

FROM THE EDITOR

BY JEAN HOUSE

The mission of Dup15q Alliance is in action every day—online; at fundraisers, national research meetings, and other events; and in hardworking volunteers' homes. This issue of *The Mirror* is dedicated to the mission:

Dup15q Alliance provides family support and promotes awareness, research and targeted treatments for chromosome 15q duplication syndrome. Through our work to raise awareness and promote research into chromosome 15q duplications, we seek to find targeted treatments so that affected individuals can live full and productive lives. Together with our families, Dup15q Alliance is working toward a better tomorrow for children with chromosome 15q duplication syndrome.

The organization handles several responsibilities for families and researchers—and we are growing and making progress in all these areas.

Family support: Planning is underway for the upcoming conference in Minneapolis. We also share information and support in other ways: online, at regional gatherings, as well as through *The Mirror*. Be sure to check out the Family Portrait, Reflections from a Grandparent, and book review in this issue to learn how other families live with dup15q syndrome.

Awareness: Dup15q Alliance recently was represented at a meeting of the National Institutes of Health. Reaching out ensures that awareness of the syndrome continues to grow among researchers, genetic counselors, and doctors alike.

Research and targeted treatments: Important studies are underway, including a mouse model and stem cell research. For more, see the articles about these studies, as well as the research report in this issue. Also, be sure to check out the registry update.

Dup15q Alliance is always looking for more volunteers to help meet all of these goals. What are you interested in? How can you help? Joining the registry is a first step we all can take. If you are interested in contributing your time and talent to put our mission into action, visit www.dup15q.org and discover how to get involved!

JEAN LIVES IN WILMINGTON, N.C., WITH HER HUSBAND, CHRIS, AND SON JONAH, 5 (DUP15Q)
E-MAILS ARE WELCOME AT JEAN@EDITORHOUSE.COM.



INSIDE THIS ISSUE

- 2 A New Way for Families in the UK to Support Research
- 3 Mouse Model Holds Promise for Unlocking Secrets of Dup15q
- 4 Research News
- 6 Organizing A Fundraiser
- 7 Siblings, You're Not Alone: Book Reviews
- 9 Dup15q Alliance Registry: A Resource for Families and Researchers

In Every Issue

- Financial Report 5
- Cheers To Our Volunteers 8
- Family Portrait 10
- Reflections from A Grandmother 11

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.

A NEW WAY for Families in the UK to Support Research

EXCERPTED FROM "BRAIN RESEARCH: THE KEY TO UNDERSTANDING," BY BRENDA NALLY



A research initiative to improve the current understanding of a range of conditions including chromosome 15q duplication syndrome has been established in the United Kingdom. The Brain Bank for Autism & Related Developmental Research will coordinate brain tissue donations in the United Kingdom and help share tissue resources through the Autism Tissue Program (ATP) in the United States to approved neuroscience projects worldwide. The U.S. and U.K. brain banks are the first to establish an international brain tissue network for this area of research. Scientists in other countries are exploring the possibility of joining this partnership by setting up further brain banks.

The key to fully understanding genetic disorders such as dup15q syndrome is through brain research. There are strong limitations on what can be learned about the brain's development during life, but a great deal can



be learned from scientific study of the human brain after death. Only postmortem research can clarify the cellular and molecular differences in the brain. When scientists better understand the differences in the brain functioning of individuals with dup15q, it may be possible to develop targeted interventions to help individuals and their families to respond most effectively.



The first of 21 donations made to the U.K. brain bank to date was from the family of a young person who had dup15q. After they had agreed to this, her parents commented: "Our beautiful daughter Alice died at age 13. She had dup15q syndrome and a diagnosis of autism. During her short life we always tried, in whatever way we could, to help her and other children like her have as good a quality of life as possible. Although we lost our beloved

Alice, we found some consolation in being able to help others by donating her brain tissue to further research."

The most difficult time to consider organ donation is at the time of death. This is a very sensitive and personal decision and nobody should feel pressure to make a pledge to donate, but families are encouraged to consider whether they would want to support research in this way. The brain bank offers sustained outreach to families to keep them informed about the research and its progress and to give them guidance and support. The bank's development is strongly influenced by their concerns and contributions.

If you live in the United Kingdom, please find out more about this research by visiting www.brainbankforautism.org.uk or contact the free U.K. helpline at 0800 089 0707. The U.S. website is www.autismtissueprogram.org.



By KAREN SALES
CONFERENCE CHAIR

The 7th International Conference on Dup15q Syndrome, **Reach for the Stars**, will be held July 25–27, 2013, in Bloomington, Minn. Planning is well underway, and we are excited to welcome our Dup15q Alliance family to this fun-filled Midwest city!

As the conference committee continues to find speakers on new and exciting topics, we are also working on different ways of presenting topics we have all come to rely on! Here are two ways you can begin preparing yourself and your family for a fantastic time in the Minneapolis area:

- Check out the hotel by visiting www.msairport.hilton.com. Watch for an announcement in early 2013 to start booking rooms at a rate of \$139/night + taxes.
- Learn about the different adventures that await you in

Minnesota! A few websites to begin your search are www.exploreminnesota.com, www.funminnesota.com, and www.mallofamerica.com.

There are plenty of sights to see and activities to do for an extended family vacation!

Mouse Model Holds Promise for Unlocking Secrets of Dup15q

BY GUY CALVERT
RESEARCH CHAIR



In February, the Dup15q Alliance Board voted to spend \$40,000 to help fund the development of a new mouse model of dup15q syndrome. Although the alliance had long supported research indirectly—for example, by sponsoring annual scientific conferences on dup15q—this direct investment for a specific research project marks a step into new territory for us.

We did so because we believe that this new project will make a critical difference in advancing research toward life-changing treatments for dup15q families, the price tag for the research was affordable, and we felt that our funding would have a major impact on the completion of this project.

How and why would this project make a difference? Animal models of genetic syndromes are a vital tool for researchers investigating the behavioral, biochemical, and neuroanatomical mechanisms of disease. These models can afford researchers an otherwise elusive understanding of how these mechanisms are altered by genetic anomalies, which is very often the key to coming up with targeted therapies to restore normal function. Among animal models, mice are prized by scientists for their similarity to human genomic structure and availability of testable analogs to human brain function and behavioral traits.

This is not the first mouse model of chromosome 15 duplication to be created. Several others have been produced, each with its own particular strengths and weaknesses. In a promising development last year that was hailed as a significant breakthrough, Harvard researcher Matthew Anderson published the results of his lab’s investigations into their dup15q mouse model of autism. According to Anderson, his mice—which were fashioned with extra copies of the 15q-critical UBE3A gene—exhibited behavior that corresponded on three key points with autistic behavior in humans.

The good news is that this mouse model holds much promise for unlocking the secrets of dup15q syndrome and laying the groundwork for research into life-changing treatments. The bad news is that this mouse model is not yet available to the wider research community and may not be for some time.

So while we applaud Anderson and his team for their extraordinary work, sometimes imitation is the sincerest form of flattery. In a bid to accelerate research by clearing a logjam at a critical juncture, we decided to fund a joint proposal by Dr. Lawrence Reiter (University of Tennessee Health Science Center, and a member of the Dup15q Professional Advisory Board) and Dr. Scott Dindot (Texas A&M University), to develop and study a new mouse model of UBE3A-duplication—similar to Anderson’s model but with additional features that allow for more finite control of UBE3A gene expression both temporally and regionally in the mouse brain. According to Reiter,

“this new mouse model of duplication 15q syndrome will allow us to control the expression of the UBE3A gene to mimic both interstitial and isodicentric duplication levels of this protein. It will also provide a way to over-express UBE3A in sub-regions of the mouse brain so that we can begin to understand how different regions of the brain respond to increased UBE3A levels.”

Reiter and Dindot hope to finalize the project by early 2013, and as a condition of funding we stipulated that all reagents and materials be made available to other investigators in compliance with National Institutes of Health (NIH) guidelines for resource sharing.

Why should the funding for this project come from Dup15q Alliance? It turns out that government sponsors like the NIH are more and more reluctant to make such investments, and in any case the NIH pool of funds is greatly diminished these days. While the \$40,000 pricetag is beyond the means of most individual families, it is possible for our organization to fund this work. That itself is remarkable—often annual research bills come to a quarter million dollars or more, amounts that are currently beyond the means of the alliance as well.

This project represented a rare opportunity for the alliance to uniquely make a difference, advancing critical research toward game-changing treatments for families living with dup15q. As Executive Director Kadi Luchsinger put it: “This is an exciting time for the Dup15q Alliance community. A mouse model is critical to propel research forward on dup15q syndrome. The entire community should be celebrating this accomplishment, and we look forward to learning more through the work of Dr. Reiter and Dr. Dindot. Thank you to all who have made donations to the alliance; you have helped to make this happen and ultimately helped to make a difference.”

Correction

The list of international representatives in the Spring 2012 issue of *The Mirror* was incomplete. The correct list of representatives for Canada and the areas they’re responsible for follows.

Lori Eisenhour—British Columbia

Denis Myers—Manitoba

Ellen Doxtator—Ontario

Nick Pomainville—Eastern Canada

Research News

BY KADI LUCHSINGER
EXECUTIVE DIRECTOR

Many exciting things are happening with our Dup15q Alliance research efforts, and we are thrilled to share them with our families:

- The dup15q clinic at Massachusetts General Hospital in Boston has seen more than 30 people, and many more are coming.
 - The 2012 Dup15q Scientific Meeting is now being supported by a \$5,000 grant from Autism Speaks.
 - The alliance received a \$1,000 scholarship from the Autism Science Foundation, allowing me to represent the organization at the International Meeting for Autism Research (IMFAR).
 - I also attended a meeting at the National Institutes of Health (NIH) regarding autism and epilepsy. Fifty of the top leaders in the field of autism and epilepsy gathered to develop a roadmap to best understand the two. Dup15q had a strong voice there.
 - We have launched the tooth study with researcher Dr. Larry Reiter. He has already collected some teeth that came from people with dup15q, which are now growing stem cells.
 - The mouse project has gotten off to a promising start for Dup15q Alliance. It is going to take some time, but things are moving along. For more information, please see the story in this issue.
 - I have been working with Orrin Devinsky and his staff at New York University as they are conducting an in-depth review of the sudden unexplained death in epilepsy (SUDEP) cases in people with dup15q, by interviewing each family. More than 10 families have participated.
 - Ron Thibert and Brenda Finucane are finishing up the written part of the seizure survey. They hope to have a draft this June and submit for publication this summer.
 - Some potential new research studies in the pipeline will be announced once they have been finalized.
- We are actively encouraging families to add their information to our registry (www.dup15qregistry.org). The registry is only as rich as the contributions from each family. Please take some time and add your child's information today!



The Dup15q Alliance Booth at the 45th Annual Gatlinburg Conference, March 7–9, 2012, in Annapolis, Md. The focus of the meeting was disorders of chromosome 15q.

MORE TEETH PLEASE!

We are really up and running on the dental pulp stem cell study. Two people are working with me on this now, and a third is coming in mid-June. We can now handle as many teeth as we can get. I suggest that parents get their genetics reports handy first before contacting us. E-mail is the best way to get in touch. Parents are also welcome to call if they wish—they may just miss me.

We have already had:

- 6 interstitial duplication 15q teeth
- 6 Angelman syndrome teeth
- 1 isodicentric tooth

Unfortunately, due to mold contamination and other issues with growing these cells, we only have frozen:

- 1 interstitial duplication 15q line
- 2 Angelman lines

We have quite a few control lines now and are working out the conditions so that we can grow each precious tooth. That being said, the number of e-mails and calls has slowed to a trickle. Please contact me regarding participating in this research study.

Lawrence T. Reiter, Ph.D.
Associate Professor,
Department of Neurology
lreiter@uthsc.edu
855 Monroe Ave., Link 415
Memphis, TN 38163
901-448-2635 (Office)
901-448-1160 (Fax)
901-448-7443 (Lab)

EDITOR
JEAN HOUSE

DESIGNER
MELISSA HAIRR

PROOFREADERS
NICOLE CLEARY
ALISON KALNICKI
KIM MARING
JANE TRUE

FINANCIAL REPORT

By TOM DOYLE
FINANCIAL OFFICER

As of the time of this writing in mid-May, Dup15q Alliance has \$28,743.19 in checking and just over \$253,000 in savings and money market accounts. We are preparing for the scientific research meeting this summer and already making plans for the biannual conference in Minneapolis in summer of 2013. Once again, we are grateful to all who have contributed to Dup15q Alliance. As we approach the end of the fifth month of the year, we are already at 50 percent of what we budgeted for income for the year. Special thanks to so many of you who have companies that provide matching funds for your donations; that really multiplies the size of your gifts.

We are also at about 50 percent of our budgeted expenses, but we decided to fund a mouse model for research after the budget was set. That 50 percent of expenses includes the \$40,000 that we set aside to kick-start some specific research, so we are in good shape on the spending side of the budget at this time as well.

In addition, if you haven't already please help us make the registry a valuable tool for Dup15q Alliance by completing the registry process for your family. This is also a major expenditure for our organization and one that will reap rewards for us down the road, so we encourage you to participate.

We continue to be grateful to the many volunteers who make Dup15q Alliance work and to our families who so generously support our efforts. Thank you on behalf of Dup15q Alliance.

The Dup15q Alliance annual report for 2011 is available on the website. You can find it on the right hand side of the home page, in the blue section; it's the last item in the list.

Announcement

The Dup15q Alliance Board of Directors is pleased to announce the election of two new board members: Lori Eisenhour, of North Vancouver, B.C., Canada, and Dana Tilton, of Tyrone, Ga. Lori is our first International Board Member and currently serves as coordinator of the International Registry. Dana is currently a regional representative and talks to many new families. We are very excited about the energy and passion that they both will bring to the board. Both will serve 2-year terms effective with the June 2012 board meeting. Congratulations to Lori and Dana for being elected and also our sincere thanks to the other candidates who submitted applications.

The seats being filled are vacancies created with the departure of Patti Rubel and Rachel Doucette. On behalf of the entire Dup15q Alliance organization, we thank both Patti and Rachel for their service to our organization.

Dup15q Alliance

CORPORATE OFFICERS & BOARD MEMBERS

LEN POORE Tewksbury, MA	Board Chair
KADI LUCHSINGER Fayetteville, NY	Executive Director
TOM DOYLE Bellevue, WA	Vice President of Finance
KAREN SALES Ankeny, IA	Corporate Secretary
MIKE PORATH La Crescenta, CA	Board Member
RYLIE McHAM Columbus, OH	Board Member
LINDA MEAGHER Sugar Land, TX	Board Member
PAUL KARCH Madison, WI	Board Member
GUY CALVERT Burbank, CA	Board Member
LORI EISENHAUR North Vancouver, BC	Board Member
DANA TILTON Tyrone, GA	Board Member
DONNA BENNETT Thomasville, PA	Co-Founder & Lifetime Board Member

PROFESSIONAL ADVISORS

AGATINO BATTAGLIA, MD, DPed, DNeuro Calambrone, Pisa, Italy
EDWIN COOK, Jr., MD University of Illinois at Chicago
BRENDA FINUCANE, MS, CGC Elwyn Training and Research Institute; Elwyn, PA
JANINE LASALLE, PhD UC Davis; Davis, CA
LAWRENCE T. REITER, PhD University of Tennessee College of Medicine; Memphis, TN
N. CAROLYN SCHANEN, MD, PhD Nemours Biomedical Research; Wilmington, DE
SARAH SPENCE, MD, PhD Department of Neurology, Children's Hospital; Boston, MA
RON THIBERT, DO, MsPH Pediatric Epilepsy Program Massachusetts General Hospital; Boston, MA

ORGANIZING A FUNDRAISER

BY TESSA AND MIKE QUINLAN

Be Determined and Follow Up

When you ask for donations for a fundraiser, you have to be ready to hear “No.” But you will be surprised at how many times you hear “Yes,” so don’t get frustrated. Be determined when you are out fundraising. Many of the members of our fundraising team kept a folder in their vehicles. If we visited a place we felt was good to solicit, we did, and if we had the motivation to fundraise, we did. Fundraising has to be on your own time and at your own pace. There will be days you don’t feel like fundraising at all, but on other days you may visit 10–15 businesses.

You may not receive an answer on your first visit to a business, so be prepared to follow up via phone or e-mail. Following up is key. A few places gave us a donation for our GQ Fundraiser on the spot, but many of our donations came from determined follow-up. If someone says “get back to me” or “you need to talk to the manager,” don’t be disappointed. Just say OK, but ask them the best time to follow up and make sure you do.

Create Unique Raffle Items:

Think of exciting ways to create a raffle basket that you would want to win. Consult your team members, and try everything! At our fundraiser, we had a wide variety of prizes. Here are a few ideas that brought in the most raffle tickets per item:

- We were blessed with generous donations of professional sporting events tickets. Then we took it one step further. With a few simple phone calls, those professional event tickets became even more enticing. Two different local limousine companies agreed to provide transportation to and from both of the Chicago Blackhawks hockey games for which tickets were donated.
- If you have lotteries in your state, create a basket valued at \$50 worth of lottery tickets. The basket can contain any dollar amount of tickets, but we found that it is best if the minimum total value is \$50.
- Items signed by professional athletes are always popular.

Log onto corporate websites and fill out the donation request forms, but do this well before your fundraiser because most have deadlines for when the requests need to be made.

Make it Special

Think about events you’ve attended. Why did you enjoy them? Make your fundraiser fun and exciting for attendees; that is what will keep them coming back. We had an Elvis impersonator for entertainment because our son Gavin, 7 (dup15q), loves listening to Elvis’s music. There was another added surprise at the GQ Fundraiser: the Chicago White Sox mascot, SouthPaw. Gavin loves the White Sox very much.

Throughout planning, fundraising, and finally hosting the event, make sure you are organized (write everything down), detailed, ambitious, and determined. At the event, take pride in how things look. Your attendees will remember this. It takes a lot of work, but in the end it will all be worth it.

Finally, through all the stages of organizing your fundraiser, remember why you are doing this: fundraising is a difficult task, but it takes a village to keep Dup15q Alliance running. We need this charity to keep us sane, gain insight, communicate with others, and make strides in research.

Best of luck on any future fundraising events you plan. We are always here to help!



TESSA AND MIKE ARE THE PARENTS OF GAVIN, 7 (dup15q), AVA, 4, AND ADDISYN, 1. THIS IS THE SECOND IN A TWO-PART SERIES ABOUT THE QUINLAN FAMILY’S EXPERIENCES ORGANIZING THE GQ FUNDRAISER. VISIT [WWW.DUP15Q.ORG/MIRROR](http://www.dup15q.org/mirror) TO READ PART 1 IN THE SPRING 2012 ISSUE.

Siblings, You're Not Alone

Books for Sisters and Brothers of Children with Dup15q

BY ALISON KALNICKI

If you have a brother or sister with dup15q, these two books might reflect a bit of what it's like to deal with the syndrome in your family. Although both books depict life with a sister or brother with autism, the kids in *Rules* and *Autism & Me* might have lives similar to yours.

"Sometimes I just wish someone would invent a pill so David'd wake up from a long coma, and he'd say, 'Jeez, Catherine, where have I been?' And he'd be a regular brother . . ."—*Rules*

Rules, a fictional account of a 12-year-old girl's summer, has a suggested reading level of 10 years old to adult. Catherine has a younger brother with autism. She loves her brother and helps guide him through "regular" life by creating rules for him. As their summer progresses, she adds to the list:

- No Toys in the Fish Tank
- A Boy Can Take Off His Shirt to Swim, But Not His Shorts
- Sometimes People Laugh When They Like You, Sometimes They Laugh to Hurt You

Catherine meets the new girl next door, avoids the boy down the street, and is befriended by a teen in a

wheelchair who uses pictures to talk. She draws pictures for his communication book and chooses the words and phrases that are important in her life: *guinea pig*, *awesome!* and *sucks a big one*. Suddenly, black-and-white rules are difficult to use. But in the honest voice of a 12 year old, Catherine navigates her summer with wisdom, humor, and a willingness to rise to the challenges in her life.

"I'm like most kids, but my sister Arie is not."
—*Autism and Me*

Autism and Me is a series of one-page essays with a suggested reading level of 5 years old to adult. Each page gives voice to a different sister or brother of a child with autism; photos give a glimpse into their everyday lives. Reading like a conversation, the brothers and sisters discuss what life is like with their siblings. It candidly explores their hopes, fears, and frustrations, as well as their love, affection and understanding of how their sisters and brothers are different.

Interested in reading these? Pick them up at your library or bookstore. You might want to share them with others in your family or neighborhood. Ask an adult if you have any questions. And remember, you're not alone.

Autism and Me: Sibling Stories, by Ouisie Shapiro. 2009. U.S. publisher: Albert Whitman. Canadian publisher: Fitzhenry & Whiteside. ISBN 978-0-8075-0487-1. Also available in United Kingdom. Suggested age: 5–adult.

Rules, by Cynthia Lord. 2006. Scholastic. ISBN-13:978-0-545-03640-5 ISBN-10:0-545-03640-2. Available in United States, Canada, and United Kingdom. Suggested age: 10–adult.



Children who live in the same neighborhood as the Quinlan family were excited to host a lemonade stand for an entire weekend. They donated the proceeds, \$157, to Dup15q Alliance. Thanks for your hard work and generosity! From left: Ava, Jimbo, Connor, and Olivia.

Cheers to Our Volunteers!

BY LEN POORE

Dana Tilton is the Volunteer of the Quarter. Thank you Dana, for organizing the first gathering of Georgia families. A description of the event, in Dana's own words:

I truly believe everything happens for a reason. As I look back on my life, interests, and strengths, I know I am right where I'm supposed to be. I was born and raised in Atlanta and went to Auburn University, where I studied psychology. I have always been interested in the mind-body connection and have always wanted to help others. My husband, Luke, and I have three amazing kids: Hannah, 5, Addison, 4 (dup15q), and Christian, 21 months. Being a mom is the greatest job in the world and is one that I take very seriously. Having a child with dup15q, I get to dive deeper into trying to help others in a meaningful, life-changing way.

I am determined to do all that I can for Dup15q Alliance. In March, we had the first gathering of Georgia families living with dup15q. With the



very welcome addition of an Alabama family, seven families attended. We had the gathering at our house to allow the kids a feeling of freedom, while still being in a safe, fenced-in, "dup15q-proof" environment. Ultimately, this created a relaxed day with continuous conversation amongst all of the families. Everyone brought a different dish, and we ended up with enough food to feed a small army. We enjoyed a beautiful day full of jumping, swinging, story sharing, and instant connections that no words can describe. The love and happiness that pours out of our kids' hearts is contagious to those around

them. Being with others who have experienced the same heartaches and joys, fears and excitement, challenges and perseverance, an unbreakable bond is formed. This is the very reason that the members of Dup15q Alliance are more than members of the same group, they are a family.

It will be my pleasure to help strengthen and grow our dup15q family here in the South with many more gatherings in the future. I know in my heart that even greater things are yet to come for Dup15q Alliance, and I feel beyond blessed to be a part of it.

DUP15q ALLIANCE REGISTRY: A RESOURCE FOR FAMILIES AND RESEARCHERS

By NICOLE CLEARY

www.dup15qregistry.org

Thank you to the 104 families who have taken the time to add your child's data to the Dup15q Alliance Registry! We are actively encouraging all families who have not already done so to add their child's registry data today!

Why Should I Register with the Dup15q Alliance Registry?

This database is maintained with the vision of bringing together researchers and families raising children with dup15q syndrome. We hope that by connecting families and researchers more effectively, we might better understand dup15q as a disorder. You should register because we need you—joining with many others—to help develop effective treatment recommendations for dup15q syndrome.

What are the benefits of adding my child's information to the registry?

Although there is no direct benefit to any individual participating in the registry, by collecting information on affected individuals, scientists can:

- study why individuals have different symptoms
- learn about which treatments work and don't work
- help medical professionals improve how they treat individuals affected by dup15q
- speed up research in dup15q by collecting information that scientists can use
- let families know when they may be eligible for clinical research studies or clinical trials

What have we learned so far?

Families who enter their children's data in the registry have access to the "Explore Responses" feature, which allows them to see graphs of the registry data on:

- diagnosis
- seizures
- other neurological
- general health
- deceased
- pregnancy/neonatal
- cognitive development
- development
- behavior
- puberty
- pulmonary/sleep
- cardiac
- gastrointestinal
- ears/nose/throat
- vision
- orthopedic
- immunology/allergy
- anesthesia
- renal/metabolic
- survey comments

These graphs can provide immediate information to families and their doctors or therapists. For example, recently on the BigTent dup15q discussion board there was a thread about speech delay that incorporated information from the registry. Our registry captures data on children's current level of speech. Among current participants, we can see that more than half are verbal. Many of the registry participants are young children, and this data will certainly change as they age.

Please help us continue to grow this resource by participating today!

Please indicate the affected person's level of verbal communication			
		RESPONSE PERCENT	RESPONSE TOTAL
No Speech		43.88%	43
Speech is a mixture of meaningful speech and some echolalia or jargon		30.61%	30
Most speech is meaningful; however, some echolalia or pronoun reversal may occur		13.27%	13
Mostly normal speech		9.18%	9
Unsure		3.06%	3
Total Respondents:			104

More Registry News: We Need Your Input!

The good news is that we have 211 people signed up for the registry. The bad news: only 104 have a completed profile, which means that they answered all of the questions and also attached their child's genetic report. We have a scientific meeting this summer so are asking families to complete their profiles by Aug 1. We will present the findings from the meeting to you all this summer, but we can't make any conclusions with only 104 completed files. At a recent meeting about autism and epilepsy at the National Institutes of Health, the importance of having a registry was a recurring theme. A registry will ensure that research propels forward and we find the answers we all so desperately want. Every family matters, and together we can make a difference.

Please visit www.dup15qregistry.org today. Thank you!

Aeriel's Story

BY BRETIGNE CALVERT

Aeriel was tiny when she was born: 5 lbs., 6 oz. Her mouth was so small that she had trouble nursing, so I pumped milk and fed her with a syringe. But we didn't suspect there was anything "wrong" with her until her third day, when I was starting to feed her with a bottle. Her face turned grey and she went limp. I looked at her neck and saw that she was breathing. Very quickly, she was moved to the well-baby nursery for observation and then to the NICU, where she would spend the next few weeks. She was four months old when she stopped having her "episodes," a pleasant term for the terrifying moment when your baby suddenly stops breathing in the middle of eating, turns blue or grey, and has to be startled back to life by a harsh pat on the back, a flick on the sole of her foot, or a burst of oxygen.

We were so relieved when these episodes stopped that we didn't worry too much when her pediatrician said at six months that she was a little behind in development. She was evaluated and started the recommended therapies, but we sort of assumed she would catch up, that whatever caused her to have the episodes had held her back a little but that she would outgrow this delay as she had her breathing and swallowing problem.

Then, just a month after she turned 1, I noticed her sitting slumped in her crib. I picked her up and she remained in a slumped position. She wouldn't make eye contact and just kind of lay there. It was frightening at the time, but I now look at it as one of the most fortunate episodes of her life so far. Had I not seen what turned out to be a seizure, and had we not taken her in for an EEG and learned that she was in fact very susceptible to a wide range of seizures, we might never have recognized the frequent head drop seizures she had been having for some time, and we might never have sought treatment for them.

About a month later, we learned that Aeriel had dup15q syndrome. We read everything we could find about it and met with geneticists and neurologists, but we still felt that we didn't know what to expect for her. All we knew was that we needed to get her as much therapy as we could, do whatever we could to stop her seizures, and learn as much as we could to find solutions.

We decided to move from Palo Alto down to Burbank, Calif., where my parents and sister live, so we could have additional family support and so Aeriel and her older brother could spend more time with their grandparents and cousin. We would also be closer to a hospital in Pasadena that had been highly recommended for administering the ketogenic diet, an extremely low-carbohydrate diet that has been found to be very effective in stopping the kind of seizures our daughter was having. We were fortunate that my husband Guy's employer, Google, was able to accommodate our move, allowing Guy to transfer to the Santa Monica offices and to work primarily from home.

We made the move and were staying in a temporary apartment when we had another stroke of good luck: Just a week before



Aeriel was scheduled to begin the ketogenic diet, she caught the flu and didn't eat well for several days. She lost over a pound and a half and—although she did not go into ketosis—her seizures stopped. It has been almost a year now, and we have not seen another seizure. Even better, her EEGs, while not yet "normal" are much "calmer" than the mass of spikes we had seen before and show no indication of seizure activity. Because there were so many variables—the flu and weight loss; switching to a pill form from liquid antiseizure medication (which had not been working and her doctors were certain would not work in the future if it hadn't yet); the craniosacral therapy she had recently begun, which had a visible and dramatic impact on her energy and awareness with each treatment—we will probably never know what caused her seizures to stop. We are just very, very grateful that they have.

Before we had a precise diagnosis, we met with a neurologist who told us that none of Aeriel's problems were our fault. We hadn't done anything wrong; we were just "very unlucky." She was not being insensitive; in fact, this was the most genuinely helpful meeting with a doctor that we had had yet. She was trying to assuage any guilt that we might feel about our daughter's condition. But her words just didn't ring true to me at all, and they still don't.

I know what "unlucky" is. Before Aeriel or her brother were born, we lost our first baby to a cord accident at 39 weeks. We know what it is to be terribly, terribly unlucky, to feel great grief and guilt over a real tragedy. I just can't feel anything remotely like that about my little girl, and I don't feel "unlucky" that she is the way she is. Yes, it is challenging, and, yes, there are concerns about her future that keep me up at night worrying. But I just can't feel regret about who she is. If anything, I feel just the opposite: that we have been blessed to be in the presence of this pure, joyful spirit whose sweet smile lights up our lives. It is almost as if she is a pure spirit who lives in a different realm from ours and needs our protection from the hard practicalities of the material world. I feel that we have been honored with the sacred task of caring for this beautiful, ethereal being so she can be here on Earth with us.

THE CALVERT FAMILY—BRETIGNE, GUY, THEIR SON, GRIFFON, 5, AND AERIEL, 2 (DUP15Q)—LIVE IN BURBANK, CALIF.

Reflections *from a Grandmother*

Reflections

BY ELAINE LOWELL

The Gift of Grace



Grace is our youngest grandchild, and she is absolutely delightful. She has a great personality. She often has us laughing at some new antic and amazes us with something new she has learned or tried. For instance, last summer, after years of all of us trying many different ways to get her to eat a more varied diet, she started eating all sorts of new foods on her own for no apparent reason. She also decided one day that swinging, something she had never really liked before, was fun. Last fall, when she saw pumpkins sitting on someone's porch she said to her mom, "Trick or Treat! Get candy!" Oh, if we could only get inside that little head of hers for even one day.

Every one of these precious children with dup15q is extraordinary. There

are wonderful stories to tell about each of them and the strides they make every single day. This brings me to the Dup15q Alliance parents who are reading this, the moms and dads who are equally as remarkable and, dare I say, courageous as their children (although I doubt they consider themselves as such):

I so appreciate your posts on BigTent. You are unflinching as you search for new ideas and better ways to deal with a particular issue, whether it's potty training, seizures, sleep issues, feeding problems, or iPad apps. You provide exceptional help and support to each other as you respond to questions and offer tips from your experiences and perspective. You are fearless and gutsy when it comes to advocating for

your kiddos with teachers, therapists, doctors, and other professionals. You often go through the day sleep-deprived, but you rarely complain. You seem to be able to find some joy in each day.

You are doing an outstanding job. Being a parent is not easy on the best of days, and undoubtedly parenting a child with special needs greatly increases the challenges. I am continually amazed at your attitude, your ingenuity, your perseverance, and your determination. You are excellent advocates for your children. You are great at juggling all of the demands of your family and keeping them in balance. You are a tremendous support for each other.

ELAINE, THE GRANDMOTHER OF GRACE LOWELL, 8 (DUP15Q), LIVES IN MAPLE VALLEY, WASH.



*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.*

UPCOMING EVENTS

NEW ENGLAND REGIONAL GATHERING BBQ & SWIM Aug. 4, 1 p.m. until . . .

181 Cardigan Road, Tewksbury, Massachusetts
Bring chairs, bathing suits, and towels.
Contact: Joanne Poore, jmpoore@comcast.net or (978) 851-5182.
Please RSVP by July 30.

CAMP PRIMETIME Aug. 10-12

Cascade Mountains of eastern Washington
Contact: Greg Keller, gregkeller1887@msn.com

Contact Karen Sales at ksales76@gmail.com or your regional representative for information about the regional gathering policy. Please see www.dup15q.org for a continually updated list of events and a list of regional representatives, who can provide more information.

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.

ILLINOIS DUP15Q GOLF OUTING Aug. 17

Bridges of Poplar Creek Country Club,
Hoffman Estates, Illinois

2ND ANNUAL KAYJAY 5K RUN/1K WALK Sept. 22

Wheeler, Wisconsin

3RD ANNUAL GQ FUNDRAISER Oct. 6, 6-10 p.m.

Morgan's Restaurant, 18700 Old Lagrange Road,
Mokena, Illinois

MIDWEST REGIONAL GATHERING Sept. 4-12

Lincoln, Nebraska

<http://www.dup15q.org>

Address Service Requested

Dup15q Alliance
PO Box 674
Fayetteville, NY 13066
855-dup-15qa