THE MIRROR

Spring•2013

The Newsletter of DUP15q ALLIANCE

DUP15Q IS ONE OF MANY RARE DISORDERS WITHOUT BORDERS

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February 28, 2013 was the sixth international Rare Disease Day organized with rare disease national alliances in the U.S. and 24 European countries. The Dup15q Alliance was one of them! As the Alliance grows, we welcome families from all over the world and do our best to connect families and provide support. We dedicate this issue of ***The Mirror*** to the theme of Rare Disorders Without Borders. Inside you will find the story of Vega, age 55 and Luca, age 8, both living in Italy. You will learn about Associación Idic 15, a family support organization for families raising children with dup15q in Spain. You will read a fun book review by a Canadian mom reviewing a book written by an Australian mom. And you will see photos of families who celebrated Rare Disease Day by Wearing Blue for Dup15q.

Nicole Cleary lives in Portland, OR, with her husband Tim and daughters Corrina (dup15q), Sierra and Jasmine. She can be reached at nicleary503@gmail.com

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.

Introducing Asociación Idic 15

By Miguel Ángel Orquín González and Fernando Gomez

Editor's Note: As part of this issue's celebration of Rare Disorders without Borders, we are launching a recurring feature article introducing Dup15q Alliance families to parent support groups for dup15q around the world.

Miguel Ángel Orquín González is the President and one of the founders of Asociación Idic 15, an organization for Spanish-speaking families living with dup15q. Fernando Gomez is the Vice President of Asociación Idic 15, and also a co-founder. Recently, Miguel Angel wrote about his family, his daughter Mara, his journey finding answers about dup15q syndrome, and finally sharing that knowledge and strength with other families. Mara's full story is available on the family stories tab at www.dup15q.org.

In 2006 my search began and, most days I was awake until the early hours of the morning on Internet forums. I remember for years entering the Google search term "partial tetrasomy of chromosome 15," and nothing at all would be returned. I contacted a person of prestige in the field of pediatric neurology, and surprisingly he answered, perhaps seeing the desperation of a father full of uncertainty and with no answers to his many questions. He said, "Syndromic autism searches. I have two papers." There, in a short article of just four pages, were six lines about a syndrome called "invdup15". Finally, what had happened to my daughter had a name that apparently only a few professionals knew about, and it became the guiding principle for my later medical training.

Then a parent came to me (who would later be in Asociación Idic 15), a father who, like me, had found no answers for many years. On that day, through the tears that this father shed, I realized that I understood why I had spent all those years searching. I could not allow any more people like us to experience such pain, and I said that we would start right there and travel this road together.

The origins of Asociación Idic 15 go back to early 2011, when Miguel Angel and Quino Renovell contacted Fernando Gomez, father to Alison (dup15q) who lives in the U.S. They wanted to learn about Dup15q Alliance and attend the Dup15q conference held in Philadelphia, PA, in June, 2011. Following the conference, a family meeting was held in Valencia, Spain in October, 2011, and soon thereafter, the Asociación was incorporated as a non-profit in February, 2012.

Fernando serves as a bridge between the two organizations, serving both as Vice President of Asociación Idic 15 and serving on the Board of Dup15q Alliance as International Liaison. In this role he works to support the creation and development of international groups which share common research projects and goals.

Miguel Angel states, "On behalf of the Asociación Idic 15, and as president of the company, I send an affectionate greeting to anyone interested in visiting our website at http://www.idic15q.es/. The main objective of our organization is to improve the quality of life of people with idic15 and their families. We have high hopes that the website will be helpful, as we are aware of the great uncertainty of newly diagnosed families who have a lack of information."

Asociación Idic 15 works to provide information for anyone interested in idic15 syndrome. We also want to provide information on all research projects, participate with professionals from any field and institution, and thus improve and ensure the information and resources are available for families.

"I hope that our website meets the expectations of all visitors. I am grateful for the vote of confidence that families and acquaintances have handed both to me and to each member of the Asociación Idic 15 Board."

From Italy: Beginning the School Journey By Francesco Jannon

Luca has started the first elementary.

I feel excited and proud, happy and hopeful. The first elementary is the beginning of a long journey, the dividing point between infancy and adulthood, between play and commitment. On the night before his first day, I stayed in his bed until one and a half in the morning because he was wide-awake, restless and moaning. I scratched his back and caressed him after having fought with him to swallow the Melatonin he has recently learnt to spit out amusingly. Luca entered his class with all his new notebooks, his pencil case full of colors, erasers and pencil sharpeners, schoolbooks with both name and cover, a bag containing his tennis shoes and lunch pack.

Luca heads off to school

Luca started elementary wearing his neat and clean overall, bringing his new blue and red rucksack, the same color as his wheelchair. The other day I laughed when I saw that he had to take the bag containing his tennis shoes because none of his shoes are ever dirty, quite simply he can't run or walk. Luca has just started to walk on all fours this year.

Luca is eight years old and is starting first elementary two years late. Luca can't speak. It is difficult to know what Luca truly understands. As parents, we are sure he understands much more than what it seems, but it's quite impossible to answer this sort of question precisely. Luca's school expectations must necessarily be special: Luca will make progress, learn many new things. He will grow up and make improvements. Nobody knows in how much time because Luca has his own time which is not our own; he cannot be judged in semesters with average or excellent marks, he cannot be classified in numbers. Nobody will expect him to learn how to read and write by Christmas. He will never get an F.

Luca is always, I mean always, happy and serene. Maybe he understands that it is possible to escape from high expectations. Maybe he understands that he has this privilege. He is the only one to decide when to move on fills our heart with joy. It is not always easy to be Luca's father, but today I am proud of him and I am happy that the school accepts him for who he is and what he can do together with his classmates. He is exactly the same in every way as his classmates: Eight o'clock, rucksack, packed lunch and bag containing his tennis shoes.

Today is a beautiful day.

Francesco Jannon is Husband to Roberta and Father of Lorenzo (12), Luca (8) idic15, and Sara (5)

Photographs of Families and Classrooms participating in Wear Blue for Dup15q on Rare DiseaseDay

Vanessa Torres Torralbo
Madrid, Spain
"My son in his school day celebrating rare diseases"

Annie Colello
Special Education Teacher to Nicholas
Ilchester Elementary, MD, USA
"We ended up making tshirts for national rare disease day with Nicholas's handprints on them. We celebrated him all day!!"

Olivia Taylor and her dog in blue, is from Kirklington, North Yorkshire, U.K.

Brynleigh rockin the blue!

Ivonne Ruiz & Denis Feterman
Hialeah, FL, USA

Katarzyna Dunin-Borkowski and children Lima, Perú

Dup15q Registry: A Half Hour for our Children

By Stephanie Barmann

It takes about half an hour to change our children's futures. It's the same amount of time spent playing games on Facebook or driving to the grocery store to pick up coffee.

It took me 30 minutes fill in the registry and send in my daughter's genetic results. I know that the lives of parents of children with dup15q are much busier than some/most people's. We live more the clock. This is such an important thing we all can do with a short investment of time. When your children were diagnosed, didn't you seek our more information? Didn't you want to know every aspect of what could happen and what your child's future might hold? I know I did. I became a research junkie, and to be honest I still am. We deal with such a rare condition, and so little is known. The registry is one of the tools provided by Dup15q Alliance that will benefit our families.

One great thing about the registry is that you will always have access. All of your child's milestones and treatments will be in one place. You won't have to open the baby book every time someone asks when your child first walked and then go through a stack of medical records to find the date of an EEG or MRI. All of this information will be in one place so you can easily get to it with a click of the mouse.

None of your information will be shared unless you want it to be. That means a doctor three countries away can't see your child's records whenever he pleases. This is your information and will never be shared without your permission. If down the road, there is a study you qualify for, one of the administrators will send you an e-mail and you can make a choice to be involved or not.

The Registry will make a difference. If you have started and not finished, please take the time to answer the rest of the questions. If you have been meaning to fill out the registry and forgot, this is your reminder.

If this is the first time you are hearing about the registry, you can learn more at www.dup15qregistry.org.

Remember, you can help your child and other people's children by taking those 30 minutes to fill out the registry. You really have nothing to lose and everything to gain.

Financial Report

Tom Doyle is the Financial Officer and Interim Chair of the Dup15q Alliance Board

The Dup15q Alliance board is pleased to keep you updated on the status of your Dup15q budget as of the end of the first quarter of 2013. As of March 31, 2013, the Dup15q Alliance has $379,930.38. With registration for the conference now under way, this total will surely rise quickly. We had a great fundraising year last year in support of our registry and preparation for the Conference in Bloomington, MN, this July.

Thanks to all of you who participated in our "$15K in 15 days" beginning on Valentine's Day 2013. We are pleased to announce that we met that goal of raising $15K with your incredible help. Thank You!

We have set our budget for 2013. Our top three investments for this year will be: 1. Our International Family Conference: July 25 – 27, 2013, Bloomigton, MN; 2. The Dup15q Registry; 3. Our Research Roundtable meeting in the fall of 2013. Your generosity will help us to put on a great conference at the Mall of America as registration fees only cover about half of the total costs. Our investment in the Registry costs $3K per month, and provides a critical database for our families and our doctors and researchers. This fall we will be hosting a Research Roundtable in the San Francisco area to bring researchers and doctors together to discuss the latest research on Dup15q. Most importantly, the budget will allow us to communicate with and support the many new families that register with us annually.

Before the end of the year, we hope to increase our resources and ability to fund research projects and support our families through the hiring of a Development Director who can help us to focus our efforts. Recognition among the medical community has led to a significant increase in Dup15q diagnoses and, therefore, to our membership. The resources entrusted to us now and in the future will help us to educate the medical community about Dup15q and allow us to provide education and support for all our present and new families.

Each year, the Dup15q Alliance completes a tax return for the IRS and one for the State of Oregon where we are incorporated. Both of those were completed and submitted by March 15. We also go through an extensive audit of our books to ensure that your resources are safe and secure. That audit, which included reviewing every transaction for the year, was also completed by March 15 and indicated that all our financial records are in good order.

Thank you for all your generous support of the Dup15q Alliance. Please contact me if you have any questions about our finances or our budget.

Sincerely,
Tom Doyle
Tom.doyle@dup15q.org

Book Review

By Alison Kalnicki

Book Review: Don't call me Honey! Call me Cherry Pie
Written and illustrated by E.C. Knox, 2011, Strategic Book Group, 40 pages, soft cover, ISBN: 978-1-60976-192-9

Reviewed by Alison Kalnicki, from Courtenay, British Columbia, Canada

Liz (E.C.) Knox is the wife of Tony and the mother of Luke and Lindsay, a thriving 22 year old man with dup15q from Australia. Tony and Liz have been happily married for nearly 28 years. Liz writes "Lindsay attends a day program Monday to Friday from 9am to 3pm. He also frequently goes with Tony to feed the cows and cart loads of rock in the truck. Lindsay has a girlfriend and they attend a supported house once a month for a long weekend. He plays basketball each Monday (his girlfriend is also on the team) coached by volunteers of a local basketball team. Last year Lindsay attended the tri-state games (sports events for people with a disability). Lindsay was entered in four events and came home with two medals (one for swimming and one for discus throw). We all joke that he probably talked the opposition out of the sport (he talks nonstop – mind you it took us three years to teach him to talk!) Lindsay's brother has moved out and is building his first house, but we see him every weekend (he works as an architectural draftsperson and is an elite cyclist). I work 3 days a week as a teacher librarian at a local primary school and I absolutely love my job. I keep myself a little fit by running or riding early in the morning."

A book that many people will want to add to their shelves is E.C. Knox's Don't Call Me Honey! Call Me Cherry Pie. It is a candid and amusing description of life with someone who has an intellectual disability. Written from a brother's point of view, it illuminates the type of situations that families affected by disabilities experience. Colour illustrations on each page show the readers the brother and mother's embarrassment, shock, and, eventually pride, as things go out of control around them.

Poignant and humorous, this book can be read to children of all ages, and shared with any adults who are affected by someone with an intellectual disability. A great tool to encourage understanding of differences, share Don't Call Me Honey! Call Me Cherry Pie with neighbours who do not understand your child, and education professionals who will find it a useful resource in guiding children's interactions. The book includes two pages where a reader can write their own story.

E.C. Knox is currently working on her next book: Your Goat's Out. She lives in Australia with her husband. She has two adult children; the youngest son has isodicentric 15 (idic15), a rare chromosomal syndrome.

Don't Call Me Honey! Call Me Cherry Pie is available from Amazon at www.amazon.com , Barnes and Noble at www.barnesandnoble.com and can be ordered at local bookshops.

Dup15q Alliance Participates in Sudden Unexpected Death in Epilepsy (SUDEP) Institute

Kadi Luchsinger, Executive Director, Dup15q Alliance

The American Epilepsy Foundation and the Danny Did Foundation invited 30 experts from all over the world to help develop a strategic plan to address SUDEP (Sudden Unexpected Death in Epilepsy) on February 26, 2013. I had the privilege to attend the SUDEP Institute meeting, in Washington D.C., on behalf of the Dup15q Alliance.

There were professionals, researchers and several advocacy groups present for this jam packed day. The morning started with a review of the current materials available for medical professionals, medical examiners/coroners and people living with epilepsy. We looked at a summary of the current materials available, reviewed suggestions, discussed best approaches, the feasibility and timeframe for each suggestion, and finally, selected priorities. The afternoon focused on current research and what supportive care options are available to families.

The Dup15q Alliance is known internationally for our support of families and promotion of research into SUDEP, and it is an honor that we are considered to be one of the model family organizations. I shared our experiences with SUDEP among our membership, including the resources that are available to our families and the process of brain donations through the Autism Tissue Program.

As we moved through the day, the discussion notes were posted on the walls of the conference room. Meeting attendees were asked to sign up for the topics which interested them. I signed up for communications, awareness, and research, and am now awaiting my assignments.

One of the best things about the day was the connections made with other organizations and people who join us in the fight against SUDEP. I sat next to a man who lost his wife and has started a candlelight concert series, which is very moving.

I look forward to being a part of the very important project and thank the SUDEP Institute for including Dup15q Alliance.

If you would like more information on SUDEP go to the Dup15q Alliance Physician's Advisory at www.idic15.org/Doctors.html. You can also email questions to info@dup15q.org or phone 855-dup-15qa (855-387-1572) between the hours of 9 a.m. – 5p.m. EST.

Other links:

Autism Tissue Program/Brain Donations
www.dup15q.org/autism-tissue-program.html
www.autismtissueprogram.org/site/c.nlKUL7MQIsG/b.5587185/k.E3A0/Overview.htm

The American Epilepsy Foundation on SUDEP
www.epilepsyfoundation.org/aboutepilepsy/healthrisks/sudep/index.cfm

Danny Did Foundation
www.dannydid.org

Cheers to Our Volunteers!

The Dup15q Alliance Cheers our Volunteers: The Supina Family

Katie and Todd live with their children Emma, Annie, and Ryan in Minnesota.

Our son Ryan was diagnosed with Dup15q on February 8, 2010, when he was 8 months old, after a couple of suspected seizures and missed milestones. We were handed a pamphlet and given some pretty horrible descriptions of kids with dup15q. We were so fortunate to have such a great group of local families - they all welcomed us and helped answer our zillions of questions. We went to the conference in Philadelphia the next year, not knowing what to expect. It ended up being one of the best experiences of our lives. We felt so connected to everyone there and loved seeing all of the happy families celebrating their awesome kids. Through the other families, we saw our future and knew that along with our twin daughters we would help Ryan continue to be who he is: one super cool dude. When Kadi announced that the next conference would be in Minneapolis we ran to Karen (who had never actually met us in person before) and told her we wanted to help!

Since then we have been working along with Deb Lindgren to help in any and every way needed: the raffle, meeting with speakers, touring the conference area, and trying to convince local businesses to donate to our group. We've loved working on this conference because we've learned so much about our kids and their needs.

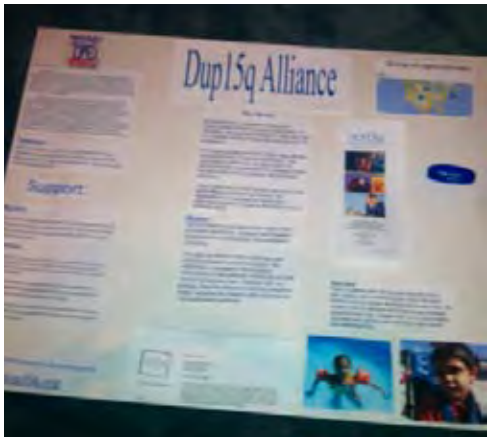
When we started planning for the conference we were a little overwhelmed by the size of our to-do lists. As soon as we were able to scratch something off, we ended up adding ten more things to the list. It just kept going and going. Luckily we have a great group of parents involved that are willing to jump in and help. Because of Deb's many connections in the area, we were able to find some amazing presenters who over the past year have spent hours and hours studying several kids with dup15q. Karen and Kadi have worked so extremely hard on behalf of all of our families to build this conference from the ground up. There are so many details involved with planning this conference - we are so excited to see it all finally coming together!

Ultimately it is all about our kids. The best part about being involved with conference planning is that we get a chance to gush about our sweet kids. We know we are totally biased, but we believe that our kids have a sparkle and zip that makes them irresistible. We are so grateful for everyone that gives up a piece of their free time to help all of our kids - we have amazing families in this group!

Raising Awareness: A college story

By Tina Delorenzo, mom to Nicholas (dup15q) and her family live in New Jersey

Kelly Dorman is my sister. She is attending Rowan University in Glassboro, NJ, and will be graduating this May with degrees in Elementary Education, English, and Special Education. As part of her requirements, Kelly had to do a presentation to her Human Biology class. The presentation was set up like an expo, in which each student had to do a presentation on a support group. Her presentation has information from the Dup15q Alliance website, pictures of our son Nick, and brochures.



Her teacher was very knowledgeable about chromosomes and the human body in general, but Kelly stumped him with her presentation on dup15q. He had never heard of it and wanted to know more about it. Kelly had hoped to teach something that her teacher and class had never heard of. Having a direct connection with Nick helped her decision to do her presentation on the Dup15q Alliance. She has a special and close connection with Nick. Her presentation helped people understand how rare Nick's syndrome is and how special kids with dup15q are.

The feedback she received confirmed that no one had ever heard of dup15q. The class was very interested in it and in the personal connection Kelly has with Nick. Kelly found the project really helped her learn more about Big Tent, the Alliance families, and the characteristics of dup15q. Helping her with this project helped us realize the love and support that is out there for our kids, especially when we have an opportunity to raise awareness!

THE MIRROR

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Hosting an Annual FUNDRAISER

By Tessa Quinlan

Without Dup 15q Alliance, we would be lost. We wouldn't know who to call for everyday life advice on dealing with a special needs child, how to find knowledgeable professionals regarding medical advice, or just a special friend who we can vent to at times. We have made some amazing friends within the Dup 15q Alliance community, and we are so grateful. Our annual GQ fundraiser is our way to give back.

Of course we do it for our son, but why not help more families who are dealing with the same syndrome at the same time? Our donations, along with many others, allow Dup 15q Alliance to carry on. Our 3rd annual GQ fundraiser raised over $13,000 for the Dup 15q Alliance. Every year we have increased our donation on behalf of our son, Gavin Quinlan. What will next year bring? We will find out in October, 2013, when we host our 4th annual fundraiser.

Every year we cringe at the idea of hosting it again, but at the same time my husband and I get excited in planning for the next year. It's a lot of work,, and a lot of call backs but it is all worth it. We are always in planning mode! 2012 was a great year. Our venue has gotten to know us. We have things under control on how to efficiently run things and we always strive to improve on the simplest things. For us, year 1 was the tough year as we were just figuring how to do a fundraiser. Year 2 is the year we worked out the quirks of year 1. By year 3, we have it almost figured out!

We never feel like we have it perfected because it may cause us to miss out on a great idea. Here was this year's great idea: we made use of the numbers that the bracelets come already preprinted with. For each attendee who purchased a bracelet, the only number they had to remember was the number that was preprinted on their bracelet. Each attendee that had a bracelet and then purchased raffle tickets wrote the bracelet number on the back of each raffle ticket. They didn't have to remember the umpteen raffle ticket numbers. Since they only had to remember one number, it made it simple. It also allowed things to run smoothly and much faster.

We have found a great venue and that is huge when planning. Make sure to find a venue that you are comfortable with and that feels comfortable with you. We plan an adult based fundraiser at a bar on a Saturday night from 6-10pm that includes food, drinks, entertainment, and a complimentary door prize raffle ticket for each bracelet they purchase, all for only $30. This year we scored with a northern suburb chamber of commerce that donated 400 $25 gift cards that could be used at a number of restaurants in the area. Each person who walked in paid $30, and in return received a $25 gift card and a comp raffle ticket. This was great advertisement for the suburb and also for our fundraiser. We hope people will remember this and want to come back next year.

When we do fundraising we are super persistent with our direct asks for donations; we don't take a no answer/no return phone call as a NO. We keep calling and emailing, telling them that we are following up and don't want to miss out on a possibly opportunity that can benefit children. If the answer is NO, then we let them know we understand but hope that they remember us because next year may be the year that works better for them. We ALWAYS send out thank you notes after our fundraiser!

If you would like to talk about running a fundraiser for the Dup15q Alliance, I am only an email away. I can be reached at Tessa526@comcast.net. If you have any questions, please contact me. I am always eager to share my thoughts and experience.

Tessa Quinlan and her husband Mike are raising Gavin (Dup 15q), Ava & Addisyn. The family lives in Illinois.

Los Angeles and New York Dup15q Clinics Open

The Dup15q Alliance is proud to announce two new Dup15q Clinics in New York, NY, and Los Angeles, CA. Dr. Shafali Spurling Jeste, head of the Los Angeles clinic, shares some of her excitement about the establishment of a Dup15q clinic at the UCLA Center for Autism Research and Treatment.

Q: How did you first become interested in establishing a Dup15q Clinic at the UCLA Center for Autism Research and Treatment?

Dr. Jeste: I had two patients in my clinic with 15q duplications, both with developmental disabilities. As an autism specialist in the UCLA Center for Autism Research and Treatment, my research focuses on using biomarkers to understand development and outcomes in children with Autism Spectrum Disorders. I was fascinated by the fact that despite their similar genetic mutations these girls had such different behavioral, cognitive, and developmental profiles. I began a dialogue with the parents of one of my patients who is an active member of the Dup15q Alliance Board (Guy Calvert) and we started the process of formalizing a clinic for these kids at UCLA.

Q: Are you collaborating with the clinics in Boston and Miami and New York?

Dr. Jeste: Yes, we will be meeting regularly over the phone to discuss clinical issues and cases. We also will be working to coordinate the gathering of clinical data so that, over time, these data can be queried for prospective studies on development, behavior, and treatment in these children.

Q: What will the UCLA clinic offer to families who visit?

Dr. Jeste: The clinic will evolve to meet the needs of our patients. Currently, all children will be evaluated by me and will receive any necessary behavioral or cognitive testing that can help better understand the child's developmental profile. This testing will be performed by a psychometrist with supervision from a psychologist. Based on the child's neurological workup, further testing such as EEG, MRI or any laboratory studies will be ordered on an as-needed basis. I will follow up with all studies. For children with active epilepsy, children will be referred to the UCLA epilepsy program and their care will then be coordinated between my clinic and the epilepsy clinic. Any other medical referrals will also be made by the clinic. We will follow children as closely as necessary based on their neurological needs. Finally, as our research efforts become more formalized, parents will be given the opportunity to be enrolled in appropriate research studies, such as those targeting biomarkers and treatment.

Q: With the establishment of these specialty Dup15q clinics, where do you imagine our understanding of dup15q syndrome may be 5 years from now? (Go ahead and dream big!)

Dr. Jeste: Children with genetic mutations provide us with a valuable opportunity to translate our understanding of disease mechanisms: from genes to brain to behavior. With a detailed understanding of the specific areas of strength and impairment seen in children with Dup15q we can start to design targeted treatments that will be most effective for these children. Our goal is to ensure that children are given an opportunity to have the best possible lives and, in the process, to be given access to all of the cutting edge research in the field of developmental neurogenetics.

Los Angeles Clinic: *Located in Los Angeles, California, the Clinic is run by Shafali Spurling Jeste, M.D., Assistant Professor of Psychiatry and Neurology and Director of Electrophysiology Core at University of California Los Angeles (UCLA).*
To schedule an appointment with Dr. Jeste, please call (310) 794-4008.

New York Clinic: *Located at NYU Langone Medical Center, New York, NY, the Clinic is run by Heather Lau, M.D., Neurogenetics Division Chief.*
To schedule an appointment with Dr. Lau, please call Elizabeth Odewale at (646) 558-0802.

Additional Dup15q Clinics can be found in Boston, Massachusetts; Miami, Florida; and Seattle, Washington.
For more information, visit www.dup15q.org/Dup15qClinics.html.

Family Portrait

Our Life With Vega

Pietro and Cinzia are the parents of Vega, age 55 (dup15q). They live in Genoa, Italy (Genova in Italian), and they have been living there since Vega was 10

My wife and I were thirty years old. We were happily building our life together. I was a physician in an Italian town and my wife was a school teacher. We had a little girl, vivacious and bright, and were anticipating the birth of our second child.

Vega was born in 1958. We chose the name of a star, but it became clear that our star was not radiating brightness. Our baby was severely disabled. She was listless and barely able to swallow. My wife could not bear the thought of the suffering that her girl was going to endure. We were desperate and hopeless, but our love for our child made us react. We started fighting, a fight that became an indissoluble element of our life. With Vega's first timid smiles we travelled all over Italy looking for someone who could cure her. The best pediatric neurologists gave the vague diagnosis of "acquired neonatal cerebral disease". My wife and I worried we had somehow caused this.

Vega's childhood was hard, but with great effort and guided by my wife's care, Vega acquired skills. Vega muttered her first words at four years old and by age six she was walking alone. At age ten, she became continent. My wife managed to sustain a cheerful energy with all the children, only to silently cry at night. Those years were full of intensity, tiredness and anxiety, with endless sessions in therapy and hopeful trips to specialists all over Europe, but they were also years that cemented our family. We realized that having a disabled child was not incompatible with raising a happy family. In her own way, Vega was bringing us closer with a sense of purpose. She loved life. Perhaps she was not the shining star we had hoped for, but Vega had graciously become the sun who made us brighter.

Vega's older sister first struggled with the demands of her helpless sibling. She gradually took a maternal role that has lasted over time. Her younger brother was born two years after Vega and they became inseparable. When he was four years old, he discovered that Vega did not understand well. He then became her protector and teacher. As they played, he taught her skills that we had been unable to teach her: saying difficult words, climbing stairs and coloring. My wife and I came to understand that by imitating other children, Vega acquired skills which were out of reach with therapy or teaching.

In the 1960s, the norm was to institutionalize disabled children. My wife said, "No, my girl will not go to an institution. If we cannot change her, we will change the world around her." We signed petitions, met with school authorities and politicians, and held public demonstrations. Italy became one of the first countries to open its schools to disabled children. Vega was enrolled initially in her mother's class and was likely the first intellectually disabled child to enter an Italian school.

Meanwhile, our careers continued, our other children did well in school and we become a much happier family than we ever hoped for. Vega discovered music, sweets and travel. Once, while visiting a Roman archeological site, Vega expressed loudly to the guide that "I have had enough of broken walls", but she tolerated them because she loved hotels, restaurants and going out with her family.

At age fourteen, Vega's adult life started to take shape. She finished school and joined a day program. She could recognize letters, numbers and could write and read some simple words. She would come back from her "work" happy and excited. Vega continued to improve her motor, verbal and intellectual skills, and became engaging and charming.

We had imagined Vega would remain an eternal child, but in her early twenties, she started asking to have her nails done. She also started to fall in love with boys. Fortunately her crushes never lasted long, and when she announced that she was getting married, we convinced her that we needed her at home. Vega loved to go to the hair dresser with her mother, shop with her sister and go to parties with her brother. This was probably the happiest time of her and our lives.

At age 23, Vega had her first generalized seizure and a new set of problems began. She took medications, but her complex-partial seizures continued and became more frequent. The drugs made her less vivacious and more clumsy.

A few years later, Vega's siblings moved away, ending years of closeness. She became depressed and less responsive, so we visited her brother. At first, Vega barely looked at him, but soon they started going out together and her depression lifted. My wife and I saw it was time to plan for when we could no longer care for Vega. Her siblings made an unprompted promise that they would care for Vega. We worked with our lawyer and accountant to provide her the best financial and legal protection.

In 1999, Vega was a part of a genetic study of adults with intellectual disabilities and was diagnosed with IDIC-15. Finally, the anguish and guilt that we had caused her disability came to rest.

When Vega was close to 50 years old, she hit her head in a terrible fall - perhaps due to a seizure – and our life tumbled. Bleeding around her brain had to be removed surgically. We feared for Vega's life and became painfully aware that we could not bear the thought of living without her. Eventually when she was able to return home, she could no longer walk alone, was indifferent to all her favorite things and would scream inconsolably for hours. Many nights we sat in despair as she cried, unable to find a way to help her. Our happiness and sense of achievement was shattered in those terrible weeks.

We had no strength left, but also no choice. We again declined the suggestion to institutionalize Vega, and cared for her as if a frail baby. Holding her, nourishing her with creamed food, singing her favorite music, she slowly improved. Vega became calmer and stronger but her happiness that so much enriched our life had given way to a wordless sadness we had no tools to reach. Two years after the accident, in Vega's slow way, she came back: funny, adorable and contagiously happy.

Now in our mid 80s, we are again her timeless parents and playmates. She continues to enjoy her "work" and has a heightened sense of self. Vega thanks her mother for her care and asks: "I am a normal girl, am I?" It is getting hard for us to do certain chores, but we now wonder if Vega is adding years to our life. She certainly continues to bring home a life affirming joy that blocks our worries for the future. We realize that life has been a never ending struggle with unexpected rewards. We cannot imagine a richer, more difficult and more rewarding life than our life with Vega.

Reflections from a Sibling

Eyes Full of Love

By Alison Orton

The burning piercing of everyone's eyes is penetrating deep into my soul while the tight grip she has on my hand gives me comfort. We are walking into a store that has groceries, any kind you could imagine. The smell of just crushed coffee beans and butter salt popcorn are invading the smell of produce. But I can feel that there is still a stench in the air. The stench of heads turning, eyes staring, and fingers pointing as my sister Mikaela and I walk hand-in-hand into Family Fare.

Mikaela walks with confidence, never looking back to see if people are staring. As we walk through the store to get the groceries our mom sent us to get, she wants to stop to talk to everyone. Everyone is important to her; no matter hair color, height, or race, she loves everyone. To some people she looks like a typical sixth grader, and to others she is something to stare at. Her short bob haircut, growing side-swept bangs and average height gives her the appearance of a "normal" middle schooler. As we walk hand-in-hand, a sixth grader and a high school senior, people stare. It is not normal for that to occur. We are just shopping, getting groceries just like any "normal shoppers."

As we go past the coffee shop, and down the aisle of reds and blues, greens and pinks, she stops. The scene that unfolds before our very eyes draws Mikaela in to help. A young mother is with two children, who stand just below her hip in height, eyes and hands wandering, ready to capture anything they can. She is trying to calm an infant with a bottle while her oldest son is trying to run back-and-forth down the aisle. As we continue to walk Mikaela bends down, picks up the pacifier that the baby had dropped during its fit to get food, and hands it to the frantic mother whose face is starting to turn red with anger and frustration. She looks at Mikaela and with all the calmness she still had in her says, "Thank you sweetie," and we keep walking. Going directly to the coffee grounds that our mom wanted us to pick up Mikaela says, "aww he was so cute," talking about baby. As I respond with, "yes he was" I pick up the coffee and walk to the self-checkout counter, with Mikaela right next to my side.

Love. If I could define Mikaela with one word it would be love. She loves to hold babies; she loves to help people; she loves everyone the same. She loved picking up that pacifier just to help that mom who felt like everything was chaos. Although some may say she's weird, I say she is perfect. God crafted her, molded her, and made her special, with an extra partial chromosome. God gave her more chromosomes the most people have. He awarded her with more than we have just so that he could showcase his love for people. God loves others through Mikaela. There is no one that she has ever come into contact with that she would not want to help, even when so many people glare, stare and point. She loves everyone unconditionally, just as we are called to do.

Allison Orton lives with her parents Tracey and Julie and her siblings in Michigan.

Reflections from a Grandmother

Peanut is Our Sweet Baby Girl

By Dee LaRocco

Peanut is a precocious five year old little girl. She loves Glee, princesses, Curious George and Sponge Bob. She also has Chromosome 15q Duplication Syndrome. And Peanut is my granddaughter that I have had the privilege of being with and raising since birth.

Almost a year ago we were informed of Peanut's new diagnosis of Chromosome 15q Duplication Syndrome. The diagnosis came as we were preparing for her Chiari 1 Malformation Decompression surgery in a matter of days. Her dup15q diagnosis was the most recent of a very long list of medical issues we have had to deal with, research, or just plain old live with over the past five plus years.

When we were told of Peanut having dup15q, I pushed the geneticist into giving some kind of predictions of what we could expect down the road. His answer was for us not to waste our time and energy by saving for college as she would never have the mental capacity to go that far in her education. What this doctor didn't know, and will learn as she teaches him, is that we don't allow the word "never" into our vocabulary.

I have always held the faith that although we don't know what it is, God has a plan for us all. I also have a rare disorder, which my mother had, and so grew up being "different". When Peanut was born and started to develop her medical issues, I finally understood why these things happened in my family.

Each day that I look into Peanut's brilliant smile, I feel blessed that I should be chosen to share in this wonderful child's life! She is an inspiration to all who meet her, even if only for a moment. She has a way of making you feel like you are the most important person on this earth and that you are so thoroughly loved. I see her do this day after day.

One of my most favorite philosophies for children is: expect everything but accept what's possible. This has helped us to refuse the negative attitude towards Peanut from doctors. We have heard too many times to count that Peanut will never be able to do things like normal children. HA! She's passed all the limitations the doctors have given us over the years! The one thing that she truly wants to do is still evading her: to hula hoop like her Auntie Barbie! But give her time, she will achieve this too! I have all the faith in the world for my lil Peanut!

THE MIRROR

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.***

REACH FOR THE STARS!!!

We are looking forward to seeing many families on July 25- 27, 2013, at our Reach for the Stars conference in Bloomington, MN, at the Radisson Blu – Mall of America. This conference is full of learning sessions and workshops, social opportunities to network with other families, and fun events!

Families can register for the conference at
http://dup15qalliance.ticketleap.com/reach-for-the-stars-conference/

Families can book their hotel rooms at the Radisson Blue - Mall of America at the conference discount price by entering DUPALL on the main hotel booking page, or using the link at http://www.radisson.com/reservation/resEntrance.do?pacLink=Y&promoCode=DUPALL&A=USABLMA

Special conference events:

Thursday, July 25th, 7:30 - 10:00 am

Nickelodeon Universe - Once in a lifetime opportunity! The amusement park is reserved just for our Dup15q children and their families! No lines, no wait! This is for you!

Friday, July 26th, 7:00 - 9:00 am

Breakfast at Sea Life Minnesota Aquarium - Thank you to our sponsor: KayJay 5K and the Ebensperger Family. We have the Aquarium all to ourselves. Join us for this fabulous adventure as we eat a light breakfast with the creatures of the sea.

Child Care Available on Thursday:

On Thursday from 12:30 -5:30 pm, babysitting services will be provided in our childcare room by Above and Beyond Nannies, Inc. for both our Dup15q children, as well as typical siblings. Above and Beyond Nannies, Inc. has provided nanny services for over 15 years in both home and conference settings, with experience in dealing with special needs children. They will provide nannies in an appropriate ratio to watch our children, including toileting as needed. The room will be filled with activities that both our Dup15q children, as well as typical children, can enjoy. With babysitting services provided, it is our hope that all adult family members will be able to participate in the family support sessions, which are typically some of the most popular and appreciated sessions of the entire conference.

http://www.dup15q.org

Address Service Requested

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