he has been such a blessing to our family and through him a lot of people have seen the power of God working and given others the hope of miracles, and allow people to realize God is still alive and working miracles today.

Never take for granted what you have and always take everyday as a blessing. I hope Aydin's story can help or bless some one

- Regina

Rylan, age 2 1/2, CES

Celebrating Our Beautiful Kids!

BRIANNA

We knew that my husband had Cat eye syndrome. He was only diagnosed in his late 20s. He has not been affected much. He had an operation for strabismus of the eyes and has dyslexia and dyspraxia. We went for genetic testing and knew what to expect or so we thought. Whilst pregnant we had blood tests and nuchal scan and found that there was a 1:8 chance of having a child with a chromosomal disorder (although the medical profession kept telling us Down syndrome). We also had a foetal heart scan done at 18 weeks and were told that the baby heart looks fine and that there would be only a 2% chance of anything being wrong with the heart. I also had a major bleed and was told that I was miscarrying. However our little fighter held in there and we refused all advice of further tests and abortion.

On 12/12/2006 our beautiful daughter was born naturally. The first thing that was noticed was that she had no anus. She went into neonatal unit and we were told to have some rest. It was night. The next morning we went into see her and was told she was doing fine and due to go to another hospital to look into her anus. The hospital team seemed reluctant to have us there and persuaded us to go for breakfast. After breakfast they wouldn't let us in. I knew in the gut of my tummy there was something very wrong and asked her to be baptised. After a very long time we were told there was something wrong with her heart and that her sats was very low. We knew straight away TAPVD and told then what we thought. They told us it didn't look good and that she had to be rushed to another hospital and might not even make it. Well she did and she is now a healthy and bouncy 5 year old. She had TAPVD and had open heart surgery at 1 week old, imperforate anus for which she had a colostomy bag at 2 weeks old, anus reconstructed at 5 months and colostomy reversed at 9 months old. She has hyper mobility, Duane syndrome and gastric reflux. She also has little pits in front of both ears. After she had heart surgery her wound got infected and into the blood stream which meant a further stay in hospital. All in all her first month was spent at a beautiful London hospital and we cannot thank the surgeons, doctors and nurses enough. The first two years was very hard with feedings, kidney infections and pneumonia as well as severe reflux. Today she suffers from constipation and sore tummy, legs and back. But her height and weight is normal and she has reached all milestones and doing very well in school. She is very confident and has a robust character (certainly very assertive) which proves that these kids grow to be very strong. We named her Brianna too because of its meaning strong and noble.

When we got home I searched for information and there didn't seem very much. I then got contacts on good old face book and set up cat eye syndrome group. From there I have met many lovely people and we share a wealth of information. I am more aware of all the risks of having a child with cat eye syndrome but will have Brianna all over again as she is such a blessing and has taught us lots.

-Sinead

TRACE

Trace is a happy, overall healthy 4 year old. He will be 5 in June. Trace was just recently diagnosed with Autism & ADHD, which is very normal for children with CES. He is very intelligent and can tell you the name of a dinosaur for every letter in the Alphabet. He can tell you what time period they are from and whether they are carnivore or herbivore. He is very active and very loving and I am honored that he chose me out of millions of other women in this world to be his mother. We have good days and bad days, but the good days are the best. I think Trace has a very bright future ahead of him, and I am sure he will grow up to be an architect an anthropologist (Well that's what he tells me anyways!) - Heather

Celebrating Our Beautiful Kids!

Famke, age 10, CES

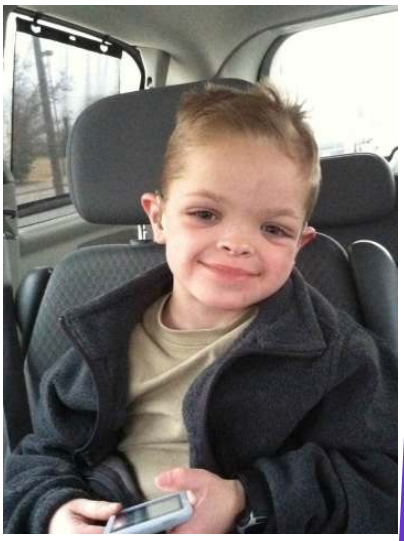
Sila, CES

Corey, 7, CES

DAIN

Dain will turn 7 March 29th! CES month! He was recently diagnosed with ADHD combo type. He started meds on a Sunday and has had good days at school ever since!

Also about once a month Dain gets to "play guitar and sing" with a local band. They are great with him and the crowd loves him! We had to not go for about 4 months due to his behavior. In this photo was his first time back. They introduced him as we would like to introduce Dain, he has been off writing a new album!



-Rochell

Celebrating Our Beautiful Kids with CES!

JOEY

Joey is four-years-old and is a preschooler at Blix Elementary School in Tacoma, WA (USA). He loves to do puzzles, play games, read, sing and hopes to be a baseball player some day.

When Joey was born he was quickly transported to Tacoma General Hospital's Neonatal Intensive Care Unit (NICU) and underwent surgery the next day to repair his imperforate anus. While he was recovering it seemed like he was checked out by almost every specialty in the hospital and was diagnosed with bilateral colaboma, ear tags and ear pits, moderate hearing loss on one side, Total Anomalous Pulmonary Venous Return (TAPVR), a tethered spinal cord, vesicoureteral reflux and one kidney that was multicystic dysplastic and had no function. All of these features and symptoms lead the doctors to believe he had Cat Eye Syndrome, a diagnosis that was confirmed by genetic testing a few weeks later. Joey had his second surgery on this three-week birthday to place a g-tube in his stomach because he was not allowed to eat by mouth. When he was four weeks old he was able to go home with a strict regimen of medication for his heart and round-the-clock feeds.

Joey still needed open heart surgery to save his life, and had frequent visits to his cardiologist to monitor his congestive heart failure. There is risk of organ failure with open heart surgery, and with his existing kidney problems the likelihood of him needing dialysis after his surgery was high. Together, his cardiologist and nephrologist determined it best that he go to Seattle Children's Hospital for the surgery. The date was set and when he was 2-1/2 months old he was once again on the operating table, this time for approximately six hours. During the operation a tube was placed to prep him for dialysis if it would become necessary. As with his previous surgeries, Joey did extremely well and within a few days was moved from the Cardiac ICU. His kidneys did not fail and he did not need dialysis. His first smiles came a few days later, and after a week he was sent home with his incredibly relieved and thankful parents.

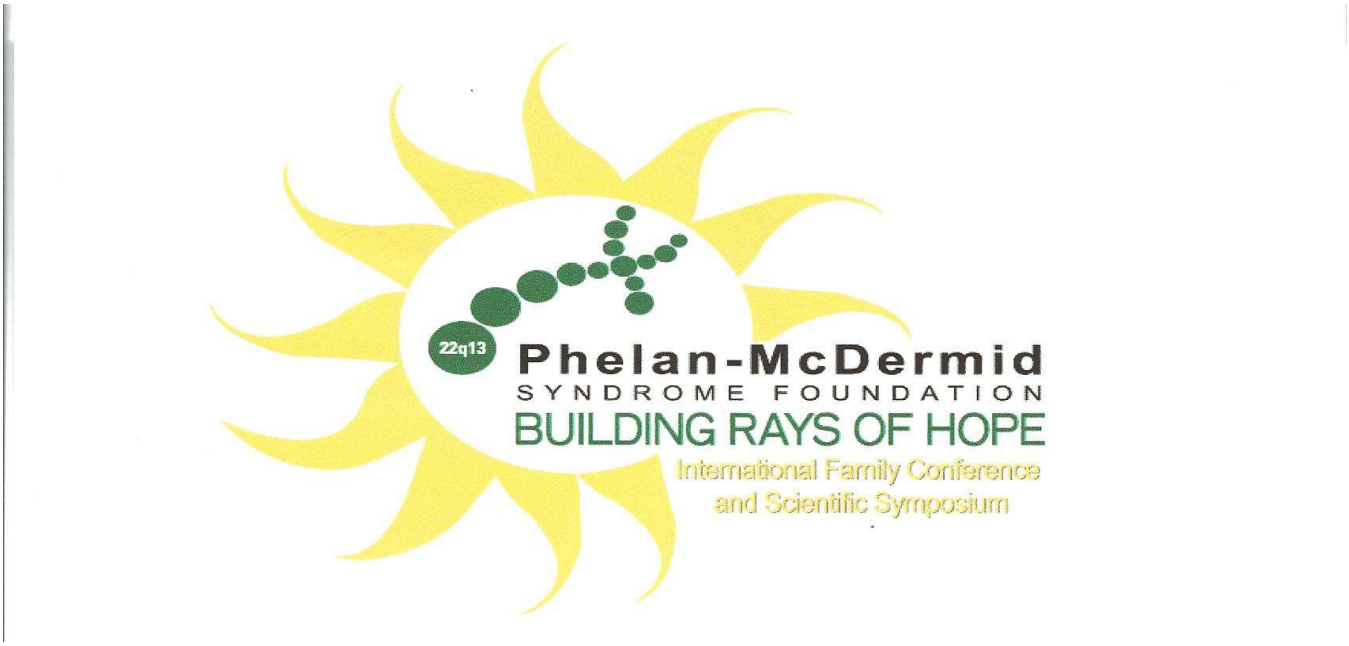
Since Joey's heart surgery he has had five more minor operations for things like placing ear tubes, taking off his ear tags, closing his g-tube site after her learned to take a bottle and most recently to repair his kidney reflux. He never ended up needing the spinal surgery that had been expected; an MRI done when he was five months old showed that the problem had fixed itself. He was also diagnosed with bilateral vocal cord paralysis and laryngomalacia, as well as other malformations of his airway. He participated in birth-three services and physical therapy, and still attends weekly sessions with his speech therapist at Mary Bridge Children's Hospital focusing on aural rehabilitation.

Joey continues to see most of the specialists he was introduced to as a baby regularly, though less frequently. He wears a hearing aid in one ear, and while his vision is very limited on one side, the other eye is in the normal range and he manages with that just fine. He loves to tell stories, and thanks to the help of his hearing aid and speech therapy most of the time he doesn't stop talking. Sometimes when Joey gets a cold or flu he needs extra help with his breathing and has been hospitalized three times with various bugs. He is always well taken care of at Mary Bridge Children's Hospital, and bounces back each time.

Joey has a whole team of amazing doctors, nurses, teachers, therapists and family members who have fought with him and celebrated his accomplishments. In turn, Joey is a bright, loving boy who brings joy to many.

-Jennie

Phelan-McDermid Syndrome Foundations 8th Biennial International Family Conference and Scientific Symposium



"Building Rays of Hope" will be the 2012 theme for the Phelan-McDermid Syndrome Foundations 8th Biennial International Family Conference and Scientific Symposium, to be held July 25th - 28th at the Caribe Royale All Suite Hotel and Convention Center in Orlando, Florida. All families affected by Phelan-McDermid Syndrome (PMS), also known as 22q13 deletion syndrome should attend. Extended family members and caregivers are also welcome and are encouraged to attend.

This year's theme symbolizes the advances our organization is making in providing a brighter future for our families. With each new advancement, we see hope for a stronger future.

This one of a kind event will feature educational speakers, special sessions for siblings and extended family members, round-table discussions for contributing and sharing information, a scientific symposium, where researchers will share their findings and discuss potential therapeutic approaches, social activities and outings and events that provide fun for the entire family. Child care will also be available.

The conference will also host an EXPO featuring vendors offering life enhancing products and services, and the opportunity to meet one-on-one with some of our speakers, Foundation committee members and Board of Directors. Vendor space is currently available, for more info contact Barbara Cruz, Associate Director, Phelan-McDermid Syndrome Foundation at (941) 485-8000.

For a detailed conference informational booklet log on to the foundation website at www.pmsf.org. To register click on the "REGISTER NOW" tab. Be sure to register before June 1st to take advantage of the best pricing available. Families that are members of the Phelan-McDermid Syndrome Foundation are entitled to a reduced member rate for the conference.

For additional conference information: Contact Vicki Camarillo: Vicki@pmsf.org

Phelan McDermid Syndrome Foundation, PO Box 1016, Venice Florida 34284, 941-485-8000, www.pmsf.org

Research

STUDY ON MICRODUPLICATION 22q11.2 SYNDROME: Invitation to Participate

Title of Study: Delineation of Clinical Features of Individuals with Microduplication 22q11.2: A Questionnaire Study

Investigators: Melissa Carter, M.D., Rosanna Weksberg, M.D., Ph.D., Irene Drmic, Ph.D., C.Psych., Leona Fishman, M.D., Cheryl Cytrynbaum, M.Sc., Andrea Shugar, M.Sc., CGC., Evdokia Anagnostou, M.D., and Wendy Roberts, M.D.

All investigators are affiliated with The Hospital for Sick Children in Toronto, Ontario.

If you or your child has been diagnosed with microduplication 22q11.2 syndrome, you may already know that there is very little information in the medical literature about this condition. We know that individuals with this syndrome can have average intelligence, no major birth defects, and no major medical illnesses. We also know that individuals with the *same* microduplication can have significant problems with learning, behaviour, growth, muscle tone, and a variety of birth defects. Individuals with the same duplication but very different problems can be found in the same family. In fact, the findings are so variable that it is difficult to determine whether the presence of this microduplication is the *cause* of these problems, or simply one of many genetic and environmental *risk factors* that work together to contribute to a given individual's physical health and mental development.

We are interested in studying individuals with microduplication 22q11.2 to learn more about the medical and developmental problems that are most likely to be associated with this condition. We are inviting you to participate in a questionnaire study in which we will collect information about individuals who have this duplication. You must have an email address and access to the internet to participate.

For more information, please contact **Dr. Melissa Carter** at melissa.carter@sickkids.ca or **416-813-7654 x28986**.

Thanks in advance for your participation! We look forward to hearing from you.

22q11.2 Conference

The 8th Biennial International 22q11.2 Deletion Syndrome Meeting – "Detection • Care • Cure – *The Magic of Making a Difference*"

When? July 6 -10, 2012
Where? *Disney's Yacht and Beach Club* Resorts in Lake Buena Vista, Florida, USA

Please mark your calendars as **The 8th Biennial International 22q11.2 Deletion Syndrome Meeting** returns to North America for the first time since 2004! This superb conference features presentations from top clinicians and scientists who devote themselves to the study of the 22q11.2 deletion syndrome; span the globe geographically; cover all aspects of the medical, psychosocial, speech, language, and cognitive characteristics of the diagnosis with an emphasis on treatment; and are especially delighted to share their expertise and experiences with families and professionals alike! The meeting entitled, "Detection * Care * Cure – *The Magic of Making a Difference*" will indeed be magical as it is set in the most inviting of settings – *Disney's Yacht and Beach Club* Resorts in Lake Buena Vista, Florida. In addition to the meeting sessions, unique activities such as Character Greetings are planned for children and adults alike, beginning with a Welcome Reception on **Friday, July 6, 2012** and concluding on **Tuesday, July 10, 2012**. For more information please visit www.22qconference.com as The International 22q Deletion Syndrome Foundation, Inc. has graciously accepted the baton from the 2010 Coventry, England host Max Appeal as lead sponsor and conference organizer. Thus, all those interested in learning more about the 22q11.2 deletion syndrome from families to healthcare providers, therapists to educators are all invited and encouraged to attend. Looking forward to seeing you there!

For More Information * To Register Online * To Make a Reservation * Please Visit www.22qconference.com

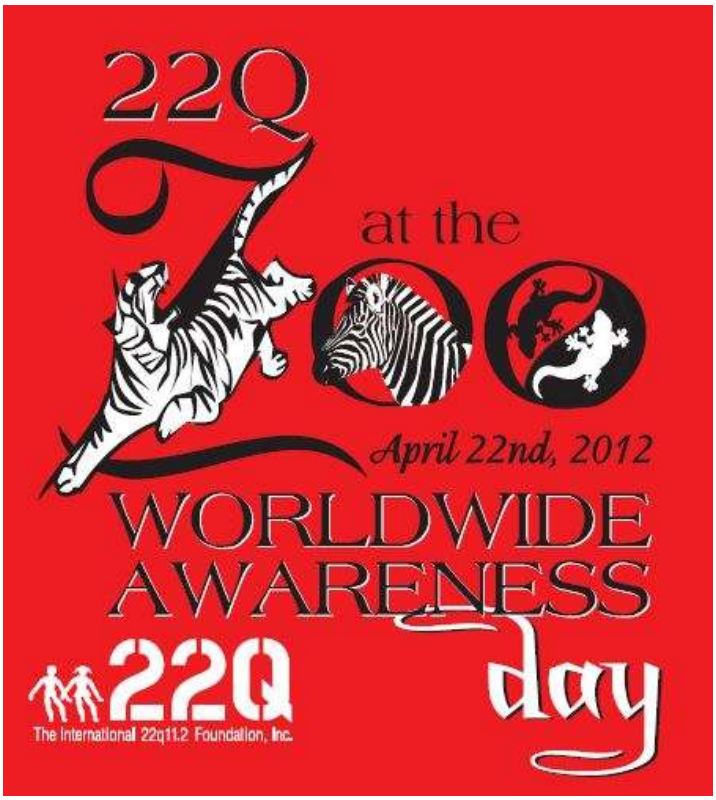
22q at the Zoo! Worldwide Awareness Day—APRIL 22!

Join thousands of 22q families worldwide to celebrate a fun-filled awareness day at zoos all over the world on Sunday, April 22, 2012!

Organized and developed by The International 22q11.2 Foundation, ***22q at the Zoo! Worldwide Awareness Day*** will take place on Sunday April 22, 2012 at zoos all around the world. This fabulous event is aimed at raising the public profile of chromosome 22q11.2 deletion syndrome. In 2011, we had the participation of 65 zoo locations in 9 countries spanning 3 continents with close to 10,000 participants worldwide! This year our goal is to double those numbers! This was a start to something very special – a ray of hope and solidarity which we have all been waiting for. DO NOT MISS OUT on a chance to participate! 2012 is shaping up to be in the largest awareness event in history. Please visit www.22q.org to find a list of this year's participants! Currently we have 50 zoos spanning 4 continents that have signed up to host this event!

This event is not for raising funds. It gives families touched by the 22q deletion a chance to socialize and network with each other, all in the name of awareness. For many, this will be the first time they have ever met another family affected by the syndrome. The event also helps us increase awareness and improves the quality of life for affected children and their families.

22q at the Zoo! Awareness T-shirts are available for purchase at www.22q.org! The 22q at the Zoo! Worldwide Awareness Day was created to engender solidarity among all patients, families and clinicians who deal with 22q. 22q at the Zoo in 2011 was truly the first time in history when support and clinical groups finally came together, forgot the name game, and accomplished something big with solidarity. This t-shirt is a celebration of this.



If you or someone you know is interested in organizing this event at a zoo near you, please contact Swati Patel at info@22q.org for more details. We encourage you to please join our *22q at The Zoo! Worldwide Awareness Day* Information Page at

www.facebook.com/groups/22qAtTheZoo2012

For more details regarding this event. Please feel free to share this site with friends and family for more information.

Don't miss out on this opportunity to spread awareness about 22q11.2DS, Make Friends, and Have Fun! Not to mention having the opportunity to own your very own highly acclaimed 2012 Awareness T-Shirt! Supplies are Limited so Don't Wait – Order Now!!!!

22q is in need of awareness!! Let's double our numbers! Together we can and will make a lasting difference!

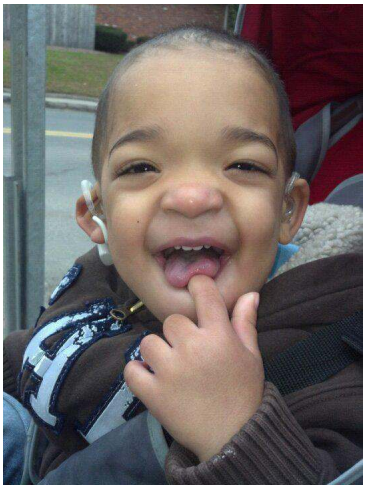
Isaiah, Kendra and Dayanara

Baby in yellow is Isaiah 2, next Dayanara 7 towards the right, then Kendra 4 in orange, and finally Luis 9 in the white. Luis does not have any genetic disorders but suffers from other problems. These kids have gone through so much in the last 2 years. From Cochlear implants, to being diagnosed with ADHD, to continuous seizures, and hard time coping.

While attending school at South University continuing my Medical Degree; I have learned so much about health with my children and not to mention with my own medical problems. I write so much about my children and never really bother writing about myself. I am finally opening up to say….I have two of three genetic disorders Isaiah has. I have not been affected like my children have, these disorders are dormant in me, but nonetheless I have had complications. I was born November 27th 1986. My name is Coralis Yessenia and I was 4lbs and stayed in the NICU. I couldn't feed from the breast or a bottle. I was jaundice and also needed the incubator. As a child my health was pretty good aside from asthma and colds. I suffered my first seizure when I was 16. I was pregnant with my second child. I have not had seizures since. I wear glasses to see for astigmatism and a floater behind my left pupil that has been closely monitored. I also have back a bit of a lower curve that goes in on my lower spine.

In 2010 I suffered multiple infections and almost lost my life not once but 3 times. My immune system is not as strong as a person that is generally healthy. It took a whole 2 years to somewhat heal from 5 surgeries. Even after 2 years I still have some problems that I don't seem to get better from. I get pneumonias frequently just like Isaiah. I have also inherited the unfortunate problem of multiple miscarriages and two stillbirths due to my genetic disorders. Anemia has also been in my life since I was about 1years old. No one knows this but I too have hearing problems. I can hear a bit more from the right side. I can hear from both ears but my left is not as good as my right side.

With my own struggles I have managed to become a mother/advocate to these four beautiful children and meet all my milestones and stepping stones in life with the help of my husband. I don't use my children's health or mine as an excuse to not progress in life.



Isaiah has 9p and 22q Microduplication also complicated by several other medical problems. Just a recap on Isaiah: he was born January 17th, 2010 severely/profoundly deaf in both ears. At birth suffered a broken right humorous at birth by C-section, he was jaundice, cyanotic, with feeding problems needing a feeding tube. Soon after birth he was found to have a sub mucous cleft palate, hypotonic and hypertonia, thin corpus callosum of the brain, seizures, GERD, frequent Pneumonias needing to be hospitalized and intubated, asthma, leg discrepancies, scoliosis, Pneumonia, glue ear and CVI (Cortical Visual Impairment) at the age of 2 months. He is also weak with his immune system.

On January 26th, 2011 Isaiah was implanted with his first cochlear implant on the right side. August 14th, 2011 his implant was activated. Isaiah is still not able to hear with his device. It has been a very hard struggle since then to get him to hear or show improvement. I as a mother to a non-hearing child that was "finally going to be able to hear" bought a bunch of musical toys.

3of 4 Siblings and Mother with 22q and 9p Micro-duplication

I even went and bought a soothing heartbeat, ocean and bird speaker since he was never able to hear my heart as a baby when I held him. I am still holding on to the hope that he may someday hear.

Isaiah has developed extreme obstructive sleep apnea, continues to be cyanotic and was recently diagnosed with Autism. It has been so hard with the new stuff happening in his life. The Autism adds so much more to everything he has going on. It has taught me so much though. He likes being in his room alone and at times has kicked Gabriel and myself out; by yelling, pointing and banging his head into the crib. He destroys his toys to play with parts, and his attention span is not very big. Isaiah tends to hide in corners, under chairs and tables when he is not in his room or wants to be left alone.

I have started to work with some people to get a Deaf Bill of Rights past in the state of CT. Isaiah has a small dog named Bella. She was part of his daily home therapy. Due to a Nurse and the Doctor that was seeing Isaiah I had to get rid of Bella. They did not see her as a type of therapy for him and did not sign papers I needed in order to keep her in my home. In our eyes she was, she comforted him and he was excited to see her.

Kendra's recap: Born May 30th, 2007. As a baby Kendra stopped breathing on me 3times forcing me to give her CPR until EMTs came. Kendra was on breathing machines and apnea monitors for the first 2yrs of her life. She is also on a bunch of meds. Kendra is still having issues with mild hearing loss and continues to have speech problems we are still working/fighting the

school system to give her services she needs. She has been seen and may be getting hearing aids soon. In 2008 she was diagnosed with 22q Micro-duplication which is the cause of her having the following: Hearing loss, frequent ear infections, bigger ears than she should have, Obstructive Sleep Apnea, Cyanosis, Rhino-Malaysia, Gerd, Strider, floppy wind pipe, Seizures, Heart Murmur, Asthma, constant colds, Leg discrepancy, Hip dysplasia, She is more like a 2 year old at the age of 4, Liver issues

Kendra was doing great in the beginning of winter until February 2012. She has been in the hospital so much and has had more seizures that are taking her back to when she was 1. The latest seizure caused her to be stiff on one side of her body and she wasn't able to talk or walk. She is still recovering from those seizures.
Kendra is also more prone to sickness and infection from weak immune system.

Dayanara has 9p Micro-duplication. Dayanara has not been as sick as the other kids and

me. If she wouldn't have been tested I would have said she is a pretty healthy child. Her disorder has caused: Large ears Hypertelorism, Small teeth, Wide spread eyes, She is slower than others her age with some stuff and it takes time for her to respond to directions. Dyslexia, Quite emotional, Bullies' siblings, Aggression, Visual issues (astigmatism), Asthma, Small fingers and toes, Body pain, Anemia, Easily influenced.

In 2011 she was diagnosed to also have ADHD/ADD. She is followed by a behavioral specialist for this. Since she was about 4 she had a small birth mark on her arm that is now bigger by a lot. It starts at her shoulder and comes all the way down to almost her elbow. She is going to see a dermatologist soon to determine if she has strip syndrome. I have never heard of this so it will be something else for me to learn. Dayanara does wear glasses and is pretty much doing well. She has been getting great grades in school and wants to join softball.

We would like to thank Steph for the opportunity she gives us every time to not only learn and see all the wonderful stories, but also for allowing me to share mine.

-Coralis

19TH ANNUAL INTERNATIONAL SCIENTIFIC MEETING OF THE VCFS EDUCATIONAL FOUNDATION

I am so excited that by the time this newsletter is published, registration for the 19th Annual International Scientific Meeting of the VCFS Educational Foundation will have been available on the website and ready for participates to make plans to join us for this event. It is somewhat surreal that we are at this point given our planning to host this event started in the Fall of 2008 after we attended the 15Th Annual meeting in Tory Michigan. As Chair of the Local Arrangements Committee I want to ensure that we pack the time with lots of activities for the kids as well as ensuring there are relevant topics for the professionals and parents. We also want to ensure that your time in Canada is enjoyable and you get a chance to experience a bit of what Canada and Toronto are all about. We will be continuing in our efforts to raise funds to assist with covering some of the costs of the meeting. The response to date has been fantastic.

The Programming Committee will also be continuing to review the papers submitted on-line and ensure that we have covered the key topics and issues that are relevant to this population. Families, individuals with VCFS and professionals are welcome to submit an abstract by following the link on www.vcfsef.org. The Foundation will be accepting submissions online up to April 15, 2012, 11:59 EST.

Currently some of the topics planned to be covered include:

- Social Cognition across the ages - the processing of social information and application to social situations,
- Transition from teen to adulthood,
- Information on feeding/ swallowing,
- Communication development,
- Velopharyngeal Function for speech,
- Psychology,
- Cardiology,
- Genetics and Immunology

And much more….

While you are engaged in the topics listed above your kids will be participating in some great activities. Nancy DeNardis, a friend of mine from grade school, will be in charge of keeping all your children involved and entertained during the weekend. She LOVES kids and spends a lot of time with her young nephew including volunteering as his soccer coach. Her two children are in high school and so she loves the opportunity to spend time with the little ones. To date we are working on organizing the weekend into blocks of time that will have different themes. We have various performers who are donating their time to entertain the kids – we are just worried about keeping the parents in the meeting room and out of kids zone! There will be crafts, games, puzzles, toys and even time to run around in the hotels squash courts. We guarantee the kids will have a BLAST.

We also want to encourage everyone to join in on the Friday night dinner that we are organizing. This event is a great opportunity for the families and professionals to mingle in an informal setting. We have engaged the “Drum Café” to entertain us on Friday night. Take a few minutes to check out their website (www.drumcafe.ca). This group emphasizes communication without words and how we are all stronger when we work together. We felt this linked to our group given the VCFS connection we all have. You will learn through the musical experience just how important it is to collaborate and listen to one another – a key theme we can relate to the meeting. Participants will also learn the value of their own individual contributions within the context of the team – how your approach to increasing awareness, or approaching treatment, etc. can have an impact for us all.

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To help ensure you see a little bit of Toronto during the weekend we will be arranging a school bus to take interested attendees from the hotel to the CN Tower area of Downtown Toronto for the low cost of $5 per person. This will be available on the Saturday night. This part of Toronto is central, close access to public transportation and an area full of things to do. You can:

- take a ride up the CN Tower and see all of Toronto from a unique perspective. The CN Tower includes a restaurant and other activities (www.cntower.ca)
- visit the Harborfront Centre which has many options (www.harbourfrontcentre.com)
- take a short ferry ride across to Toronto Islands. This is a great place for some family activity and includes Centreville – an amusement park, with over 30 rides and attractions as well as a number of restaurants and food vendors. A lot of fun – many rides are appropriate for smaller children (www.toronto.ca/parks/island/index.htm)
- enjoy dinner at one of the many restaurants in the area
- union station is nearby which can link you into Toronto's subway and street car networks that can take you to many other parts of Toronto.

There are many other activities available….

The Local Arrangements Committee will have maps and details of attractions available for attendees.

We look forward to meeting all of you during the weekend. Email: info@vcfs.org

The story of my life

I was born Michelle Leigh Padilla-Hanna. I was born in Tuscan, Arizona at Davies Monthin Airforce base on April 15, 1976. I was supposed to be born on March 18,1976. I almost died due to lack of oxygen to the brain. The doctors on the base had to transfer me to a different hospital in Arizona. It was June 1976. My parents and I came out to California. I was seeing a cardiologist who diagnosed me with severe Tetralogy of Fallot. When I was two years old I had my first heart operation. It was a leaking value. When I was 5, I had my second open heart surgery. Also, I had tubes put in my ear. Finally another surgery that I had was for pharyngeal flap. My last open hear surgery was when I was 13. I had to go back in because I had another leaking value. I was held back in first grade because I had problems reading, writing, and doing math. When I was 18 I had a grand mal seizure. I was in a coma for a couple days. Because of that I suffered brain damage. I have been in special education classes all through my life. When I had my seizure in high school, the teachers were afraid that a ball would it me on the head and cause me another seizure. I finally graduated in the summer because I had a hard time

passing math. After high school, I was working different jobs but I always got sick so they had to let me go. It was a few years later that I wanted to go back to school. I started out going to a trade school and I got a certificate for computers. After that I decided that I wanted to go to college. I started going to Harbor College in the year 2003, until this year 2012. I will be graduating this summer. I had my sinus surgery in 2010. I found out that I have two dislocated plates in my lower back. I also have a hernia in my stomach. I found out that I had VCFS in 2010. I never knew that I had it. This year I did a study for UCLA which told me that I suffer anxiety disorders as well as ADHD. I was told that there is a 50-50 chance that my kids could have this syndrome. I got this syndrome by accident. This is my story.

-Michelle

Recently Released Journal Articles

22q11 MICRODUPLICATION

Chromosome 22q11.2 duplication is rare in a population-based cohort of Danish children with cardiovascular malformations. Agergaard P, Olesen C, Ostergaard JR, Christiansen M, Sørensen KM. Am J Med Genet A. 2012 Feb 2

22q13 DELETION / PHELAN-MCDERMID SYNDROME

Growth in Phelan-McDermid syndrome. Rollins JD, Sarasua SM, Phelan K, DuPont BR, Rogers RC, Collins JS. Am J Med Genet A. 2011 Sep;155A(9):2324-6

Association between deletion size and important phenotypes expands the genomic region of interest in Phelan-McDermid syndrome (22q13 deletion syndrome). Sarasua SM, Dwivedi A, Boccuto L, Rollins JD, Chen CF, Rogers RC, Phelan K, DuPont BR, Collins JS. J Med Genet. 2011 Nov;48(11):761-6.

22q13 DUPLICATION

Mitochondrial Disease in 22q13 Duplication Syndrome. Frye RE. J Child Neurol. 2012 Feb 28.

CAT EYE SYNDROME

A 600 kb triplication in the cat eye syndrome critical region causes anorectal, renal and preauricular anomalies in a three-generation family. Knijnenburg J, van Bever Y, Hulsman LO, van Kempen CA, Bolman GM, van Loon RL, Beverloo HB, van Zutven LJ. Eur J Hum Genet. 2012 Mar 7.

RING 22

Azoospermia and paternal autosomal ring chromosomes: case report and literature review. Rajesh H, Freckmann ML, Chapman M. Reprod Biomed Online. 2011 Oct;23(4):466-70

22q11 DELETION

Computerized neurocognitive profile in young people with 22q11.2 Deletion syndrome compared to youths with schizophrenia and at-risk for psychosis. Goldenberg PC, Calkins ME, Richard J, McDonald-McGinn D, Zackai E, Mitra N, Emanuel B, Devoto M, Borgmann-Winter K, Kohler C, Conroy CG, Gur RC, Gur RE. Am J Med Genet B Neuropsychiatr Genet. 2012 Jan;159B(1):87-93.

Prenatal diagnosis of a 22q11 deletion in a second-trimester fetus with conotruncal anomaly, absent thymus and meningomyelocele: Kousseff syndrome. Canda MT, Demir N, Bal FU, Doganay L, Sezer O. J Obstet Gynaecol Res. 2012 Mar 2

Is there a core neuropsychiatric phenotype in 22q11.2 deletion syndrome? Baker K, Vorstman JA. Curr Opin Neurol. 2012 Apr;25(2):131-7.

Genotype and cardiovascular phenotype correlations with TBX1 in 1,022 velo-cardio-facial/DiGeorge/22q11.2 deletion syndrome patients. Guo T, McDonald-McGinn D, Blonska A, Shanske A, Bassett AS, Chow E, Bowser M, Sheridan M, Beemer F, Devriendt K, Swillen A, Breckpot J, Digilio MC, Marino B, Dallapiccola B, Carpenter C, Zheng X, Johnson J, Chung J, Higgins AM, Philip N, Simon TJ, Coleman K, Heine-Suner D, Rosell J, Kates W, Devoto M, Goldmuntz E, Zackai E, Wang T, Shprintzen R, Emanuel B, Morrow B; International Chromosome 22q11.2 Consortium. Hum Mutat. 2011 Nov;32(11):1278-89.

Chromosome 22q11 in a Xhosa schizophrenia population.Koen L, Niehaus DJ, Wright G, Warnich L, De Jong G, Emsley RA, Mall S.S Afr Med J. 2012 Feb 23;102(3):165-6

Successful Treatment of Sepsis Caused by Staphylococcus lugdunensis in an Adult with 22q11.2 Deletion Syndrome. Hirasaki S, Murakami K, Mizushima T, Ohmori K, Fujita S, Hanayama Y, Kanamori T, Yokota R, Ebara H, Kusano N, Kudo C, Yamaguchi T, Akagi T, Koide N. Intern Med. 2012;51(4):377-80. Epub 2012 Feb 15.

Preliminary structure and predictive value of attenuated negative symptoms in 22q11.2 deletion syndrome. Schneider M, Van der Linden M, Glaser B, Rizzi E, Dahoun SP, Hinard C, Bartoloni L, Antonarakis SE, Debbané M, Eliez S. Psychiatry Res. 2012 Feb 27.

Atypical presentations of 22q11.2 deletion syndrome: explaining the genetic defects and genome architecture. Tuțulan-Cuniță AC, Budişteanu M, Papuc SM, Dupont JM, Blancho D, Lebbar A, Viot G, Lungeanu A, Arghir A. Psychiatry Res. 2012 Feb 22.

Cortical gyrification in velo-cardio-facial (22q11.2 deletion) syndrome: A longitudinal study. Kunwar A, Ramanathan S, Nelson J, Antshel KM, Fremont W, Higgins AM, Shprintzen RJ, Kates WR. Schizophr Res. 2012 Feb 22.

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A second look: No effect of the COMT Val158Met polymorphism on conflict adaptation in youth with chromosome 22q11.2 deletion syndrome. Stoddard J, Takarae Y, Simon TJ.Schizophr Res. 2012 Jan 13.

Recently Released Journal Articles

2q11 DELETION

Detecting 22q11.2 deletion in Chinese children with conotruncal heart defects and single nucleotide polymorphisms in the haploid TBX1 locus. Xu YJ, Wang J, Xu R, Zhao PJ, Wang XK, Sun HJ, Bao LM, Shen J, Fu QH, Li F, Sun K. BMC Med Genet. 2011 Dec 21;12(1):169

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Computerized neurocognitive profile in young people with 22q11.2 deletion syndrome compared to youths with schizophrenia and At-Risk for psychosis. Goldenberg PC, Calkins ME, Richard J, McDonald-McGinn D, Zackai E, Mitra N, Emanuel B, Devoto M, Borgmann-Winter K, Kohler C, Conroy CG, Gur RC, Gur RE. Am J Med Genet B Neuropsychiatr Genet. 2011 Dec 13.

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Dental development and tooth agenesis in children with velocardiofacial syndrome. Heliövaara A, Rantanen I, Arte S. Int J Paediatr Dent. 2011 Nov;21(6):446-50.

Sex differences in reproductive fitness contribute to preferential maternal transmission of 22q11.2 deletions. Costain G, Chow EW, Silversides CK, Bassett AS.

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Shape-Based Classification of 3D Facial Data to Support 22q11.2DS Craniofacial Research. Wilamowska K, Wu J, Heike C, Shapiro L. J Digit Imaging. 2011 Nov 16

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Immunological aspects of 22q11.2 deletion syndrome. Gennery AR. Cell Mol Life Sci. 2011 Oct 9.

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Collaborative Group Marches on Washington, DC, USA to Promote Newborn Screening for 22q11.2 Deletion Syndrome!

In an historic moment on January 26, 2012, groups from throughout the United States, Canada, and the UK had the opportunity to present before the US Secretary of Health's Committee on Heritable Disorders and Newborn Screening to rally support for newborn screening for 22q11.2. These representatives came from The Children's Hospital of Philadelphia, The University of Toronto, The International 22q11.2 Foundation, Max Appeal, Children's Hospital of Wisconsin, and The Ryan and Jenny Dempster Foundation.

In an interesting turn of events, the Committee had already predetermined that the application to add 22q11.2 screening to the current federally mandated list of newborn screening studies be denied, prior to any public comments or proponent group presentation, based on a lack of pilot study evidence to support the proposal. However, following an extremely convincing and eye opening presentation by the Newborn Screening Proponents, the Committee agreed to readdress the proposal once such pilot data is submitted into evidence. To this end, the collaborating parent organizations (The International 22q Foundation, Max Appeal, and the Dempster Family Foundation - with hopes that other such organizations will join in the very near future) have agreed to begin a rapid and restricted fund raising campaign to allocate the necessary funds for such an endeavor.

Ninety-eight percent of individuals who are dealing with the 22q11.2 deletion support Newborn Screening for the 22q11.2 deletion as 92.9% of symptoms in children started less than one year old. With that, the Newborn Screening Proponents are working very hard to design and implement a pilot study to obtain more evidence and re-submit the proposal to the Secretary's Advisory Committee on Heritable Disorders in Newborns and Children.
By way of background, the 22q11.2 deletion syndrome (22q11.2DS), present in 1 out of every 2000 - 4000 live births, is the most common of rare genetic disorders. It is present in 1 of every 68 children born with congenital heart disease. It is the most common cause of syndromic palatal anomalies, and it is a leading cause of developmental disabilities. 22q11.2DS is very serious and can lead to long-term issues especially when it goes unrecognized as children may have low calcium, for example, which can cause subclinical (silent) seizures. This deletion has the potential to affect almost every system in the body and can cause a wide range of health problems from mild to severe. As the number of children with this deletion grows and as awareness efforts continue, it is important to take action now.