



Together we are Moving the Science of Dup15q Forward

NICOLE CLEARY, EDITOR

In the last issue of *The MIRROR*, we explored how our dup15q family has grown. In this issue we will look at how the scientific understanding of dup15q is growing as well, both in depth and breadth. Promoting awareness, research and targeted treatment is part of the core Dup15q Alliance mission, and rate of growth in research is exciting. We all want targeted and effective treatments for individuals living with dup15q. Scientific discovery points the way to these targeted treatments, and it is exciting to read about the rate of scientific discovery in dup15q.

In this issue Alliance Board Chair, Paul Karch, provides a glimpse into the meeting of 60 researchers studying the genetics, biochemistry and clinical characteristics of dup15q syndrome who gathered at the UC Davis MIND Institute in September for the 2013 Dup15q Alliance Scientific Meeting. Dimitrios Arkilo, MD introduces you to the 7th Dup15q Clinic at the Minnesota Epilepsy Group in St. Paul, Minnesota. Our Volunteer of the Quarter, Jane Kim, MD, shares her story of becoming the dup15q clinics point person. Laura Piredda tells readers about the Nonsolo 15 annual gathering in Italy, and their hopes for moving research forward there. Rylie McHam, Alliance board member shares updates from the Dup15q Registry, a critical piece of the efforts to move the science of dup15q forward. Finally, please take note of the increasing number of scientific advisors as we welcome

Shafali Jeste, MD and Scott Dindot, PhD to the Dup15q Alliance's Professional Advisory Board.

This is a really exciting time for dup15q research. Scientific interest in dup15q continues to grow, due in large part to the efforts of the Dup15q Alliance board. Our families are essential partners in this effort, providing both critical information about the effects of dup15q in our registry, and the financial support that allows the Alliance to have a registry, hold scientific meetings, and invest in promising new discoveries such as the dup15q mouse model. Together we are all moving the science of dup15q forward.

NICOLE AND HER HUSBAND TIM ARE RAISING CORRINA (IDIC15), SIERRA, AND JASMINE IN PORTLAND, OR.

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Moving Dup15q Forward in
SCIENCE

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.



Dup15q Science Meeting Report

By PAUL AND ANNE KARCH, MADISON, WI
PAUL IS THE CHAIR OF THE DUP15Q ALLIANCE BOARD.

Over 60 researchers studying the genetics, biochemistry and clinical characteristics of dup15q syndrome gathered at the UC Davis MIND Institute

in September for the annual Dup15q Alliance scientific meeting. Even with all that scientific firepower, it was clear that **WE** the families of the Alliance, are the most critical resource to accelerate the breadth and speed of understanding dup15q. **We contribute vital information to the Dup15q Registry!** We hold in our collective heads, file folders and medical records the most important key to making progress in understanding and treating dup15q.

The dedicated and talented doctors and scientists working on dup15q generally fall into three camps: (1) **genetics**: scientists working to determine which genes are in the duplicated sections of chromosome 15 and how they are expressed in cells, particularly in nerve cells in the brain; (2) **biochemistry**: scientists figuring out how the chemical changes caused by the 15q duplications in cells, particularly brain cells, affect cell and organ function; and (3) **clinical**: clinicians observing, describing and treating people with dup15q. The first two camps are often called “bench science” because the work is done in labs (on lab benches). The third camp is often called “clinical science” because the doctors work directly with patients in clinical settings.

Because dup15q was first identified through genetic testing, the bench science of dup15q is ahead of the clinical science, but it is slowed by a lack of extensive and detailed data on the clinical characteristics of people with dup15q. One of the keys to further progress on dup15q is better data on our children, which we can provide through the registry and dup15q clinics.

The overall goal of both bench science and clinical research is the development of targeted treatments which could make a real difference in the lives of people with dup15q and their families. Clinical research is already yielding some useful information for families. For example, the registry and dup15q clinics are providing some sense of what anti-epileptic drugs are effective for seizures caused by dup15q. Exciting treatments based on understanding other rare genetic disorders, in particular Fragile X, provide a basis for hope that we will find more successful treatments for dup15q. However, each genetic disorder is unique and dup15q is more complex than most, so it is likely to take many years to develop targeted treatments. The Alliance is in this for the long run. We support and encourage basic research by providing opportunities for researchers to share findings, methods and possible paths for future inquiry at our scientific meetings.

CLINICAL SCIENCE

Sleep – or lack thereof. Some kids with dup15q do not sleep well. In a panel discussion, clinicians Janine LaSalle (UC Davis), Ron Thibert (Massachusetts General Hospital), Orrin Devinsky (NYU) and Alessandro di Rocco (NYU) reported that many individuals with dup15q are helped to sleep with melatonin, but they then wake up in the night and stay awake. Clinicians confirmed what parents already know: disrupted sleep can cause other problems. In fact, sleep deprivation is often used in seizure observations to trigger seizures. Common sleep drugs often have a paradoxical effect on kids with dup15q, revving them up instead of calming them down. Finally, the panel discussed possible connections between seizures and sleep. Better characterization of seizures and adding more sleep studies to the Dup15q Registry might help establish, explain or disprove connections between seizures and interrupted sleep.

Autism. Two separate panels discussed the relationships between dup15q and autism. Panelists included Larry Reiter (Univ. of Tennessee), David Segal (UC Davis), Ian King (UNC), Grant Belgard (UCLA), Sarah Spence (Harvard), Shafali Jeste (UCLA), Christa Lese Martin (Geisinger), Kathryn McVicar (Univ. of Tennessee), John Spiro (Simons Foundation) and Sarah Porath (parent). Autism in people with dup15q is different in some ways from other autism, including high levels of beta activity (brain function seen on EEGs) and greater levels of social engagement. The panel discussed different ways of creating better descriptions of autistic behavior, which might provide more consistent treatment plans for clinicians. Dr. Spence described autism tool kits on sleep, transitions and other issues available from Autism Speaks at <http://www.autismspeaks.org/family-services/tool-kits>.

Epilepsy. Orrin Devinsky described some of the efforts and challenges in creating databases of epilepsy patients, seizures and treatments. The real power of data is to provide families and health providers with practical advice for prognosis and treatment. Once again, increasing the size and detail of the Dup15q Registry, including accurate descriptions of seizures, is critical to our understanding of seizures and their causes in people with dup15q.

Alessandro Di Rocco, a neurologist at NYU who has a sister with dup15q, described the dramatic increase in the severity of his sister’s symptoms when she began taking GABA-related drugs. It took her medical team and family some time to realize the drugs were making her condition worse rather than better.

Early Impressions from Dup15q Clinics. Representatives of the dup15q clinics participated in a discussion led by Brenda Finucane, co-founder of the Dup15q Alliance, and Sarah Spence, member of the Professional Advisory Board. The first patients seen at the dup15q clinics are likely not a good representation of people with dup15q. Those with more severe symptoms are more likely to bear the expense and effort of travelling to a clinic. The physicians noted that though GI issues are present in the patients they’ve seen, determining the causes and treatment remains a challenge. As more clinics are opened, current clinics see more patients, and the clinic doctors share more information, we expect the clinics to be a great source of information in advancing the clinical description and understanding of dup15q.

Moving Dup15q Forward in SCIENCE

GENETICS

It's Getting More Complicated. The genetic story of dup15q grows ever more complex, as shown by Janine LaSalle's work. While the actions and interactions of the extra genes in dup15q are already quite complicated and still not well understood, the genes are only part of the story. Dr. LaSalle found that some of the segments of the chromosome between the genes affect when the genes are turned on and how they work. It's also possible that these "noncoding" portions of the chromosome act differently at various stages of life and different times during the day and night.

UBE3a – A Gene of Interest. Ben Philpot (UNC) discussed his work with the Gene Ube3A, which is *deleted* from the paternal chromosome 15 in Angelman syndrome and of which there are *extra* copies in core dup15q. He described a drug discovery process to explore if any compounds could prompt paternal expression in the cells. They found one compound that did force expression of Ube3a and are working to see if what works in the lab will work on animal models. The same logic could be used to look for a treatment for dup15q, although he cautioned that the effects of the extra genes in dup15q are likely to be more complicated to isolate.

Next Steps. A panel of scientists discussed possible areas for further investigation in fundamental research. Panel members suggested: further work on RNA transcription, analysis of rare patients, and possible causes of common conditions in kids with dup15q, such as frequent ear infections. The panel emphasized, once again, the importance and value of adding people to the registry and discussed ways the researchers could help recruit families to join the Alliance.

BIOCHEMISTRY

This is Also Complicated. Joachim Hallmayer (Stanford) described the genetic links to autism. Over 200 genes have been identified as having a connection to autism. Many of those genes impact autism through copy number variants (CNV), having too many or too few of the genes. The connection between a CNV and autism can be very complex as genes create proteins, proteins affect neurons, neurons interact in the brain, and the brain influences behavior. In dup15q, the likely section of chromosome causing the most severe problems is the Prader-Willi syndrome imprinting center. Because it is not possible to examine or test cells in the brain of a living person, these complex connections are studied through animals, post-mortem analysis of the brain, and peripheral cells.

Studying Cells and Animal Models. Understanding and testing the effects of dup15q on cells is done through study of various animal and other models of dup15q which allow controlled experiments on specific chemicals and cell functions. Stormy Chamberlain (UConn) explained the painstaking techniques required to use pluripotent stem cells to create nerve cells and then to confirm that these cells share all of the important characteristics of neurons in people with dup15q. Larry Reiter grows cells to study from another source, dental pulp stem cells, thanks to the families who have carefully preserved and sent teeth to Larry's lab. Other scientists learn from animal models rather than cells. Scott Dindot (Texas A&M), who developed a mouse model for the Alliance, and Matt Anderson (Harvard), who works with a separate mouse model, described their work in creating and characterizing those mouse models. Robert Nicholls is working to develop pigs with dup15q characteristics to see what insights can be gained from these models, which are close to humans in many ways.

Scientific Conference Take Home Message The message we took home from the scientific meeting is that there is a great deal of interesting and important work being done on dup15q! Much of it is difficult for a non-specialist to fully grasp. One parent, himself a scientist, said that listening to the experts talk to each other was like watching a good foreign film, you get a general sense of the story and the characters, but you miss many of the details.

The 7th Dup15q Clinic Opens in St. Paul, Minnesota

The Dup15q Alliance is proud to announce the opening of the newest dup15q clinic located at the Minnesota Epilepsy Group in St. Paul, Minnesota. The clinic is led by Dimitrios Arkilo, MD. We asked Dr. Arkilo to introduce his clinic to our readers.

How did you first become interested in establishing a dup15q clinic?

As a pediatric neurology fellow at Tufts University-Floating Hospital for Children, I had the opportunity to work with Dr. Ronald Thibert at Massachusetts General Hospital. During these rotations I was impressed by the dedication of the families with children diagnosed with dup15q and amazed by the care they received. That experience made it clear to me that I would like to play an active role in the management of children with dup15q.

How will your clinic interact with the other established dup15q clinics?

My goal is to maintain a high level of communication with all the other dup15q clinics, as I believe this will benefit our patients. I want to learn from the experience of others and apply successful clinic models at the Minnesota Epilepsy Group. Collaboration amongst all the clinics will be important for expanding the already significant research in the field of dup15q.

What will the Minnesota Dup15q Clinic offer to families who visit?

At the Minnesota Epilepsy Group we want to offer comprehensive care to patients with dup15q and efficiently support their families. In order to achieve that, we established a clinic without walls involving the divisions of genetics and gastroenterology at the Children's Hospitals and Clinics of Minnesota in St. Paul. As an epilepsy and neurology practice we have an excellent team of psychologists, neuropsychometrists, social workers and pharmacists that we will utilize for the care of our patients. The collaboration with Dr. Atkinson, an adult neurology and epilepsy specialist in our group with interest in dup15q, will ensure the smooth transition of care of our adolescents with dup15q that approach adulthood. Jennifer Young, our very enthusiastic clinic coordinator, and I will ensure that the referral goals are established prior to the child's visit, so as optimize our patient's experience in our clinic.

With the establishment of these specialty dup15q clinics, where do you imagine our understanding of dup15q syndrome may be 5 years from now?

Many advances have been made in the diagnosis and care of children with Dup15q in the last 5 years. The awareness of the condition increased and the global networking improved immensely. I am very hopeful that we will be able to build on this success and benefit our patients and their families. The ongoing vigorous research in the field will give us better understanding of the relationship between deletions and duplications in chromosome 15 and disease-related aspects of brain development and autism. Furthermore, I am very hopeful that with the ongoing sharing of clinical data from our Dup15q clinics and the registry, we will be able to provide new targeted recommendations for treatment. We actually have already 5 patients scheduled and are very excited to see them.

To schedule an appointment with Dr. Arkilo, please call Jennifer Young at (651) 241-5290.

Additional Dup15q Clinics can be found in:

Boston, Massachusetts

Miami, Florida

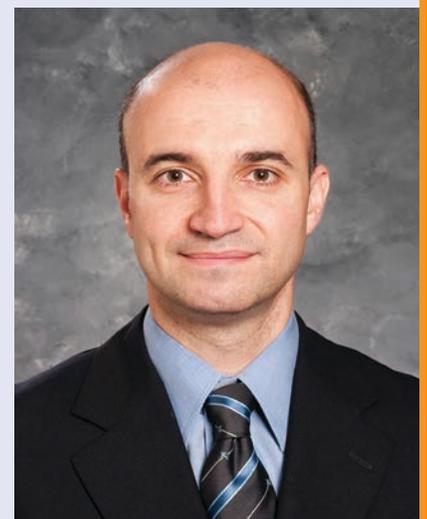
New York, New York

Seattle, Washington

Memphis, Tennessee

Los Angeles, California

For more information on all the clinics, visit www.dup15q.org/Dup15qClinics.html.



Nonsolo15 Annual Gathering



By LAURA PIREDDA

THE CASULE/PIREDDA FAMILY LIVES IN
 SASSARI (SARDINIA), ITALY

I've always thought that our angels give us lots of opportunities and special emotions. In July, my sweet Greta gave me the possibility to visit for the first time in my life the United States for the occasion of the Dup15q Alliance conference. I left with Gloria, Federico's mum, from Naples, Italy to New York. We spent a lovely time there and then we reached Minneapolis! How many emotions! It was great to meet other families from all over the world and everything was so interesting!

This great experience has been the main topic of our annual national gathering of families held in Tuscany from the 6th to the 8th of September, 2013. This year we decided to spend a holiday all together on the beach, in Versilia, near Lucca. It was not a scientific meeting, just a vacation, and we really enjoyed it! Thirteen families participated in the gathering.

Gloria and I talked about the projects of Dup15q Alliance and about the importance of the registry. We translated into Italian some slides that can be found on our website:

<http://www.idic15.it/info-per-familiari-e-specialisti>.

We also discussed our projects and the achievements of our little association, Nonsolo15. This year has been a great year for us. Our scientific committee was created on the 15th of July. At the moment we are working on the realization of a research project (epidemiologic and genetic project) whose aim is

- to distinguish the symptoms and physical features characterizing dup15q patients from other patients with similar neurological conditions such as motor and cognitive delays
- to create a regional registry on the 15q11-13 duplication syndrome
- to create a protocol in order to quickly recognize a rare disease in a regional context.

The genetic aspect of the project seeks to find a connection between the symptoms/physical features and the genes involved.



Nonsolo15 Comitato Scientifico



In order to realize this project titled "Genotype – phenotype correlations for the early diagnosis of 15q11-13 Duplication Syndrome", we have applied for funding to support exploratory projects promoted by Telethon, an Italian organization working on research on rare genetic diseases.

Nonsolo15 co-operates also with the Telethon Genetic Biobanks, so we are collecting our kids' blood samples in order to help the research.

We are now planning our next meeting that is going to be a scientific meeting with the presence of all the doctors of our scientific committee. We are also organizing fundraising activities to promote research and spread awareness on the condition of our special angels.

DUP15Q ALLIANCE REGISTRY UPDATE: FILTERS NOW AVAILABLE FOR CHART DATA

BY RYLIE McHAM, COLUMBUS, OH, DUP15Q ALLIANCE BOARD MEMBER

The Dup15q Alliance is working to make the registry an even better tool for families. We are excited to announce that filters have been added to the reporting feature! Filters will allow families who have filled out their child’s information in the registry to filter their registry searches, making the graphs more specific and relevant for individual users.

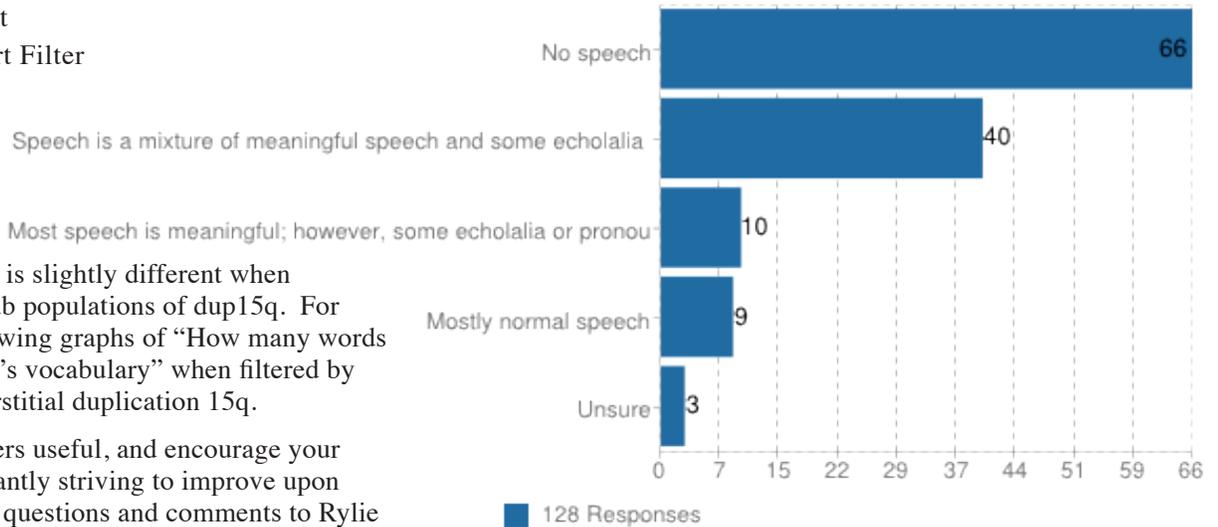
For example, users are able to select a specific diagnosis, like isodicentric 15q, and compare their responses to others diagnosed with Isodicentric 15q. Currently, participants can filter information by diagnosis or gender. To utilize the filters:

- Log in to your registry account at <https://www.dup15qregistry.org/>
- Click on the Explore tab
- Click on a subject
- Click Select Chart Filter

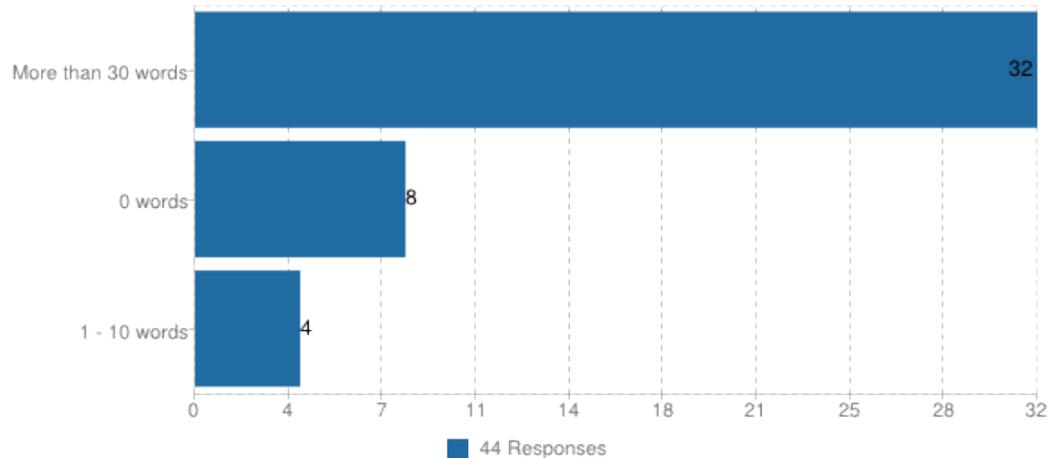
You will see that the data is slightly different when you filter two different sub populations of dup15q. For example look at the following graphs of “How many words are in the affected person’s vocabulary” when filtered by isodicentric 15q and interstitial duplication 15q.

We hope you find the filters useful, and encourage your feedback as we are constantly striving to improve upon the registry! Please send questions and comments to Rylie McHam at ryliemcham@gmail.com.

How Many Words: isodicentric 15



How Many Words: interstitial dup15q



How can you continue to help?

Please update your family member’s registry data **annually**. These updates are important and help our registry data reflect the current strengths and challenges your family member is experiencing. Please update all sections where there have been changes over the last year. New medication? A new seizure type? New skills? Your family member’s birthday can be a perfect reminder that it is time for an annual update.

For those of you who are filling out the registry for the first time, please complete ALL of the questions on the registry, and please do not leave any question blank. If your answer is “no” or “unsure” indicate that rather than skipping a question. If you find certain questions confusing or you need assistance we are happy to help! Email us at coordinator@dup15qregistry.com.

If you have not yet uploaded your genetic reports, we encourage you to please either upload such documents or email them to coordinator@dup15qregistry.org or fax to 515-964-1512. We are happy to help with uploading documents, so please email us with any questions or concerns.

DUP15Q Alliance FINANCIAL UPDATE

By TOM DOYLE, ALLIANCE FINANCE OFFICER

The third quarter of 2013 was filled with events and excitement. The conference at the Mall of America was outstanding. Families from all over the world had a great time at the amusement park and the aquarium, and the presentations and speakers provided wonderful information for our families. Thanks to Karen Sales and all her able assistants for a great conference experience.

In September, the Dup15q Alliance held its annual scientific conference in Sacramento, CA, and had more than 75 attendees from around the world. The meeting included the first face to face (rather than teleconference) meeting of representatives of our growing clinics throughout the United States. Great information was shared by the doctors, researchers, and professionals brought together by Guy Calvert.

As far as the specifics of our finances, at the end of September, the Dup15q Alliance balance sheet includes \$350,025 in current assets. Our largest expenditures this year have been for the conference (nearly \$90,000 – which

included sponsoring the Nickelodeon Universe and the Aquarium), the Registry (\$36,000 annually), and our scientific meeting (\$10,000 annually). In the next year, we will be adding significant costs (\$10,000 or so) for a redevelopment and upgrade of our website. We are also in the process of defining a new position and interviewing for a development director to help us with furthering our mission and goals.

We continue to be grateful to the many volunteers who make the Dup15q Alliance work and to our families who so generously support our efforts. None of this could be done without your tremendous participation. We are supported by many family fundraising events (too many to name them all here) as well as your wonderful donations. We are already budgeting for the next two year cycle and hope to be able to continue to raise awareness, promote research and targeted treatments, and provide support for our families. Thank you for all your efforts on behalf of the Dup15q Alliance.

Holiday Shopping NOW OPEN IN THE DUP15Q ALLIANCE STORE

<http://www.dup15q.org/Store.html>

Great teacher gifts - mugs, blue bracelets and the cause bracelets too!

A few fleeces left- warm and cozy!

Holiday gifts for family - mugs, stickers, beautifully-made cause bracelets, magnets and rings!



KLARA BELLE



MICHELLE BERES LIVES IN ROSENDALE, WI WITH HER HUSBAND STEVEN, KLARA AND HER SISTERS.

Family Portrait

The doctor broke my water and measured five liters of amniotic fluid. They had never seen that much before. When Klara was born, there were 22 people in our room. She was completely black. The doctors sucked gross looking water out of her chest as other doctors pushed air into her lungs. She began to pink up from the chest outwards...it was the most incredible thing I have ever seen. She had beautiful black hair and looked just like my brother Joe and my niece Sara.

Klara was sent to the NICU and lived there for two months. She was diagnosed with cerebral palsy, chylothorax, failure to thrive and many other things including isodiscentric 15 syndrome. However, two babies died in the two months we lived there. It was the first time I said, "I'll take the Klara version anytime!" When she turned two months old, the doctors came to us and said they needed to do something. She was accumulating fluid in her chest (chylo-thorax) and they needed to eliminate that fluid. A surgeon talked to us and convinced us to allow exploratory chest surgery. My

My dear Klara Belle was one sick baby when she was born... actually even before she was born. I was 27 years old and had a very serious car accident. I was also three months pregnant with Klara. My doctor was a muscle specialist learning to deliver babies. His diagnosis of my injuries was severe muscle spasms and torn muscles. We found out much later that I was in pre term labor. I was put on bed rest on and off until June because of pain and fatigue.

I was admitted to the hospital to stop labor at 33 weeks. It was a wild ride. I was taken by ambulance from St. Luke's to St. Joseph's (the neonatology center for sick babies). On the way, they gave me a dose of magnesium sulfate to stop my contractions. The medication was effective but made me feel high and sick at the same time. For the next 11 days, I sat in a hospital bed with an IV of meds to stop contractions, and was very pregnant! Klara and I developed a condition called hydrops, which means she stopped using my amniotic fluid. I managed to stay pregnant so Klara's lungs could develop more. We had ultrasounds almost daily. She was very cute on screen!

On June 24th, they stopped the medication and told me it was Klara's birthday. They also told me (repeatedly) that she would not be able to breathe when she was born and would probably die soon after she was born (if she wasn't born deceased). I was developing an infection and was too sick to be pregnant anymore. Klara's lungs were not ready...it was such a sad morning.

husband signed the papers and they transferred her to Children's Hospital. I was too emotionally drained to make a rational decision and I did not want my baby cut open. She had surgery when she was two months old.

One week later, Klara was nursing and gaining weight! I thought she would die and she lived, thrived and got better. We took her home on September 12, 1992. I needed to quit my teaching job as she was still in the hospital when school started. I knew maternity leave would not be enough time to take care of my very special baby girl. My older daughter, Hannah was two (my sister, Susan helped her toilet train while I was at the hospital...bonus!), and we started our lovely routine of taking care of Klara. Hannah went to therapy. Hannah waited. Hannah learned to love everyone. Hannah became a loving, caring, supportive, big sister. Today, Hannah is a recreational therapist taking care of sick babies!

I wanted to share Klara's rough start to provide a possible silver lining for all

of us. The doctors told us she would not live. Now Klara is 21 and has two jobs. They told us to plan for a life of wheelchairs and therapies. She walks, skips, runs and has endless energy. The therapists told us she was globally developmentally delayed with atypical features. She tells a mean joke, has more empathy than anyone I know and tells EVERYONE they are beautiful, handsome, smart and talented!

Don't believe the negative things you hear. Believe in your child! Don't stress about the massive fails and inabilities. Stress ONLY about things you can control like your attitude, your lifestyles and your choices. Even as Klara tells me, "I want to have babies... I want to be like my sisters, they can have babies...it's not fair...I don't want to be special anymore. Even as I grieve the babies Klara can't have, I celebrate who she is and who she has allowed me to become. I am Klara's mom! "The prettiest, bestest, most beautiful, most talented mom EVER", Klara says. "You are the perfect mom for me!"

Cheers to Our Volunteers!

The Dup15q Alliance Cheers our Volunteers: Jane Kim, Dup15q Clinic Coordinator



My son, Zachary, is currently 18 months old and was diagnosed with idic15 less than one year ago. I still remember the phone call we got the day after Christmas, telling us that the genetic testing showed Zach had something called idic15. It is ironic because I am a neurologist and my husband is also a neurologist, but neither of us had heard of idic15 or dup15q. The first thing I did after Zach's diagnosis was to google "idic15" and I came across the Dup15q Alliance website. It was so helpful to get more information on dup15q, read family stories, and know that there were others out there with dup15q. Over time, I have come to find the Dup15q Alliance to be a wonderful organization that does so much for our kids.

When the e-mail went out looking for volunteers for the Dup15q Alliance, I knew that I wanted to use my neurology background to help. I was introduced to Guy Calvert, who has been doing amazing things with moving research forward. I attended the scientific meeting in Sacramento, CA this past September and I had the chance to meet with several of the clinicians involved with the dup15q clinics. It was great to see the exchange of ideas between all the dup15q clinic physicians. Even though I was already busy as a practicing neurologist with two young kids, I knew that I wanted to be more involved.

I have taken on the role of being the "dup15q clinics point person." We currently have 7 dup15q clinics open around the country. It is our goal to get all the clinics on the same page, as far as evaluation and treatment of our kids. We will be setting up a clinics registry, where all the data and information gathered from the various clinics can be brought to one central database. This is going to be important as we learn more about the common characteristics and medical issues our kids have and also how to treat them.

We will be having conference calls at least once a quarter with all of the dup15q physicians so that we can discuss any issues, new ideas, interesting cases, etc. We are also thinking about having webinars or other types of forums so that parents can ask our dup15q specialists questions, even if they can't make it to a dup15q clinic. We really hope to make the clinics dup15q centers of excellence, where there are knowledgeable doctors about dup15q syndrome who can provide the best care possible. I am open to any suggestions or comments about the clinics, so please feel free to contact me anytime at janeckim@hotmail.com.

Dup15q Alliance

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Reflections *from Siblings*

BROOKE RIVARD RECENTLY INTERVIEWED HER BROTHER KYLE, AND TOGETHER THEY WOULD LIKE TO SHARE THEIR SIBLING EXPERIENCE

Our Sister Megan



Describing Megan, the words that come to mind are easy going, always happy, and always smiling. She could get along with practically anyone. My brother and I have never once gotten into a fight with her, so she's pretty great to have as a sister. At home a typical day for her would be to watch her videos, especially Barney and Elmo. She'd be happy just with that, but weekdays are even better because she gets to go to school, her absolute favorite place to be. All summer she asks if she's going to school. Most kids aren't that excited to go to school but Megan's a senior this year and she's just happy to be there.

What's different about living with Megan?

Megan needs some extra help from us when she does certain things. Kyle (13) may be asked to push her in the stroller when we go to places with a lot of walking or just out to walk the dogs. He might also put in new videos for her. I, being 15 and also a girl, could be asked to help her get dressed for the day or help her go to the bathroom because she's still in pull-ups and won't always go on her own. I may be asked to do other simple things like watch her when my parents aren't home and make sure she has videos and goes to the bathroom. Other times I may have to make her dinner or lunch depending on the time of day. For my brother and me, it is different because usually you would take care of a younger sibling but we help take care of her and, being 17, she's older than us.

What do your friends think?

Both Kyle's friends and my friends are very accepting of Megan. We make sure to tell people about her before they meet her, so they have a better understanding of how she may act towards them or around them. When we tell our friends about her we will generally just say she has special needs and there aren't usually any questions after that. If there are questions of the more specific type I can answer them but Kyle isn't really sure how to explain it. I am very open about talking about Megan being special needs--it has never bothered me. She's just like you and me except she needs a little more support.

What's it like going to school with Megan?

Kyle doesn't go to school with Megan yet, but I do. It's my second year going to school with her. She's a senior and I'm a sophomore. I see her from time to time in the hallway. Sometimes I'll say hi to her aides while she's off walking to where she needs to be. Most people don't know we're related until any sibling questions come up. Partly it's because we don't take the same classes and we look very different for sisters. She has curlier hair than me. Overall, going to school with her is just like going to school with any other sibling--you don't see them much and you both do your own thing.

How does living with Megan change your views of disabilities/ people?

Living with Megan definitely changes how my brother and I view people with disabilities and even people who don't have them. Kyle says it gives him a better understanding of how everyone is different and even disabled people can be talented or special in their own way. Living with Megan changes my views of people, because it shows me how some people are either uninformed or misinformed about people with disabilities. The biggest thing I see, as an example, is people who use the "R" word. Before I wouldn't think anything of it, but now when I hear someone say it I'll tell them not to use the word because it's offensive to me. Some people will stop and then give me a funny look. Others will ask why. I tell them that I have a sister with special needs and that's why it's offensive when they use the "R" word. Lastly, it teaches me to be understanding towards different people with different disabilities, how hard it can be for them, and with those people to have more patience. If I did not live with Megan I would probably have a much more difficult time learning this, so I'm grateful to be learning it this way.

Reflections from Grandparents



We are Pepe and Marga, Noa's grandparents. Noa is our first grandchild, born in May, 2009. From that day we stopped being the couple and parents known as Pepe and Marga, and became the grandparents to Noa, our Number One.

One of the most beautiful feelings we have experienced in our long life is the love of grandparents, which is comparable only with the love of parents. It might be equal in intensity, but it is a more serene love, very

patient, more understanding. It's perhaps more innocent and full of hope, without fear and without the burden of parental responsibility.

We act in very silly ways. We get to the child's level to sing like we never have before, We play lying on the ground. We believe that having grandchildren at our age is the most beautiful thing that can happen to us.

As the days passed, Noa began to develop. Despite not wanting to breast feed, she was growing beautifully. She was calm and slept a lot, especially when held in our arms.

We noticed that Noa was not developing like other children her age. At early stimulation classes, we began to notice things that made her different. We worried, but we knew that every child is different in their development. We saw it in our own three children.

Finally, after many consultations and various tests, the dreaded diagnosis came. Our son called to say genetic testing revealed a duplication of chromosome 15. All we saw was a broken heart and parents' pain, inconsolable. We had never seen our son so desolate and despairing. The news broke our hearts. We did not want to ask, so as not to exacerbate their pain. We did not know what to do to ease that pain.

We felt the weight of the world coming down upon us--the diagnosis of a rare disease, the question of what we could do, and wondering how this disease might impede our little girl's happiness.

The letter by Emily Pearl Kingsley, mother of a child with Down syndrome, clarified a lot of our feelings. She compares the situation with a trip you have planned to the smallest detail and have been anticipating with joy. Your destination is supposed to be Italy, but you find yourself in Holland. Suddenly your brochures about beautiful Italy become useless. You cannot visit those wonderful

The Love of Grandparents

PEPE AND MARGA LIVE IN MADRID, SPAIN.

places you've dreamed of. What to do now? It is not so bad. Holland is beautiful in its own way. It is in your hand to discover all the beauty there.

That's when the stage of acceptance and adaptation began. Initially there was more pain than hope. It was worse when Noa wasn't with us; when with her, the joy moved sadness aside. Slowly we began discovering her beauty, charm, personality, achievements and struggles.

We also had to learn from Noa's parents how to act: to understand and spot her needs and how they manifested, what caused her to become irritable and calm, how to dialogue and not impose on her. We gain confidence as we learn. We keep searching for new strategies through the Asociacion Idic-15, at lectures, and in books. Now our biggest challenge is trying to know at all times what she is asking for, what is needed, and to not get Noa very angry if she doesn't get what she wants.

We found it important to join the Asociacion Idic-15 and meet other couples in the same circumstances. Another thing that we think is important is to maintain ties with old friends.

The trust that our son and daughter-in-law place in us by allowing us to look after Noa whenever they have the need gives us further confidence. Thus both we and the maternal grandparents can enjoy our wonderful little girl.

Nowadays there are many more joys than sorrows: her happiness, tenderness, loud manners, her unique form of dance. She can remember and sing an amazing number of songs. She loves water, even when cold!

Noa's cuddling gives us a unique joy.

Best of all is witnessing the adoration her parents have for their child, their ability to fight, their infinite patience, understanding, adaptation and acceptance. Noa has exactly the parents that she needs! This makes us joyful, hopeful and proud.





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

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

Visit www.dup15q.org/calendar.html.

UPCOMING MEETINGS AND ANNOUNCEMENTS

2013 Dup15q Scientific Meeting

Videos and presentations are posted where available.

<http://www.dup15q.org/scientificmeeting2013videos.html>

This meeting was financed in part through the generous support of our sponsors. We gratefully acknowledge the support of:



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SIMONS FOUNDATION

2014 is the 20th Anniversary of the Dup15q Alliance

Stay tuned for events

The 8th International Conference will be held in Orlando, Florida

July 30 - August 1, 2015

More details will be available in 2014.



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