



The Newsletter of **DUP15q ALLIANCE**



BY ALISON KALNICKI

2015 CONFERENCE COMMITTEE MEMBER, VANCOUVER ISLAND, CANADA

On the eve of the Dup15q Alliance's 2015 Stronger Together Conference, volunteers gathered in a spacious Orlando, Florida hotel to greet conference participants with name tags, schedules, and smiles. Out in the courtyard, kids from different countries played in the pools together. Upstairs, the room for the New Family Orientation filled with adults, kids and strollers. Adults chatted with those seated around them - strangers, yet already connected.

This issue of *The Mirror* builds on the momentum of the conference. In these pages, you will see the heart of the conference – the connections between families, affected individuals, research and medical practitioners, educators and specialists.

The Dup15q Alliance has now grown to 1,017 families, with over 300 families from outside the United States, including Saudi Arabia, Peru and Austria. We celebrate and support our international families with a dedicated session for conference participants from countries other than the United States to discuss current international activities and country based groups.

This conference had at least five families who attended with a service dog at their side. These calm dogs seemed to naturally lower anxiety and encourage social interactions with everyone around them.

Scientific interest and progress into dup15q is gaining momentum! Guy Calvert provides an excellent summary of the Dup15q Annual Scientific Meeting, and Carolyn Schanen

introduces the first two Dup15q Alliance research grants. At the conference, rooms were also set up for researchers and technicians. Individuals with dup15q syndrome and family members attending the conference participated in research studies on site, instead of having to travel to specific research sites.

We appreciate everyone who contributed to the conference related fundraising successes! Links of Love, which ran from June to early August, raised more than \$17,000. The Hometown Dinner Raffle and Silent Auction raised \$9,000. Twenty people volunteered at the conference store over four days, and sold 220 t-shirts, the entire inventory of silver bracelets, and much more. You'll find a financial update from the Dup15q Alliance in this issue.

Thank you to all who made the 2015 conference such a success: every participant, presenter sponsors, raffle donors and ticket salespeople, room monitors, store and registration clerks, Board and Conference Committee members, our DJ (who drove his sound system 3,016 miles), and many others!

Special thanks to the K4 Team: Kim, Kadi, Karen and Katie, for their long-term commitment to our international family conference.

For families touched by dup15q syndrome, our lives are not always filled with ease and comfort. From the strength of 353 conference participants and presenters who came together in Orlando to all of you, enjoy this issue of the Mirror.

Each person's journey's with dup15q syndrome is different, but we are stronger together.

Hoping to see you in 2017 in Los Angeles, California.

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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.

<http://www.dup15q.org>

Since 2011, I’ve had the privilege to organize and chronicle four annual Dup15q Alliance research meetings, each time bringing together 50-100 of the best minds engaged with dup15q research. As both an armchair biologist and parent to a child with dup15q syndrome, I’ve been both delighted at the accelerated pace of progress over these years, and frustrated that it still takes so long to identify targeted treatments. Every year we hear of new incremental insights, each tantalizing in their promise, but often raising more questions than they answer.

This year’s meeting felt different somehow. It wasn’t that the work from past years was any less valuable, nor that this year brought with it any definitive answers for targeted treatments. The thing that impressed me most was that suddenly many of those incremental insights seemed connected to each other. It was as if everyone had decided to sit down and fit together the pieces of the dup15q jigsaw puzzle and, this year, at long last, had finally gotten past laying out the edge pieces. The final picture remains unclear, but at least it’s starting to take shape.

## UBE3A

Jason Yi, a postdoctoral researcher in Mark Zylka’s lab at the University of North Carolina, presented his findings on properties of a key gene called UBE3A, which is located in the critical 15q11.2-13.1 duplication region. Dr Yi’s work (which has since been published in the journal *Cell*) represents a substantial advance in the understanding of how UBE3A is controlled at the protein level. Although many genes are in the dup15q11-q13 region, there is intense interest in UBE3A because deletion of the maternal UBE3A copy causes Angelman syndrome and duplication of the maternal UBE3A copy occurs in most cases of dup15q syndrome. This study showed that UBE3A can be modified to be either active or inactive. This was not previously known, and is big news. If ongoing over-expression of UBE3A leads to hyperactivity of UBE3A function in dup15q, then some kind of chemical off-switch or tuning dial would certainly come in handy. Yi and his collaborators discussed a medication, previously studied as a treatment for depression, that may help to regulate UBE3A activity and potentially impact some symptoms of dup15q syndrome. A natural next step would be to try this and similar medications out on well-characterized model systems of dup15q syndrome, to see if those systems can be “rescued” (i.e. restored to normal).

Regarding the model systems, the good news is that there are several alternative systems under development - ranging from animal models (flies and mice) that have been doctored to over-express UBE3A, to human brain neurons that were harvested from the stem cells of actual people with dup15q syndrome (and therefore have the same genetic duplication that those individuals had). True, none of these models are fully characterized yet, meaning no one has established definitive phenotypes that are caused by the duplication and which scientists can then attempt to rescue. But some models are at least partially characterized, and can offer up critical insights into the molecular mechanisms of dup15q.

# 2015 Annual dup15q Scientific Meeting

By GUY CALVERT, Dup15q ALLIANCE RESEARCH CHAIR

## Dup15q Neurons

Stormy Chamberlain’s team at the University of Connecticut produces neurons from dup15q stem cells from donated skin and blood – those who donated blood at our family meeting this year directly support this work. The ideal control system for comparison would be neurons with the same genetic make-up, without the duplicated material. So - with the help of the Dup15q Alliance registry - Chamberlain discovered and reached out to the families of individuals who are mosaic for dup15q syndrome (i.e. have some dup15q cells and some normal cells) and received generous skin and blood donations. She is now attempting to derive neurons from matched pairs of normal and dup15q cells.

Meanwhile, Chamberlain has also identified some compounds that restore UBE3A protein levels to normal in her dup15q neurons. Two of her UConn colleagues, Eric Levine and Les Loew, are now attempting to characterize the dup15q neurons. They have found some encouraging leads. Levine and his team observed that a current passed through dup15q neurons elicits a different electrophysiological response than Angelman neurons or normal neurons. That may sound rather abstract - far removed from the seizures, autism, and so on that individuals with dup15q syndrome experience - but at the cellular level it is the beginning of a possibly rescuable phenotype.

## Mathematical Models

Olena Marchenko, a graduate student working in Les Loew’s lab to fit mathematical models of cell dynamics to dup15q neurons, dropped a quiet bombshell while describing her equations. Marchenko was interested in the neuron’s dendrites - branched tendril-like projections that send electrochemical signals between neural cells. Such neural networks are the basic building blocks of thinking, learning, dreaming, pretty much everything the brain does. The dendrites themselves, if you zoom in closely enough, are lined with a multitude of tiny spines. We could leave it at “multitude”, but the relative number of spines sometimes turns out to be quite important. So the bombshell: Marchenko and Loew found that young neurons grown from dup15q cells have a higher than normal density of dendritic filopodia - think of these as baby dendritic spines. Time will tell whether the filopodia mature into spines in the same proportion, but abnormal spine density has been noted in other syndromes linked to autism, so this could be an important piece of the puzzle.

## Animal Models

Scott Dindot, of Texas A&M, gave an update on his lines of dup15q mice, some of which he has sent to the mouse repository at Jackson Laboratories so that they are available to other researchers. Larry Reiter, at the University of Tennessee in Memphis, presented some surprising insights from his studies of fruit flies. For flies that had an extra copy of UBE3A in their cells, Reiter was able to induce something like a seizure. After spinning them around in a device called a vortex, the dup15q flies would be initially stunned, then start twitching around violently. Control flies were also stunned, but then recovered without the twitching. Moreover twitching was dependent on having extra UBE3A copies in glial cells (another kind of brain cell, distinct from neurons). Having an extra UBE3A in neurons was unnecessary - the twitching flies expressed extra UBE3A in glia but not neurons. This is intriguing because much of the initial interest in UBE3A derives from the fact that only maternal UBE3A is expressed in neurons. In glia the parent of origin doesn’t appear to matter, but the dosage of UBE3A does.

There were also several presentations about clever ways to characterize animal models of dup15q. Christian Hansel, from Chicago is planning to run tests on Dindot’s dup15q mice in his own lab. So is Peyman Golshani, a UCLA researcher. So is Mark Zylka, at UNC. Do you see now why I think the research seems more aligned?

## Clinical Research

The second day of the research meeting was focused on clinical research. Dr. Franck Kalume, from the University of Washington, presented his study of SUDEP (sudden unexplained death in epilepsy) in a mouse model of Dravet syndrome - a rare and intractable epilepsy disorder. Dr. Kalume was able to induce seizures in the mice while tracking their biorhythms. In many cases it was observed that the mice died after a characteristic period of bradycardia (slowed heart rate). For the clinicians in the audience, who had patiently sat through the entire previous day of molecular research presentations, it was as if someone had fired a starter pistol - a spontaneous and energetic discussion ensued about the findings and their relevance to dup15q.

Professional Advisory Board members Dimitrios Arkilo (who runs the Minneapolis dup15q clinic) and Ron Thibert (of the dup15q clinic at Massachusetts General in Boston) reported on their joint work regarding VNS performance and characteristic sleep patterns on EEG in dup15q patients.

Dr Arkilo reported survey findings on the efficacy and safety of vagus nerve stimulator (VNS) treatment in children with dup15q syndrome and medically resistant seizures. Seventeen families responded to the survey, and the overall conclusion was that VNS was safe and well tolerated. Dr Thibert presented a deeper analysis

of sleep EEG patterns in dup15q. First reports on sleep patterns were presented in 2012 based on an initial group of children seen at Dr Thibert’s clinic. The latest data was gathered from individuals seen at dup15q clinics all around the country as part of a multi-center study. The most unique findings were three separate patterns present in a certain percentage of children with idic(15). One pattern is continuous spike and wave of sleep (CSWS), which is when spikes take up more than 50% of the sleep record – this finding has been associated with developmental regression and autistic features in the general epilepsy population. The second pattern is alpha-delta sleep, which is an uncommon pattern of fast and slow activity on EEG present at the same time and has been associated with autonomic dysfunction in the general population. The third pattern is one of very high amplitude, very high frequency spikes – this pattern is of unclear significance, but very likely associated with seizures, and may be unique to dup15q.

Shafali Jeste, from UCLA, also presented analysis on EEG patterns - in this case using a more advanced technology to look at the characteristic beta oscillations found in dup15q kids. Her preliminary work shows that this pattern is more intense for isodicentric duplications compared to interstitials, and that as a group, children with dup15q show an EEG pattern that is distinct from children with non-syndromic autism. That means this intense beta wave pattern is so far a true biomarker of dup15q syndrome and not autism, per se. Jeste noted that the pattern was most pronounced in the kids who were more challenged in verbal and motor skills, but this relationship may be related to the type of duplication. Dr. Jeste’s group collected EEG data from 25 more children with dup15q at the family meeting in order to better understand this biomarker. Meanwhile Connie Kasari, also at UCLA, presented insights from her pilot study on JASPER-based interventions (intensive one-on-one therapies) with some of the same kids. The interventions were generally and measurably beneficial, but it was clear that a handful of sessions was insufficient.

One of the finest and possibly most valuable moments of the research meeting was an entirely off-menu event. Midway through the second morning, the meeting adjourned briefly so that the researchers and clinicians could walk downstairs to watch the opening parade of the family conference. In an emotionally charged scene, the crowd erupted in cheers as 63 individuals with dup15q and their families marched through the room to take their place at the front. For many of the basic science researchers, this was their first exposure to individuals with dup15q syndrome. Some were moved to tears. One scientist later told me that throughout his career, this was his first opportunity to work on something with the potential to directly improve human lives. For him, like many of us, the dup15q puzzle has a very personal feel.

# Dup15q Alliance Funds Research

By CAROLYN SCHANEN, MD, PhD, Dup15q Alliance Professional Advisory Board

In May, 2015 the Dup15q Alliance announced its first Request for Applications (RFA) for grants to fund research specifically for dup15q syndrome. The grant program supports scientists early in their research careers by providing training grants to graduate students, post-doctoral fellows or medical students pursuing research into dup15q-related fields.

The response to the RFA was excellent. We received 10 applications that were very competitive and focused on a number of topics directly related to dup15q. A group of seven scientists and clinicians reviewed the applications and selected the top two for funding. It was a tough decision because of the high quality of the proposals. We extend our congratulations to the winners, Kevin Hope and James Fink (and their mentors)!

## Kevin Hope: Investigation of Synergistic Interactions Among Genes in Dup15q

Kevin is a neuroscience graduate student working with Lawrence T. Reiter, PhD at the University of Tennessee Health Science Center, in Memphis, TN. He was awarded 4 years of funding for his ongoing studies on dup15q syndrome.

Kevin's PhD thesis project aims to find out which genes in the duplication act together, resulting in the major clinical features of dup15q syndrome. Many genes are located in the segment of chromosome 15q duplicated in both idic(15) and int dup(15). So far, most of the research on the genetic basis of the syndrome has focused on a single gene, UBE3A (the gene that causes Angelman syndrome when it is not present). Kevin suspects that other genes in the region contribute to the complex spectrum of symptoms seen in people with dup15q syndrome. He has selected 4 genes commonly duplicated in most individuals with dup15q syndrome, and likely to impact the function of the nervous system. He wants to understand how these genes influence learning and memory, autism behaviors, and seizures by duplicating and deleting them in *Drosophila* (fruit flies). Why fruit flies, you ask? First, it is relatively easy to change the dosage and part of the body where the genes are active in flies. Second, they reproduce quickly, so Kevin can create many strains of flies that have different combinations of gene duplications/deletions to test for potential interactions between the genes. Third, you might be surprised to know that fruit flies display complex social and learning behaviors, and can even be triggered to have seizures like mice and people. Thus, Kevin can measure how changing the dosage and combinations of these genes affects the nervous system.

The review committee was very excited about the potential that this study holds for creating a model system that can be used to screen for new therapeutics - potentially by modifying gene function - noting that the fly model of Fragile X syndrome has been a powerful approach for identifying new drugs to use to improve symptoms for that disorder.

## James Fink: Hyperexcitability in Human Stem Cell-Derived Neurons from 15q Duplication Syndrome Patients

James is a graduate student working with Eric Levine, PhD, in the Department of Neurosciences at the University of Connecticut Health Science Center in Farmington, CT. The grant will provide funding for 3 years of this research.

James' proposal focuses on understanding what causes the increased risk for seizures in dup15q syndrome. Brain neurons are electrically active cells that send highly coordinated impulses among complicated networks of cells. Seizures often result when regulatory processes are not functioning normally, or when connections are not formed properly. Although seizures are a major problem for many individuals with dup15q syndrome, we don't understand how the duplication chromosome is influencing the brain to make it more vulnerable. James' work will use a technique that allows him to convert skin cells from dup15q patients into neurons that he can grow in the lab. Because seizures involve the whole brain, you might wonder how James can study seizures 'in a dish'. He can do this by measuring the electrical activity (excitability) of the neurons, how they interact, and whether they are accurately regulating the signals transmitted between cells to determine whether there is something fundamentally different with the dup15q neurons compared to normal neurons. James' work has broader implications, because accurate regulation of the interactions among brain neurons is critical for learning and memory, language, autism, and even muscle tone.

James' research project also got the review committee excited because using neurons grown from actual dup15q patients to understand the processes that could be causing seizures is truly innovative. This research could provide the groundwork to eventually test different types of compounds or drugs to see if they can correct processes that are functioning abnormally, which could potentially lead to new treatments for dup15q syndrome.

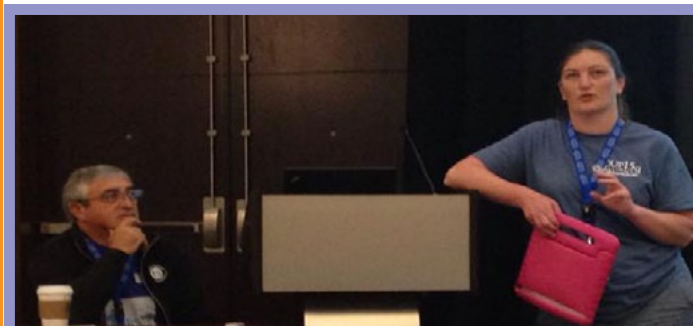
Directly funding research empowers the Alliance to drive work into areas that are important to dup15q syndrome. In the review process used to select the winners we asked – would the work impact our understanding of the syndrome? Could it potentially generate model systems to screen for new treatments? Both of the grants awarded meet those goals. The grant winners are at an influential stage in their careers, and these grants could spark long-term commitments to dup15q research. Training grants allow the mentor to invest in new dup15q projects, and generate results that can be used to apply for large-scale funding by agencies such as the National Institutes of Health. It is exciting that the Alliance is now in a position to drive the direction and quality of research that hold promise for improving the lives of individuals with dup15q syndrome.

# Cheers to Our Volunteers!

*International family conferences provide information, connection, and hope to families affected by dup15q. They are only possible because of the many volunteers who step forward to help with planning and then carrying out the thousands of tasks associated with a successful conference. We cannot individually recognize every conference volunteer in this article, but if we could, we would! Instead, we would like to honor several volunteers who provided foundational support for Stronger Together with everything from supporting international families, to coordinating the Alliance store, to playing major roles in the planning and preparation. The Alliance thanks these and all our conference volunteers!*

## FERNANDO GOMEZ ROBBINSVILLE, NEW JERSEY

In preparation for the 2015 conference, I was most excited about touching base with all the dup15q folks coming in from overseas, and coordinating the input and feedback from those who could not travel to Orlando. It was so great to have the opportunity to hear the voices from the international families who have so much energy and determination to help their children reach the next milestone, as well as the energy they devote to help other families in their home countries - from Australia to Switzerland! Volunteering with the Alliance gives me an opportunity to help other families "see the forest from the trees", the forest being our global dup15q family!



Fernando Gomez looks on as Anne Tiplady from Victoria, Australia presents at The International Families Meeting

## KATIE SUPINA NEW BRIGHTON, MINNESOTA

I really enjoyed helping with the speakers for Stronger Together. We had so many speakers this conference that presented for much less than they could get for speaking at other international conferences. They did it because they believe in our kids and the organization. I learned so much by working with all of them. The best part of the conference for me was the opening parade. It made my heart so happy to see all our beautiful kids walk through the huge audience and get cheered one by one. They all deserve to be honored in that way. Volunteering with the Alliance makes me feel like I'm doing something that has value. Like everyone, I'm busy with work, family and life but this group is important to me, so I make time to help out.

## PAUL KARCH MADISON, WISCONSIN

I was most excited about helping with being a room monitor so I could meet, talk with and introduce speakers. My favorite part of conferences is sharing experiences, struggles, and laughs with old and new friends. Volunteering with the Alliance makes me feel that I am helping families and people with dup15q feel the support of a community, and I get to learn about research, treatment and services that will help them live fuller and happier lives.

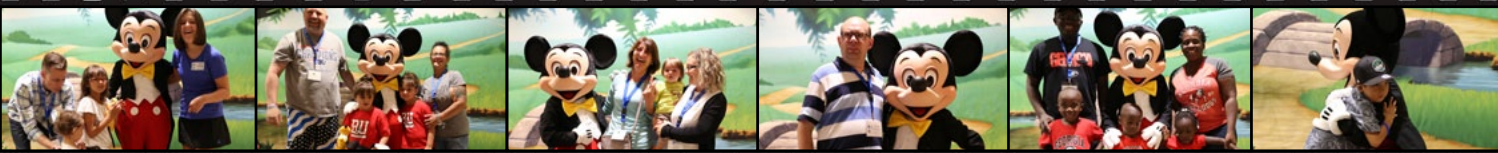
## ALISON KALNICKI VANCOUVER ISLAND, BRITISH COLUMBIA, CANADA

One of my favorite ways to help during this conference was organizing a table for some of the older youth and adults with dup15q syndrome to sit together at lunch. I always love meeting so many kids and adults with dup15q syndrome. Volunteering with the Alliance makes me feel positive about my son's life. I meet people who inspire me, and I learn so much about idic(15). I really enjoy donating my time and energy to an organization that has done so much for my family, and I hope it helps Dup15q Alliance to be there for many more generations of "dupers".

## KAREN SALES ANKENY, IOWA

For the 2015 conference, I was most excited about helping with EVERYTHING! As one of the conference co-chairs, I had a hand in everything. I love everything about conference, and I love making the whole conference experience the best I can for everyone. The best part of the conference for me was seeing all my old friends, and making new ones! And watching other families make and renew lasting friendships. Volunteering with the Alliance makes me feel like I have a purpose! I love knowing that I'm helping make William's life and the lives of all the other kiddos with dup15q a little bit better.

The first **Believe Walk** raised \$60,000!  
If you are interested in hosting one next year, contact us at [info@dup15q.org](mailto:info@dup15q.org).



## The Beauty of dup15q

The Dup15q Alliance is fortunate to have a longstanding relationship with Rick Guidotti. Rick is the founder and director of Positive Exposure ([www.positiveexposure.org](http://www.positiveexposure.org)), an innovative arts, education and advocacy organization. Rick's photos capture the beauty and joy of people living with genetic, physical, cognitive and behavioral difference. It is always a treat to see the beauty of dup15q as captured by Rick, and here are some of the *Stronger Together* conference photos from our remarkable friend.



# Dup15q Alliance Financial Report

By TOM DOYLE  
Dup15q Alliance Treasurer

The Dup 15q Alliance takes the responsibility of handling the resources that you have entrusted to us very seriously. As of this writing, our financial position is strong. Our balance sheet at the end of August has slightly over \$496,000. We still have the large conference hotel bill to pay as well as several other conference related expenses, but we remain in good financial standing because of the generosity and persistent fundraising of so many of our families and friends.

You have heard, I am sure, that we committed resources to fund “priority projects” including two \$25K per year research grants awarded at the conference (see Carolyn Schanen’s Dup15q Alliance Funds Research article for more on this). One of these is a three-year grant, the other a four-year grant. In addition, we are investing \$40K in funding the development of a data base for our dup15q clinics so that we can share information to understand and treat individuals with dup15q more effectively. We are excited about the possibilities of this research and data.

For 2015, our annual budget income goal is \$367K. We have raised around 50% of that goal, with several Believe Walks and end of the year fundraising to come.

The Alliance works on a two year budget cycle as one year we have a conference and the next we don’t. So our 2015 expense budget is \$498K, of which the conference will account for nearly \$100K in expenses. We are currently at about 30% of our projected expenses, so we expect to come in under budget. In 2016, our expense budget will be significantly lower.

We are transitioning from being a family run organization to one that can clearly sustain itself and maintain professional leadership.

We are blessed to have Kadi as our Executive Director, but know that, at some point, we will transition to a full-time director. Kim is awesome providing support and leadership behind the scenes, making sure that everything from the website, to fundraising mailings, to conference details are all taken care of in a professional manner. In addition, we are engaging other people to assist with tasks including mailings as well as more complicated projects like our walks and fundraising events. All of these require additional funds, but if we are to grow as an organization we need to think differently about leadership, funding, and succession plans. We are budgeting with that in mind, and currently have nearly \$100K budgeted for such positions. We don’t have them all staffed so we won’t spend that much, but we felt we needed to be ready to make those moves when the time was right.

We budgeted \$15K this year to focus on our International Families as well. Those funds will be used to support dup15q related organizations in other countries as they develop their organizations and websites. Government and medical decisions are made very differently throughout the world, so we need to be prepared to assist those organizations as they grow rather than presume that our model works everywhere.

If you have any questions about the budget or our thought processes on developing it, please feel free to contact me at [tom.doyle@dup15q.org](mailto:tom.doyle@dup15q.org). I would be happy to answer any questions.

Thanks so much to everyone who has and who continues to support the Dup15q Alliance through your generous donations, fundraising, and gifts of time and talent. We obviously couldn’t do it without you. Together we can make a difference!

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## Reflections from a Sibling Stronger Together

By CHAS KARCH, BROTHER OF RACHEL (dup15q)

I had not expected to come face to face with younger versions of myself. As I sat behind the table on the stage at the Sibling Panel, I had expected to speak about childhood memories and experiences, not be speaking to them. As siblings on the panel and in the audience shared their experiences of having a brother or sister with dup15q, I saw in them my own past.

There was me in 7th grade talking to my friend on the phone, asking if I could come over to his house, because I didn’t want to go through the effort of explaining why our VHS collection was more likely to feature *101 Dalmatians* than *Dude, Where’s My Car*. There was me in 4th grade biting my tongue in shame as one classmate stutters and flails his arm “like a retard”, while others look on and laugh. There was me in my first year of college arguing with a friend about how my sister was no different than any other, even though I never texted her and wasn’t her Facebook friend.

There are happy memories too, of course, like going to Cheeseburger in Paradise on Rachel’s birthday to celebrate her favorite food and one of her favorite songs. Walking up to the front of the line at Space Mountain twelve years ago with my sister Rachel and the rest of our family to ride the roller coaster she loves, but hates waiting in line for. Unfortunately, Disney has changed their policy on tag-along younger brothers skipping lines, but Rachel was better at waiting in line this time. There were also memories, feelings and thoughts that I had forgotten I had at that age, just as one forgets what it’s like to truly feel the excitement of Christmas morning as a six-year-old.

Perhaps I am nostalgic because at 24, I am practically ancient by dup15q sibling standards, but it was a nice trip down memory lane. I had not expected to learn so much on a panel where I thought I would be the one to share with the audience the real scoop on having an older sister who really acts younger, has seizures, needs to be taken care of, and thankfully never criticizes my fashion choices. I am grateful for the siblings and parents who attended the panel, as well as Dr. Naseef for leading the discussion. In just one short session we uncovered the complicated mix of the highs and lows, the single word conversations and multi-syllabic medications, the worries and joys, and the alienating differences of having a family member with extra pieces of chromosome 15 that brought all of us in that room together.

I cannot summarize the entire experience of what was shared at the sibling panel, but I will share this. Many of the parents in the audience said something like, “I can’t believe our kids are worrying about the same things we are”. The fact is that while caring for a sibling or child with dup15q is very different than a genetically typical child, it is very similar to the experiences anyone caring for another child with dup15q has had. Like I said before, I didn’t remember many of the points shared by other siblings until they reminded me I felt that way once too.

There were many siblings in that room from different backgrounds and even different countries, but we shared many of the same experiences and thoughts. Walking around the halls of the Hilton after the panel, I talked with other siblings about experiences I have only ever talked about with my own family, even though I had just met them. This is why I will try to keep coming to dup15q conferences, and I know why many families return. The sense of community is priceless and is hard to find. There was talk about reviving the sibling Facebook group at the panel and I hope it happens. If there are siblings who are looking for someone to talk to who knows what it’s like, please put them in touch with me. I might not have any answers, but I can at least say, “I’ve been there”. I highly recommend that there is another sibling panel in two years and that all siblings attend.



Rachel, Lydia and Chas Karch enjoying each other during Stronger Together

# I Had No Idea...

By ROBERT SWIRSKY, RAISING HIS SON IN SCOTTSDALE, AZ



ROBERT, HOELDON AND SUSAN AT STRONGER TOGETHER

I thought it would be wonderful to have another child. I had no idea....

I had no idea how far from any dreams of what I considered 'normal' I would find my life. I had no idea of the joys that I would experience in not 'normal'...

Susan and I each had two children from prior marriages. Susan had a daughter (9) and a son (12) while I had sons (10 and 11) at the time we discovered Susan was with child. I came from a family of 5 children; we had both been single parents for most of our older children's lives. This seemed to be the opportunity for a wonderful new chapter for us and our children.

Hoeldon Day Swirsky was born in September, 2009, at 5 pounds and 10.2 ounces, and 18 3/4 inches long. He needed help with oxygen right after birth as he was not breathing. While this sent my shock and fear to levels I did not know I could attain in two seconds time, the medical staff quickly resolved the situation, and all was well within a few minutes. Hoeldon was a beautiful baby. I was excited to be a father again. At the hospital, before completing the birth registration, the staff tried to convince me that the common spelling was 'Holden'. I insisted on spelling it 'Hoeldon', not realizing it was a unique name for a very unique baby. I had no idea...

Hoeldon was a good eater. He slept in Mom and Dad's room in the same bassinet that his sister Lauren and brother Nick slept in as babies. Hoeldon was a happy baby, and enjoyed his baths and being held and talked to. At his 4 month well check, the pediatrician noticed that he was not rolling over and seemed to have very low muscle tone. We were referred to physical therapy. We were also referred to a pediatric neurologist. Hoeldon appeared normal, so we were not anxious.

The physical therapist suggested to Susan that she call DDD (Department of Developmental Disabilities) for Hoeldon. Mom said, "No, that's for people in wheelchairs with a disability." A month later the therapist suggested calling DDD again. Susan knew that she was serious this time. Hoeldon was given a diagnosis of torticollis, meaning that he preferred to only look in one direction and sleep with his head in one position. Every time Hoeldon's diaper was changed, Susan did neck stretches with him. Hoeldon developed plagiocephaly or 'flat head syndrome' and was fitted for a DOC band. A DOC band is a funny looking white helmet used to reshape the skull. He wore this helmet for a few months. In the interim, Hoeldon saw the neurologist and had some basic blood tests run. Everything came back normal, so the neurologist suggested continuing physical therapy and scheduled a follow-up visit three months out.

The next visit had the neurologist asking, "why is this baby not able to hold up his head?" We were to go back in another three months to decide what to do next. At 12 months, although able to sit independently, Hoeldon was still nowhere near typical developmental milestones for a one year old. Hoeldon was eligible for early intervention services through DDD, and started Occupational Therapy and Developmental Special Instruction in addition to Physical Therapy.

At 12 3/4 months of age, the Pediatric Neurologist suggested that a chromosomal microarray analysis be obtained. At the time, neither I nor Susan had any idea that the blood test would give us real answers. Rather, we thought it just another routine test. Everything else seemed not ideal but at least 'ok'. He was a cute and chubby little baby, just a late bloomer we thought.

I will never forget the phone call from the pediatric neurologist at 9 a.m. on a Monday morning in November 2010, when he said that the chromosome test which he expected to come back normal was in fact abnormal. The rest of the brief conversation was a blur, except I remember asking, "Is there any way that this is going to just be ok?" He replied, "No, this will never just be ok." He said we needed to see a geneticist as soon as possible...crying, tears running down my face, I took down the

number of a geneticist he gave me. I immediately called and she had a cancellation in an hour and half...we were on our way.

The geneticist explained that Hoeldon's diagnosis was very rare. She gave us some material from John's Hopkins website that stated there were at the time 267 known cases of idic(15), that there was a 70% chance our son would be severely mentally retarded, that he would never be able to live on his own. She gave us a printout from the website that explained the main characteristics. We were shocked to say the least. We had no idea...

The geneticist suggested we visit the Unique Rare Chromosome website. Unique eventually led us to the Dup15q Alliance, although we did not become much involved with it. It seemed to us at the time that this was a condition which was not going to change and that there was little we could do. It seemed that therapies would do little and with both parents working full-time while battling our own demons, Hoeldon made little or no improvement. He did attend two years of a public, mixed class (typical and non-typical) pre-school with regular visits from speech, occupational and physical therapists.

The next few years saw Susan and I struggle individually with our own personal emotional, mental, and career difficulties while at the same time facing an apparently irreparable domestic situation. Through this all, God has looked after Hoeldon in ways that can only be described as miraculous. I had no idea...

In August 2014, I assumed responsibility for Hoeldon on a full-time basis. He was barely speaking, stimming a lot and spent his after pre-school hours at a typical daycare, much of it stimming on his own. It was quite a challenge for me. Due to my own struggles, and our domestic situation, I had rarely

seen Hoeldon for more than court ordered Saturday afternoon visitation for the prior three years. I had not played an active role in his caregiving nor advocacy. I wish I had seen the importance of advocating for my son, for his therapies and for his opportunity to progress past what I thought was a static prognosis. I had no idea of his potential to bring purpose and joy to my life, and to grow and thrive in his own life. I had no idea of the tremendous support of the myriad individuals who together actually are the Dup15q Alliance, the caregivers, friends and therapists that God would send to help me help Hoeldon....I had no idea....

Happily, Susan and her older children are in Hoeldon's life. He loves and needs them very much. We communicate about Hoeldon's schooling, therapy and medical needs.

This August, Hoeldon started 1/2 day kindergarten at the Arizona Autism Charter School. The school is the only charter school in Arizona for autistic students. It is in its 2nd year of operations; admissions are by lottery, and we were on a wait list. The curriculum is delivered via Applied Behavior Analysis (ABA). Board Certified Behavior Analysts supervise teachers and staff; class size maximum is 9; and 3:1 student-to-staff ratio. The school provides PT, OT and Speech therapy. Through a combination of DDD funding, and insurance I have arranged for Hoeldon to also receive outside speech therapy, physical therapy, occupational therapy, and will soon begin outside ABA therapy.

These days, Hoeldon talks and laughs a lot, approaches and says hello to strangers everywhere he goes (yea, yea, I know...) enjoys his trampoline, Legos (does not build anything yet), Sponge Bob, playing with landscape stones, and most of all swimming.... I had no idea.....

## Reflections from a Grandparent Our New Ohana

By CRIS MERIA, URIJAH'S TUTU AND NEW ALLIANCE FAMILY MEMBER

Attending the Dup 15q "Stronger Together" conference was an overwhelming experience. I had many warm and touching moments as I met with new "Ohana" (Hawaiian for "family") and friends who were willing to share their tender stories with me. I also need to mention how joyful my daughter-in-law, Aiisha was when other families reached out to her and my grandson, Urijah, to welcome them to join the activities.

How joyful it was to see the families come together and celebrate "themselves". It was a time we could all put aside how overwhelming and stressful it can be to care for a child with dup15. Seeing the other children with their parents was such a comforting time, and eased the heartache I kept so long inside of me. I recall the moment Tom Doyle encouraged the opportunity to share stories from the extended families in the

room, and how the young woman (also from Washington) put her arm around me to comfort the pain I was feeling. I felt it was my time to share my anxiety and concern as a grandparent. I felt he could sense my anticipation of wanting to talk, and so I did! Out flowed my feelings and an overwhelming amount of tears that soon made me feel like a whole lot of weight was lifted off my chest.

As the conference continued, I found other sessions to be very informative and educational. I now can articulate Urijah's condition more clearly and confidently, knowing he is going to be okay. Aiisha and I also discussed other concerns we did not think of initially about finances and trusts. We now have a clearer understanding and can make better decisions for Urijah's future. This finance class was truly an eye opener for me and Aiisha as well.

In closing, I want to express my deepest gratitude to all of the staff members and to our new Ohana, for the warm welcome and special gifts bestowed upon us while at the conference. Our family has been truly blessed by your acceptance of us, and Urijah now has "forever friends" to look forward to meeting again. We have shared our experiences with all of our family in Hawaii (and me in Seattle) and have plans to have more of our extended family experience the next conference in California! Mahalo Nui Loa.



*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.*

## HOLIDAYS ARE COMING!

And what better gift than one that supports the Dup15q Alliance and helps raise awareness of dup15q syndrome? The Alliance store has several new items. Check them out!



- Silver Believe Bracelet
- International Believe Shirts (youth and ladies)
- Fleece Zip Ups
- Baseball hats
- Golf Shirts

- License Plate Covers
- Drawstring Bags
- Water bottles

Order your Alliance merchandise at  
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