

# THE MIRROR

The Newsletter of **DUP15q ALLIANCE**

Support is a  
*Verb*

Nicole Cleary, MIRROR Editor

Support is a verb. So when we say the Dup15q Alliance is a family support organization, we mean that there is action, energy and intention of many individuals combining to create help and hope for families affected by dup15q syndrome. This action, energy and intention flow through every article in this issue of *The MIRROR*, and frankly, it is pretty awe inspiring. I am inspired by the individuals who find ways, both big and small, to lend their energy and intention to the Alliance while they are raising a child with dup15q. Equally inspiring is the involvement of extended family, clinicians, and researchers who contribute their energy and expertise to the growing effort to understand dup15q syndrome and develop targeted treatments.

In this issue, you will learn about changes coming for the Dup15q Alliance in 2016. Tom Doyle, Dup15q Alliance board chair sums it up perfectly in his introductory quote by John Maxwell: Change is Inevitable; Growth is Optional. The Dup15q Alliance has experienced phenomenal growth since its beginnings in 1994, and Tom provides a glimpse into the growth opportunities that the Alliance Board is focused on this year. Kadi Luchsinger announces her transition, which will bring a change to the leadership of the Alliance.

You will read updates on the work of the dup15q clinics and the international dup15q medical registry. The registry is a prime example of how families have invested their energy and shared their information to help our community better understand dup15q syndrome, increase awareness, and promote research.

You will read about a published article on gastrointestinal issues in dup15q. The work of the dup15q clinics is translating into journal articles which helps raise awareness of dup15q and provides pediatricians and other medical professionals with increased understanding of some of the treatment issues in dup15q.

As the Alliance ages, so do the children whose families got involved in the early days of this organization. Because the support needs of Alliance families now span all the way from prenatal diagnoses through adulthood, *The MIRROR* will be featuring articles relevant to both young children and to adults. In this issue, you will read about guardianship and how this process differs depending on where you live.

As always, you will meet a dup15q family through the family portrait article. You will also be invited to remember Michael, a young man with dup15q who passed away recently. These stories bring to life what it is like to raise a child with dup15q.

It is my hope that as you read this newsletter, you too will be inspired by the breadth of action, energy and intention represented here. Perhaps you will reflect on whether you are in a place to add to this effort, and help create even richer support for families. Check out the article about getting involved with the Alliance. It is the combined energy of each of us that make the support of this family support organization possible.

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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>

# Change is Inevitable; Growth is Optional ~ John Maxwell

TOM DOYLE, DUP15Q ALLIANCE BOARD CHAIR

The Dup15q Alliance mission is to provide family support and promote awareness, research and targeted treatments for chromosome 15q11.2-13.1 duplication syndrome.

Change is in the air and seems to be the only thing we can count on for sure. The real question is how we adjust and grow when that occurs. People often compare the age of organizations to the age and development of a person. If that is true, the Dup15q Alliance is quickly approaching our mid-twenties with the life and energy of those years, but also with the changes and challenges that come with that stage of life as well.

We are growing and represent around 1,000 families now. We are expanding our involvement in the science and genetics of dup15q as we grow our clinic relationships, fund research projects, and bring scientists, doctors, and families together at our bi-annual conferences and our annual science meetings. The organization, like a twenty-something, is trying to manage new income levels, resources and growing donations with the new expenditures required to support all the needs associated with our mission.

We see change in our board make-up as we brought on three new members this fall, including Tessa Quinlan, Elaine Lowell, and David Gifford. They all bring great passion and expertise to the organization. We recognize that we had more applications and more qualified candidates for the board openings than ever before, and are so grateful to those who applied and whom we hope to engage in committee work and eventually board work as more positions open up. We said goodbye to board members, Karen Sales, Paul Karch, and Linda Meagher who gave so much of themselves, their time, talent and wisdom to all of us.

We know there will be changes in the near future in our administrative team (see Kadi's transition notice below). We are going to the west coast for a conference for the first time in 2017. We are working with our Italian counterparts to help facilitate a science meeting this fall in Milan, and with our Australian families to work through the process of setting up their country's version of a non-profit.

There is life and vitality and the excitement of a growing family, new opportunities, and visions of what we can be that comes with age. We have hopes and dreams of what we will yet become, and a commitment to pursue that.

We aren't perfect. We make mistakes as we go, but we keep plugging on, trying to learn lessons from our errors and choosing to embrace the change and to grow. We thank all of you, our families, for supporting us in our mission, for reminding us when we are getting off track, and for volunteering and participating in so many ways. We are all walking this together, maturing and growing together. Thank you for your hours and hours of time, for your wisdom and suggestions, and for your financial commitment to help us move into our thirties!

Sincerely,



Tom Doyle  
Board Chair



## Transitions KADI LUCHSINGER, EXECUTIVE DIRECTOR

To my dup15q family,

This is the hardest article I have ever written for the Alliance. I have been involved in the Alliance board since 2004, and started serving as the Executive Director in 2007. It is with much thought and reflection that I have decided to end my term June 30, 2016. The time has come where I need to step back and focus on my family. My children are getting older and I want to be able to spend uninterrupted time with them.

I am so proud of all that we have accomplished - the clinics, the registry, the science meetings, family conferences, the growth of our family, and too much more to name. This has been a journey that has been driven by you all. I have felt your passion and drive to help our children and that has fueled me to work around the clock for you all. I have made some lifelong friendships between the professional advisors, researchers and with you all. It has been an honor and a privilege to be your voice. We have done great things together, and I know we will continue to do the same.

The board has started on a search for a new Executive Director and I have promised to guide the new Executive Director in their role. I will always be supportive of the Alliance and will be here to help as needed. Thank you all for helping to make the Alliance so successful. I am proud and humbled to be the one to lead you all for the past 9 years - we have accomplished so much. I am still a part of the family always and excited to continue supporting this great cause. I still believe with all my heart that together we can make a difference.



# Issues in Early Adulthood Guardianship

Anne Karch interviews her husband and lawyer about how guardianship works in Wisconsin. Anne and Paul Karch, are parents of Rachel (29 yrs old, idic(15)). In the United States guardianship is part of state law, so the rules vary from state to state.

**A: What is guardianship?**

**P:** Most children under the age of 18 have very few legal rights or powers. The law assumes they are not capable of making decisions on their own, so their parents have the power to make those decisions for them. Once a child turns 18, he or she automatically gets all the rights and powers of an adult. Those include the ability to sign contracts, such as enlisting in the military, borrowing money, getting a cell phone or credit card, or owning property, and the ability get married, to vote, to sue or be sued, and to apply for and receive government assistance.

**A: I am Rachel’s mother, why did I also need to become her guardian?**

**P:** Because the law doesn’t automatically know that Rachel is disabled. Once she turned 18, being Rachel’s mother gave you very little power over decisions made about her. Many people do not understand this about their other children. If your typical child decided to get married on her 18th birthday, she could do that, even if her parents thought it was a very bad idea.

**A: Why did we need to hire a lawyer to become guardians for Rachel - didn’t I marry you to get this kind of stuff for free?**

**P:** Because guardianship is a specialized court proceeding for which you want to have a lawyer who knows and has experience in the applicable law. Setting aside whether a lawyer should ever represent himself, this area of law is far from what I do or know, so we hired a lawyer to do this for us. It might be possible in some states to work without a lawyer and some states might also provide lawyers or support to people who cannot afford to hire a lawyer on their own.

**A: Are there deadlines parents need to be aware of?**

**P:** You want to have the guardianship process completed before your child turns 18. You need to begin the process with enough time to get it done, and you need to make allowances for the processing time in your local court. You can do it after your child turns 18, if you haven’t done it yet. However, if you wait till after your child with dup15q is 18, you run the risk of not being able to make decisions for him or her when you need to, if there is an emergency.

**A:** I remember someone telling us to start on Rachel’s 17th birthday because it’s always a temptation to say, “I’ll take care of that later,” and then forget, or become wrapped up in other aspects of her care. Suddenly she’s 24 years old, and needs some sort of medical care, and you’re not able to authorize it.

**A: What does it mean to be a guardian?**

**P:** It means that you act on behalf of your adult child in legal matters. So, when Rachel is being admitted to the hospital and would normally have to sign consent forms, you or I sign the forms on her behalf. When Rachel accepts Medicaid Waiver services, we sign all the forms for her.

**A: Wasn’t there another lawyer involved?**

**P:** Yes, in Wisconsin a person subject to guardianship proceedings must have a “guardian ad litem”. This is a lawyer who represents the child in case the parents are seeking unnecessary control. The lawyer interviewed Rachel, reviewed reports and talked to others to determine her ability to make decisions for herself.

**A: You said Rachel’s guardianship proceeding was pretty straightforward. What would make a more complicated case?**

**P:** We are Rachel’s guardians for all purposes because the court concluded that Rachel did not have the capability to exercise any of the adult powers we talked about before. She couldn’t make an informed decision in an election, for example. Sometimes a person with disabilities might need a guardian for some reasons but not for others. A person might need help managing their money but not for deciding whether or not to get married. Those kinds of judgments would be more difficult than the decision about Rachel.

**A: How does a judge decide whether to appoint parents as guardians?**

**P:** The judge considers the information put before her, including what the parents say, a doctor’s report about the person’s cognitive abilities, and what the guardian ad litem reports. The judge may also ask a few questions of the person for whom the guardianship is being requested.

# Guardianship Around The World

The legalities of appointing someone to make decisions for an individual with intellectual disabilities as they move into adulthood differs from country to country and region to region. Have a look below for some examples.

	<b>Australia</b>	<b>Italy</b>	<b>Canada</b>
<b>What's it called?</b>	Decision Maker Adult Guardian	Administrative Support Trust	Adult Guardianship Trust Legal Representation
<b>Regional rules?</b>	Different for each state	Same process for whole country	Different for each province/territory
<b>Age of adulthood?</b>	16 - 18 years old	18 years old	17 – 19 years old
<b>When to start?</b>	Before it is needed	At least 6 months before 18 <sup>th</sup> birthday	Before it is needed
<b>Who does it?</b>	Centrelink, the gov't welfare system	Charitable institutions Lawyers	Charitable institutions Lawyers
<b>Where to get more info?</b>	Office of the Adult Guardian	Charitable institutions Internet	Government and advocacy organizations internet sites

*Thanks to the following for providing information, compiled by Alison Kalnicki*

Laura Piredda, Italian mom of Greta Casule (5 yrs, idic(15)) and President, Italian Association Nonsolo15

Joan Lammas, Australian mum to Stew (23 yrs, dup15q)

Alison Kalnicki, Canadian mom to Logan (14 yrs, idic(15))

## Remembering Michael John Hauler



On December 2, 2015, Michael John Hauler, 35, died peacefully in his sleep. For the past 28 years, Michael lived with his mother and stepfather, Jeanine and Bill Rossbach, for the last 8 years in Traverse City, Michigan. Michael is also survived by his sister, Julia Hauler of Garden Grove, California, birth father John Hauler of Anaheim, California, his step-brother Ben and nephew Nick Rossbach of Ramona, CA.

Though he lacked the power of speech, Michael conveyed his kindness and loving nature with a warm smile, a gentle touch, and the kind of enthusiasm parents pray for. He was much loved by all who took the time to know him. He was known at his adult day program as a silent jokester with a funny sense of humor. Michael was the light of our family's life and our greatest teacher. He showed us how to be present in every moment and enjoy life to the fullest.

Michael never met an adventure he didn't want to pursue. He traveled extensively with his teacher parents, visiting most U.S. states and many Canadian provinces. He camped at many National Parks from Hawaii to Maine. He loved seeing new places. He hiked around the Grand Canyon Rim, rainforests, beaches, geysers, waterfalls, etc. Two weeks before he passed, he was determined to follow his visiting step-brother and nephew up the steep sand dune climb at Sleeping Bear Dunes National Lakeshore, a challenge even for the able bodied. He always was curious to see what was down the road, beyond the door, over the horizon.

Michael's life was made richer by the caring staff at his adult program who gave him art, music, dance, took him bowling and to the theater weekly as well as to the beach, on picnics, on scenic drives, horseback riding, to local fairs and festivals, to museums, adapted skiing, and more. His fellow riders, aides, and drivers on his daily bus ride became a little family to him. Traverse City medical and dental professionals gave caring and expertise that did much to ease his pains and improve his quality of life.

Michael's family will arrange a memorial gathering in late spring. We will arrange to have his final resting place be at The Sleeping Bear National Lakeshore on Lake Michigan, a fitting location to honor our dear, adventurous and gentle spirited young man. May his spirit rest in peace.

# Dup15q Clinics Update

JANE KIM, DUP15Q ALLIANCE BOARD

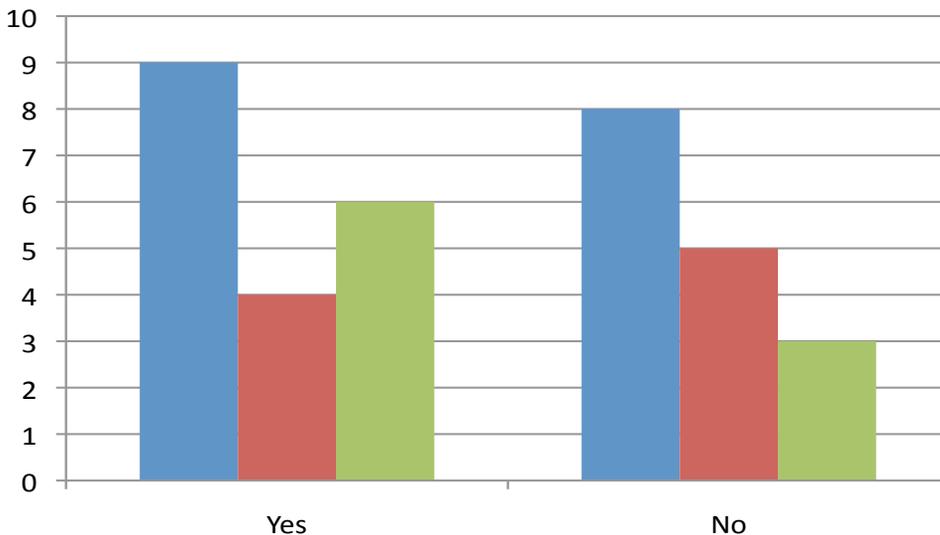
The dup15q clinics are thriving. The Memphis clinic is seeing 15 to 17 families each year, the Pennsylvania clinic is seeing 15, and Boston is seeing around 15 to 20 new families per year. Ron Thibert estimates he has seen about 90 families total in Boston, and he sees a good number of return patients in addition to the new families. In 2015, there were at least 50 new families seen in dup15q clinics. This is fantastic both for the level of expertise these families received, and for the increased understanding about dup15q that our clinicians get from each visit.

There has been a big focus to advance the dup15q clinical database. The dup15q clinic database was successfully launched at the Memphis site in 2014. This database complements the dup15q registry that parents complete. The clinics' database allows patient information from the various dup15q clinics to be pooled and collected into a central database. With a rare condition like dup15q syndrome, combining data from multiple clinics allows for

more robust findings and allows us to better characterize dup15q syndrome and find the most effective treatments. The Memphis clinic has been the first to enter data into this central database. The St. Paul Minnesota clinic is also in the initial stages of entering data. The Pennsylvania, UCLA and Boston clinics have completed or nearly completed all the necessary paperwork and will begin using the database in 2016. The Dup15q Alliance has funded the creation of this clinical database with \$80,000 given over 2 years.

The clinic database, like the dup15q registry, is starting to help identify the challenges faced by individuals with dup15q. For example, statistics extracted from the Memphis database using the 17 participants enrolled there show 9 of the participants struggle with insomnia and also struggle to stay asleep and over 50% struggle with falling asleep.

Sleep Habits



- Does your child have sleep problems?
- Does your child have trouble falling asleep?
- Does your child have trouble staying asleep?

As more patients are being seen in the dup15q clinics, the clinic heads have identified additional funding needs to help with ensuring a good clinical experience for families and streamlining research efforts. The Boston and UCLA clinics are two of the busiest clinics, and the Dup15q Alliance has pledged \$10,000 per year to each of these two clinics to help hire a clinic coordinator. The clinic coordinator will help schedule appointments with the appropriate specialists, make sure proper testing is arranged, set up follow visits, help with arranging transportation and housing, as well as enrolling patients into various clinical research studies at each site. The clinic coordinator will also help input data into the centralized database and help answer any questions from patients about the database.

The dup15q clinicians are very motivated and passionate about learning more about dup15q syndrome and helping individuals with the condition. There continue to be conference calls roughly once a quarter and an in person meeting once a year. In the near future, we hope to designate a small proportion of the dup15q clinics as “centers of excellence.” This will help identify those clinics who are participating in the dup15q central database and the lead clinicians actively engaged in dup15q research and collaboration.

# Gastrointestinal problems in 15q duplication syndrome...

EUROPEAN JOURNAL OF MEDICAL GENETICS, VOLUME 58, ISSUE 3, PAGES 191-193

ELIAS A. SHAAYA, SARAH F. POLLACK, SUSANA BORONAT, SHELBY DAVIS-COOPER, GARRETT C. ZELLA, RONALD L. THIBERT

The association between dup15q and neurodevelopmental disorders including intellectual disability, motor delays, autism spectrum disorder, language problems, and seizures is relatively well documented at this point. One of the less understood issues frequently affecting individuals with dup15q is gastrointestinal (GI) problems.

In the hopes of raising awareness of GI issues in dup15q among physicians and health care providers, the Dup15q Center at Massachusetts General Hospital recently published a study of gastrointestinal problems in 15q duplication syndrome in the European Journal of Medical Genetics. The aim of the study was to examine the frequency and severity of gastrointestinal problems in a group of children and young adults with 15q duplication syndrome.

A total of 38 individuals were included in the study. Thirty individuals had a diagnosis of idic(15), and 8 had int dup(15). Among this group of children and young adults, 79% experienced gastrointestinal symptoms. Of those affected by GI symptoms, 57% had gastroesophageal reflux disease (GERD), 60% had constipation, and 30% had both. The range of GI problems and the severity of symptoms varied.

Treatment for constipation consisted mainly of stool softeners and stimulants such as polyethylene glycol and bisacodyl. Proton pump inhibitors were the preferred treatment for reflux. These methods effectively treated the GI symptoms in most of the children and young adults with dup15q. Importantly, treatment of GI issues improved behavior, and the authors suggest that untreated GI symptoms may cause discomfort that causes irritability and, in some cases, aggression. They stress the importance of considering GI disease as a possible cause for behavioral difficulties in dup15q.

Ron Thibert, Director of the Dup15q Center for Massachusetts General Hospital for Children and one of the authors of the study answered a few questions about this study.



**1. In the article, the authors recommend considering GI disease as a possible cause for behavioral difficulties in individuals with dup15q. Based on the findings of frequent GI problems in this sample of individuals with dup15q, would you recommend parents seek GI evaluation as part of the routine well child care for a child with dup15q?**

*I don't necessarily think that each child needs to see a GI specialist for routine care, but I do believe families should be discussing GI issues with their primary care physician (or with doctors at a dup15q clinic if they go to one) at routine well visits. At that point there should be a low threshold to refer to GI if potential issues are identified.*

**2. Is there any plan to expand the research on GI issues in dup15q beyond this sample of individuals seen at the Dup15q Center at Massachusetts General Hospital?**

*Yes, we are in the process of building a large central database for all the dup15q clinics. The MGH case series was a nice first article to increase awareness of GI issues in dup15q but a larger, more systematic approach should lead to even better data.*

**3. It is very exciting to see new research being published out of your Dup15q Center!! Are there other papers in the works??**

*Yes, there are definitely more papers on the way. Among other upcoming publications, we expanded our EEG data at MGH to include data from at least 4 other centers. There are also publications upcoming from other centers, especially UCLA. We meet frequently as a clinics group to discuss ideas and collaborate.*



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## Alliance Welcomes Three New Board Members

DEIRDRE POPP, DUP15Q ALLIANCE ADMINISTRATOR

We are pleased to welcome three new members to the Dup15q Alliance's Board of Directors. David Gifford, Elaine Lowell and Tessa Quinlan joined the board in October. We give a big "thank you" to Paul Karch, Linda Meagher, and Karen Sales for their many years of service to Dup15q Alliance.

When David Gifford's granddaughter, Annabel Porath, was diagnosed with dup15q syndrome, his daughter, Sarah and her husband Mike shared information from the Dup15q Alliance with David and his wife. David found that the level of support the Alliance provides for families was something special, and this is the reason he wanted to be involved with the Board. When David was growing up, close family friends had a daughter with down syndrome. The advice at the time was to institutionalize her. However, her mom and dad believed she would be best cared for in their family. The family persevered, with a great deal of effort, and the last time David saw Vallie, she was in her mid-50s, living in a care home, working, and as always, happy. There were no support groups at the time like Dup15q Alliance.

Elaine Lowell's connection to Dup15q Alliance is her granddaughter, Grace, who was diagnosed with dup15q syndrome before she was one year old. Shortly thereafter her son and daughter-in-law (Jeff and Colleen Lowell) learned about IDEAS (now Dup15q Alliance). Grace is now 12 years old. She is a sweet, happy little girl who continues to struggle with seizures. The Alliance has provided an enormous amount of support to Grace and her parents as they have navigated the previously unknown world of dup15q syndrome, including autism, developmental delay and epilepsy. Grace's family is very grateful for the work of the Alliance and for all the support it provides. Elaine wants to be involved as a way to help Grace, her parents and all of the other children and families affected by dup15q syndrome.

Tessa Quinlan is the mother of 11 year-old Gavin, who has dup15q syndrome. She and her husband quickly accepted Gavin's diagnosis, but had a "hit in the gut" feeling. They soon realized they could tackle what came before them and found Dup15q Alliance. Gavin is verbal and highly energetic, full of life, no sign of seizures, but has a developmental delay. Tessa has been connected with Dup15q Alliance since 2009, and from the start she wanted to give back to a community who has given their family so much hope and inspiration. Their family has learned from the families who have traveled this path before them, and they hope to be that same inspiration to those who will join Dup15q Alliance in the future. She looks forward to serving on the board and aiding in the success of the Alliance for years to come.

## Dup15q Alliance

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# *The lives a very full life*

## *Family Portrait*



DEBBIE (MOTHER) AND  
SHANNON (DAUGHTER) NELLER  
ORANGE CITY, FL

Shannon wants to start this article by saying, “I’m incredibly thankful that God gave me my disability. I’m very blessed. It gave me an opportunity to go to Special Olympics, and to get Steve (my service dog), and to meet friends, and go to Camp Challenge and be BFF’s with my friend Maggie. It helped me meet other people with disabilities at my conference. People like me. That was another opportunity.”

Shannon was born 21 years ago. There were already 3 girls in the family and all of them were busy, happy, screaming girls. Life was busy. I had no idea how life was going to change for more reasons than adding another girl into the mix.

I remember thinking when they laid her on me right after she was born, just how ‘squoshy’ she was. I’m an Occupational Therapist, and after having 3 babies already I just.....had a feeling. I couldn’t go anywhere without her, and as an infant she screamed when she was in the car. Shannon had awful panic attacks even as a baby if I wasn’t there. When she was little if I was standing at the sink with my sneakers on, she would come up and start taking my shoes off. She would panic if she heard my keys jingle. Then she threw them in the trash.

Shannon didn't really speak clearly until she was almost six years old. She started out with a sort of throaty sound as an infant, and I soon realized she wasn't hearing us. Three sets of tubes, speech therapy, and asking God to "please let her talk" finally culminated in the fact that now Shannon rarely stops talking! She has more stories, questions, and comments than any child I have ever known. Did I say questions? Sometimes I marvel at how she even comes up with some of her questions, and I certainly find myself stumped in finding an answer in my limited repertoire.

We knew something wasn't right early on though. Shannon had torticollis as a tiny infant, and I took her to Easter Seals for therapy. They started OT, PT, ST and there began the long journey of trying to get our pediatricians to listen to me about my child. "Oh, she's fine!" "She'll catch up", "You're just over-thinking it". As time went by, she had CAT scans, MRI's, hearing tests, blood work, on and on, but still "she's fine, she'll catch up."

By the time Shannon was 5, she had been attending a Montessori preschool for 2 years. The teachers there were wonderful, and always let Shannon be Shannon. But one day Miss Mary told me that they didn't know what to do with her. She was still not able to fully communicate with her teachers and her peers, and we still saw the anxiety. I really prayed about it and I quit my job. I knew we had to do something different, so I brought her home and started home-schooling. After a couple of months, I called my mom in tears. Something was not right. In the way only a mom can, she said "Debbie, you call her doctor, and insist on genetic testing". Previously, we

had gone to a developmental pediatrician for one visit. He told us that she would never progress past the cognitive age of 9. He mentioned genetic testing and yet when I called back to have it scheduled he stated he had not suggested it, and that it was unnecessary. Now, after my mom's direction, we visited our regular pediatrician and I poured out all of the things chronologically that we had observed and been through with our little girl. That did it. Off we went to Nemour's Children's Hospital in Orlando to see a wonderful geneticist who ordered all the tests and ultimately told me with more compassion than I could have imagined that, "here we found something". I'll never forget sitting there as he spoke. Hearing, but not really hearing. Shannon had fallen asleep with her head on my lap, and I was stroking her beautiful brown hair. He told me Shannon had something called Isodicentric Chromosome 15. He said he really didn't have a lot of information on it, but gave me all he did have. He didn't know anyone else with the diagnosis, and I certainly didn't either.

I continued to home school Shannon. It was the hardest thing I ever did. One daughter in high school, one in middle school, one in elementary school, and

one at home. To be honest, I learned more about being a mom, being a teacher and definitely about being a therapist during those 3 ½ years than I ever could have imagined. Ultimately, entering school became the best option for Shan, and she was beyond blessed by the teachers she had over the years. She has had her challenges, but nothing she and I and her service dog Steve can't handle. Shannon has never had seizures, and my heart goes out to all the parents I met at the dup15q conference this summer in Orlando who live with those frightening episodes with their precious children.

My Shan is now 21. She has lots of friends, Steve (her dog), family who love her, and she volunteers at one of my elementary schools (yes, I'm still a school-based OT!). She takes the bus for special needs individuals from our home to 'work' each day, and lives a very full life. Shannon needs routine, but then I seem to thrive on it. Shannon likes to have things to do, but then I could just hibernate given the opportunity. She marvels at simple things, she keeps me grounded. She loves God, and she shows me unconditional love every day. I can't imagine life without her.



# International Dup15q Medical Registry Update

RYLIE MCHAM

As parents and caregivers of children and adults with dup15q, you have critical information that can help our community better understand dup15q syndrome, increase awareness, and promote research. Each family’s active participation in the dup15q medical registry makes the registry a more powerful resource. The goal is to publish the registry data to raise awareness within the medical community and open avenues for future research.

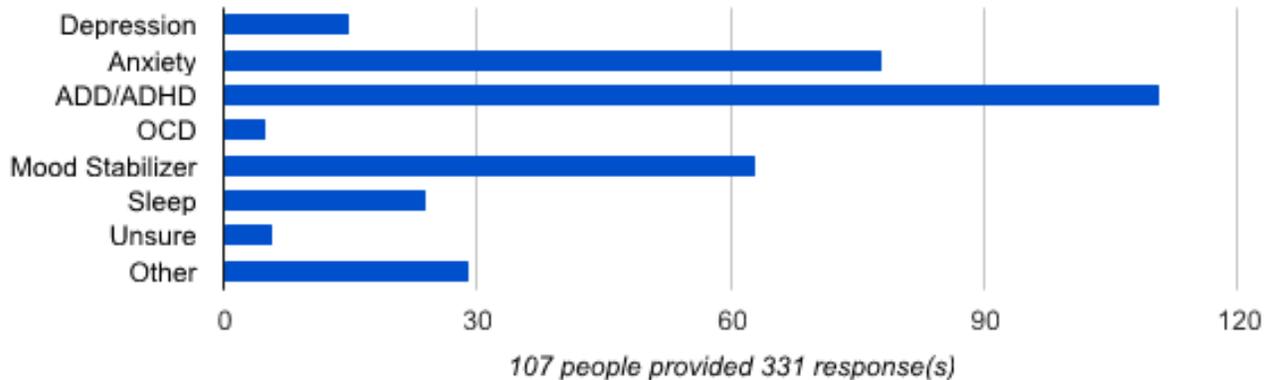
For those of you who are not familiar with the dup15q medical registry, the registry is an online parent/caregiver driven database that stores important information about the characteristics of those affected by dup15q syndrome. It can be accessed at <https://www.dup15qregistry.org/> The registry allows us to collect data to look for similarities, differences, and areas that need to be further studied. The registry provides researchers and scientists with accurate

and firsthand information, and as a result assists in developing a better understanding of dup15q syndrome. The registry allows families to organize and store medical and developmental information in one location. Storing information on one online database allows participants to easily access their information. The registry provides helpful information about dup15q syndrome which equips families to better educate

those who work with and assist their affected family member. For example, participants can print the registry graphs and charts and share this information with teachers, doctors, family members, aides and therapists. Registry participants can access such information by logging into their account, clicking on the “Explore” tab or by clicking on the pie chart icon next to the questions on the clinical questionnaire to view graphs like this one.

**Has the affected person ever taken any of the following behavior medications? (Please only select the medications that the affected person IS taking/HAS taken and leave the other medications blank.)**

Reason for taking - All Responses



To create an account families complete a clinical questionnaire and upload a genetic report. When completing the clinical questionnaire, it is important to answer every question. If you are not sure of an answer mark “unsure”, but please do not leave any question blank. If you are confused by any terminology used in the questionnaire, please use the glossary of terms which provides easy to understand definitions.

It is important for those with a completed account to update their account yearly. It is essential that the registry have

current information. To update your account simply log into your account, click on clinical questionnaire tab, click on a questionnaire subject, click on the paper and pencil icon and update your information.

Should you have any questions please email [coordinator@dup15qregistry.org](mailto:coordinator@dup15qregistry.org). The registry is a wonderful way in which parents and caregivers can help the dup15q community and researchers gain a better understanding of this complex syndrome!

# Get Involved



Want to help Dup15q Alliance, but don't know where to start? Check out our committees. There is something for everyone.

**FUNDRAISING:** This committee meets monthly via conference call. Help strengthen our fundraising plan and develop new fundraising ideas so we can invest even more deeply into family support and research.

**SUNSHINE:** This committee sends out birthday cards to our members with dup15q syndrome.

**CONFERENCE PLANNING:** Planning for 2017 in Los Angeles is beginning now. Help us develop our speakers, events and many of the details that go into making our family conferences such a wonderful event.

**REGIONAL GATHERINGS:** We are always looking for individuals to host a regional gathering in their area. The Alliance helps promote your gathering and can offer small stipends to help with the cost of hosting. These gatherings are especially welcome in 2016 since this is not a conference year.

As board member and regional gathering coordinator, Rylie McHam notes, "A regional gathering can be really simple...it does not need to be complicated or require a ton of work. Send out an invite to families in your region, pick a location...can be someone's house or other community location...consider the likes and abilities of the children/adults with dup15q (we have met at a recreation center where the kids could swim, my house and then the City Club), have people help with bringing food etc. The best part really is catching up with dup15q friends and meeting new families. It is such an opportunity and so worth the planning. Everyone seems to be thankful to have the opportunity to connect with other dup15q families. We have found that it is helpful to have a few people there to help out with the kids (extra set of eyes and hands) so the adults can actually have a conversation."

**FINANCE:** This committee meets quarterly to go review the finances of the Alliance. We are looking for individuals with a background in accounting, finance or banking.

**MEDICAL REGISTRY:** This committee meets quarterly to discuss the registry and ways to utilize the data as well as promoting the involvement of families.

**RESEARCH:** This committee focuses on promoting research into targeted treatments for dup15q syndrome. This includes organizing scientific meetings and monthly calls with the Professional Advisory Board, structuring research awards and fellowships, securing external funds for dup15q syndrome research via relationships with other organizations, explaining research to families, explaining families to researchers, updating the Dup15q Alliance research wiki, and generally fostering cross-fertilization between researchers in disparate disciplines. Some background in science is helpful, but most important is an inquiring mind and thirst to keep abreast of dup15q syndrome developments across many fields.

**FAMILY:** The Family Committee provides Dup15q Alliance families with support by way of regular communication regarding topics of interest. We recently surveyed our community to find out what topics they would like to hear more about. We conduct research on these topics and provide a regular column in our newsletter, *the MIRROR*.

**PUBLIC RELATIONS:** This committee meets via conference call quarterly to discuss ways to raise awareness about dup15q syndrome.

**WEBINAR:** This committee meets quarterly to help develop the webinar schedule of topics.

**WEBSITE:** This committee meets quarterly to review our website and look for areas that need to be updated or resources to be added.

**BELIEVE WALK:** Help to organize our largest fundraising walk on September 18, 2016.

**The MIRROR Newsletter:** Suggest topics, find authors or write articles, help with proofreading.

If you have a special talent that you can offer, we would love to engage you in one of our committees. To join, email us at [info@dup15q.org](mailto:info@dup15q.org).

*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.*

## ANNOUNCEMENTS...

### **\$15,000 IN 15 DAYS**

This annual fundraiser begins Valentine's Day and ends on Rare Disease Day. We are so blessed that we have a generous donor who has agreed to match our donations up to \$15,000!

### **RARE DISEASE DAY**

**FEB 29, 2016**



Bring awareness to dup15q syndrome and all rare diseases on February 29th 2016. Join other rare disease groups and wear a blue shirt, make a blue sign or get creative and post a picture of your group in blue and your location on the Dup15q Alliance Facebook event page: <https://www.facebook.com/events/1662432367361923/>

### **ANNUAL DUP15Q WALK**

**SEPT 18, 2016**

Save the date for the second annual dup15q The Alliance is hoping to have 10 host sites around the country. There is a 10 min video we can share to help you understand how to organize a walk. Please contact [info@dup15q.org](mailto:info@dup15q.org) if interested in helping. We will help all site coordinators in setting up and running the event. Be a part of the second annual Dup15q Walk - together we can make a difference.

<http://www.dup15q.org>

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