



Summer • 2013


The Newsletter of **DUP15q**
ALLIANCE

FROM THE EDITOR: OUR DUP15Q FAMILY HAS GROWN

NICOLE CLEARY, PORTLAND, OR

Attending the 2013 conference this summer, I was struck by an inescapable conclusion: our Dup15q family has grown! With 409 attendees, we had more people attending than ever before. More international families traveled to the conference as well, 36 individuals from 9 other countries. It was amazing to meet families from all over the world raising children with many different forms of dup15q. It is always a special delight to spend time with families who are attending the conference for the first time, and we bring you their voices in this issue.

Growth often comes hand in hand with increasing complexity, and that is certainly the case for the Dup15q Alliance. Some of our recent growth comes from families raising children with small “edge duplications” of chromosome 15, sometimes referred to as microduplications. These families came to conference and helped the Alliance better understand their unique needs and concerns. You will learn more about edge duplications and how the Alliance umbrella is expanding to support these families in the article by Guy Calvert.



One of my personal highlights at each conference is hearing an update from Rick Guidotti of Positive Exposure. Rick is a regular fixture at Dup15q Alliance conferences, taking pictures and spreading joy. Rick's work takes him around the globe to promote a more inclusive,

compassionate world where differences are celebrated. You will see a celebration of the beauty of dup15q in the photographs Rick took.

The 2013 conference reached a new level of family fun. Held in the Mall of America, families enjoyed time together at the Nickelodeon Universe Amusement Park and the Sealife Aquarium in addition to the conference itself. What could be more magical than this? We'll find out in Orlando in 2015!

NICOLE CLEARY LIVES IN PORTLAND, OR, WITH HER HUSBAND TIM AND DAUGHTERS CORRINA (DUP15Q), SIERRA AND JASMINE. SHE CAN BE REACHED AT NICLEARY503@GMAIL.COM

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Chromosome 15q duplication syndrome (dup15q) is a clinically identifiable syndrome which results from duplications of chromosome 15q11-13. 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.

<http://www.dup15q.org>

Celebrating the Beauty of Dup15q

Positive Exposure's Rick Guidotti uses photography and video to transform public perceptions of people living with genetic, physical and behavioral differences. Rick has spent the past fifteen years working internationally with advocacy organizations, medical schools, universities and other educational institutions to effect a sea-change in societal attitudes towards individuals living with genetic difference. His work has been published in newspapers, magazines and journals around the world. The Dup15q Alliance is fortunate to have a longstanding relationship with Rick.

This year Rick brought 5 medical students from the University of Minnesota who were enthusiastic to learn about dup15q. It is exciting to be introducing the next generation of doctors to dup15q! We are thrilled to celebrate the beauty of Dup15q, as captured by Rick at our 2013 conference. To learn more about Positive Exposure, visit www.positiveexposure.org.



FIRST CONFERENCE PERSPECTIVES *from Around the World*



My name is Victoria Lee. I found out in March that my daughter Ashley has a one of the small dups on chromosome 15q13.2-13.3. I also found out that she got her dup from her daddy. Ashley has a verbal and nonverbal learning disability, sub mucous cleft palate, speech and language disorder, and anxiety. I attend my first conference with the Dup 15 Alliance to find out more information about the duplication on chromosome 15q13.2-q13.3.

At the conference, I met so many nice people and got a lot of good advice. Taking the different sessions helped me think about how to work with my daughter and my other children with their disability. I also learned at the conference that the duplication that my daughter has is still new and that Dup 15 Alliance do not know much about it yet. I am hoping that maybe the Dup 15 Alliance might have more information on chromosome dup15q13.2-q13.3 or have more research studies on that duplication by the next conference. It was the best conference that I ever been too. I recommend that everyone should go to the dup 15 conference in 2015.

Victoria Lee
BILLINGS, MONTANA

We traveled from our home in San José, Costa Rica to attend the Dup15q Alliance conference.

It was a very positive experience because we could clarify some questions we had regarding our daughter's condition. For example the concepts given to us by the geneticist Brenda Finucane about the difference between the diagnosis of Duplication on Chromosome 15 and the various effects this condition can have. Another useful presentation was David Geslak's on the exercise as a way to promote the development of our children. There were also very good presentations by Jennie Delisi on Technology and Music Therapy, and the presentation on "Potty Training" as well as what was shared in the session "Parenting Siblings".

For us it was very valuable to meet other parents who face challenges similar to ours every day and we are excited to think that we could meet again at a future conference. Since the number of Spanish speakers attending the conferences is increasing, we are hoping future conferences have the service of simultaneous translation.

Juan Manuely Mercedes Campos
SAN JOSÉ, COSTA RICA

The recent 2013 Dup15q Alliance conference was the seventh International Dup15q conference but the first that my son and I have attended. Why, you might ask, with a 21-year-old son with IDIC 15 have you never attended a conference? We've known our son's diagnosis for 20 years, could have attended all of the past conferences, learned so much, been more educated on seizure control, diet issues, genetics, wills and guardianship, coping techniques, met many amazing families, shared experiences with them. But I couldn't. I just couldn't handle meeting that many Dup15q families and their children all at once. I feared that my heart would break in too many pieces.

Over the years our family has met Dup15q families one-on-one, heard about their child's health and developmental issues, the families' struggles, saw the similarities between our children. I found that when I had only one family to think about I could handle the emotions of meeting a child very much like my own son, understand what the family was going through, know what that small child's future would be like and not be overwhelmed with sadness.

Then I got brave and helped to organize a small gathering of 7 Dup15q families in Ireland and it got easier to meet several families with children like my son all at once. My heart had to recover after that gathering, but I felt more confident that I would not break down and cry upon meeting other children like my son. Later I attended a meeting of 8 Dup15q families in Florida and, you know what? My heart didn't shatter in to a million pieces! I found the experience interesting and helpful. Many of our children had similar issues and we were able to share helpful ideas plus I loved getting to know the families, and felt such a kinship with them. We only met for a few hours. It was rushed and too quick, so I was hungry for more sharing of information, ideas and support and I knew that I was ready to finally attend the 'Mother Ship' meeting of Dup15q children at the 2013 Reach for the Stars conference. I knew that I was finally ready to meet my 'extended family'.

By attending this most recent conference I feel more empowered, as though I was given a transfusion of ideas to help me get through the next two years with my son until I can return for a new infusion. The support and acceptance is amazing, but the education that these conferences present is invaluable. My hat is off to all the presenters and the organizers of the conference. It was a job very well done. A humble thank you for allowing my son and I to participate, basking in the warmth of our extended family.

Mari & Christopher O'Connor
RED WING, MINNESOTA

We had a fantastic time coming to the U.S. and attending the Dup15q Alliance conference. We were able to visit the Mackling family and feel blessed to have finally met. Fran and I got matching tattoos! She is amazing, like a sister and best friend.

Everyone here is amazing. It is an awesome feeling knowing you don't have to worry that your child is happily chattering so loudly, or giggling uncontrollably, or screaming their heads off. They all get it.

If you are thinking of making the long haul to Orlando in 2 years time, do it! You will not be disappointed. We are going to try to get back here, and hopefully bring/help another family get here too. It would be cool to see more than 1 Aussie flag in the mix.

Belinda McDonald
BAHRS SCRUB, QUEENSLAND, AUSTRALIA



LIVING ON THE EDGE: DUPLICATIONS OF 15Q AT 11.2 AND 13.3

By GUY CALVERT, Dup15Q Alliance

The July 2013 Dup15q family conference in Minnesota was the biggest of its kind in many ways. One notable growth trend was the fast emerging subgroup of families with duplications of 15q localized at the 11.2 or 13.3 coordinates. The rapid upsurge in the number of families with these types of duplications brings with it several challenges as well as much opportunity. It also prompts a fair number of questions. Why are we suddenly seeing so many more individuals with these conditions? How are their duplications different from the isodicentric or interstitial duplications of 15q with which most Alliance members to date are diagnosed? How do those differences play out in expectations for the affected individuals? And what do the various classes of 15q duplications stand to gain from banding together under the umbrella of the Dup15q Alliance?

CORE DUPLICATIONS AND EDGE DUPLICATIONS

To correctly characterize the underlying genetic issues, it helps to visualize the landscape of chromosome 15 where all of these duplications occur. In the top-left quadrant ("A") of the figure below is a greatly simplified representation of a normal version of chromosome 15. The centromere (a point on the chromosome which is engaged during the process of cell division)

divides the chromosome into the small (or "p", for petite) arm and the long (or "q"... for letter after "p") arm. Genes – each of them little factories for producing proteins – are strung out end to end along each arm of the chromosome like words in a sentence. Geneticists refer to the locations of genes on the chromosome using coordinates, as one might when pointing out a destination on a map.

CORE DUPLICATIONS

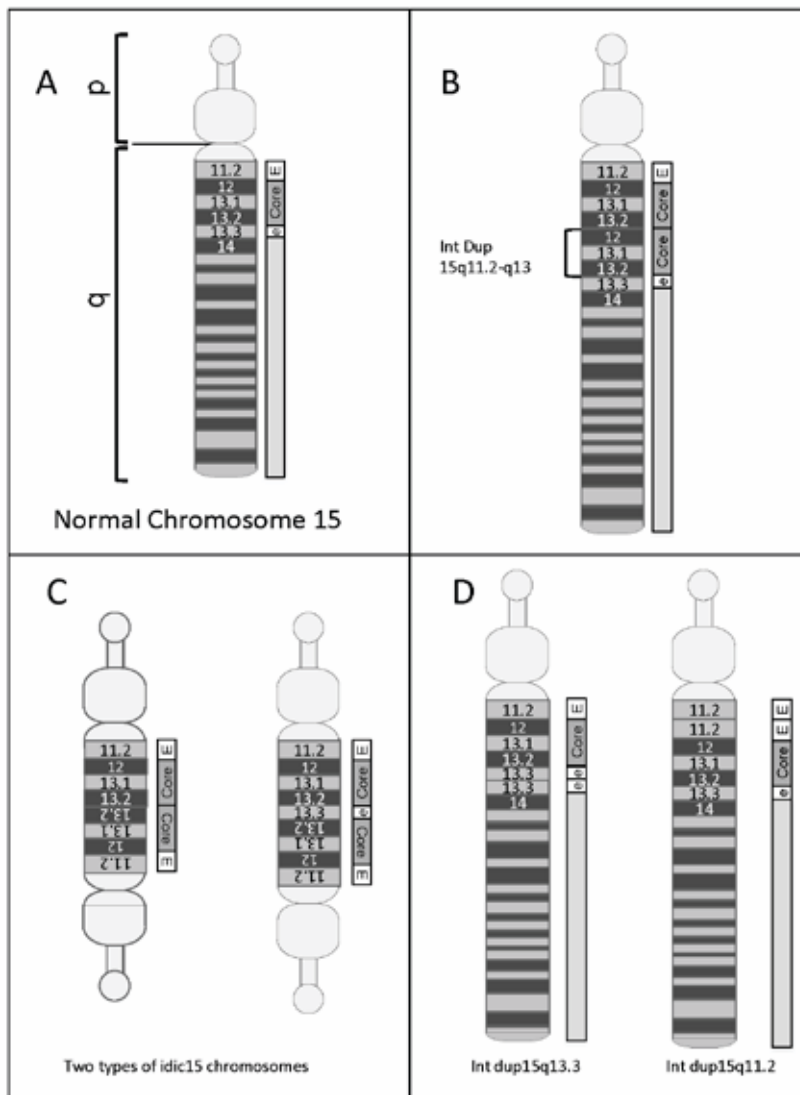
can be either interstitial (int dup 15, in which the extra copies of genes are laid out within the chromosome next to the originals - see top-right quadrant, "B"), or isodicentric (idic15, in which the extra copies of genes appear by themselves in a separate, 47th chromosome - see bottom-left quadrant, "C").

EDGE DUPLICATIONS

are generally of the interstitial variety - see bottom-right quadrant, "D". Some of the core interstitial duplications, and all of the isodicentric duplications, span a range of genes that goes beyond the PWS/AS to include some of the genes that are copied within the edge duplications. Sometimes people refer to edge duplications as "microduplications" since they span a relatively short stretch of genes, easily missed by older genetic screens like FISH. But while this term explains why the edge duplications have until recently evaded detection (advances in genetic testing technologies, e.g. microarrays like aCGH, can more readily pick up edge duplications), it does nothing to distinguish these diagnoses from core interstitial duplications on 15q, some of which are also microduplications. The location of the duplicated genes matters far more than the size of the duplicated region, so edge should prove a more apt adjective than micro.

Copy number variants (CNVs) sometimes occur, in which there are too many, or too few, copies of a certain gene or range of genes (duplications or deletions, respectively). Of particular interest to us is the stretch of genes marked by coordinates 15q11.2-13.3, also known as the Prader-Willi Syndrome / Angelman Syndrome (PWS/AS) critical region due to the disorders that result from deletions of certain key genes that reside there.

We can distinguish the core 15q duplications, those that span most or all of the PWS/AS critical region, from the edge duplications at 11.2 or 13.3, in which only the genes that are flanking the PWS/AS critical region on each side are duplicated. The PWS/AS range of genes spanned by the core duplications are marked in the figure as "Core", whereas the 11.2 region is marked "E" and the 13.3 region is marked "e".



HOW DO THOSE DIFFERENCES PLAY OUT IN EXPECTATIONS FOR THE AFFECTED INDIVIDUALS?

The question of expectations associated with edge duplications is complicated. Most families coming to the Dup15q Alliance are already experiencing serious issues – presumably the very issues that prompted them to run a genetic screen in the first place. But overall the issues associated with edge duplications appear to be relatively less severe than those that come with core duplications. This should surprise nobody: it is well known that different classes of genetic duplication (“genotypes”) can result in different clinical or developmental issues (“phenotypes”), e.g. core interstitial duplications produce a generally milder phenotype than core isodicentric duplications.

In the case of the 15q edge duplications, however, many scientists question to what extent clinical or developmental issues are actually caused by those specific genetic anomalies. The problem is that for many issues (e.g. autism spectrum disorder), the same 11.2 or 13.3 duplications have turned up – with the same frequency – in population samples of unaffected individuals, so the causality is unclear. It is uncertain whether the issues these individuals are experiencing are truly an associated phenotype for their given genotype, or rather the result of some other – still unknown – factor. That’s not to say that the edge genes play no role at all – much is still not understood – or that the issues being experienced by any individual are any less real. But the question of causality naturally complicates any effort to set expectations for the edge duplications – if the issues aren’t actually caused by the duplication, then the specific fact of the duplication can’t really be used to chart a course for the future, or to seek solutions.

How does the Alliance umbrella expand to provide relevant support for all families? This is something the Board has been thinking deeply about. In addition to the research or clinical angles, many dup15q families - regardless of the nature of their duplication - come to the Alliance hoping to meet others with similar experiences, to share stories and maybe get some answers. There is always a concern that, by broadening the Dup15q Alliance umbrella to include duplication classes with widely disparate clinical characteristics, we risk diluting the value of such peer-to-peer information sharing.

Fortunately, this concern is addressable. To the extent that the social conversation becomes unhelpfully broad in our existing forums, we are actively working

to facilitate and encourage the formation of more nuanced forums. This has already begun to happen, as with the recently created 13.3 page on Facebook ([Dup15q Alliance: 15q13.3 Microdup Parent Page](#)), which is now moderated by Alliance volunteers similarly to the larger [Dup15q Alliance Parent Support](#) Facebook group.

Certainly there is much the Alliance can do - and still needs to do - to accommodate the increasing diversity within our community. For clinicians and researchers alike, we need to ensure that whenever we present insights from dup15q data (such as Registry data or data collected via the growing network of dup15q clinics), that those insights are appropriately filtered by diagnosis type. For families - especially new

WHAT DO THE VARIOUS CLASSES OF 15q DUPLICATIONS STAND TO GAIN FROM BANDING TOGETHER UNDER THE UMBRELLA OF THE DUP15q ALLIANCE?

Causality aside, there’s no question that the edge and core constituencies alike have much to gain from working together toward common goals within an organization such as the Alliance. From a research perspective, the genes within the domains of the edge duplications are also duplicated in many of the individuals with core duplications, so the conclusions of scientific investigations into edge genes benefit everyone. There currently is not as much research into edge duplications as stand-alone syndromes (in part because the causality of these duplications is uncertain). The Dup15q Alliance has invited a leading 11.2 researcher (Brett Abrahams, of the Albert Einstein College of Medicine) to present his work at our 2013 annual scientific meeting in September. By the same token, we hope to learn not just from each other, but also from related syndromes – for example in 2014 for the first time we’re planning to run our annual scientific meeting jointly with the Angelman Syndrome Foundation.

There are also many economies of scale when it comes to funding research, so it’s in everyone’s interests to build as large a consortium as possible for the purpose of funding or otherwise driving research. It also doesn’t matter that the various classes of 15q duplication don’t resemble each other all that much. Very often in such research, the differences – properly accounted for – provide as much insight as the similarities.

Likewise, from a clinical perspective, every family – regardless of the nature of their 15q duplication – is well served by a network of clinics that is familiar with the variety of issues presented by individuals with dup15q. And the clinical insights gleaned across duplication classes could be valuable both for immediate treatment and long run research. Insights from the clinics are potentially of benefit to individuals of all dup15q varieties, and that those insights become all the more valuable as clinicians encounter more cases, and more cases of different types.

families - we need to provide better and more diagnosis-specific information for each class of 15q duplication, to help people understand their diagnosis and its implications as best we can. To this end we are developing updated “diagnosis triage” content for our website. We’re even planning “curbside consults” from genetic counselors at the next family conference. More generally regarding future family conferences, we’ll attempt to schedule both broadly relevant presentations on issues faced by all families raising a child with special needs, and more nuanced breakout sessions for particular flavors of dup15q. As much as possible we want to ensure that everything we do in every forum - from the website to the family conference - is as relevant as possible to all of our members.

DUP15Q ALLIANCE REGISTRY UPDATE

By RYLIE McHAM AND KADI LUCHSINGER

What does the Dup15q Alliance International Registry do?

The Dup15q Alliance International Registry is an online database that stores and organizes information about the characteristics of those affected by dup15q syndrome. As information about each person with dup15q syndrome is added or updated, the picture of this rare chromosome condition gets clearer. Our registry does several important things:

- creates a better understanding of the characteristics dup15q syndrome
- determines areas that need further research
- will help develop future targeted treatments to improve the lives of those affected by dup15q syndrome
- organizes information for families and interested medical professionals while protecting all individuals' identity

How do I join the Dup15q International Registry?

Only people who have dup15q syndrome and/or their guardians or parents can add information to the registry. The registry's clinical questionnaire consists of approximately 100 questions on various topics including development, seizures, sleep and general health. The questionnaire is easy to fill out, and as the data entered it is saved so the survey can be completed over time. The information is de-identified, so no one will have access to identifiable information other than the administrators of the registry.

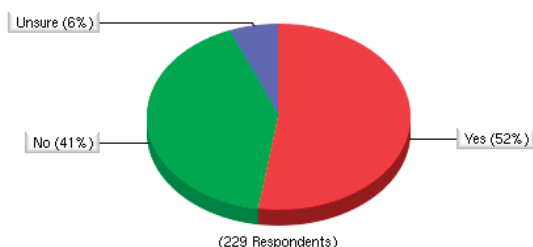
To join the registry, go to www.idic15registry.org and click on the Register Now button. To access or update your account information, go to www.idic15registry.org and click on the Login button.

If you are a medical professional who would like to access information for research purposes, please contact coordinator@dup15qregistry.org

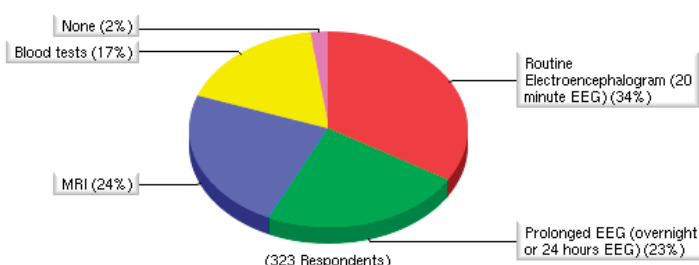
Can I get information about Dup15q from the International Registry?

The registry can be a valuable resource for families that complete it! There are charts and graphs that allow families to compare their results to the results of other participants. To do this you go to www.idic15registry.org and click on the Login button, click on the Clinical Questionnaire tab and then click on the section you want to view. Clicking on the colored chart icon enables you to see how your responses compare to other registry members. Clicking on the pencil and paper icon enables you to see your responses and to update your responses. There is also an Explore tab that includes various charts and graphs of the survey questions. Use this information for your own curiosity and knowledge and share with doctors and therapists when needed. Below are two examples:

Has the affected person ever had seizures?



Has the affected person had any of the following tests to diagnose seizures? (check all that apply)



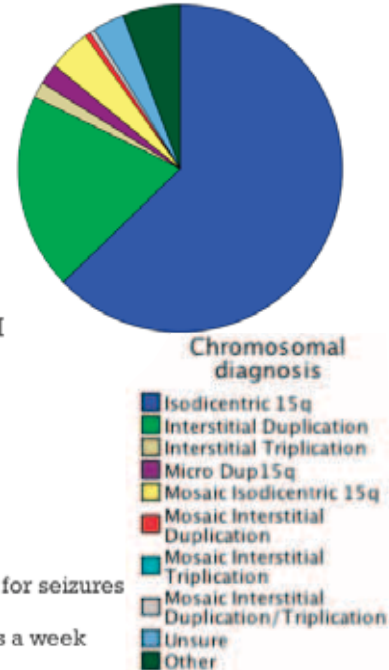
What's in store for the Dup15q Alliance International Registry?

The Dup 15q Alliance registry continues to improve. We feel passionately that the registry can be a valuable tool for individual care givers and families, as well as for the medical community and researchers. Recently, David Evans and his colleague, Laina Lusk, of Bucknell University analyzed some of the registry data and are planning on publishing their findings. We presented their information at the Dup15q Alliance conference. This information was included in the conference binder, which is currently available at the Dup15q Alliance store. A few slides from David are included below:

Subtypes

■ In our sample of 191...

- Isodicentric 15q
- Interstitial duplication
- Interstitial triplication
- Micro dup15q
- Mosaic isodicentric 15q
- Mosaic interstitial duplication/triplication
- Other



Neurology

- 49.0% have been hospitalized for seizures
- Average frequency is 1-6 times a week
- Average age of onset is 29-34 months old
- 16.2% experienced loss of a skill due to seizures
- Includes motor skills (fine and gross)
- complex skills (e.g. toilet training, reading)
- and speech

Filters will soon be added to the registry so that registry participants will be able to make searches more specific and relevant for individual users. For example, users will be able to select a specific diagnosis, like isodicentric 15, and compare their responses to others diagnosed with isodicentric 15. We are also hoping to have webinars and/or instructional videos to help those navigate the registry website.

Dr. Ron Thibert announced that the six Dup15q Clinics in Boston, Miami, Los Angeles, New York, Seattle and Memphis will create a clinician driven registry, which will be separate from the Dup15q Alliance registry. A clinician registry is created by the clinics, where clinicians directly enter data in to a registry. This is an exciting additional tool which will assist our community in better defining and understanding Dup15q Syndrome. We will share more on the clinician driven registry as it develops.

At the end of August, 2013 there were 220 completed registry surveys and 178 completed surveys with an uploaded genetic report. Keep the information coming! Create a registry account for each person with dup15q syndrome and update existing accounts at least annually. Should you have any questions, please contact us at coordinator@dup15qregistry.org.

DUP15Q Alliance ANNUAL BOARD MEETING REPORT

BY LINDA MEAGHER, BOARD MEMBER FROM SUGARLAND, TX

The Dup15q Alliance Board meets by phone once per month and also conducts business via email. It's rare we have the opportunity to meet face to face, but the 2013 *Reach For the Stars* conference allowed us to do just that!

This year, instead of having the annual board meeting prior to conference opening, we chose to hold it after the conference closed. This provided a dual advantage by allowing board members to connect with old friends and meet new families, as well as discuss conference results in detail after the close, while the experience was still fresh.

Approximately 90 minutes after the conference closed, the board gathered. Having a full quorum, Tom Doyle, Interim Board Chair, opened the meeting with a review of the conference – what went well, was the schedule agreeable, did the families and children enjoy the special events, was the food and hotel accommodations what everyone expected, etc. Board members shared their insights on conference details, and notes were taken for the 2015 conference.

Based on initial feedback from our Alliance families, the many extended family members that attended, as well as our Dup15q professional advisory members, the board members were pleased that the 2013 *Reach for the Stars* conference surpassed expectations and could not have gone better! The board members expressed their sincere thanks to Karen Sales (Conference Chair) and Kadi Luchsinger (Executive Director), as well as to the Conference Committee, and all the families that held fundraisers, donated to the auction, or otherwise volunteered their time and efforts to ensure the success of the conference. Kadi advised that a follow up survey would be sent to all families that attended that would provide additional conference feedback and ideas for 2015.

The meeting then turned to a discussion of the needs of families whose children have the smaller edge duplications on chromosome 15. The board agreed that the Dup15q Alliance would continue to work to support these families. The board further agreed that additional guidance and education was needed in order to better understand the small edge duplications and how best to support the families affected. Please see Guy Calvert's article on edge duplications in this issue of the MIRROR for more information.

The Board also addressed the Registry and an action item was taken by Kadi and Rylie McHam to have a discussion with registry personnel about the different forms of dup15q experienced by Alliance families. Kadi and Rylie further discussed ideas they had to get more Alliance families to complete the registry.

The upcoming Scientific Meeting in September, to be held at the MIND Institute in California, was next discussed and Guy Calvert and Kadi provided an overview of the proposed schedule of events.

Mike Porath discussed ideas and thoughts on fundraising for 2014, including a 5K event. Mike also discussed status of the website project.

Fernando Gomez discussed current activities underway as well as future plans with the international members, and Linda Meagher discussed several projects in the PR area. Paul Karch provided an update on Governance Committee activities and Dana Tilton provided an update on Sunshine Committee and regional reps.

In accordance with the Dup15q Alliance bylaws, the board then considered the election of board members whose 2-year terms expired at this meeting. Each of the board members expressed a desire to continue their work on the board. After a motion duly made and seconded, the following were elected to 2-year terms expiring in June 2015: Kadi Luchsinger (board member and Executive Director), Karen Sales (board member and Secretary), Guy Calvert, Paul Karch, and Linda Meagher. The Dup15q Alliance co-founder, Donna Bennett, serves a continual board role.

The board next turned their attention to the status of various ongoing and future projects of the Dup15q Alliance. A new tracking system identifying each project, description, person(s) responsible, proposed timing, and priority status. This will allow the board to better track the various projects that are necessary to maintain the Alliance's legal status, to enable the Alliance to continue to run smoothly and efficiently, and to provide for its continued growth to meet membership needs.

All too soon, our time together was at an end and the 2013 annual board meeting was wrapped up and adjourned. I have no doubt that each board member walked away with a renewed sense of purpose and his or her mind spinning with all the exciting plans and projects we each have ahead of us to accomplish on behalf of the Alliance. It was not without sorrow that we hugged each other goodbye, knowing it might very well be another 2 years before we have the opportunity to meet face to face again!



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Stepping Forward with Mason



By JAIME MURPHY, GREENSBORO, NC

I never thought that I would meet an angel in my lifetime, at least not during my earthly existence, but God sent one directly into my life. My son Mason was born in June, 2009, a wonderful birthday present for my father-in-law who will now forever share his birthday with his first grandson.

The moment Mason was born the delivery room turned into a fury of activity so we knew something wasn't right. Mason had looked perfect in all of my ultrasound photos but there was no way to see that at some later point he had positioned himself with his left leg backwards across his chest with his left knee hyper-extended toward his right shoulder. It should have been a shocking sight, but I was so excited that my first child was here and I was still feeling the effects of the drugs. Also, I was immediately reassured by the pediatrician (plus the drugs) that "that sort of thing tends to happen and it is easily corrected" so I took it all in stride (did I mention the drugs?). It seemed like forever until they brought my beautiful boy to my room, and I was shocked that he appeared perfect. The pediatrician told me that Mason had hip dysplasia and the second they put the security tag on his ankle he straightened his leg on his own.

The day Mason came home from the hospital he was fitted for a Pavlik harness to hold his hips in the correct position. He wore it for the next three months. At Mason's three month checkup the doctor was surprised that Mason was not holding his head up on his own but everyone attributed it to him resting his head on the shoulder straps of the harness, so we waited. It felt like an eternity.

At six months of age Mason was referred for genetic testing and an MRI. His Occupational Therapy and Physical Therapy began immediately while we waited for the results. The MRI showed a definite neurological problem that we were told was low myelination. The neurologist then told us we had the symptom but the condition or disease was still a mystery. We would know more when the genetic testing results came back. So we waited, and waited, and...well, you know.

I will never forget the call I received the day we finally got the results, and not for the reason you would expect. I was calm and open minded when we were told that our baby boy had a duplicate of his 15th chromosome. "Idic Dup15q," the neurologist spelled out to me. But what sticks in my mind the most was what he said to me next: "It is very rare; more rare than trisomy 21 which is Down's Syndrome, but I knew it wasn't that because Mason is too pretty." My jaw dropped. I couldn't believe that those words came out of a medical professional's mouth and that was the sum total of information he had to share with our family. Our follow-up appointments with neurology and genetics really weren't any better. Our geneticist told us, "I looked on the Internet this morning and didn't see any new studies or research to share with you." We did, however, check out the one and only resource provided to us which at the time was called IDEAS and is now Dup15q Alliance. The organization and its members were God sent. We read every bit of information we could and for once we had at least something more than lame jokes and placations to wrap our minds around.

Mason continued with his therapies and added speech therapy to the mix at two years old. Mason was able to sit on his own at one year old, crawled at 18 months and was just starting to walk at two-and-a-half years old. Mason is now four years old and he is still a little unsteady at times, but his balance is much better. Mason loves to walk everywhere and runs half the time.

On Mason's first birthday we found out we were pregnant again and eight months later we welcomed his brother, Asher, into our family. Asher is absolutely in love with his brother and is constantly hugging and doting on him. Mason, on the other hand, is more physical and loves to tackle and wrestle his brother to the ground. When Mason was 3 years old we started taking both boys to swimming lessons and they love anything that has to do with the water. As much as Mason loves swimming, it still can't hold a candle to music. Music is honestly Mason's passion and I don't think a day goes by without singing or playing any instrument he can get his hands on.

Mason is still non-verbal, but he can use the sign for "more". Most of the time he lets us know with body language or by taking us by the hand and leading us to what he wants. Mason starts pre-school through our public school system this year and I think, no, I know it will be harder on me than on him. He has never met a stranger and gets along well with adults, but I'm just not sure how well he will do with other children.

Mason is the most lovable and huggable little man in the world and he works so hard for every little step forward he takes. He truly is an Angel here on Earth.

Cheers to Our Volunteers!

The Dup15q Alliance Cheers our Volunteers: **Rachel Pomainville**

Rachel Pomainville, from Ottawa, Ontario (Canada) recently started a blog to help families connect and share stories. Her initiative in launching this blog is a perfect example of how anyone can contribute to the Alliance in ways that feel natural or allow them to share a passion or interest. Thank you Rachel for creating another online resource that helps families feel connected!

We interviewed Rachel so that families can learn more about her dup15q blog.

What is the intention of the dup15q.wordpress.com site?

The dup15q.wordpress.com blog was created to give families affected by the syndrome a voice to express their experiences, and share life with dup15q with the general population. It is a very public blog that is meant to both raise awareness for this chromosome anomaly, and to humanize the syndrome with stories and pictures from real families.

How did you get the idea to create this site?

I got the idea to create the blog because I am a blogger. I know the work that can go into creating, and maintaining a blog by yourself. I figured that there were families within our organization that really wanted/needed to get their stories out there, but they might not know how or they might not want the hassle of having their own blog. This way the work is done for them- they simply need to contribute their thoughts, and stories, and I do the rest.

How do families make a contribution?

Anyone can contribute a blog post. You can email it to be me personally at cottagetremlant@hotmail.com and I will transfer it to our blog.

What are your plans for this blog in the future?

In the future, I'd like for this blog to continue to be used as an outlet for family members, but to also raise more awareness for the Dup15q Alliance's initiatives, fundraisers, board responsibilities, and committee projects. The more people that know about our organization, and participate in its projects, the stronger this international non-profit organization will be in terms of family support, and research.



Dup15q Alliance

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Reflections *from a Sibling*

Little Bear

BY REBECCA SEGARS, PALO ALTO, CA



I have two brothers: Nathan, who is eight, and Luke, five. I love being an older sister, and I have an amazing relationship with both my brothers. Luke especially has shaped a large part of who I am. Luke is the 'little bear' of the family. Contrasting with the light hair of my family, he has soft dark hair and big brown eyes that look like they go on forever. When Luke was seventeen months old, he was diagnosed with a genetic disorder called Chromosome 15q Duplication Syndrome.

Chromosome 15q Duplication syndrome (dup15q), also known as Isodicentric 15q (idic15q), is a relatively rare genetic disorder that can affect people in different ways. Those diagnosed vary in ability, and have different ways of functioning. Some physical characteristics can include having low muscle tone, which can make people appear very floppy, and having a flattened button-like nose. Typical medical concerns for children and adults with dup15q can be severe, such as seizures, or milder like anxiety disorders and ADD. We are so blessed to be on the less severe side of the spectrum, but there will always be fears of medical and physical difficulties with Luke.

Luke is very typical looking and is growing at his peers' rate, but his cognitive ability is delayed. Luke cannot speak and is still in diapers, but he has his own way of telling you what he wants. One of the most amazing things to see him do is take your hand and lead you to what he wants or needs. Luke is a very happy little boy and he has always been content playing with his loud and colorful toys. Many people visiting our house will notice the noise as soon as they walk through the door. There is always music playing and usually more than one toy making some sort of obnoxious sound. Waking up in a house with Luke is like waking up in a house with a baby elephant running and jumping around the house with heavy feet. There is never a quiet moment in my house, and I am perfectly happy with it that way. I, along with the rest of my family, have gotten to the point where we don't notice the noise anymore, and as anyone who has been around children will say, it is more scary when things are quiet.

Like many children on the Autism spectrum, children with dup15q can go through phases of certain obsessions. Luke's latest way to entertain himself is by watching nursery rhymes on YouTube. We recently bought a new desktop for the family to use. Luke has definitely taken advantage and has used it more times than the rest of us put together. He has figured out how to use the touch screen and will spend hours sitting on the desk navigating his way around the YouTube menus. He knows exactly what he wants to listen to, what the icon looks like and where to find it. He has even figured out what other videos he has to select for the one he wants to come up on the menu screen. It is an absolutely amazing thing to watch. This child who cannot speak has suddenly found his own way to navigate around the internet.

My brother and I are very close, and we have always enjoyed doing certain things together. He loves music, so recently he will sit on my bed and watch me play my guitar, or I will help him hold the ukulele and we play together. When I was about twelve, I would get in from school around the same time he would nap, so I would sit on the couch and let him fall asleep while I sang to him or talked him off to sleep. One thing we have gotten in the habit of doing now that we are both older is taking walks around our neighborhood. I push him in his stroller and we look at all the interesting things around Palo Alto. Luke enjoys sticking his hand out and running his little chubby fingers along fences and plants within his reach. He has always been fascinated by anything that spins or is very colorful, so we watch the cars going by us. We may walk for about fifteen minutes, or sometimes we may be out wandering for hours. Usually Luke will fall asleep at some point along our walk, so it gives me time to listen to music or clear my head. The amount of times people have thought he was my child is hilarious. I may have only been asked a few times, but I am definitely aware of the questioning looks I get pushing this small boy around Palo Alto.

Being an older sister is something I take on as a huge role, maybe because it was handed to me in that sense. I was an only child for seven years and always wanted a younger brother to explore with. I play the protective older sister to both brothers, and have always wanted to make sure they are both doing well. Maybe it is the age difference, but I do not fight with my brothers as much as I have seen friends argue with their siblings. In the past few years Nathan and I have found ways to annoy each other, but not nearly as much as in many other families. I want the best for both my brothers, and I love them both more every day.

Luke being the way he is has changed the way I look at the world. I love this boy so much, and I have truly learned how to be patient in tough situations, which has changed the way I deal with people. I am now a lot more understanding of the physical and mental challenges in people's lives, and I have actually become good at listening and understanding people. It seems ironic learning this from a child who cannot tell you his problems. I have dedicated a lot of my time to helping others see the world and learn to respect unique learning differences.

Reflections from Grandparents

BY DENNY AND JUDY LAGER, GROVE CITY, OHIO



We have a new perspective on life and its meaning since Avery came into our lives. She was the first "Special Needs Child" in our family, and so we entered into a different type of behavior that was previously unknown to us. We had to learn how to react to some of her behavior problems which are exhibited by special needs children when they try to deal with their frustrations and anxieties.

She exhibits a special uninhibited love that reveals how God would have us view love for one another. She has enjoyed swimming in the pool at our Condo during the summer months, and exhibits no fear of the water. She wears a flotation vest and loves to splash and interact with other kids. She treats other kids the

same whether they are strangers or friends. Avery recently went up to a lady she didn't know, sitting at the pool side, and gave her a big hug. Most people react favorably when she interacts with strangers in this manner.

She can be very difficult at times, as we try to understand her emotions and frustrations when she is dealing with some of life's problems.

Because of Avery, we have a new perspective on the meaning of love and the values we place on this life.» We also have a new appreciation for the patience and love that our daughter Connie and her husband Gary have shown as they are trying to give Avery every chance to live a happy and productive life.



Dup15q Alliance is a nonprofit organization that provides family support and promotes awareness, research and targeted treatments for chromosome 15q duplication syndrome. Dup15q Alliance offers help and hope for chromosome 15q duplication.

Great news! We now have a calendar on our website so you can keep track of all of the upcoming events. If you have an event to put on the calendar, please let us know!

Visit www.dup15q.org/calendar.html.

UPCOMING MEETINGS AND ANNOUNCEMENTS

November, 2013: Melbourne, Australia

We are having a group meeting for Dup15q families who reside inside/outside Melbourne Australia. Everyone is welcome.

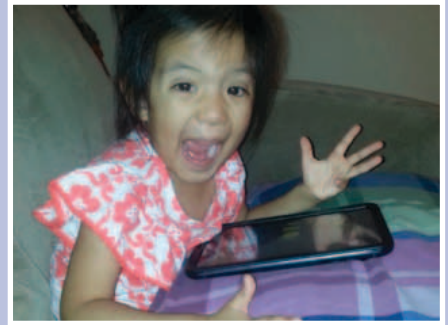
Contact: Belinda McDonald Belinda.mcdonald@gmail.com

2015 Conference: Orlando, FL

The 8th International Conference will be held in Orlando, Florida on July 30 - August 1, 2015

The Dup15q Alliance Store recently received a major renovation! Increase awareness of Chromosome 15q Duplication Syndrome by purchasing our new items, and old favorites, today! Missed the 2013 Reach for the Stars Conference in Minnesota? Get your binders, t-shirts and other items shipped to you!

Visit the Dup15q Alliance Store and place your order!
www.dup15q.org/store.html Shipping is included on all US orders. For international orders, email info@dup15q.org for rates.



The Dup15q Alliance thanks all parents who have completed the registry. Congratulations to Kristina Hernandez who is the winner of our iPad drawing for the registry. Looks like someone is excited!!!

Congratulations also to Rae Wertz, Nancy Barnett and Jacqueline Hayada who won \$50 iTunes cards. The Alliance will run another promotion in the future.

<http://www.dup15q.org>

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