

THE MIRROR

The Newsletter of **DUP15q ALLIANCE**

Learning Together



Fall is the season that families with school age children focus on learning. Families who are impacted by dup15q need this commitment to learning to be a lifelong one. There are many things about dup15q syndrome that remain unknown, and we are all learning together. We are challenged to learn about the genetics of this disorder, the best treatment options, the best strategies for academic, social and emotional growth of our affected children, and effective ways to support the social and emotional needs of our family.

This issue of *the MIRROR* provides a rich array of how families, clinicians and scientists are learning together about dup15q. Guy Calvert and Jane Kim provide updates from this year's science and dup15q clinics meetings. These summaries are exciting, not only for the breadth of work underway but also for the energy and momentum moving forward into the future.

Paul Karch writes about the Dup15q Alliance Board's strategic planning and the importance of the Alliance membership's feedback provided through the July survey. The Dup15q Alliance Board is learning along with scientists, clinicians and families. This learning is translating into clarity about the organization's mission and long term goals.

The family portrait in this issue, written by Ed Gibbons is a beautiful testament to the learning opportunities that exist for both parents and children, and for school communities when our children affected by dup15q are introduced to new school environments.

Learning acceptance and a sense of peace about dup15q is a challenge every family faces, especially for the siblings and grandparents who aren't as directly involved in the care of a child with dup15q as the parents. The sibling story by

BY NICOLE CLEARY
EDITOR

Emma O'Reilly relates the heartfelt experience of learning to accept her brother's disability and appreciate the incredible gift he is in her life. Margery and David DeVilder describe the journey of grandparents who work hard to learn what words like "isodentric" mean, and to understand nature of their grandchild's condition.

The Dup15q Alliance is committed to learning together with families, scientists, and clinicians. To that end, the Alliance is launching a series of webinars that will allow up to 100 people to learn about topics of interest to families. You can find information about these learning opportunities on the back page of this issue.

The need for lifelong learning is part of the dup15q experience. Our families are fortunate to have so many bright, caring, articulate, passionate people within and associated with the Alliance, making the road to learning a little easier for all of us.



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Chromosome 15q11.2-13.1 duplication syndrome (dup15q) 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 isodentric 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.

Science Meeting: Two Sides of a Coin

BY GUY CALVERT AND JANE KIM, DUP15Q ALLIANCE BOARD

In the spirit of scientific collaboration, our annual scientific meeting - held in Boston this August - was something of an experiment. For the first time, we partnered with the Angelman Syndrome Foundation to put on a joint scientific meeting. Dup15q and Angelman have much in common at the molecular level: dup15q syndrome is caused by too many copies of genes in the critical 15q11.2-q13.1 region of the maternal strand of chromosome 15, whereas Angelman syndrome results from the deletion of those genes. However, the clinical manifestations of these syndromes include both striking similarities and stark differences. For example seizures, intellectual disability, and profound challenges to the development of language are common characteristics of both syndromes, but individuals with dup15q syndrome have different sorts of motor challenges, are much more likely to develop features of autism, and are at far higher risk of SUDEP. The hope is that both the similarities and differences can provide insights to help researchers tease out the mechanisms of action for these disorders. So our "Two Sides of a Coin" meeting was intended to facilitate the cross-fertilization of ideas between professionals that are focused on one syndrome or the other, raising awareness and bringing more scientists and clinicians into the field of dup15q research. At least in terms of the raw count of attendees, the experiment was a success with 88 people registering for the meeting, significantly more than turned up to either individual organization's past scientific meetings.

Research into 15q11.2-13.1 is moving forward at an exciting pace, both in the realms of bench science and in clinical science. The first set of presentations focused on new developments in

bench science, and many of those were concerned with *UBE3A*, a key gene in the critical region whose deletion is known to be sufficient to cause Angelman syndrome (AS) and which is also suspected of driving at least some aspects of dup15q syndrome. This gene is *imprinted*, meaning that its expression depends on the parent of origin, and it turns out that only the maternally inherited copy of *UBE3A* is active in neurons. It provides instructions to produce a specific protein - also written as UBE3A, but without the italics. (In case you're curious, the long form scientific name is "ubiquitin protein ligase E3A".) One known function of UBE3A is to mark other proteins (the so-called *substrates* of UBE3A) for degradation. The substrate theory of these disorders, as articulated by Seth Margolis in his talk, holds that at least some features of dup15q syndrome are caused by too few substrates in the cell, whereas Angelman syndrome is caused by too many substrates in the cell. It's a simple and elegant way to look at the problem, and it enables scientists to frame questions whose answers are very relevant to families: as Larry Reiter summed it up later in the meeting, the development of biochemical treatment options comes down to identifying the right targets - druggable targets - and substrates are an important clue within the wider search for those targets.

If the substrate theory holds, then it is natural to ask: which proteins are targeted by UBE3A for degradation and, of those substrates, which ones matter for each syndrome? Scientists at the meeting approached these questions from several angles - animal models, post mortem brain tissue and human stem cells.



Animal Models Margolis presented his data from experiments with Angelman mice, highlighting the interaction between UBE3A and the transcriptional regulator ATRX, as well as speculating on the role of another protein (called Ephexin-5), which is expressed in the hippocampus and acts to decrease the density of dendritic spines in neurons. Ugo Mayor, working with fruit flies, gave an update on the data he presented at last year's dup15q meeting in which he had identified a substrate of UBE3A in flies. Mayor is now eager to repeat his experiments using mouse models of dup15q.

Post Mortem Brain Tissue Grant Belgard gave an update of his data from RNA-seq gene expression analyses of post-mortem autism and dup15q brains. According to Belgard, there's a significant signature of gene over- and under-expression in brains from individuals with idiopathic autism, a signature which is even more pronounced in dup15q brains.

Human Stem Cells Larry Reiter described his work with neurons derived from dental pulp, whereas Stormy Chamberlain discussed insights from neurons induced from pluripotent stem cells and cord blood. Reiter found notable differences between neurons from dup15q versus Angelman donors, as well as 123 genes that are differentially regulated in dup15q neurons versus controls. Chamberlain compared the protein expression profiles in neurons derived from Angelman and dup15q, as well as controls. Of the differentially expressed

proteins encoded by genes outside the critical region, many were expressed in the same direction for both syndromes. Also working with Chamberlain's iPSC neurons, Eric Levine discussed their electrophysiological properties in Angelman syndrome, and Olena Marchenko outlined her work to characterize the neuron structure. This is all exciting and promising work - echoing Carolyn Schanen's complaint from the dup15q science meeting two years ago: nobody has any idea yet what dup15q synapses look like. With the help of stem cell technology, that could be about to change.

Scientists had speculated that the imprinting of *UBE3A* developed through natural selection as a mechanism to regulate gene dosage. Taking an evolutionary approach Scott Dindot ultimately rejected this hypothesis, presenting the opossum as a counterexample. There were also several presentations that touched on other genes in the critical region, or on novel models of behavior. Stephen Moss discussed mouse model experiments focused on differential expression of GABA-A receptors. Janine Lasalle explored the complex interactions of genes involved in brain development with organic environmental pollutants. Richard Mooney presented insights from songbirds as a model of language learning that could be applied to molecular changes in humans for proper expressive language. And in a tantalizing preview of what lies ahead for Dr. Dindot's dup15q mice, Jill Silverman walked us through the cutting edge battery of behavioral tests that she uses to characterize mice sent to her lab.

Clinical Research

On the clinical front, Shafali Jeste discussed her new pilot study, which was funded by the Dup15q Alliance. She is characterizing behaviors and development in children with dup15q syndrome through cognitive and behavioral testing, as well as EEG testing. She hypothesizes that individuals with dup15q syndrome have expressive language delays and motor impairment with relatively preserved nonverbal social activities. After characterizing around 20 children, some of the children will be chosen for a targeted intervention study to be conducted by Dr. Connie Kasari, a clinical psychologist at UCLA. Dr. Kasari also spoke at the conference and described her JASPER model of intervention, which stands for "Joint Attention, Symbolic Play, Engagement, Regulation." Depending on the results of this pilot study, Dr. Jeste may be able to apply for additional funding to spread this model to other dup15q clinics, many of which were represented by individuals at the meeting.

Orrin Devinsky gave a fascinating talk on the history of and potential future use of cannabinoids in the treatment of intractable epilepsy. He also presented early insights from open-label trials of Epidiolex, mentioning that one dup15q individual had a decline in seizures of more than 90%.

Ed Cook presented data clarifying the very small increased risk for autism in individuals with small duplications on 15q just proximal to the critical region duplicated and deleted in most AS or dup15q cases. These so called "edge duplications" may be more accurately described as variants of unknown significance, but they do appear to contribute, if only slightly, to increased risk for autism.

Ron Thibert gave an important and revealing talk that exemplified the reason for this joint meeting. Dr. Thibert - a clinical neurologist specializing in epilepsy - directs the Angelman syndrome clinic and Dup15q Center at Massachusetts General and serves on the advisory boards of both the Dup15q Alliance and the Angelman Syndrome Foundation. Thibert compared and contrasted the features of epilepsy in both syndromes which he himself had been working on independently. Infantile spasms are much more common in dup15q syndrome, and more generally there also seems to be a larger range in the severity and tractability of seizures, whereas in Angelman syndrome there is not as much variability. The type of medications used to treat seizures in dup15q syndrome and Angelman syndrome also differ. Seizures in Angelman syndrome respond well to benzodiazepenes, but these types of medications have almost no effect of seizure relief in dup15q syndrome. Seizures in dup15q syndrome often respond to antiepileptic medication that is directed towards focal seizures, such as carbamazepine, while this can exacerbate seizures in Angelman syndrome. Clearly there are two faces to the coin when one considers seizure treatment and management in AS versus dup15q syndrome, but they are effectively the same coin (i.e. they respond in opposite ways).

We asked speakers for consent to post videos of their presentations on our website. Some of those videos have now been uploaded, and more will be coming soon. Check them out here:

<http://www.dup15q.org/events/scientific-conferences/two-sides-of-a-coin-deletions-and-duplications-on-15q-2014/videos-and-schedule-of-events/>



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Dup 15q Clinics Meeting Report

By JANE KIM, DUP15Q ALLIANCE BOARD

The dup15q clinics' meeting was held on the morning of Saturday, August 16, 2014. There were clinicians that flew in from 6 out of the 9 dup15q clinics, including Boston, Los Angeles, Memphis, Minnesota, New York and Seattle. One of the main topics of discussion was the new dup15q clinical database. The clinical database will be piloted at the Memphis clinic first since that is where the database is housed and the medical bioinformatics experts are located. Afterwards, the database will roll out in a few additional sites, where further refinements can be made. The new database will allow for physician reviewed data to be collected and shared among different sites. We hope the database will be in use at some of the clinics by the end of the year.

Best practice measures were also discussed, such as EEG guidelines and neuropsychiatric testing recommendations. Clinicians also talked about common findings throughout the clinics. One observation has been the common presence of alpha delta waves in sleep. Another finding has been continuous spikes and waves during slow wave sleep, as well as high amplitude bursts of rhythmic activity during sleep. These common findings are of uncertain clinical significance but may play a role in SUDEP, sudden unexplained death in epilepsy. Future studies need to be done to see what these findings may mean. Other new research ideas were discussed as well, such as exploring the science of epileptogenesis at a microRNA level.

It was exciting to have so many clinicians in the same room discussing best practices and research ideas for dup15q syndrome. Many of these busy clinicians flew in for a half day meeting, and it really showed the dedication and commitment of our dup15q doctors. Going forward, there is a lot of excitement for further collaboration among the dup15q clinics.



From Left to Right
Heather Lau (NYU), Marcio Sotero DeMenezes (Seattle, WA), Naga Nagisetty, Clinics Database Developer (UT Memphis), Larry Reiter (Alliance Professional Advisor), Ed Cook (Alliance Professional Advisor), Dimitrios Arkilo (St Paul, MN), Jane Kim (Alliance Board), Shafali Jeste (UCLA), Ron Thibert (Boston, MA Clinic and Alliance Professional Advisor), and Kathryn McVicar (Memphis, TN).

THE ALLIANCE BOARD WELCOMES ADRIENNE CAMPOLMI



Adrienne Campolmi joined the Dup15q Alliance Board. We want to give our families a chance to learn a little bit about the Campolmi family.

Our son, Jackson (age 12) was recently diagnosed in January 2014 with Dup15q. Jackson is a wonderful little boy, and we call him our "Little Buddha" as he possesses a go with the flow personality

and is full of love and joy that he expresses through his smile and enduring affection.

As soon as my husband and I were given the diagnosis of dup15q we knew immediately we wanted to be involved with the Alliance in some capacity. Upon receiving the diagnosis we joined the family support network and familiarized ourselves with the Alliance. In an effort to become involved we decided to host a fundraiser this coming fall where the proceeds will benefit the Dup15q Alliance. Our intention is to have the success of our inaugural event become an annual event. Additionally, I was honored to be asked to join the Board. As a new board member I hope to utilize my skills

and experience to provide value and input to the Dup15q Alliance Board.

Our family lives in Charlotte, North Carolina. My husband Chris and myself, in addition to Jackson, have a second son named Caden (age 9). Our family enjoys doing all things active and social. We love the outdoors and we make sure to include Jackson in all of our family adventures to the best of his abilities (hiking, biking, swimming, horseback riding, surfing, white water rafting) you name it and we try it. We love to host parties and can use any holiday or event as a reason to celebrate with friends and family.

Professionally, I have been employed with IBM for the past 16 years. Currently, I'm a Project Manager and mentor in the Global Services division.

Dup15q Alliance Board Report

By PAUL KARCH, DUP15Q ALLIANCE BOARD CHAIR

The Dup15q Alliance Board met on August 16 and 17, 2014, first for a strategic planning session and then for a regular board meeting. The board held its meeting following the very successful scientific symposium sponsored by the Alliance and the Angelman Syndrome Foundation which is described on page 2 and 3 of this issue of *the Mirror*.

The Alliance Board is made up of 11 parent volunteers, one of whom, Kadi Luchsinger, is also paid for part-time work as the Executive Director of the Alliance. The Board meets monthly by conference telephone call but finds it very important to meet in person at least once each year in order to know and understand each other face to face. We welcomed two new board members this year, Jane Kim and Adrienne Campolmi.

In the strategic planning session on August 16th, the Board reviewed information provided by Alliance Members through the survey conducted in July. Major findings from the survey include:

Alliance members rank providing family support as our most important goal.

The Board then developed five-year goals as a way of helping us think about action plans.

These goals include:



1. Supporting research through at least \$500,000 per year in direct grants and hiring a research director.
2. Expanding and promoting a comprehensive registry to include both family and clinic information.
3. To continue to support and expand the international family conferences and regional gatherings.
4. To continue to provide information and support through direct communications, such as the Mirror, and social media and expand offerings to include educational webinars.
5. To expand collaboration with international organizations of dup15q families.

We also considered how best to develop and allocate resources to meet those goals. The Board will develop, with the help of families, and adopt specific action plans for staffing and fundraising to ensure the continued growth and effectiveness of the Alliance.

In the Board meeting following the strategic planning session, we addressed general organizational issues that are current right now. The Board confirmed the theme for next year's Family Conference in Orlando as Stronger Together. The Board also determined to maintain the same \$150 registration fee for the 2015 Orlando Family Conference in order to allow more families to attend. The conference will include presentations by parents, families and professionals sharing their experience and learning. Further details will be provided in future issues of the Mirror.

The most important issues to families are seizures and behavior, followed by school and family support

Members read Alliance communications - over 75% read every issue of the Mirror, 50% carefully.

Members like regional gatherings and conferences, although cost, distance and travel with a dup15q child make attendance difficult for many.

Around 25% check our Facebook pages regularly while over 50% visit them rarely or never.

Members find most Alliance activities helpful.

This information was helpful to the board and we appreciate all families who took time to provide feedback.

The Board confirmed the current mission of the Alliance to provide family support and promote awareness, research and targeted treatments for chromosome 15q11.2 to 13.1 duplication syndrome. The Board agreed that all three parts of the mission, family support, awareness and research, are important and interrelated.

We also discussed continued support needs for the Dup15q International Registry and development of the Dup15q Clinics Registry. These registries are complementary and both fill an important role in helping characterize dup15q syndrome and effective treatments.

Finally, the Board discussed the need for the Alliance to focus its limited resources on projects and activities that will fulfill its mission of providing family support and promoting awareness, research and targeted treatments for chromosome 15q11.2 to 13.1 duplication syndrome. The Board described this focus in a letter to the Members of the Alliance, which will be sent to Members by mail and email.

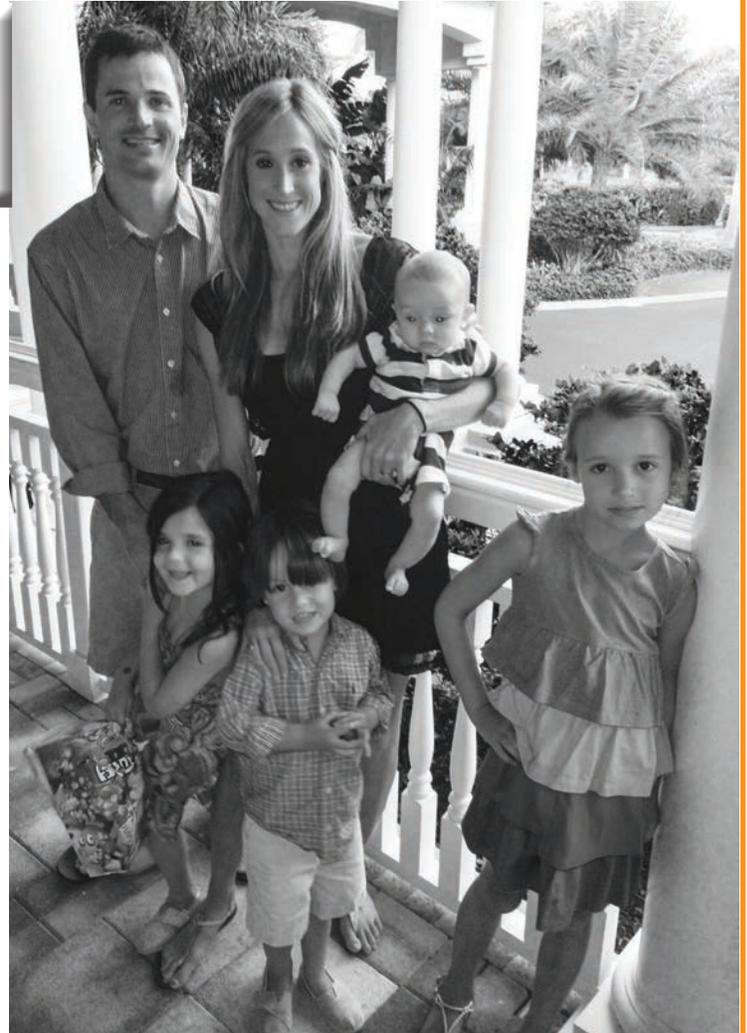


Cheers to Our Volunteers!

Thank you Dana Tilton for your service on the Dup15q Alliance Board, and your continuing role as the southeast regional representative.

It's a bittersweet feeling, wrapping up my time served on the board of directors. I pictured myself serving on the board indefinitely...as long as they would have me. From the moment I found Dup15q Alliance, I knew I wanted to be involved on a greater level. Just being a part of this group inspires me and makes me want to be a part of the difference we are surely making for our children, and the children diagnosed in the future.

I served on the board from June, 2012 through June, 2014. When I joined, I thought I was going to have more time on my hands since our youngest was soon to be headed to preschool. As we all know though, life throws us curve balls and I found myself expecting another child around the one year mark of my time served on the board. While I would love to still be a part of such an inspiring board of directors, I simply don't have the time....at least for now.



In my time on the board, I realized just how amazing this group really is. The board of directors is where decisions are made, ideas are presented, concerns are dealt with, conferences are planned, research is guided, fundraising goals are put in place, family support is discussed, and a group of passionate and devoted dup15q parents pour their heart and soul into a better tomorrow for our families and children. I gained more knowledge about dup15q and where we are headed than I could have ever imagined. When it comes down to it, our board of directors is simply changing the future, every day, for families affected by chromosome 15q duplication syndrome. Why wouldn't you want to be a part of that?

I, for one, couldn't resist. We need more help though. We need more parent volunteers that are ready and willing to make a difference. The time it takes benefits your child and the rest of the dup15q family. Whether you want to send birthday cards, raise money, or help drive research, there's something for everyone. Be a part of it. Be a part of creating a better future for our children. The more volunteers we have, the faster we will attain the goals we have put in place.

I will treasure my time spent on the board. I will continue to serve as the regional representative for the southeast and raise funds for our group in any way possible. I hope to be back on the board in a couple of years when life has settled down a bit. Where will you be? I hope we can all make a difference together. After all, that's what this group is all about. I'm beyond grateful for my experience. It's simply irreplaceable. Thank you to all that continue to serve; you are a blessing to us all.

The Reasons We Smile

We celebrate the life of Tyler Kobuszewski with the reprint of this poem written by his brother, Collin.

We smile when you say “oh no” after making a mess
 We smile when you ask for your 5th breakfast sandwich
 We smile when you sing and whistle
 We smile when you throw your stuffed animals all over the place
 We smile when we hear your laugh
 We smile when you pick out your favorite movie to watch
 The neighborhood smiles when we take you on an ATV ride
 We smile at how fearless you are with everything you do
 We smile when you splash and swim
 But most of all, we smile because we know you are in a better place where you will continue to give people a reason to smile; just the way you made us.

We love you Tyler.



Dup15q Alliance

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A Moment To Remember

ED GIBBONS AND HIS WIFE MARY ELLEN ARE THE PROUD PARENTS OF KATHRYN.

Family Portrait

After many years of our nineteen-year-old daughter Kathryn attending a school that educated students with special needs, my wife and I had to find a different placement for her. Our choice was between selecting a similar school that only educated handicapped children or a traditional one that was open to all. Eventually we chose to send Kathryn to Union High School, a comprehensive public school, where I just happened to be the principal. The change would bring stressful moments but our decision turned out to become a wonderful experience for Kathryn.

On Kathryn's first day, I asked one of the security officers to escort her off the bus and ensure that she get to her classroom. I clearly remember hearing over the radio, "Mr. Gibbons, package delivered." He also said, "Mr. Gibbons, she looks very happy." From that moment, I knew we made the right decision. There were still challenges to conquer and valuable lessons to learn, but Kathryn and I were ready.

Union High School is culturally diverse. There are 2,500 students and approximately 250 staff. Kathryn and her classmates were scheduled for physical education, music, and art with the "regular" population. She ate lunch in the cafeteria with all of the other students and that meant waiting in line, paying for lunch, and cleaning up when finished. During the lunch period, Kathryn was exposed to the typical behaviors of high school students; some good, and some not so good. The cafeteria was noisy, busy, and filled with activity. Lunch time for Kathryn exemplified the real world.

Kathryn's program included community based instruction that had her leaving school and working at Kean University's cafeteria where she cleaned tables, going to a pizzeria where she folded pizza boxes, and travelling to Stop & Shop where she stocked shelves. Her days also included self-contained instruction in language arts, math, computers, and life skills. Her teacher arranged many field trips that exposed

the students to the abilities needed to handle the challenges of everyday life. Using public transportation, ordering food at a McDonalds, and dealing with strangers, were some of the lessons taught on the trips.

Kathryn knew that being the daughter of the principal would not afford her any privileges. She was instructed to acknowledge me as Mr. Gibbons, not Dad. Similar to any student entering a new environment, there was a period of adjustment. The students in her class had been together for many years and Kathryn was an outsider. Navigating the halls was a challenge and she was expected to get to her classes on time. One day she was startled by two students shouting at each other in the cafeteria. I heard about those students for several days during dinner.

Gradually Kathryn gained confidence. She even improved at calling me Mr. Gibbons...although I would smile when she would error and call me Dad. Eventually cafeteria events did not alarm her and she enjoyed walking throughout the crowded building. Kathryn's successful transition to her new school resulted from the nurturing and accepting environment that existed. Union High School's students and staff had plenty to do with that.

Kathryn and I made an agreement. If she had a good week, she would leave with me on Fridays rather than take the bus. On those afternoons, before we headed home, Kathryn and I would walk the halls together. Kathryn loved being in the halls saying hello to everyone. As we made it from one end of the building to another, students that I did not even recognize would say hello. We would hear, "Hello Kathryn...Hey Kathryn, good time in gym today." Teachers would wave to her and say, "Have a nice weekend Kathryn!" As we continued these Friday afternoon jaunts, the greetings became more frequent. They were also directed at the same person - KATHRYN! I learned that my daughter was more popular than the principal!

The welcoming environment of UHS added to Kathryn's increasing self-confidence.

Kathryn turned twenty-one years old during her second year at Union which meant that graduation was approaching. Before the year started, I reminded Kathryn that she needed to succeed in school and work hard at everything, if she wanted to graduate in June. Preparing herself for the big day, Kathryn concentrated on all her responsibilities. At Kathryn's final IEP meeting, she was informed that she earned the right to graduate with the Class of 2014!

On graduation day, the seniors assembled in the gym before the program began. The excitement escalated as the seniors displayed pride in their accomplishments. Six hundred students started chanting the year: "1-4, 1-4, 1-4, 1-4!". I looked to where Kathryn was sitting and, with a big smile on her face, she joined the chorus of "1-4, 1-4, 1-4, 1-4!".

I announced to the soon to be alumni that it was time to march onto the field. Again the chant of 1-4 echoed throughout the gym! As I led all of the students out, I could not help but feel a little extra pride for this class. Not only did it include my daughter, but also a group that accepted her and loved her.

After the singing of the National Anthem, my welcoming remarks, and a few speeches, the goal that Kathryn worked so hard to achieve was about to happen. One by one, the students were called and presented their diploma. When the speaker announced, KATHRYN GIBBONS, cheers permeated the air as I awarded my daughter her diploma. It was an instant that will forever be seared in my heart.

As the graduates made the traditional toss of their caps into the air, my sunglasses hid my proud tears and I know that my wife, who was sitting in the stands, had a few of her own. As Kathryn left the field after graduation, her warm and endearing smile was testimony to the wonderful experience she had at Union High School.



Reflections *from a Sibling*

Cathal fills your heart with so much love

BY EMMA O'REILLY, SISTER OF CATHAL O'BRIEN

In September 2009, my brother Cathal O'Brien was born. I was 13 years old. I think it's quite honest to say that I had been dreading the day I would become a not-so-only-child anymore. I was so used to being the only young one around, being spoilt, and having all the attention.

Anyway, I walked into the local maternity hospital, dreading to see the new happy family, and I automatically fell in love. I was absolutely smitten with this gorgeous, amazing creature that I would now forever call my little brother. All of a sudden, I felt so proud and happy to say, "Yes, I have a brother. His name is Cathal!". I would brag about how he was the most amazing little cuddle monster. I think the love of a sibling is one you can most definitely not describe in words. When I first held him, I felt so warm and content that he was MY brother. I never ever wanted to let him go. Ever.

On day three of our monster being in this big bad world, we got some bad news. Cathal was sick. He was having trouble breathing and you could see he was struggling. I can never explain how devastating and destroying it was to find out he was seriously ill and in pain. I don't know the ins and outs of what went on, but I know that the doctors said it could be fixed. Cathal just needed to grow and put on a bit of fat to have surgery. I blamed myself for wishing to be an only child forever. I felt so selfish. Now maybe, after falling in love with my little brother, he might be taken away. I regretted it so much, but after his open heart surgery at only six months old, we found out he was a perfectly healthy child again. I was over the moon. We could finally be normal brother and sister again. Cathal was the bubbliest, funniest, cutest little guy (maybe I'm a bit biased, but he was our angel!). He learned to say my name, Mam, Dad, and he particularly loved the word cow for some reason (maybe the effects living in the countryside). We would sing songs and laugh our faces off at our crazy mother who loves to make a holy show of herself.



Cathal got to about nine or ten months old, and Mam suggested he wasn't progressing anymore. She said he wasn't doing what normal kids his age should be doing. In fact, he was kinda' losing a bit of what he had learnt already. I'm sure Mam went to loads of specialists and stuff, but when she tried to tell me he might have

some condition, I thought she was actually ready for the loony bin! She needed to give the guy a chance! I was convinced there wasn't a blessed thing wrong with the little guy. Just because my brother wasn't walking and couldn't hold his spoon as good as the next kid, doesn't mean he had something wrong with him! I got really angry when she would talk about it. I just wanted everyone to stop talking about Cathal and leave him alone! It wasn't fair. He didn't do anything to anyone and now he had to take time out of having fun to go see doctors and be poked and prodded all the time! I denied the fact that Cathal was born with a rare chromosome condition called idic(15). I think it could have been on his second birthday - maybe a bit before - when I went to my room and cried. People would keep talking about what was wrong with my little monster. I wanted it to stop. I wanted it to be fake. But when I realized that other kids his age were running and racing and going

to the normal potty, I saw that maybe Cathal was a little behind. I think for a while I took a step back. I wanted everyone to figure out what was going on so I could help Cathal.

We eventually realized that no matter how severe Cathal's condition may be, how far behind he is, or whether he's in a special or a mainstream school, my brother is the happiest child you will meet. Yes, sometimes things are difficult. It's hard to know what he wants. Is he sick, in pain or hungry? Does he want to play or sing? He can't tell us. Yes, he does get frustrated, cries, and has tantrums like every other kid I know. But, I know for a fact, he is loved by absolutely everyone he meets. He is so adorable, lovable and has the kindest little heart. When he looks at you and smiles or decides to sing "Twinkle Twinkle Little Star", Cathal fills your heart with so much love. In my opinion, my brother is the best gift God could have given to my family and me.

Reflections from Grandparents

She has a winsome way of relating to people

BY MARGERY AND DAVID DEVILDER ARE GRANDPARENTS OF ANNELIESE MORGAN

Our eagerly awaited tenth grandchild entered the world without major complications. My husband David and I joined our daughter Elizabeth and her husband in the birthing room to admire 9 lb. 11 oz. Anneliese Noelle as she was poked and prodded and measured and weighed. Proud Daddy Bryan, keeper of important facts, happily announced the high Apgar score.

She was beautiful and delightful to hold, and we loved her unreservedly. “She sure holds her head up well,” I remember thinking. “That’s a good sign.”

Living four hours away as we did, our times with Anneliese were infrequent. After a couple of visits in the early months, I began to have concerns. It was the pediatrician who recognized and confirmed that something was NOT quite right. At three months she still held her head up well, but we found out it was a peculiar symptom of her condition and that she lacked muscle tone and strength. Finally, genetic tests put a name to it all.

As we learned the pronunciation and meaning of terms like “isodicentric” and “dup15q”, we struggled to grasp the nature of our granddaughter’s condition and tried to imagine her future. We felt compassion for these young parents in their struggle to assimilate the diagnosis. It was only the beginning of their ongoing effort to reconcile their dreams for their little girl with the emerging realities of her condition. At times we could see that their feelings were akin to grief.

We cuddled and played with Anneliese on our occasional visits; we prayed for the best possible future. We watched Bryan lavish love on his “Babygirl.” When he got home from work he held her constantly and was actively involved in her care—changing her, talking to her, coaxing her smiles.

Our hearts ached for them during the times when the challenges were overwhelming. Having reared four children and helped rear several grandchildren, we appreciated the value of their willingness to share cooking and other household responsibilities. Elizabeth enrolled in a course of study at Missouri University designed to equip her to deal with the challenges of the dup15q syndrome and potentially help other families dealing with similar situations. The course work was squeezed into an already full schedule of homemaking and mothering and several therapist visits each week.

The services of therapists were secured when Anneliese was five months old. I watched their tireless work as they put our sweet granddaughter through exercise after exercise. After observing several sessions in the second year, I asked one therapist whether Anneliese would ever walk unassisted. “Of course she will!” was the emphatic answer and what I needed to hear. And walk she does.

From the beginning my husband embraced this child totally. He discovered early on how to awaken her giggles by making his own sounds of laughter and holding her close as they both shook with merriment. It was Grampa who went rustling through



a pile of leaves to show Anneliese how much fun it could be. I treasure the video that shows her studying his actions and then decidedly plowing into the leaves. How we cheered her on!

At this writing our beautiful Anneliese is approaching her fourth birthday. We thank the Lord for the privilege of being her grandparents and for the ways she enriches our lives. While she will live contentedly in her own world if allowed, she has a winsome way of relating to people. We relish the warm hugs that she gives so freely. The miles separating us mean we aren’t there to hear her speak occasional words and prevent us from being able to practice signing with her. Computer options help us keep up with her progress through photos and videos on Facebook. We greet every bit of progress with thanksgiving. However her future turns out, we will love her, and support her and her family on the journey.

Dup15q Alliance is a nonprofit organization that provides family support and promotes awareness, research and targeted treatments for chromosome 15q11.2-13.1 duplication syndrome (dup15q).

Dup15q Alliance offers help and hope for chromosome 15q duplication.

UPCOMING MEETINGS AND ANNOUNCEMENTS

Webinars

The Dup15q Alliance is launching a webinar series to help extend information and support to our families. All times are Eastern Time. Webinars will be recorded pending approval of presenter. Spots are limited to the first 100 people and you must sign up ahead of time. There is no fee to attend the webinar.

Please note: the schedule is subject to change. See the website for details <http://www.dup15q.org/family-support/webinars/>

September 23	1:00 pm EDT	Seizures and dup 15q
October 6	8:00 p.m. EDT	Registry 101
October 16	1:00 p.m. EDT	Genetics of dup15q
October 24	1:00 p.m. EDT	Stem cells and dup 15q
October 28	8:00 p.m. EDT	Dup15q Alliance year in review and future plans

2015 Family Conference



We have been working hard to plan Stronger Together, Dup15q Alliance's 8th International Family Conference which will be held in Orlando, Florida from July 30 - August 1, 2015. Preliminary details, including the link for hotel reservations, are now available at <http://www.dup15q.org/events/family-conferences/stronger-together-2015/>. Please check the site occasionally as we will be posting new information as it is confirmed.

<http://www.dup15q.org>

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