2015 Vol 1

THE MIRROR

The Newsletter of DUP15q ALLIANCE

This Is Your Conference

By Alison Kalnicki, Conference Committee Member

The 8th International Dup15q Alliance Family Conference, **STRONGER TOGETHER**, will be held from July 30 to August 1, 2015 in Orlando, Florida. This conference only happens once every two years and is a one of a kind event where the Dup15q Alliance community gathers together.

The Dup15q Alliance conference has something for everyone. Children and adults with dup15q syndrome, parents and siblings, grandparents and friends, uncles and aunts, caregivers and professionals: anyone can register. For some people the opportunity to meet other people from around the world whose lives are affected by dup15q syndrome is of upmost importance. For many people acquiring first-hand knowledge is the reason they attend the conferences. What is dup15q syndrome? What does interstitial mean? How do I know if someone is having a seizure? What are the latest and best interventions? What happens in adulthood? In this issue of *The MIRROR*, we give readers a glimpse of the hot topics for our 2015 conference and provide an introduction to the presenters.

For up-to-date information on registration, the conference schedule, hotel booking and other conference details go to www.dup15q.org/events/family-conferences/stronger-together-2015/.

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Chromosome 15q11.2-13.1 duplication (dup15q) syndrome is a clinically identifiable syndrome which results from duplications of chromosome 15q11.2-13.1. These duplications most commonly occur in one of two forms. These include an extra isodicentric 15 chromosome, abbreviated idic(15), which results in an individual having 47 or more chromosomes instead of the typical 46. Individuals with an interstitial duplication 15 are born with the typical 46 chromosomes but have a segment of duplicated material within their 15th chromosome.

Hot Conference Topics

There will be over twenty presentations, moderated panels and information updates during the three day conference. Remember to peruse the conference website often to get up-to-date schedules and session information: www.dup15q.org/events/family-conferences/stronger-together-2015/

New Family Welcome and Conference Info Session

This is an opportunity for everyone new to Dup15q Alliance conferences to prepare for the next few days, gather some helpful hints on the conference, ask questions and connect with other conference participants. At press time, this session is tentatively scheduled for the evening of Wednesday, July 29, 2015.

Being Present: The Families Journey To Acceptance And Resilience

This year's keynote presentation by Dr. Robert Naseef will encourage us to do our best as a family living day-by-day. Dr. Naseef, a psychologist and a father of an adult son with autism, investigates how acceptance refuses to give up. Acceptance is not a route to making war or peace with challenging conditions. It means taking care of everyone's needs while finding the elusive balance of changing what we can and letting go of what we cannot.

Seizures

The complexity of seizures in dup15q syndrome is immense. At this point, we know that about 60% of people with dup15q syndrome will experience at least one seizure in their life. Dr. Ron Thibert and Dr. Dimitrios Arkilo are pediatric epileptologists who have in-depth knowledge of dup15q syndrome. They will explore the world of seizures including a thorough description of what a seizure is, recent findings from research, the lessons being learned from their dup15q clinics, current medications of choice and how to have seizures diagnosed. Bring your questions and be ready to learn about one of the most worrisome topics for many families.

Moderated Sessions for Our Lives: Fathers, Mothers, and Marriage

These separate sessions are what some attendees say they find the most rejuvenating. Take the time to attend the sessions that relate to your life and get together with others in a similar situation as yourself. Session titles and moderators will be outlined on the conference website.

Cannabinoids

You may have heard about the use of derivatives from marijuana to treat seizures in the news lately. Here's your chance to find out what the buzz is about from someone who treats children with dup15q syndrome and is running a trial of Epidiolex (cannabinoid) that involves some children with dup15q syndrome.

Vision in Children with Dup15q: Encouraging Efficient Use of Vision

Many children and adults with dup15q syndrome are visual learners yet they have visual challenges that are neurologically based. Join Donna Richards in an exploration of educating children with neurologically based visual processing concerns. Expect to leave this presentation with an increased understanding of the visual challenges of someone who has dup15q syndrome, and a familiarity with the visual skills needed for visual efficiency. Come and arm yourself with simple techniques and approaches to enhance the visual world of someone who has dup15q syndrome.

Transitioning to a Career

This presentation may open your eyes to new possibilities for how life looks after high school for people with intellectual disabilities. Pathways to Careers is an employment program that works towards individuals with disabilities having quality employment options. An individual's strengths are identified and matched to a paid internship. Support is provided to smooth the path to ongoing and rewarding employment. Your presenter, Therese Fimian, is the aunt of someone with idic(15). She will introduce you to the Pathways to Careers programs which is being piloted and evaluated in four American locations. Therese will review case studies where providing job exploration and career opportunities for youth and adults with autism, intellectual and/or developmental disabilities has provided a meaningful future.

Extended Family Session

Are you a grandparent, aunt, uncle or other extended family member of a child or adult with dup15q syndrome? This is a special time for you to get together with others in a similar situation, explore ways to help your families succeed, share insights, and ask questions.

Research, Clinics and Registry Updates

Join these sessions to get up-to-date information on the scientific advances in the study of dup15q syndrome. The scientists and physicians that will be on hand to answer questions include Dr. Jill Silverman, Dr. Stormy Chamberlain, Dr. Scott Dindot and Dr. Shafali Spurling Jeste. Rylie McHam will be on hand to present updates on the Dup15q Alliance International Registry. It is recommended that you attend the Genetics 101 presentation first, in order to get the most out of these informational updates.

Financial Planning for Dup15q Families

This session will guide you and your family toward financial planning that takes the financial pressures of a child with special needs into account. International conference attendees are more than welcome to attend, but be prepared for some U.S. tax law discussions. Expect to hear Mike and Jeff Rozovics, a father and son duo, cover topics, including different programing and funding sources, that will support your financial stability as your child moves through his/her life and as your finances are affected by life events. They will introduce you to long-term saving strategies for all income levels and review items such as trusts, wills, power of attorney and guardianships. They ask that you keep in mind that this presentation does not take the place of professional legal advice, but will help point you in the right direction.

Sensory Solutions

Sensory issues are common for children and adults with dup15q syndrome. At this session, you can learn about intervention methods and gather tips from both an occupational therapist (OT) and from the perspective of families affected by dup15q syndrome.

Communicating with PODDS (Pragmatically Organized Dynamic Display System)

Join speech and language practitioner (SLP) Carrie Dinneen and learn how the PODDS communication system organizes and selects symbols that will encourage communication for children and adults with communication needs. Hear also how this AAC (augmentative and alternative communication) system has had success for a boy with dup15q syndrome.

From Pull-ups to Potty: A Parent Panel

Hear from other parents which toilet training techniques are successful. Bring your questions, vent your frustrations, and share accomplishments from the bathroom with people who understand.

Positive Exposure Photography

Rick Guidotti's photos, published work and presentations are filled with upbeat images that counter stereotypical images of people with disabilities. Read more about this amazing man's work in the "Who's Who" article. Come meet Rick and let his journey shape yours.

Your Conference Presenters

Conference Who's Who

By Alison Kalnicki, Courtenay, B.C. Canada

Here is a preview of some of the people you can expect to hear from at the conference. The presenters have a wide variety of training, education and interests. They range from professionals to family members, but all have one thing in common. They want the best for individuals with dup15q syndrome.

Dr. Robert Naseef

Back by popular demand, Dr. Naseef participated at the 2011 conference in Philadelphia where attendees benefited from his expertise, humor and empathy. One father, having attended a session for fathers moderated by Dr. Naseef said, "That was the first time that I really felt heard. He understood what I am living through." Dr. Naseef is a psychologist who has practiced for over twenty years working with families of children with autism and other special needs. He has a special interest and expertise in the psychology of men and fatherhood. He is also the father of an adult son with autism.

Dr. Naseef has written several books and presents regularly on family life with special needs. He also blogs, moderates the online Google Hangout series "Guy Talk: Autism Brainstorming", and publishes a monthly e-newsletter that is subscribed to worldwide.

Dr. Naseef will be presenting at several sessions including one on being present, stress management, raising siblings of children with special needs, and he will be moderating the fathers' session.

Dr. Edwin H. Cook Jr.

Dr. Cook is a mainstay on the Dup15q Alliance Professional Advisory Board. He is Director of both the Center for Neurodevelopmental Disorders, and the Division of Child and Adolescent Psychiatry at the Department of Psychiatry at the University of Illinois at Chicago. He will be presenting on behaviors and current research findings.

Dr. Orrin Devinsky

Dr. Devinsky is the author of over twenty books including, *Epilepsy* and *Alternative Therapies in Epilepsy*. He is a professor of Neurology, Neurosurgery, and Psychiatry at New York University (NYU) School of Medicine and is the Director of the NYU Comprehensive Epilepsy Center. Dr. Devinsky will be discussing his preliminary findings and will share other facts about the use of cannabis in the treatment of seizures.

Dr. Ron Thibert and Dr. Dimitrios Arkilo

Both presenters are involved with multidisciplinary clinics for children and adults with duplications of chromosome 15q. Dr. Arkilo is a team member of the dup15q center at the Minnesota Epilepsy Group in Saint Paul, Minnesota. Dr. Thibert is the Director of the Dup15q Center at Massachusetts General Hospital and participates on the teams as a pediatric epileptologist. He is also is a member of the Dup15q Alliance Professional Advisory Board.

Dr. Shafali Spurling Jeste

Dr. Jeste, a pediatric neurologist specializing in autism and related neurodevelopmental disorders, is coming to Orlando from Los Angeles where she runs the dup15q clinic at University of California, Los Angeles (UCLA). She will be speaking about autism and social communication in children with dup15q syndrome. She will also reveal findings from her clinical work, including how children with dup15q syndrome can make meaningful developmental gains when interventions appropriately target their specific areas of delay. Dr. Jeste is a member of Dup15q Alliance's Professional Advisory Board and has published many articles on autism, genetics and biomarkers in peer reviewed journals..

Brenda Finucane

Brenda is an Associate Director and Clinical Investigator at Geisinger Health System and is the co-founder of IsoDicentric 15 Exchange, Advocacy and Support (IDEAS) which grew into Dup15q Alliance. While her background is in genetic counseling, Brenda worked for almost three decades specializing in special education, psychology and support services for adults and children with intellectual and developmental disabilities. Brenda will give conference participants the foundational understanding of genetics at the Genetics 101 presentation.

Rick Guidotti

Rick Guidotti uses the art of photography to celebrate the lives of those living with genetic, intellectual and physical conditions. Rick is the founder and director of Positive Exposure, an arts, education and advocacy organization which works with individuals living with genetic differences. Positive Exposure works with advocacy organizations such as Dup15q Alliance, medical schools, high schools and governments to encourage a positive shift in the way individuals with genetic disorders are viewed.

Kadi Luchsinger

Behind the scenes and at the podium, Kadi is a guiding force of the conference. She has been a member of the Dup15q Alliance Board since 2004 and has served as Executive Director since 2007. Expect to see her welcoming everyone at the opening ceremony, speaking on the present state of Dup15q Alliance and the plans for the future, and wrapping things up at the closing ceremony.
As well as being Executive Director of Dup15q Alliance, Kadi works as a pediatric physical therapist. She lives near Syracuse, New York with her husband Todd and their three children.

Rylie McHam

Rylie is on the Dup15q Alliance Board. She is a social worker and works in a private practice counseling adults with developmental disabilities. She's the go-to person for the Dup15q Alliance International Registry.

Donna Richards

For over thirty years, Ms. Richards has been a teacher of visually impaired students with ocular or neurological conditions, including ten years of experience with someone with dup15q syndrome and a cortical visual impairment (CVI).

Jeff Rozovics and Mike Rozovics

Mike and Jeff are a father and son team from the Chicago area with a lot of financial planning know-how. Both are partners in a certified public accountant firm and are also certified financial planners. They want to share with you their expertise about financial planning as it relates to special needs children and their families. Jeff says, "This is something I can do for the dup15q families; I can give back to Dup15q Alliance after all it's done for my eleven year old daughter, Riley, who has idic(15)."

Research Opportunities at the Conference

Conference Research Passport

By Kadi Luchsinger, Executive Director

Our 2015 conference will have several opportunities for families to get involved in dup15q syndrome research. We will be posting specific information about individual studies and sign ups on our website at www.dup15q.org/events/family-conferences/stronger-together-2015/. Please continue to check the website as we have several in the works.

Dup15q Syndrome Cell Lines

In 2003, we arranged to have blood draws at our family conference. This is how we started our collection of dup15q syndrome cell lines at the Genetic Cell Repository at Coriell Institute for Medical Research. We will be offering blood draws again at this conference. You are probably wondering why this is important. Here is what Stormy Chamberlain, Dup15q Alliance professional advisor, said,

"The blood draws provide an important resource to current dup15q researchers and will likely draw new researchers into dup15q studies. Patient samples (DNA or blood cells) that have some accompanying information about the patient's symptoms (by applying to receive such de-identified information through the Alliance or by collaborating with one of the dup15q clinics) are a rare treasure in research. That's why I think this will bring in new researchers. Some of the possible studies that could be done include determining the precise structure of the duplication, determining the variability of the different breakpoints and how this might affect gene expression or disease severity, or determining whether other genes in the genome affect disease severity. Furthermore, some samples may be used to produce induced pluripotent stem cells. These cells are like embryonic stem cells and can be coaxed into becoming brain cells. These brain cells can be used to study dup15q as well as test potential therapies. My lab plans to make stem cells from some of the samples. There also may be uses for the DNA or cells that we just haven't thought of yet. It's nice to have the blood resource there for these new ideas"

There will be no cost for the families and you can sign up in advance on our website. It is certainly not required, but we are trying to make it convenient for families to provide samples to this valuable resource. For those who do not plan to attend the conference, you can still send a sample to Coriell. Information is available at:www.dup15q.org/research/genetic-cell-repository/

Dup15q Registry

Please bring copies of the most recent EEGs, sleep studies and genetic reports. We will be uploading them on site to the Registry. We will also offer curb side counseling to go over your genetic report and help you understand it better. Start collecting your documents today.

Tooth Collecting Kits

Tooth collecting kits will be available for families who are interested in donating newly lost teeth for cell research to take home with them. More information is available on the study at www.dup15q.org/research/current-studies/neuronal-stem-cells-from-dental-pulp-for-study-of-neurogenetic-diseases

Face Base

A group of doctors and scientists at UCSF is carrying out an international study to better characterize genetic syndromes that include craniofacial abnormalities. The aim is to develop a system to help physicians diagnose these disorders in the future. They are building a database of special three-dimensional (3D) facial photographs of people with different syndromes that can be used to precisely measure and define facial abnormalities and understand the underlying biological problems.

The images will become part of the National Institutes of Health "FaceBase" database www.facebase.org, and will be available to assist other qualified scientists who are working to better understand these syndromes. For conference attendees, participation involves meeting with the study representative to discuss the study, sign consent forms, and take about fifteen 3D photos of the patient. The entire process will take about 20 minutes.

Dup15q Syndrome Research Update

Dup15q Alliance Registry Rylie McHam, Board Member

It's a perfect time to create a Dup15q Alliance Registry account or update an existingRegistry account! The Dup15q Alliance International Medical Registry is an important tool that allows families to help increase our understanding of dup15q syndrome. The Registry is a comprehensive data base that can provide a better understanding of the characteristics of dup15q syndrome, determine areas that need further research, and contribute to the identification of targeted treatments to help improve the lives of those affected by dup15q syndrome. Registry participants have found various components of the Registry to be very helpful. For example, it is possible to print off information and graphs to share with doctors and therapists. Participants can also compare their results to those of other participants.

Laina Lusk, a research assistant at Geisinger ADMI, is currently analyzing the Registry data for an article, helping our community to better understand the data. It is an exciting time for the Dup15q Alliance Registry! Every account matters so please consider creating an account and please update your existing account if you have not done so within the last year. Visit the Registry website at dup15qregistry.org and should you have any questions, please email Rylie McHam at ryliemcham@gmail.com

Dup15q Clinics Jane Kim, MD Board Member

We have exciting news to share. The dup15q clinics database is up and running! The dup15q clinic in Memphis, Tennessee is the first dup15q clinic to start using the database. As more patients are seen, there will be refinements and adjustments that will need to be made. We hope to start using the clinical database within the next few months at the Boston, Los Angeles and Minnesota clinics. This database will help our dup15q clinics combine their information and findings together so that we can learn more about dup15q syndrome. Our clinicians remain hard at work trying to make sure the appropriate data is being collected. We hope to have enough data to report some preliminary findings later this year.

Rare Epilepsy Network Kadi Luchsinger, Executive Director

Dup15q Alliance is hoping to recruit at least 100 people with chromosome 15q11.2-13.1 duplication (dup15q) syndrome for the Rare Epilepsy Network (REN). The REN is a patient-powered and patient-centered research network that will expedite research into the rare epilepsies. The REN is led by the Epilepsy Foundation in partnership with 10 rare epilepsy organizations, including Dup15q Alliance, and Columbia University, New York University and RTI International. All individuals affected by dup15q syndrome who have had at least one seizure are eligible to participate. If you are a parent or legal guardian of a person with dup15q syndrome, please consider participation in this important research project. So far 34 individuals with dup15q syndrome are enrolled in the REN so we are 1/3 of the way to our goal. Please help us get to 100 participants.. You can access the REN at ren.rti.org.

Coming Soon: Dup15q Documentary

Jo Ann Santangelo is an Austin, TX based documentary photographer and film maker who spent four weeks documenting four New England families and their children with dup15q syndrome for a documentary short film and public service announcement (PSA) for Dup15q Alliance.

During those weeks Jo Ann captured the daily life of the children in and out of their homes (school, doctor, and therapy appointments), interactions with family, friends, strangers and themselves. The day in the life footage will also be paired with interviews of the parents. The videos will provide a look into daily life of these families, the struggles, the highs and lows of a having a child with dup15q syndrome, and the different ways this little known syndrome affects each child. The goal of the film and PSA will be to help put a human face on and expand societal awareness of dup 15q syndrome and the Alliance. The PSA and film will be available to view online (YouTube, Jo Ann's website and on the Alliance's site).

Dup15q Alliance

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Cheers to Our Volunteers!

Sandy James

Sandy James ran the JFK50 Miler in November 2014 to benefit Dup15q Alliance. This was her third event of 2014. We asked Sandy to share a little bit about what motivates her to combine her passion for running with her support for the Alliance.

What is your connection to Dup15q Alliance?

My 8 year old nephew, Nicholas Stokes, is a child affected by dup15q syndrome.

What activity did you do to benefit the Alliance?

In June, I ran a 50 mile ultramarathon to raise money for the Alliance. I also hosted a virtual race where anyone could participate to raise additional funds. Participants could run a 5K, 10K, or a half marathon (13.1 miles) at their own pace, on their own time and receive an awesome medal for doing so. On November 22nd, I ran another 50-mile ultramarathon in the historic JFK50 race to try and raise more money for the Alliance.

How do you hope your activity will make a difference?

Every time I incorporate running with raising awareness for the dup15q syndrome community, I help educate people on what dup15q syndrome is. So many people are unaware and slowly, those around me are becoming more and more aware. I have also utilized my "So What? I run." blog to raise awareness of dup15q syndrome. I feel that the more people hear about it, the more the Alliance can get support for research and services.

Would you encourage others to volunteer in this way?

I think running or walking is a great way to raise funds as people are becoming more active to stay healthy. However, I encourage families to utilize whatever they are passionate about to raise awareness. I am very passionate about running. Even those who know me who are not runners are always interested in hearing about my running activities because I'm passionate about it. I feel that if you are excited about something and use that excitement to raise awareness for the Alliance, people are more willing to listen and either participate in the activity to help raise funds or donate if they are unable to participate. For me, it's about making it fun for me and those who participate. Fundraising should never be a chore.

Reflections from a Grandparent

Ella Rearranges My Life

By Geraldine Puttick, Nana to Ella Lawson

Geraldine lives in Brisbane City, Queensland, Australia

I am thrilled to be asked to provide some of my thoughts on being the grandmother to a child with this genetic imbalance.

From the minute Ella was born I have loved her, and my love for her grows immeasurably. She has made me a better person. Ella's journey hasn't been easy for her, but each day she accomplishes new skills. She is a quick learner and can mimic and recall almost all that she sees. She has the most amazing musical ability and can grasp and repeat any little tunes that you sing to her. The earliest song that we sang to her was her Granny's song, "Daisy, Daisy, give me your answer do." She loves this song and often we sing this as we go off to sleep.

Ella is a very assertive little girl and this is one of her many strengths. She is determined in everything that she does, like rearranging my pantry, my scarf draw, my shoe rack, the bathroom and even my underwear drawer. She just delights in this and at first it seemed she would never tire of dismantling things. But now she is learning that there are more interesting things to do like drawing, helping with the washing, and watering the garden.

She loves water and has taken to swimming in the pool whenever she visits. What is amazing is that I have found I am now enjoying the water too!

Ella is so affectionate. She has moved on from the scratching/biting thing in a big way and only occasionally do we see this. It is usually with a stranger who gets too close, but even that has almost disappeared. Now she greets everyone with a big kiss. She visits her great grandmother on a regular basis and when she enters her room insists on climbing onto her Granny's bed to give her a big hug. She knows who Granny is and understands how special she is. You can see the empathy in photographs of the two of them!

The most astounding thing I have noticed with Ella is that it's almost like an adult is within her little mind. She has a deep sense of affinity and understanding and her grasp of certain bits of vocabulary is a mystery. I said to her "You're a funny girl" and she said "Hilarious"; when I said "That's silly" she said "Ridiculous".So, what do you make of that? I am truly in awe of her and I have great confidence that with continued love from those around her she will go far.

Reflections from Siblings

She Always Shows People She Loves Them

By Kelley and Lucas Mullen

Kelley Mullen

My name is Kelley Mullen and I am 25. My parents met in the Peace Corps; I spent my first two and a half years living in West Africa. I got malaria with high fevers which caused seizures. I took strong medicine, which may be why my brain doesn't process things very quickly today.

When we came home, Mom was pregnant with twins. I found out I was going to have a brother AND a sister. When we brought them home from the hospital, I asked Mom when they were going to be returned BACK to the hospital. Soon I learned that it was great to have a big family.

My brother Lucas could do many things early on, but my sister Kathleen was much slower. I stayed home with Dad and the twins when Mom would teach. I played with them, swang them, and hid them from Dad under all my toys. When Kathleen was two we found out she had something wrong inside her. For me nothing changed; she was my sister and Lucas was our brother.

When Kathleen started having seizures, after Mom and Dad divorced, I helped out a lot. I was going to a special school and knew many of the teachers my sister would have as she got older.

Now my brother lives in San Francisco, so we only see him a few times a year. I share a room with her because I need to help Mom when I hear her having a seizure in the night.

I love my sister, but wrote my parents a letter asking for a break now and then from her so I can have one on one time with Mom or Dad. I try to accept that whenever we are out in public or at the mall she is very loud and everyone stares. At church I am patient with her, too.

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Mom is impressed with how I take care of myself, as she is always busy with Kathleen. During vacation I let them sleep in and got up on my own, made my lunch and waited for my taxi. When I'm home I love to get Kathleen off the bus.
Kathleen loves to go to McDonald's for a Happy Meal box when she has a good day at school or at church. She loves to go to family functions. We have to keep it secret or she will drive us crazy talking about going to see Grandma and Grandpa. It's fun when Mom says where we are going, because she gets so excited.

My sister loves Barney videos, so I play them for her at the end of the day.

Though she can embarrass me, I love her no matter what, because she is always so happy and loves me. She is a lot like me, with the seizures, although hers are more often and strong and violent. I need to help my mom when Kathleen goes onto the ground, to get a pillow or help hold her legs down. I am always happy when she comes out of it and smiles. I can't imagine my life without her. I don't see her disability or mine, just that we are sisters!

Kelley and Lucas are sister and brother to Kathleen. The family lives in California.

Lucas Mullen

Growing up in a house full of women gives a young man perspective; a sibling with special needs opens your eyes even more. Kathleen, my twin sister, has always exuded a childlike personality. To me, it was completely normal when we were young. As I got older, my age caught up with me. With Kathleen it was different; she's still young at heart. Her innocence of the world around her grants her a blissful life.

I now live away from home in San Francisco. I knew Kathleen missed me lots after I moved out. Mom said she explained that Lucas had gone to live at school. I was amused when Mom said they drove by my old elementary school and Kathleen thought I was there. They would drive by and Kathleen would yell out, "Hi LUCAS!"

Once I returned home from college for the holidays. My mom helped me hide under a blanket to surprise Kathleen. She was told a present was waiting underneath, so she whipped the blanket off revealing me, prompting her to say, "Hi Lucas, where is the present?" She always makes us laugh, and often shares that with everyone. We grew up attending St. Lawrence Church in Sacramento every Saturday night. Kathleen was always excited to see who she called "the king" (our pastor). Kathleen's voice outshines the choir during the reading of the Psalms, and you can always hear her chant over everyone, "the king, the king". After mass, she would run up to hug him. Kathleen's emotions allow her to appreciate things even more, and she always shows people she loves them. If you think you know Christmas cheer, you don't know the meaning until someone says "Santa" around her. Our family has found much joy in having a child with idic(15).

We've had our share of struggles; my family has been thrown a lot of curveballs. But just like Kathleen, we've learned to appreciate the fortunate life we have been given, and the happiness it grants us.

Family Portrait

Arielle's Story

By Angelic Yao, Atlanta, GA

Who is Arielle? She is a twin and my only daughter. She is the shining star that greets me every morning with the most beautiful and innocent smile! Arielle cracks me up with her silly laughter and makes my day when she looks at me with her beautiful brown eyes and mumbles "ma ma ma" with all her strength. She is sweet. She is strong. She is determined. She is mommy's sweet little angel!

My sweet angel and her twin brother were born in September, 2011, with minimal complications. The only concern was an excessive amount of amniotic fluid found in Arielle's sac. This condition is known as polyhydramnios, and sometimes indicates a birth defect. The doctor consulted with my husband and me and asked for consent to do an amniocentesis. We declined, given the associated risk of miscarriage. For the remainder of the pregnancy, I was monitored closely until I successfully delivered Arielle and her twin.

At home, I enjoyed watching my babies develop and grow as expected until their third month. This was the time when my husband suspected that something was not quite right with Arielle. She was very reserved and often stared into space unbothered by her surroundings. As a protective mom, I was annoyed that he would think that something was wrong with my sweet girl. However, as time went by I also started noticing that she was not developing at a similar pace as her twin brother. She was not making any attempts at sitting up or crawling, nor was she displaying much interest in age appropriate toys. This prompted us to discuss our concerns with her pediatrician. She did not seem to be concerned and told us not to worry because Arielle was in good health, and that multiples tend to have different developmental pace.

By Arielle's ninth month, my husband and I, along with the pediatrician, agreed that it was time to figure out why Arielle was not meeting her developmental milestones as expected. We started with the state's early intervention program. After her evaluation, she qualified and started benefitting from PT, OT and speech therapies. She was also referred to what seemed like every specialist in the medical field. She was subjected to a series of medical tests including an MRI and CT scan. However, all the results came back negative or normal. We were quite frustrated and could not understand what was happening. By all accounts, these series of doctors' visits were overwhelming for Arielle and the rest of the family. The going back and forth with three little ones was very exhausting. I remember having to always maintain a strong positive composure by day, but I cried myself to sleep every night.

Things started looking up as Arielle was making great progress in therapy and the follow up home sessions. Within a month of PT sessions, she was sitting in an upward position. She started crawling around 13 months. In her OT sessions, she learned how to grasp toys, make eye contact and respond to various noises. With speech/feeding therapy, she started making more sounds and managed to pick up Cheerios and other finger foods at 18 months. Once I started noticing the progress she was making in therapy, I discussed with my husband and we decided to cut back on the testing and doctors appointments after the recommended genetics testing.

A breakthrough to our long quest of finding answers to our daughter's medical issues came in the summer of 2013, when I received the call from her neurologist. She informed me that the report was in and it indicated that Arielle had a duplication on her 15th chromosome (Isodicentric 15). As any parent would be under this circumstance, I was devastated by the news. I had never heard of this syndrome, and learned that there is no cure. This was the beginning of a journey to a new life, which continues to unfold without having much control over its potential destination.

This situation has shaped the lifestyle of the entire family. We all have had to adjust schedules and sometimes we are unable to attend events and gatherings. Adjustments have been made to our diets due to her allergies. Right now we are dealing with many nights of "happy" insomnia, which often lasts from two to six hours. In general, Arielle wakes up happy during the night and ready to play. She is also suffering from allergy related issues despite having her tonsils and adenoids removed. She requires thickening powder for foods and drinks. At three years old, Arielle is currently functioning in the 6-12 months developmental stage, therefore requiring constant attention. We have altered our home to ensure that she is safe and secure in her familiar surroundings. She often bites as a way of communicating or expressing her frustration. She enjoys music, dancing, and riding the bus to school. She loves eating a variety of foods. She enjoys standing by the family room window looking outside at the sunlight. She loves cuddling.

Overall, Arielle has a fighting spirit and has been teaching us how to be resilient and foster our family bonding. She is protected and loved by a host of family and friends, especially her three brothers. I do not know what the future holds for my little sweet angel, but I strongly believe that the Almighty God will supply all her needs and allow her to live a long happy life filled with love!

THE MIRROR

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Dup15q Alliance is a nonprofit organization that provides family support and promotes awareness, research and targeted treatments for chromosome15q11.2-13.1 duplication syndrome **(dup15q)**.
Dup15q Alliance offers help and hope for those with dup15q syndrome.

UPCOMING MEETINGS AND ANNOUNCEMENTS

$15,000 in 15 Days
This annual fundraiser begins on Valentine's Day and ends on Rare Disease Day.

Rare Disease Day February 28, 2015
On Rare Disease Day please wear blue for Dup15q Alliance, and consider planning an awareness event in your school or community. Brochures and flyers are available online at www.dup15q.org/fundraising/publications/.

8th International Family Conference : *STRONGER TOGETHER* July 30 - August 1, 2015
Hilton Orlando Lake Buena Vista, Orlando, FL
Everything you need to know about making reservations is available at
www.dup15q.org/events/family-conferences/stronger-together-2015/.

First Annual Dup15q Alliance Believe Walk September 13, 2015
The Alliance is hoping to have 10 sites around the country for the first annual Dup15q Alliance walk. Please contact events@dup15q.org if you are interested in hosting a walk. We will help all site coordinators in setting up and running the event. Be part of the inaugural Dup15q Alliance Believe Walk. Together we can make a difference!

http://www.dup15q.org

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