2014 Vol 1

THE MIRROR

The Newsletter of DUP15q ALLIANCE

Celebrating 20 Years

By Kadi Luchsinger
Executive Director of the Dup15q Alliance

I will never forget our first conference in 2003 in Philadelphia. We had just received Ethan's diagnosis, and had no idea what it all meant. We walked into the room and then walked out several times, not sure we were ready to face this. Walking into that room was one of the best decisions of our lives - we have amazing family, but this was a family unlike anything we had experienced. Being in a room of people who "got it", did not judge, and were so welcoming was a feeling I will never forget. We were not alone. The instant bond prompted us both to jump in with two feet to find a way to help. I have been involved in the Dup15q Alliance for over 10 years now. As we celebrate our 20th anniversary, I want to first thank Donna Bennett and Brenda Finucane as the founders of what was formally known as IDEAS, and Nicole Cleary and the early board members who really helped this group become a true non-profit organization. What started out as a handful of families has evolved into a membership close to 1,000!

There have been so many things to celebrate over the past 20 years! My favorite highlights are the family conferences, the registry, the annual scientific meetings, the mouse model (our first science grant), the incredible support network that we have created for our families, and the nine dup15q clinics we have helped create. We have not spent any money on the clinics to date, but we plan to raise $100,000 for a clinical network database. This database will ensure all clinics are following best practices, performing the same tests, using standardized forms, and looking for common findings. There will be more information coming on this as it is in the development stage, but we are excited to share this as an example of how we are moving forward in our 20th year with the same momentum we did in the early years.

So how do we celebrate such an anniversary? We are going to have an event every month! We hope you all will join in and help us celebrate 20 years of support for families affected by dup15q. Please make sure we have your correct email and mailing address as information will be coming via email and some through mail. You won't want to miss out!

Here are some highlights for the next 6 months.

January: We focused on promoting the registry. We will be publishing the early findings and wanted to make sure we had captured as many families as possible. We acknowledged our families' efforts by holding a Kindle Fire drawing for families with completed registry data. The winner, chosen Jan 31st, was Maria Leo! It is still not too late to complete the registry, www.dup15qregisty.org. Every story counts.

INSIDE THIS ISSUE

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.

Celebrating 20 Years

Continued

February: We held our annual $15,000 in 15 days starting on Valentine's Day and ending on Rare Disease Day. We surpassed our goal this year and have raised $55,000 with more coming in the mail! Thank you friends and families, and thank you to the two families who made challenge grants! On February 28th, Rare Disease Day, we celebrated by asking families to ***Wear blue for dup15q***. We will create a video on our You Tube channel with Rare Disease Day photos. Check it out at http://www.youtube.com/user/chromosome15q.

March: We will "Spring into Fundraising" with a list of ideas and tools to help families come up with a great event. The fundraising incentive program is a way for families to defray the cost of our next conference. The more you raise, the less you pay, and who doesn't want to go to Disney World? The next conference is July 30-August 1, 2015 at Disney World. Finally, in March, we also hope to launch our new website.

April: We will launch a large awareness/PR campaign. The Alliance needs families to help be a messenger and a voice for the Alliance!

May: 50 people 50K. We are looking for 50 people to walk/run in a race and try to raise $1,000 each for Dup15q Alliance. If anyone is interested in holding a race/walk please let us know. We can help.

June: 15q at the Zoo. We encourage families to hold a gathering at your local zoo. We also want to hear about any regional gatherings this year. We hope that every region has one, so if you are interested in helping please contact info@dup15q.org. The Alliance offers small grants to defer some of the costs for families. We think it is critical for us to get together, so let's make it happen.

This is the road map for the first 6 months with more exciting activities planned for the remainder of 2014, including the first joint scientific meeting with the Angelman Syndrome Foundation.

Thank you for being a part of this journey, because together we can make a difference.

Financial Update

Investing in the Next 20 Years

By Tom Doyle, Alliance Finance Officer

After 20 years, the Dup15q Alliance is well established as an organization. In just the last decade, we have grown from about 300 families to almost 1,000. We fulfill our mission by providing family support through the Mirror, biannual conferences, parent contacts, regional representatives, an informative website, Big Tent, social media, and a First 100 Days Handbook. In addition to family support, we have funded research, created a searchable registry, provided annual scientific meetings, established and promoted clinics in nine cities throughout the U.S., and developed relationships with other organizations around the world, both in the dup15q community and in related areas.

We have gone from an all volunteer organization to one that now pays a small stipend to our Executive Director and to an Executive Assistant who manage every aspect of the organization. Imagine the number of phone calls, emails, letters, meetings, grant proposals, donation responses, and other organizational tasks that have multiplied many times over the last decade. We are grateful to Kadi and Kim for their expertise, professionalism, and for their continued donation of their time and talent as their hours and their compensation do not match.

We have further invested in the continued growth of the Alliance by contracting part time with two experts in the field of fundraising. This contract will to help the board and our staff develop a big picture view of where we need to go to fulfill our organizational mission over the next 20 years. Deep down, we don't want our organization to get bigger as that means there are more families receiving a diagnosis of dup15q syndrome or duplications of chromosome 15q outside the critical 15q11-13 region. However, we know the reality is that there are many, many more individuals who will be diagnosed and referred to the Alliance as doctors and geneticists help direct families to us. We are growing exponentially and need to be prepared for that.

We have many dreams of how the Dup15q Alliance can help our families in the next 20 years, and are trying to position the organization to be able to do that financially. Your support and financial gifts do make a tremendous difference to all our children. Thank you so much for your dedication and participation in the Dup15q Alliance. We look forward to the next 20 years and to many new, exciting, and helpful advances.

What's in a Name?

By Guy Calvert, Member of the Dup15q Alliance Board.

Families often ask us, "What is the "official name" for dup15q syndrome?" After all, other syndromes have official sounding names. Mutations in the MECP2 gene lead to Rett syndrome. Individuals with trisomy 21 have Down syndrome. Deletions of the critical stretch of genes located on chromosome 15q11.2-13.1 results in either Angelman syndrome or Prader-Willi syndrome (depending on the chromosome's parent of origin). But too many copies of genes from the same region results in ***what*** exactly?

Over the years there have been many names. Families receiving a diagnosis for their child 20 years ago may have been told that their child had "partial trisomy 15", "satellite marker chromosome 15", "duplicated material from chromosome 15", "inverted Duplication of Chromosome 15", or "interstitial duplication of Chromosome 15". chromosome 15q duplications come in many shapes and sizes, isodicentric duplications (idic 15) or interstitial duplications (int dup 15), with various copy numbers and areas of duplication. Many of these names are just different ways of describing the same thing. As one might expect, this proliferation of names has long been a source of confusion.

Recently, we've been consulting with our Professional Advisory Board for a sensible naming convention. What they proposed is **"chromosome 15q11.2-13.1 duplication syndrome",** or **"dup15q syndrome"** for short.

Why did they pick such a coordinate-heavy name? In part, it was a desire to clearly describe the regions of 15q whose duplication will result in comparable clinical issues. This is a difficult task since the various types of chromosome 15q duplications play out clinically in slightly different ways, and even within the same class of duplication the clinical outcomes can vary quite widely from individual to individual. In defining which of these duplications to include in the syndrome, our advisors had to decide where to draw the line; lumping together the classes of duplications that produce sufficiently clinically similar outcomes and splitting out those that do not. The view of our professional advisors was that the cytogenetic coordinates "15q11.2-13.1" define the common denominator: that stretch of genes (also known as the Prader-Willi/Angelman critical region) whose duplications, either isodicentric or interstitial, are thought to disrupt normal development in related ways and produce recognizably similar clinical effects.

The short-form name is of course far easier to articulate, albeit less descriptive. The main virtue of "dup15q syndrome" is that it is already a part of existing usage. Increasingly researchers and clinicians writing grant applications, research papers, or speaking at professional conferences use the term "dup15q syndrome" to encompass the various types of duplications of genes in this region. Such name recognition in the research community is an extremely valuable thing from the perspective of families.

But if dup15q syndrome includes only those "core" 15q duplications spanning the Prader-Willi/Angelman (PWS/AS) critical region at 15q11.2-13.1, what should we call the 15q duplications that lie outside of that region? Individuals with these duplications generally exhibit distinctly different clinical characteristics than the core 15q duplications. It is, in many cases, uncertain whether the 15q duplications are even the cause. There are individuals with these duplications who report no clinical problems of any sort. That's why the recommendation of our Professional Advisory Board was to describe these duplications simply as just that - *duplications.* So, for example, for the microduplications near coordinates 15q11.2 or 15q13.3, just over the edge of the PWS/AS critical region, we would write **"15q11.2 duplication"** and **"15q13.3 duplication"** (or **"dup15q11.2"** and **"dup15q13.3"**, for short).

Some parents have asked, why not name dup15q syndrome after one of the researchers who first identified and characterized the syndrome, as with Rett or Angelman syndromes? We did float this idea, but our Professional Advisory Board, which is comprised of many of those dup15q pioneers (such as Brenda Finucane, Carolyn Schanen, Edwin Cook, and Agatino Battaglia) was of the view that naming syndromes after people is generally a bad habit, and particularly inappropriate in the case of dup15q syndrome. Such names do little to convey the nature of the syndrome in question, and the hoped-for mnemonic benefits are often fairly minimal. Descriptive names, such as dup15q syndrome, dup15q11.2 and dup15q13.3, adopted according to standard naming conventions are increasingly preferred by medical professionals and our Professional Advisory Board.

THE MIRROR

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20 YEARS of Family Support

In 1994, Donna Bennett, mom to Joshua (idic 15), and Brenda Finucane, Genetic Counselor, decided to collaborate and form a support group for families affected by the rare chromosome disorder then known as inverted duplication chromosome 15. There were initially 24 families in this young support group. These families have supported each other and the Alliance over 20 years. We asked them to share with us what 20 years of family support feels like.

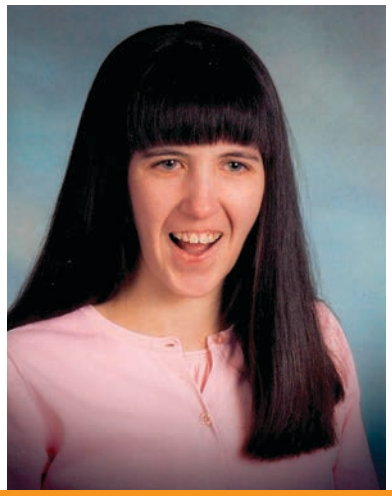
Donna Bennett, Mom to Josh, age 37

I was very reluctant to start this support group when Brenda Finucane suggested it, but it has been the best adventure of my life. The support is always available from the Alliance family. All you have to do is email your concerns or questions, and you will get an answer. I have talked to many of our families over the last twenty years and asked how they have handled different situations, and have found this information useful to pass on to the staff at Josh's group home and day program. It has been very beneficial, and has made Josh's everyday life a lot easier. While at conference this year, I was amazed how our families communicate even when they do not know our language. We had a family from Chile, two families from Italy, a family from Canada, and a family who could speak French, and they were communicating with each other even though I couldn't understand their languages. I have made friends from all over the world and thank each of you for your support to each other because you have been willing to share your lives with us.

"The support is always available from the Alliance Family"

Joanne & Len Poore, Parents of Lisa, age 23

Lisa was diagnosed with inverted duplication 15 when she was 18 months old. At the time she was 1 of only about 50 people in the world diagnosed with this chromosome abnormality. Lisa's speech therapist suggested that we register with NORD (National Organization for Rare Disorders) and through NORD we were contacted by Donna Bennett. Later, we were ecstatic to meet three local families at a regional dup15q gathering (Dawn & Paul Rivard, Kathy Swiderski and Paula Davis), each with daughters a few years younger than Lisa. Even today, Lisa still tells family and friends, that her friends Megan, Julia and Emma have the same diagnosis as her. We will never forget our first conference in Boston which we attended with many of our family members, and feeling overwhelmed by all of the information. It was amazing for Lisa and us to meet so many other children and their families. It was truly a dream come true. We have always felt blessed with the support of our dup15q families, but the support we received when Lisa had scoliosis surgery and again when she started having seizures at age 18 was truly unbelievable. We have been active volunteers because it has been important to us that we give back to such an amazing group which has provided our family with so much support and love. Happy 20th Anniversary Dup15q Alliance!!!



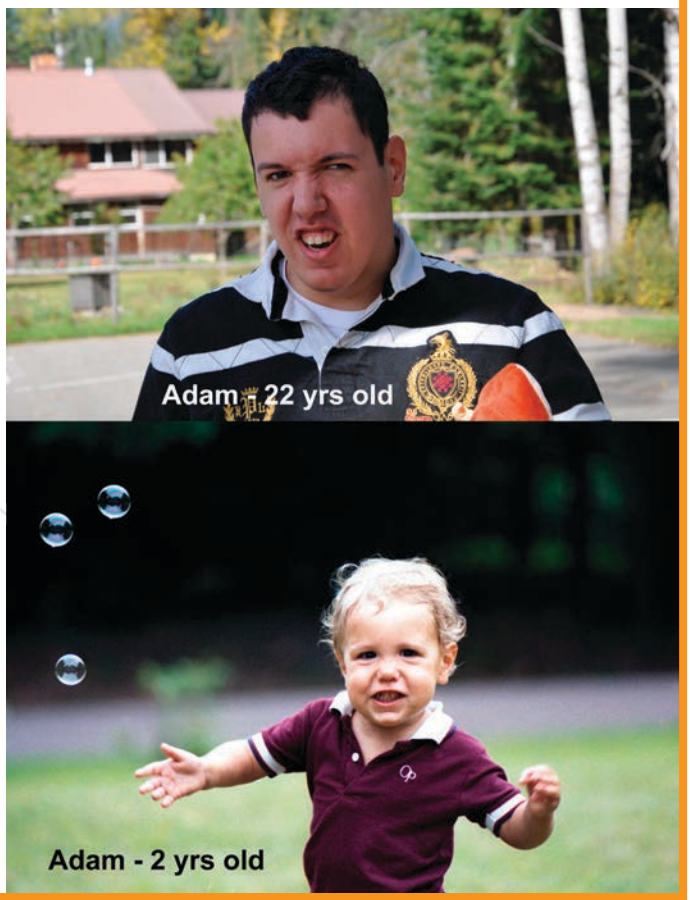
Deb and Bruce Lindgren, Parents of Chad, age 38

Twenty years ago, our family was searching. We were constantly trying to find connections with other families, as it seemed that would be the best opportunity to find solutions and best practices for our child. We never seemed to quite "fit", as we had more issues than the typical kid with autism. This search led us to the newly formed IDEAS. What a find! Looking back, we can honestly say it has changed our lives. We found immediate camaraderie. We all wanted research conducted to understand our kids' very complex medical, social, behavioral, and educational needs. We know there is much more to be discovered, but we cannot believe the difference this organization has made for our kids in just 20 short years. The conferences every two years have provided invaluable information, and the chance to reunite with families and meet new families from across the world. Dup15q Alliance has given us a solid base of information, family support, and the beginnings of amazing research. We feel so fortunate to have been bonded to this group with so many wonderful families all searching together.

" We all wanted research conducted to understand our kids' very complex medical, social, behavioral, and educational needs."

Patti and John Rubel, Parents to Adam, Age 22

Long before there was a Dup15q Alliance, there were families struggling to find a place to "belong". When our son, Adam, was diagnosed with idic15, there was a total of two short articles that had been published describing the syndrome. The geneticist could tell us very little about what we could expect in future. At all subsequent doctor visits, we were the ones telling the doctors about dup15q. We searched high and low to find support. I found Donna Bennett through a rare disorders organization, and we no longer felt alone. We finally felt as though we had a home, so to speak. It truly felt like a lifeline. We began to correspond with a few other families, and a group of us eventually met in Atlantic City at a conference where Brenda Finucane was speaking. We formed a bond at that meeting that still holds to this day. Thanks to Donna and Brenda's efforts, more and more families found our group. It began to grow and people started volunteering to help form the start of what we now know as the Dup15q Alliance. I was fortunate enough to work closely with many wonderful people, having served on the board. Proud doesn't seem to adequately describe the way I feel about our organization. You are family to us, and we are grateful beyond words.



Sheryl Kaminer, Mom to Elana, age 25

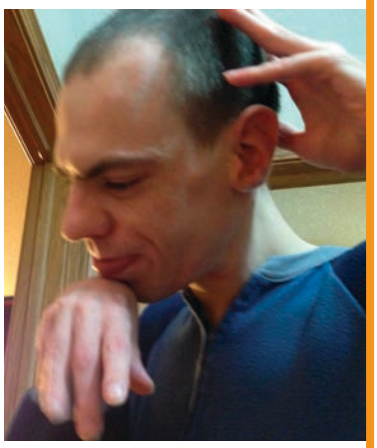
Our family has been involved with this wonderful group for 20 years. When Elana got diagnosed, there was not much known about this disability. I remember a doctor telling me to do everything I possibly could to get her out in the community and hope for the best. I discovered a few people, who I am still in contact with, raising children with the same disability! Needless to say this had helped so much. In the past 20 years I have met so many beautiful families and beautiful children. No one can understand what it is like to have a child with a rare disability unless you have one yourself. It has not been easy these past 25 years raising Elana, but with the help and knowledge of this wonderful group it has been easier. I have attended get togethers and some conferences, and just love seeing everyone. I have beautiful memories of these events! Unfortunately some of the beautiful children have passed, and one of the beautiful moms, but they have left an imprint in my heart and will never be forgotten. I look at their pictures with a smile.

Margaret Kelliher Gibson, Mom to Suzy

For our family, finding other children with inverted dup 15 was a godsend. We didn't fit into the Prader/Willi syndrome group, which was Suzy's first diagnosis at age 1 year. We were finding a fit with the autism group for symptoms and teaching at age 3 or 4, but many people did not think she was autistic because she was so loving. I was very interested in the biological basis of Suzy's problems. Being part of this group has helped us raise Suzy into the beautiful, happy, young woman she is. I learned how to make decisions without knowing everything. When people share within the Alliance, I can tuck away the knowledge that may be relevant later. For example, Suzy didn't have any seizure until she was 12 years old, but I was better able to cope since I had heard about them for years.

"Being part of this group has helped us raise Suzy into the beautiful, happy young woman she is."

Jeanine and Bill Rossbach, Parents of Michael, age 33



My family's connection to the Dup15q Alliance started about 30 years ago through an ad I saw in Special Parent magazine. The ad was from a medical researcher who was looking for children with unusual genetic anomalies. They led me to Donna Bennett. How wonderful to find another family whose son had an extra chromosome 15! And this woman was a lovely and helpful friend. We lived in Southern California and they lived in Pennsylvania so we have never met, but we called each other and wrote. This was before internet, email, and Facebook. None of our geneticists had seen a person with an extra chromosome 15q, and the only studies about it they could find were in Italian. They had very little information to tell us so we didn't know that seizures, bowel issues, autism, and no speech were a possibility, and they came as a complete surprise. We had to learn it all as we went. Knowing Donna was a breath of hope. Even though she was far away, we always knew we were not the only ones dealing with our seemingly "mystery child." We knew she had similar issues with her Josh. We knew their family was surviving so this gave us hope, moral support, and comfort. Today it gives me great pleasure to see other families coping and reaching out. It is wonderful to share my experience with newer parents, and I learn from them too. We are forever grateful to the parents who support the Alliance, and certainly to dear Donna.

Cheers to Our Volunteers!

Volunteers Paint the Town Blue

My name is Traci Ward, and I live in Hokes Bluff, AL. Our son Davis, age 6, was diagnosed at 4.5 years with dup15q. When we found out, our geneticist was no help at all. I started researching and came across the Dup15q Alliance website. Without this tool and the Facebook page, I would have been lost. It was my one source to learn, ask questions, vent, and not feel alone. So last December, 2012, my family and I got together and decided to have a fundraiser to support the organization that had helped me understand Davis.

We decided to plan a 5k run and 1k fun run. Being my overachiever self, I also decided to do a dinner with band and silent auction the same night. We decided to call it Paint the Town Blue. We had a great turn out, and lots of sponsors helped us out.

To do something like this, you have to have a great support system around you. My advice is plan and plan early. Especially important is to hit the sponsors early. We told them that their name would be on the shirt, and they could give us business cards or brochures that we would stuff in goody bags that we handed out. That is all free advertising that they like a lot. Also we told them that it was tax-deductible, which helped to seal the deal. We had a shirt printed up so they could see the design. There were smaller sponsors who gave door prizes and auction items. We let them put stuff in our goody bags, and their name went on our banners.

One of the other things that raised great money was auctioning off a Yeti cooler. One of the local stores put a picture of it with the tickets and we sold hundreds of them. We sold shirts and bracelets with our logo and the dup15 website on the other side from www.wrist-band.com. We donated all our funds to the Alliance because we want it to grow and we wanted to raise awareness. It was very hard work, but the payoff was worth it. In this last year the Alliance had opened the dup15 clinic in Memphis, and we were the first to attend. The service was great and so informative! Painting the Town Blue was well worth it to ensure that dup15q clinics and all the great things the Alliance does will be resources for others who struggle with no answers.

Dup15q Alliance

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Family Portrait

An Exciting Time is in Your Immediate Future

Our adventure with dup 15q began after 2 years on the adoption waiting list, when our beautiful son Timothy was born. After waiting all day at the hospital, we were told that it would be at least another 5 hours before Timothy would enter the world. We decided to run across the street from the hospital and get some dinner at Panda Express. Rod's fortune cookie said, "An exciting time is in your immediate future." We laughed because we knew that this was true. What we didn't know was how immediate or how exciting our future would be. When we got back to the hospital, we were told that Timothy had been born. We ran upstairs to meet our son. The first thing that I noticed were Timothy's beautiful teardrop shaped eyes. I wondered about their unique shape, but thought maybe he inherited them from his birth father, who was unknown. Then Timothy started having infantile spasms. A pediatrician was consulted and felt that they would go away within 48 hours, which they did.

Timothy was a very easy baby who was sleeping 6 hours at night by the time he was 2 weeks old, and sleeping even longer soon after. The only real issue that he seemed to have at first was that he would take a very long time to take a bottle, and he would spit up or throw up very easily. He seemed to be developing fairly normally, though most of his milestones were on the long end of normal. By the time Timothy was 4 months old, I started to notice that he seemed obsessed with spinning his toys. I became concerned that he might be autistic. He wasn't able to sit up without support until he was 8 months old, he crawled at 10 months, and walked at 16 ½ months. His feeding issue became more noticeable when he started eating chunky solids.

In our church, there were several babies born around the same time as Timothy. As he got older we saw him slipping further behind his peers. When we moved him to the toddler nursery at church, our easy baby suddenly became the screaming toddler who was overwhelmed by all the activity in

Rod and Lanae are parents to Timothy (idic 15), age 9, and Isaiah, age 7

the toddler room. He started having horrible meltdowns. At 2 ½ we finally convinced our doctor that he needed to be tested to find out what was going on, so we had him evaluated. After several hours with several specialists, the team told us that they felt that Timothy was probably autistic, but they wanted to re-evaluate him again as he got older. The Pediatrician in the group also recommended genetic testing for what he called "mildly dysmorphic features". We were devastated.

We didn't have Timothy tested right away because by this time we had also adopted our younger son Isaiah, who needed to have lung surgery. During that summer, between our two boys, we spent most of our time going to different specialists in 3 different cities. The day after I got home from 4 days in the hospital with Isaiah for his lung surgery, we put our little Timothy on a school bus to go to early intervention. Timothy did great riding the bus, but had a horrible time at school. He could not even handle being in the same room with the other kids in his class. With a lot of patience and hard work he was able to be with the other kids, but needed an aide with him at all times. The staff kept saying, "Yes, Timothy is autistic, but he is different than any other child we have worked with." We finally had the genetic testing done and found out that Timothy has isodicentric 15. We immediately found the website for the Dup15q Alliance. Reading the information there was like a big light bulb being turned on because it described Timothy perfectly. I contacted the group and was put in touch with Nicole Cleary who lived in our state. She told me about a weekend camp for the NW group coming up soon. Meeting other families at Camp Prime Time was amazing. They were full of great information and looked at our son like he was perfectly normal to them.

Timothy has come a long way from where he was back then. Now 9 years old, Timothy has been gaining many words and has started reading and spelling. He can run and climb very fast, which he does all day long. He still has meltdowns and has problems with being aggressive and biting, but he has a smile that will light up a room, and he is full of hugs and kisses. Timothy faces many challenges, but we are so proud of all that he has accomplished.

A Visit to the New Seattle Clinic

By Colleen Lowell

My 10 year old daughter Grace (idic15) and I recently visited the Seattle dup15q clinic. The Seattle clinic is located at Swedish Medical Center, in their Swedish Neuroscience Institute. The clinic is run by Dr. Marcio Sotero de Menezes. Dr. Sotero has been Grace's neurologist for several years, and we are so happy that he has taken on the role of the Dup15q Alliance's new Seattle clinic director. I was so pleased to walk in the door and see the new sign identifying it as the "Dup15q Alliance Clinic"!

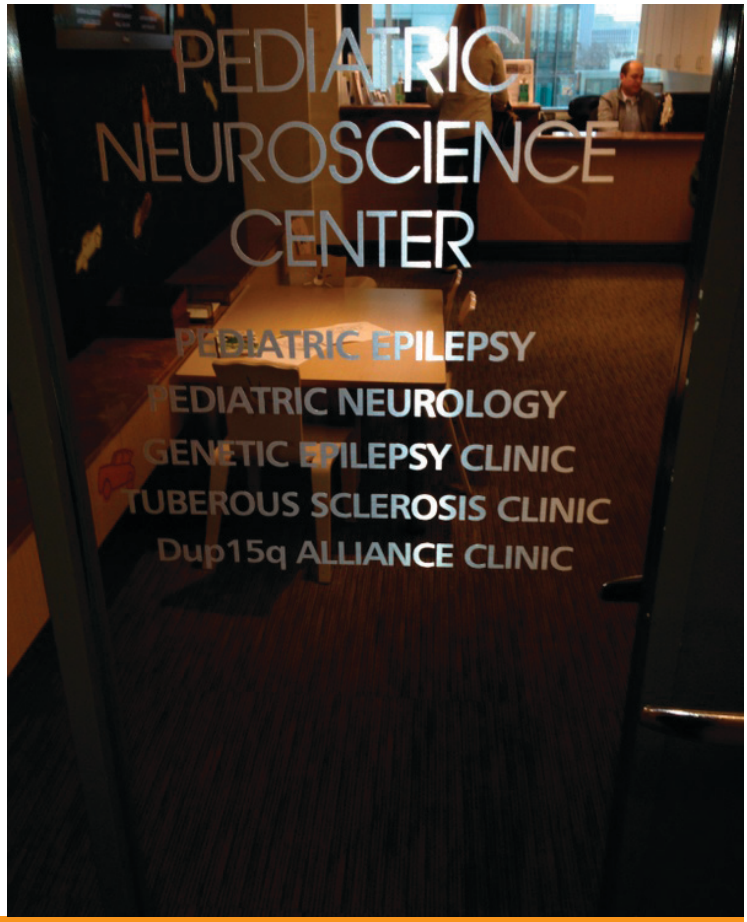
We started our visit with the clinic's nurse practitioner and the dietician, who work with patients on the Ketogenic Diet. Grace is currently on the Modified Atkins Diet, and we spent about 45 minutes talking with them about Grace's seizure control, lab work, tweaks to the diet and supplements. They answered all my questions and I so appreciate having them as part of the team.

Next, we met with Dr. Sotero for about an hour. He is a fabulous neurologist! He asked all sorts of questions about Grace, discussing not just her seizures, but her academic skills, social skills, autism, etc. I've always felt like he wanted to know who she is as an individual, but this time, in addition to that, I felt like he was trying to figure out how she fit into what he knows and is learning about chromosome 15q duplication syndrome. Of course we talked about Grace's seizure meds, up-and-coming treatments and possible things to try in the future, but I won't go into all the details here!

He mentioned several times the dup15q clinic doctors in Boston, New York City and Los Angeles; he is definitely communicating and working with them on a regular basis. It's exciting to have a doctor that doesn't just rest on what he learned way back in med school; he is actively learning and growing as a physician along with the fast-paced scientific world. He commented that in December he was at a conference in Washington DC about genetic epilepsy, and a presenter was apologizing that her slide was "out of date", and it was only three months old!

We also see a pediatric nephrologist (kidney doctor) at Swedish and Dr. Sotero and I chatted about having her be an official part of the dup15q Seattle team. It's wonderful to have a team of doctors learning and working together to help our kids. He referred us to Dr. Kim Soo, a psychologist who is part of the Seattle dup15q team. We don't have any pressing needs in that area, but I think we may go see her anyway for the sake of growing the dup15q knowledge base in this area!

We are thrilled to have a dup15q clinic in Seattle. If you live near or can travel to one of our clinics, I highly recommend it. Even though some of the clinics are new and just getting off the ground, the doctors are knowledgeable and our participation helps grow their knowledge base. I love the idea that we have a team of doctors around the US and the world working together to help all of our kids with dup15q, now and for the future.



Reflections from Siblings

Living with Andrew

By Geoff Keller (age 14), big brother to Andrew (Roo), idic 15, (age 12)

Living with Andrew is a complicated story to tell, because a lot of the time his behavior is normal for a kid of his age. For instance, he watches TV, plays with cars, and is a little bit hyper at times. Andrew's condition (idic15) impacts learning and language. He has had somewhere between 1-3 seizures in his life, so not as bad as some other diagnosed children. Andrew is 65 pounds and 4 foot 3 inches – he's kind of small for his age of 12, and he is in 6th grade. He also gets around on his own physically and does not require the use of a wheelchair.

Andrew can have tantrums every once in a while and he gets up really early, sometimes as early as 4:30 a.m., and stays up as late as 11 p.m. Andrew is still in pull-ups, no matter how hard we try. We've been potty training for years now and he can be pretty good for like a month, and then the next day he could have 3 accidents.

An interesting behavior I noticed is that Andrew has tons of cars and he lines them up as if in traffic, and moves the brigade throughout the whole house. This will entertain him anywhere from 20 minutes to an hour.

Some of Andrew's favorite shows are Mickey Mouse Clubhouse, Jake and the Neverland Pirates, and Doc McStuffins, all kid cartoons, and once in a while he watches Sponge Bob. Andrew has a short attention span with some activities, so a movie we may try to sit through does not always happen because if Andrew is not entertained, he can be very disruptive to the movie.

Andrew has something in his brain that does not always register. For instance, he could ask you, "Are you OK?", and then say your name, but after you answer he will continue to ask you the same thing over and over. This process can go on for 3 minutes, so conversations are always interesting. He will pick up words you don't want him to repeat; that has made for some interesting conversations in the past.

Since Andrew has been born, we have had a few different pets (but now we have none since we live in an apartment), and Andrew has held, poked and generally annoyed all of them when we had them. Sometimes they would nip at him, but he usually deserved it. We would warn him "Be Careful Roo! They will bite you!" He would usually stop annoying them only after a warning nip from the dog. However, none of the pets were aggressive as we would not have kept them.

Andrew gets some good reports at school. He also has a delayed reaction to pain so usually there are bumps and bruises that show up, and we have no idea where he got them. Only if something is real painful and intense will he say "ow".

Andrew also makes a lot of weird noises. When he is playing with his toys and the truck is backing up, he will make a beep sound. He also will take some of his bigger trucks, bend forward at his waist, and run with them. He can go quite a distance. He will also flip a real bike or a toy truck over and entertain himself by spinning the wheels, and its fun for him just spinning it for like 30 minutes. Just spin spin spin – it's a bit unusual at times.

Andrew's pills make him not eat much at all, so at dinner his food just sits out for a while so he can come back to it later. We give him his pills through yogurt and pudding.

Andrew has his days where he is fairly calm and easy to take care of, and at other times is almost insane. Life with Andrew has some action, but definitely a ride that I'm on and hope to stay on for many years to come.

Reflections from Grandparents

A Special Grandchild

Sue and Tom Thibedeau, Grandparents of Ryan Supina.

This photo was taken at the recent wedding of Ryan's aunt.

We have been blessed with seven grandchildren, ranging in age from 6 months to 10 years old. While each one is special and unique, this is about our four year old, "Handsome Dude" Ryan.

Ryan loves Dr. Seuss, music, merry-go-rounds, swings, fabrics with bold patterns, and toys with lights, sounds and movement. He loves snuggling with his mom and being turned upside down by his dad. Ryan, who is frequently a serious guy, gives the best belly-laughs when his dad makes weird noises for him.

When Ryan visits us, he has his favorite things to do: play with a colander, investigate each heat register, check to make sure the kitchen cupboards still open and close, and closely examine the wicker on the porch rockers. When he visits at our family cabin, Ryan loves to run on the dock (a bit scary for the rest of us), ride in the pontoon, and run the sand through his fingers (tasting a few rocks to keep the rest of us on our toes).

Ryan was less than a year old when he was diagnosed with "Isodicentric 15q", or dup15q, as we now say. None of us in Ryan's extended family had ever heard of this chromosome abnormality. There were many frightening statements in that original medical report from the genetics counselor, referring to concerns about delays in motor skills, seizures, vision problems, communication difficulties, developmental delays, and autistic characteristics. Oh, how frightening that report was to read, describing possible scenarios for our darling 9 month old grandson.

Now Ryan is four years old, and yes, all of those things are still areas of concern for Ryan. As we have gotten to know our Ryan, we realize it is easy to take care of him: he goes to bed well, he loves all food (following a gluten-free and dairy-free diet), and he keeps very busy with his favorite toys. Ryan enjoys riding in the car, is easy to take to a restaurant, and is typically a calm, content little guy. These everyday things put those frightening concerns into perspective.

What is frightening for us has changed a bit. Ryan is getting too big for us to pick up and carry. He walks/runs well, but what will happen as we age and he can run faster than we can? Will we ever solve the riddle of communication skills for Ryan? Is he happy at school when no one in the family is there with him?

Will Ryan's seven year old twin sisters experience any difficulties as the result of having a brother with significant special needs? It is our belief that any difficulties will be over-shadowed by what they are gaining in compassion, resourcefulness, responsibility and a strong family spirit.

We have met many other families with children with dup15q, and attended the 2013 Dup15q Alliance Conference. These experiences have been very helpful for us. We learned many things at the conference (diet considerations, importance of a special needs trust, communication ideas) but most of all it was delightful for us to see families having fun together and developing friendships with people who share this common bond. We were happy we were able to attend the session for extended family members. Also, it was inspirational to see people donating time and energy to make the conference successful, and being very generous at the silent auction to raise money for dup15q research.

Yes, our Ryan has special needs. But to us that just means he is a special grandchild, just like the other six grandchildren, each in their own way. We are happy that our main job is just to love them all!

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Dup15q Alliance is a nonprofit organization that provides family support and promotes awareness, research and targeted treatments for chromosome15q11.2-13.1 duplication syndrome **(dup15q)**.
Dup15q Alliance offers help and hope for chromosome 15q duplication.

UPCOMING MEETINGS AND ANNOUNCEMENTS

It's All Happening at the Zoo in June, 2014

Dup15q Alliance encourages families to hold a local gathering at your zoo this June. We hope that every region has one, so if you are interested in helping please contact info@dup15q.org and we can help you with a list of families in your area. The Alliance offers small grants to defer some of the costs for families. Let's launch our summer with a day of fun and connection all over the world!

Regional Family Gatherings are in the planning stages now! You can check to see if there is a gathering near you at www.dup15q.org

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

More information coming later in 2014

http://www.dup15q.org

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