


# DUP15q ALLIANCE



*Providing family support and promoting awareness, research and targeted treatments for Chromosome 15q Duplication Syndrome.*

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## Message from the Board Chair

Tom Doyle, Interim Board Chair

Our organization was initially formed in 1994 by Donna Bennett and Brenda Finucane. They brought together thirteen families, each raising a child with Dup15q, in order to provide support and encouragement. This simple beginning led to the incorporation of the organization in 2004 as a 501(c)(3) non-profit known as IDEAS (IsoDicentric 15 Exchange, Advocacy, and Support). In 2011, the name of the organization was changed to the Dup15q Alliance to better describe who we are and what we are about.

Dup15q Alliance exists to fulfill its mission of providing family support and promoting awareness, research, and targeted treatments for Chromosome 15q Duplication Syndrome.

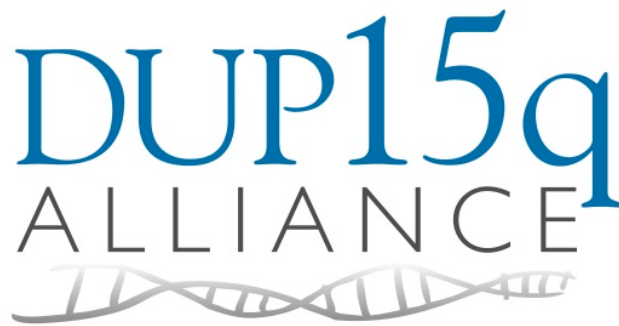
We grew from the 13 original families to nearly 800 known families in 2012. We know that this is a result of more accurate diagnoses and education of the medical community. We also know that there are many more families who haven't received the appropriate diagnosis yet. We are committed to working with our scientific advisors to spread the word about Dup15q and to help families understand the diagnosis better and find support from other families and our organization.

2012 was a year where we supported the creation of a mouse model to enhance the study of Dup15q and encourage researchers to study the resulting information. We held a successful scientific conference in Boston in July – you can find many of the presentations and research available on our website. We are well on the way to hosting our next international conference in July in Minnesota and hope to break a record again for the number of people attending. Our fundraising exceeded all of our goals for the year as a result of the hard work, dedication, and love of so many families

that participated simply through a donation or through their efforts in hosting a family fundraiser. We have begun to develop strong international relationships with our sister organizations in other countries including Spain and Italy and to contact individually members in other countries. We look forward to working closely with the new Canadian organization as it kicks off this year as well.

We have much to be grateful for every day. Thank you to everyone who participates in our mission in so many ways. Thanks to those who have donated their time, talent, and treasure. Thanks for your gifts to Dup15q Alliance to support further research, outreach, education, and conferences. Thanks to those of you who spent a great deal of time and energy hosting a family fundraiser. Thanks for those of you who provide encouragement and support for other families, too, whether it be in person or through BigTent or our new Facebook page. Thanks to our scientific

*Message continued on page 7*



## Board of Directors

In February 2012, Rachel Doucette advised the board of her resignation as Director and VP Communications. In March, Patti Rubel advised the board that she would not seek reelection in June 2012, the end of her two-year term. The Alliance then solicited applications from interested members for the open board seats and was very appreciative of all those who submitted their names!

At the board meeting held telephonically on May 20th, the board elected Dana Tilton from Georgia and Lori Eisenhaur from British Columbia, Canada (the Alliance's first international director) as directors to fill the two open board seats. Dana's and Lori's two-year terms began in June 2012.

On June 24th, the board held a telephonic meeting during which the following Group 1 board members were reelected for two-year terms: Len Poore, Board Chair; Tom Doyle, VP Finance; Rylie McHam; and Mike Porath.

In October, Len Poore advised the board of his resignation as Director and Board Chair and Tom Doyle agreed to step into the role of Interim Board Chair, in addition to his VP Finance role. Thank you, Tom! Also in October, Lori Eisenhaur resigned from the board.

To all the board members who served in 2012, we express our sincere thanks and appreciation for your time, dedication, hard work and sound guidance. The

Alliance would not be where we are today without your contributions!

In November 2012, Dup15q Alliance also added two members to the Professional Advisory Board. We welcomed Stormy Chamberlain with the University of Connecticut Health Center in Farmington, CT and Orrin Devinsky with NYU Comprehensive Epilepsy Center at NYU Langone Medical Center in New York City. We look forward to all they will do to help us better understand Dup15q.

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### Officers:

Interim Board Chair - Tom Doyle (WA)  
 Executive Director - Kadi Luchsinger (NY)  
 VP Finance - Tom Doyle (WA)  
 Corporate Secretary - Karen Sales (IA)

### Board Members:

Donna Bennett (PA) - Co-Founder  
 Guy Calvert (CA)  
 Paul Karch (WI)  
 Rylie McHam (OH)  
 Linda Meagher (TX)  
 Mike Porath (CA)  
 Dana Tilton (GA)

### Scientific Advisors:

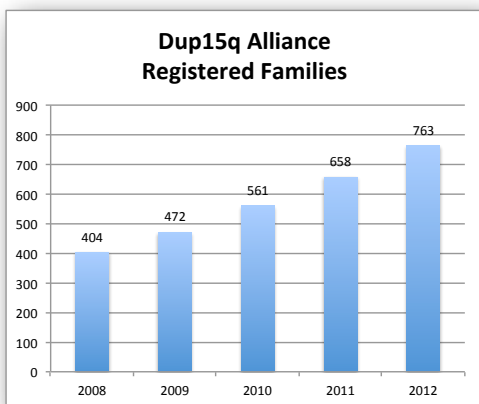
Agatino Battaglia, MD, DPed, DNeuro  
 Calabrone, Pisa, Italy  
 Stormy Chamberlain, Ph.D.  
 University of Connecticut  
 Edwin H. Cook, Jr., MD  
 University of Illinois at Chicago  
 Orrin Devinsky  
 NYU Langone Medical Center  
 Brenda Finucane, MS, CGC  
 Elwyn Training and Research Institute  
 Janine LaSalle, PhD  
 UC Davis School of Medicine

Lawrence T. Reiter, PhD  
 University of Tennessee Health Science  
 Center  
 N. Carolyn Schanen, MD, PhD  
 Nemours Biomedical Research  
 Sarah Spence, MD, PhD  
 Children's Hospital Boston  
 Ron Thibert, DO, MsPH  
 Massachusetts General Hospital

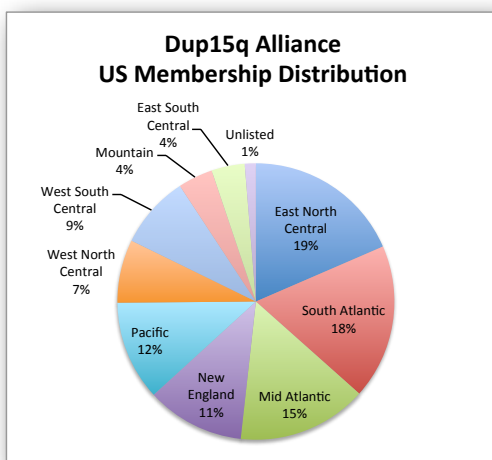
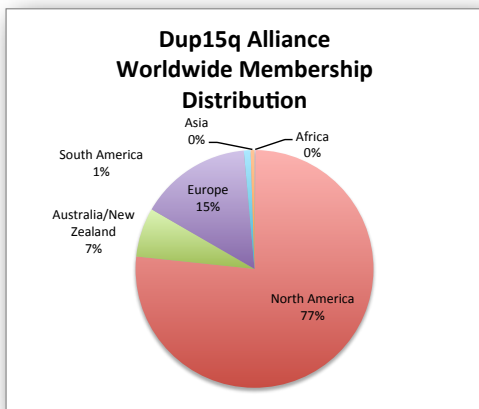
# Dup15q Alliance Membership

Dup15q Alliance serves members around the world. While the organization is incorporated in the United States, international families affected by Chromosome 15q Duplication Syndrome also rely on the support and advocacy provided by the organization. The Alliance is able to provide a private online forum for members to communicate regardless of their location. The organization also facilitates family matches allowing families to connect with other members in their general geographic area.

As of December 31, 2012, Dup15q Alliance had 763 registered families, continuing its steep growth trend with a 16% increase in members during 2012.



The chart below illustrates the Dup15q Alliance immediate family membership by international distribution. Ninety-eight percent of the membership served by Dup15q Alliance is in North America, Europe and Australia.



More than seventy percent of the membership served by Dup15q Alliance is located within the United States. The chart below illustrates the Alliance's membership distribution by US Geographical Division. The US divisions are defined as:

**East North Central:** Wisconsin, Michigan, Illinois, Indiana and Ohio

**South Atlantic:** Delaware, Maryland, District of Columbia, Virginia, West Virginia, North Carolina, South Carolina, Georgia and Florida

**MidAtlantic:** New York, Pennsylvania and New Jersey

**New England:** Maine, New Hampshire, Vermont, Massachusetts, Rhode Island and Connecticut

**Pacific:** Alaska, Washington, Oregon, California and Hawaii

**West North Central:** Missouri, North Dakota, South Dakota, Nebraska, Kansas, Minnesota and Iowa

**Mountain:** Idaho, Montana, Wyoming, Nevada, Utah, Colorado, Arizona and New Mexico

**East South Central:** Kentucky, Tennessee, Mississippi and Alabama

**West South Central:** Texas, Oklahoma, Louisiana and Arkansas

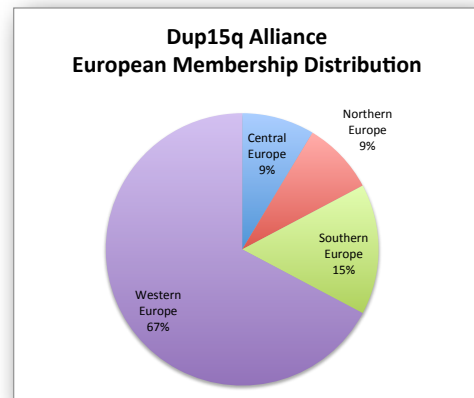
Outside of the United States, more than half of the remaining Alliance membership is located in Europe, primarily in Western Europe. The chart below illustrates the Dup15q Alliance's membership distribution by European Geographical Division. The European divisions include:

**Central Europe:** Germany, Poland and Austria

**Northern Europe:** Denmark, Norway and Sweden

**Southern Europe:** Spain, Portugal, Croatia, Israel and Italy

**Western Europe:** Great Britain, Ireland, France, Belgium and Netherlands



European organizations supporting Chromosome 15q Duplication Syndrome are also a significant support to affected families. Dup15q Alliance collaborates with these international organizations to better serve all members of this community.

# Dup15q Alliance Events

*Dup15q Alliance events bring together families, raise funds and increase awareness of Chromosome 15q Duplication Syndrome.*

## Regional Gatherings

2012 was another great year for regional gatherings! There were six get-togethers held throughout the country. Regional gatherings are a wonderful way to spend time with other families affected by Dup15q. They allow families to share their unique experiences, all the joys and challenges that go along with raising a child with Dup15q. Regional gatherings are opportunities to spend time with old friends, meet new friends, share stories and exchange information. The feedback is always extremely positive, and everyone seems to truly value the time that is spent together.

In August, Joanne and Len Poore hosted a gathering for Region 1 (ME, VT, NH, MA, CT, RI) at their home in Massachusetts. Fifteen families enjoyed a fun and relaxing day. Region 4 (OH, IN, MI) held a gathering at a recreation center in Columbus, Ohio in June. Families chatted over Mexican

food and spent the afternoon swimming, which the children really enjoyed. The Horvath family hosted a lunch in October for six families from Region 5 (IL). Respite workers entertained the children, so the adults could talk. Greg Keller coordinated a weekend gathering for Region 7 (AK, WA, OR, ID, MT, WY) at Camp Prime Time at Clear Lake, Washington. Nine families attended the relaxed weekend where they shared stories and enjoyed good company. Sarah and Mike Porath hosted a barbeque for five families in October. Three of the families were new to the Alliance and were able to share their stories and meet new friends. In March, Dana and Luke Tilton hosted a gathering for Region 10 (AL, GA, TN, NC, SC) at their home in Atlanta. Seven families attended the gathering and enjoyed a relaxing day where the adults shared stories and the kids delighted in jumping, swinging and playing in the yard.

## Sophie's Challenge

Dup15q Alliance member Katey Miller took a different approach to fundraising. Rather than planning a physical event, she launched Sophie's Challenge, an online fundraising effort. Her goal was to raise \$1,000 for Dup15q Alliance, but she ended up with over \$3,200!

Katey used Crowdrise, a fundraising website, and sent a message to her friends and family. In her email she wrote, "I work hard to give my Sophie

a better life every day, but there is a bigger picture that she is part of. During the holiday season many people look for causes to donate their hard-earned money toward. I thought this year, and maybe every year, I could take a little extra time and try to bring awareness and a little help to Sophie's cause."

## Plugged-In Concert

In January 2012 the first rock band concert to benefit Dup15q Alliance was held in Boston, Massachusetts, raising \$10,000.



Nelson Barnett, inspired by his brother Roland, dup15q and member of a non-profit teen rock band program called Plugged-In, convinced his fellow band members that their next benefit concert should be devoted to Dup15q Alliance. Holding a poster of him pushing Roland, he talked about the rare occurrence of dup15q and explained the severity of Roland's profile, being non-verbal, frustrated, anxious, behaviorally challenged and constantly interrupted by daily seizures.

The concert was incredible! About 350 people attended the two-night concert. The three song-writing bands performed live versions of their tunes. Even more incredible was five dup15q families from Massachusetts who met for the first time.

Even though Roland wasn't well enough to attend the concert, Nelson's band helped to celebrate Roland's ninth birthday that week by singing "Happy Birthday" to him as only rock stars can. It was truly an inspiration to see everyone rocking out for our kids and Dup15q Alliance!

# More Dup15q Alliance Events

## McHam Family Cocktail Party

Rylie McHam, along with her family and a very good friend, hosted a Dup15q Alliance fundraiser at her mother's house in October. It was a collective effort and the outpouring of generosity was amazing. The event was a cocktail party that brought people from across the community



together to support Dup15q Alliance. Everyone was genuinely interested in learning more about Chromosome 15q Duplication Syndrome and Dup15q Alliance. Rylie received a lot of really positive feedback about the event. She says, "It was truly a wonderful, heartfelt evening!"

The idea of hosting a fundraiser can be a bit overwhelming, but as Rylie learned it turned out to be really fun and everyone enjoyed being able to contribute to Dup15q Alliance. Local restaurants, distributors and grocery stores donated all of the food, drinks and party supplies. People were more than generous with their donations and there was a seemingly endless amount of delicious food and desserts. Invitations that were sent out for the event asked for a minimum of a \$75 donation and the event raised just over \$10,000!

## Tilton Holiday Fundraiser

Luke and Dana Tilton held their first fundraiser on December 14, 2012. The fundraiser was a holiday party to benefit Dup15q Alliance in honor of the season of giving. The party was hosted by Dana's parents, Gerald and Vonnie Wilson, and was held at their home in Atlanta. It turned out to be a night to remember; over \$40,000 was raised for Dup15q Alliance.

Dana's goal for the evening was to spread awareness, raise money, and touch people's hearts by giving them a closer look inside life with Dup15q. As people walked in, they were greeted by framed photos of some of the beautiful children of Chromosome 15q Duplication Syndrome. These were also the last faces guests saw as they left. During the party, several people were intrigued by these children and wanted to know more about the different stories behind their sweet, almond shaped eyes and contagious smiles.

After enjoying a delicious variety of food that was donated by some of Atlanta's finest restaurants, the guests sipped on wine and gathered around to view a video that was created for the fundraiser. The video was made to explain Chromosome 15q Duplication Syndrome and all that it entails, but in a

simple way that anyone could follow. The video touched people's hearts and filled many eyes with tears.

After the video, several people asked more questions about Dup15q and the answers were discussed amongst the whole group. Dana was asked to describe a day in the life of Addison, to which she began by saying, "Every day with Addison starts with a huge smile on her face as she jumps or runs into our arms. She's just so happy to see us and hug us." Many tearful eyes joined Dana's as she told her story.

The Tiltons raised the money through the use of a First Giving page, an RSVP card that included the option of donating while declining to attend, and an evening of awareness that portrayed a closer look inside life with Dup15q. Luke and Dana were overjoyed by the support shown. They plan to grow the fundraiser and make it an annual event.

In closing, Luke and Dana said, "This fundraiser was like a breath of fresh air. Making a difference while speaking so freely, with all guards down, about life with our precious Addison and Dup15q, is simply priceless and life changing."





## Research Update

The past year was a momentous one for the Alliance's research efforts. In February, the Board voted to spend \$40,000 to help fund the development of a new mouse model of Dup15q Syndrome by Dr. Larry Reiter and Dr. Scott Dindot. Although the Alliance has long supported research indirectly—for example, by sponsoring annual scientific conferences on Dup15q—this direct investment for a specific research project marked a step into new territory. Dup15q Alliance believes 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 the funding would have a major impact on the completion of this project. According to Dr. 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 in 2013, submitting a research paper on mice characteristics and distributing the animals to other scientists to spur research into the underlying mechanisms of Dup15q and potential treatments.

On August 9-10, Dup15q Alliance held its annual scientific meeting in Boston.

Researchers and clinicians gathered from around the country for the two-day meeting in an auditorium at Children's Hospital Boston (CHB). While the Alliance has organized scientific meetings in the past, they were closed, one-day



affairs of perhaps a dozen professionals - normally, our Professional Advisory Board (PAB) members plus a handful of invited researchers. This Boston meeting was the first of its size and scope, attracting more than 70 participants, including members of the press, to the CHB auditorium to hear 20 scheduled speaker presentations and four moderated panel discussions. Slides and videos from the meeting are available on our website, [www.dup15q.org](http://www.dup15q.org).

Lastly, we are thrilled that Stormy Chamberlain and Orrin Devinsky agreed to join our PAB. Stormy is the Raymond and Beverly Sackler assistant professor of Genetics and Developmental Biology at the University of Connecticut Health Center and a member of the University of Connecticut Stem Cell Institute. Her lab studies chromosome 15q11-q13

imprinting disorders, and has been focusing on induced pluripotent stem cell models of Dup15q and Angelman Syndrome. Stormy received a B.A. degree in Molecular Biology from Princeton University and a Ph.D. in Medical Sciences-Genetics from the University of Florida, where she studied mouse models of Prader-Willi Syndrome.

Orrin is Professor of Neurology, Neurosurgery, and Psychiatry at NYU Langone School of Medicine. He directs the NYU Epilepsy Center and Saint Barnabas Institute of Neurology. His epilepsy research includes phenomic-genomic relations, sudden unexpected death, surgical therapies, tuberous sclerosis, new medicines and devices, collaborative translational research, quality-of-life, cognition and behavior, and neuro-inflammation. His behavioral neurology research includes hyperfamiliarity, Klein-Levin Syndrome, delusions, autonomic nervous function, and anterior cingulate gyrus functions. Orrin has authored more than 300 peer-reviewed papers and written or edited more than 20 books. He founded FACES (Finding A Cure for Epilepsy and Seizures) and co-founded [epilepsy.com](http://epilepsy.com) and the Epilepsy Therapy Project. He serves on the boards of these organizations and has also served on the board of the American Epilepsy Society and Epilepsy Foundation. Other interests include the history of neuropsychiatry, evolution, anthropology, and animal intelligence.

*Message continued from page 1*

advisors who work so diligently with us to study Dup15q and help us to understand it better. Thanks to those doctors in Boston and Miami where we already have Dup15q clinics and to those who are trying to set clinics up in other cities around the world. Thanks to those who I don't mention directly who are quietly providing loving support in so many other ways.

Thanks to your Dup15q Alliance Board. These people put in countless hours voluntarily to keep this organization true

to its mission. I spent years in the athletic realm in our high schools and I would equate being on a Board to being a referee in a basketball game. You have to make tough decisions instantaneously. Half the crowd thinks your right; the other half thinks you're crazy! Your board is made up of human beings who do their best to make decisions that further our Mission. Are we perfect? Obviously not, but they can't be faulted for not trying or caring. Thanks to all of our board members who give so much of themselves to further the aims of our organization. Thanks especially to Kadi Luchsinger for all she

does to promote Dup15q Alliance throughout the world and with other similar organizations. Thanks to Karen Sales for her expert leadership on the 2013 Minnesota Conference. Sorry to not name everyone for lack of space, but each has an important role and fulfills that with grace and class.

Thanks for your support throughout 2012. We look forward to making even more significant strides in 2013. See you at the Conference.

**I N C O M E                      2 0 1 2  
S T A T E M E N T**

**Income**

Conference	\$ 5,693.02
Fundraising	218735
Interest	406.27
Memorials	870.00
Store	2533.15
Total	\$ 228,237.44

**Expenses**

Independent Contractors	\$ 20,600.00
Administration	5,827.50
Bank Charge	214.00
Conference	3,668.00
Education and Outreach	505.96
Fundraising	17,407.74
General Administration	4508.56
Legal/Professional Fees	686.14
<i>The Mirror</i>	3302.88
PR and Marketing	6409.11
Registry	37,175.00
Research Grants	43,000.00
Research Roundtable	7,829.13
Store	1,481.42
Website	514.99
Total	\$ 153,130.43

**Net Income** \$ 75,107.01

**B A L A N C E                      S H E E T  
A S O F D E C E M B E R 3 1 , 2 0 1 2**

**Assets**

Checking/Savings Accounts	\$ 105,217.24
Money Market Account	246,824.19
True Family Restricted	10,806.07
Total Assets	\$ 362,847.50

**Liabilities and Equity**

Opening Balance Equity	\$ 287,740.49
Net Income/Loss	75,107.01
Total Liabilities and Equity	\$ 362,847.50

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B U D G E T

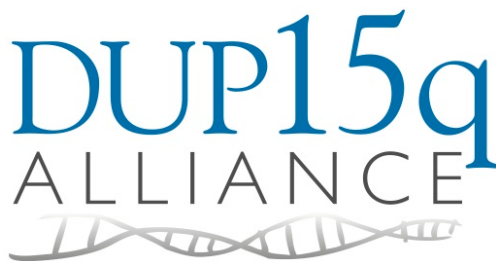
**Income**

Conference	\$	90,000.00
Fundraising		245,000.00
Grants/Memorials		10,000.00
Interest		400.00
Memorials		500.00
Store		5,000.00
<b>Total</b>	<b>\$</b>	<b>350,900.00</b>

**Expenses**

Independent Contractors	\$	80,000.00
Administration		4,000
Conference		90,000.00
Education and Outreach		2,000.00
Fundraising		20,000.00
General Administration		10,000.00
Legal/Professional Fees		1,000.00
<i>The Mirror</i>		5,000.00
PR and Marketing		10,000.00
Registry		45,000.00
Research Grants		3,000.00
Research Studies		12,000.00
Research Roundtable		10,000.00
Store		2,500.00
Supplies		1,000.00
Website		10,000.00
<b>Total</b>	<b>\$</b>	<b>305,500.00</b>

**Net Income** \$ 45,400.00



## Communication

Dup15q Alliance uses various methods of communication to reach both internal and external interests and members.

Member focused information is delivered via the quarterly newsletter, *The Mirror*. This publication is available on the Dup15q Alliance website, is emailed to members and is available by mail, if requested. Dup15q Alliance also provides an online community to members on a social network platform, BigTent. This is a community allowing Alliance members to discuss issues, plan events, share stories and provide support to others.

Dup15q Alliance provides a website for both internal and external partners. The website can be found at [www.dup15q.org](http://www.dup15q.org). Other online social networks include Twitter and both a parent support group and public page on Facebook. Dup15q Alliance is able to share organizational information and interests with individuals and other organizations using these social networks. The use of these social media has increased awareness of the Alliance’s mission as well as significantly assisting in fundraising efforts.

## Contact Information

Dup15q Alliance  
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 855-dup-15qa (855-387-1572)  
[info@dup15q.org](mailto:info@dup15q.org)

## Social Media

**Website:** [www.dup15q.org](http://www.dup15q.org)  
**Facebook:** [www.facebook.com/pages/Dup15q-Alliance/245152512165738](https://www.facebook.com/pages/Dup15q-Alliance/245152512165738)  
**Twitter:** [www.twitter.com/dup15q](https://www.twitter.com/dup15q)  
**BigTent:** [www.bigtent.com/groups/dup15q](http://www.bigtent.com/groups/dup15q)