



*Providing family support and promoting awareness, research and targeted treatments for chromosome 15q11.2-13.1 duplication syndrome.*

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## Message from the Board Chair

Tom Doyle

The Alliance celebrated 20 years of growth and service in 2014 by continuing to fulfill our mission of providing family support and promoting awareness, research and targeted treatments for dup15q syndrome.

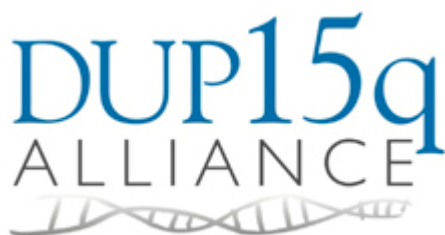
The depth and breadth of Alliance research activities grew in our 20<sup>th</sup> anniversary year as we held a joint scientific meeting with Angelman Syndrome Foundation which enabled researchers and clinicians to share discoveries and approaches to research and treatment of these related conditions (Angelman syndrome is caused by the deletion of the same critical area on chromosome 15 that is duplicated in dup15q syndrome). Research into stem cells and fruit fly and mice models helped

us learn more about the cellular and molecular impacts of dup15q syndrome, while our International Medical Registry and construction of our new dup15q clinics' database will help better define the clinical characteristics of dup15q syndrome. Our network of dup15q clinics expanded, helping many families gain a better understanding of and treatment for a condition which only a small portion of health providers will see in their practices.

Our membership continued to grow, as did support for our successful fundraising initiatives, including those which focused on our 20<sup>th</sup> anniversary celebration and continued development of the dup15q syndrome mouse model. Dup15q Alliance families continued to provide support for each other by sharing stories, challenges and joys through our thoughtful and well-written quarterly newsletter, *The Mirror*; our beautifully revitalized, award-winning website; our active Facebook page and Facebook group; and in-

person meetings at many regional gatherings. In addition to providing oversight to the various activities of the Alliance, the Board this year added Jane Kim and Adrienne Campolmi as directors and developed a strategic plan to guide us in our growth.

You will find further details of this activity in this Annual Report. I was honored to serve as chair of the Alliance Board this year and enjoyed working with a dedicated and enthusiastic group of parents, grandparents, scientists, clinicians, and caregivers to provide the 20<sup>th</sup> year of connections, support and services. I would like to thank our Professional Advisory Board, dup15q syndrome researchers, the Board of Directors, our dedicated staff, other family volunteers and our generous fundraisers and donors for this exciting progress. We look forward to continued growth and service in 2015.



## Board of Directors

In 2014, the board filled two board seats. Dana Tilton's two-year term ended in 2014, and Dup15q Alliance is very grateful to her for her years of service.

In March, the Board elected Jane Kim, from California, to the Board of Directors. She is an adult neurologist and the mother of a two year-old son with dup15q syndrome and a four year-old daughter. Jane is interested in working with the dup15q clinics.

In August, Adrienne Campolmi also joined the Board. Adrienne is the mother of a twelve year-old son with dup15q syndrome and a project manager and mentor at IBM. She is interested in working on fundraising for the Alliance.

To all the board members who served in 2014, 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!

### Officers:

Board Chair – Paul Karch (WI)  
 Executive Director – Kadi Luchsinger (NY)  
 VP of Finance – Tom Doyle (WA)  
 Corporate Secretary – Karen Sales (IA)

### Board Members:

Donna Bennett (PA) – Co-Founder  
 Guy Calvert (CA)  
 Adrienne Campolmi (NC)  
 Fernando Gomez (NJ)  
 Jane Kim (CA)  
 Rylie McHam (OH)  
 Linda Meagher (TX)  
 Mike Porath (CA)

### Professional Advisory Board:

Agatino Battaglia, MD, DPed,  
 DNeurol  
 Calambrone, Pisa, Italy

Stormy Chamberlain, PhD  
 University of Connecticut

Edwin H. Cook, Jr., MD  
 University of Illinois at Chicago

Orrin Devinsky  
 NYU Langone Medical Center

Scott Dindot, PhD  
 Texas A&M University

Brenda Finucane, MS, CGC  
 Elwyn Training and Research  
 Institute

Shafali Spurling Jeste, MD  
 UCLA Center for Autism Research  
 and Treatment

Janine LaSalle, PhD  
 UC Davis School of Medicine

Lawrence T. Reiter, PhD  
 University of Tennessee Health  
 Science Center

N Carolyn Schanen, MD, PhD

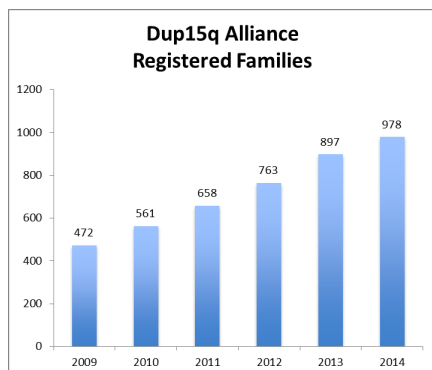
Sarah Spence, MD, PhD  
 Children's Hospital Boston

Ron Thibert, DO, MSPH  
 Massachusetts General Hospital

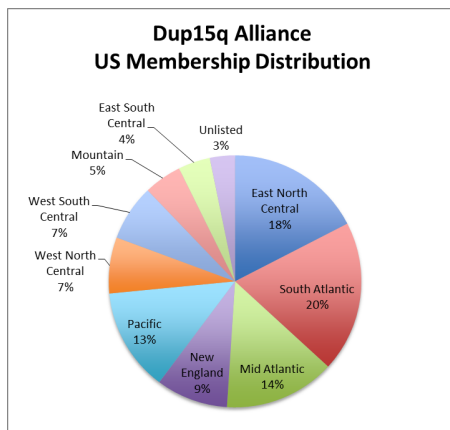
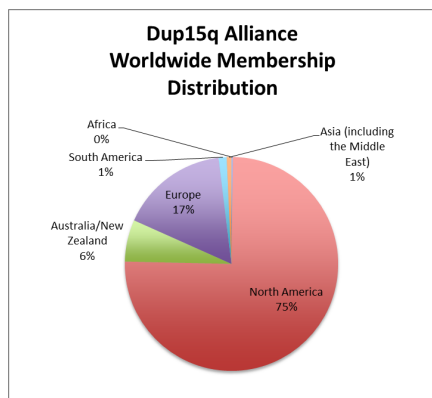
## Dup15q Alliance Membership

Dup15q Alliance serves members around the world. The Alliance is incorporated in the United States, but international families affected by dup15q 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, 2014, Dup15q Alliance had 978 registered families, continuing its growth with a 9% increase in members during 2014. By the end of 2014, Dup15q Alliance had expanded its reach so much that membership has more than doubled since 2009.



The chart below illustrates Dup15q Alliance's affected 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 above 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, 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, 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

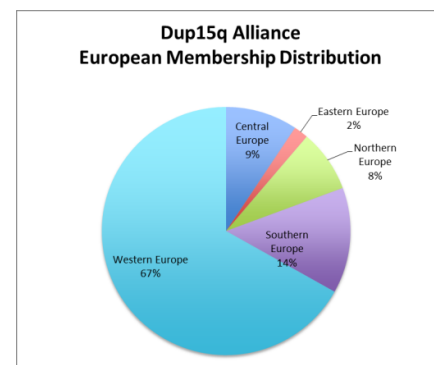
Outside of the United States, more than fifty-five percent of the remaining Alliance membership is located in Europe, primarily in Western Europe. The following chart illustrates Dup15q Alliance's membership distribution by European Geographical Division. The European Divisions include:

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

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

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

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



European organizations supporting dup15q syndrome are also a significant source of support for affected families. Dup15q Alliance collaborates with these international organizations to better serve all members of this community.

# Happy 20th Anniversary to Dup15q Alliance!

2014 was a very special year for Dup15q Alliance. We celebrated 20 years of advocacy, family support and research for dup15q syndrome. A lot happened in the first 20 years of Dup15q Alliance. What began in 1994 as a group of two dozen families grew to almost 1,000 families by the end of the year and thousands more supporters!



Highlights from the year included:

- In January, we highlighted the International Medical Registry, which provides a never-before-seen collective look into the lives of those with dup15q syndrome.
- In February, we celebrated Rare Disease Day with \$15,000 in 15 days for Dup15q Alliance. We raised over \$57,000!
- Our Spring into Action Fun-Raiser began in March. Volunteers hosted a variety of parties in honor of Dup15q Alliance. The fun was limited only by participants' imaginations!
- April featured an extensive dup15q syndrome awareness campaign.
- In May, we celebrated spring by encouraging participation in a walk or run to benefit Dup15q Alliance during our May Day 5K event.
- In June, families held regional gatherings at local zoos or other family-friendly locations.
- The Mouse Trip was a big highlight in July!
- In August, Angelman Syndrome Foundation and Dup15q Alliance hosted a joint scientific meeting to bring together researchers and clinicians studying aspects of both syndromes.
- In September, we hosted our first webinar with Dr. Ron Thibert of the dup15q clinic in Boston. We

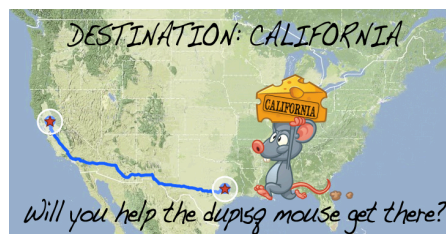
also filmed a video with several dup15q families, which we hope will be used for many years to raise awareness of dup15q syndrome.

- October celebrated the dup15q clinics! Nine clinics are now located in major medical centers around the United States.
- In November, we gave thanks for Dup15q Alliance. We asked members how has Dup15q Alliance helped your family? What would you like to see in the future?
- December brought the end of our 20th anniversary year. We looked back at all we've accomplished this year and looked ahead to many more years to come

## Mouse Trip

This year Scott Dindot and his team at Texas A&M University successfully created a group of mice with increased levels of UBE3A, a key gene in dup15q syndrome. The next step of the research was to take place at Jill Silverman's lab at the University of California at Davis, where her researchers will study the behaviors of the mice. Our hope is that continued study of this mouse model will help us to better understand the role of UBE3A in dup15q syndrome and eventually will lead to targeted treatments for dup15q syndrome.

To make raising the money for the travel expenses and preliminary research fun, Dup15q Alliance created a Mouse Trip Fundraiser. Dividing the cost of the project by the number of miles the mice would travel made the fundraising easier to track, and as hoped, more fun. Members sponsored miles—for each \$25 donated to Dup15q Alliance, the mice traveled one mile. We featured regular



updates of how far the mice had traveled. The journey was sped up when a generous dup15q family agreed to match donations during a nearly three-week timeframe. We are happy to report that we were able to fund the next step of this promising project. Thank you to all of you who helped with this journey!

## \$57,000 in 15 days for Dup15q Alliance

Each year from Valentine's Day, February 14<sup>th</sup>, to Rare Disease Day, February 28<sup>th</sup>, Dup15q Alliance sponsors a campaign to raise \$15,000 for support and research into dup15q syndrome.

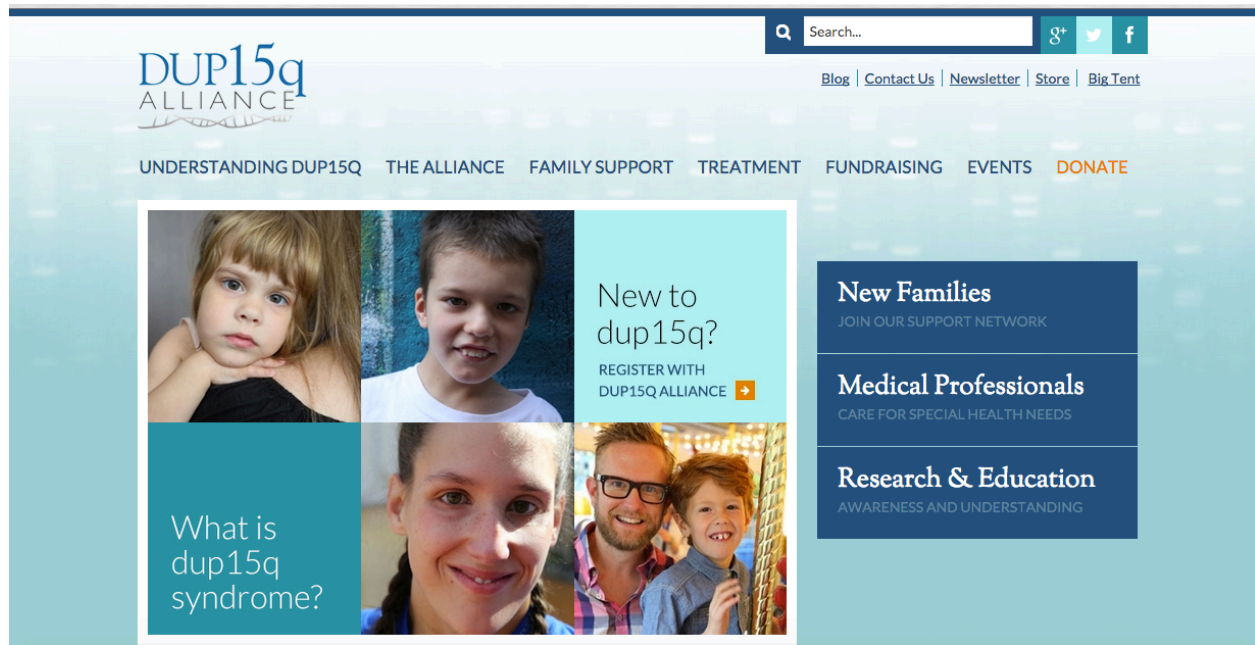
2014 was our best fundraiser to date!



Including matches made by two generous Dup15q Alliance families, we raised over \$57,000 in 15 days! We couldn't have done it without each of our supporters!

Money raised will go a long way to help us improve our network of dup15q clinics, support our annual scientific meetings and next year's family conference, and expand research into chromosome 15q duplications.





## Dup 15q Alliance Launches Redesigned Website

In 2014, we unveiled our new website, which was the result of several months of hard work. The new site with its user-friendly, modern design is hosted by GlobalCloud, and has many new features, as well as updated content from the old site.

The website is used by Dup15q Alliance families, researchers, clinicians, educators, therapists,

donors, and the general public. Each of these groups looks at the site in a different way and needs to be able to find a wide array of information. That presented us with quite a challenge. The new site contains over 200 pages to meet the needs of our multiple audiences.

Some of our favorite features include the beautiful pictures that showcase the many faces of dup15q syndrome;

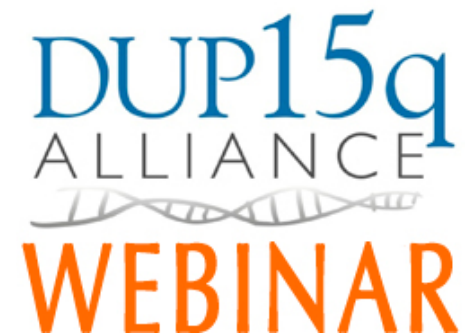
the ability to imbed videos; a search feature; past issues of The Mirror; and a reorganized calendar section.

As is often the case for Dup15q Alliance, this website thrives because of our membership. Thank you to all of you who have shared your stories, your love of Dup15q Alliance, and the photos of your special kids. Our families are what make Dup15q Alliance successful.

## Dup15q Alliance Begins Offering Webinars

In 2014, Dup15q Alliance began offering webinars, which are seminars that take place over the Internet, allowing participants in different locations to see and hear presenters, ask questions, etc. In September, we hosted our first webinar on seizures with Dr. Ron Thibert of the dup15q clinic in Boston. In 2014 we also hosted webinars on the genetics of this complicated region with Brenda

Finucane and a lesson on stem cell research with Stormy Chamberlain. Visit [www.dup15q.org/family-support/webinars](http://www.dup15q.org/family-support/webinars) for details, recordings and dates for future webinars.



## Science Meeting: Two Sides of a Coin

In August, Dup15q Alliance partnered for the first time with Angelman Syndrome Foundation to put on a joint scientific meeting. Dup15q syndrome and Angelman syndrome have much in common at the molecular level: dup15q syndrome is caused by too many copies of genes in the critical 15q11.2-q13.1 region of the maternal strand of chromosome 15, whereas Angelman syndrome results from the deletion of those genes. The hope is that the similarities and differences between the two syndromes can provide insights to help researchers better understand the mechanism of these disorders.

Presentations on new developments included information about UBE3A, a key gene in the critical region whose deletion causes Angelman

syndrome and is suspected of causing at least some aspects of dup15q syndrome. Further study of UBE3A has high potential for possible biochemical treatment options for dup15q syndrome.

Research presentations included information from several angles: animal models, post-mortem brain tissue, and human stem cell models. There were also presentations about clinical research, including Dr. Jeste's autism study; the potential use of cannabinoids in the treatment of intractable epilepsy; the increased risk of autism for individuals with edge duplications; and a presentation by Dr. Thibert that compared and contrasted the features of epilepsy in both syndromes and different responses to the same treatments.

## Dup15q Clinics Database



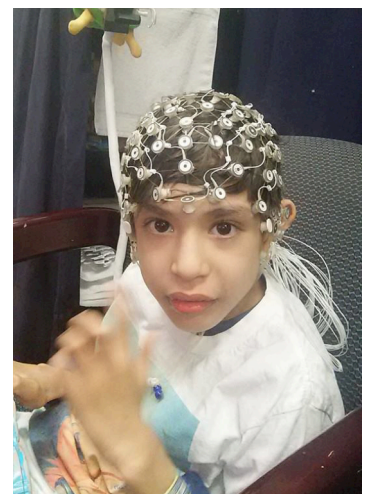
We are currently working to develop a database that will be used by the nine dup15q clinics to store and share information with each other. The goal is to have standardized care across the clinics and create a single database where information can be uploaded and aggregated. Each time a

child with dup15q syndrome visits a clinic, information will be collected about development, seizures, behavior/autism, sleep, GI issues, etc. Data management will be centered at the University of Tennessee Health Science Center. We estimated that it will take two years to get the database fully functional. The information collected from this project will help scientists better describe the effects of dup15q syndrome and determine which treatments are most effective. It is hoped that the results will eventually be published in a medical journal so we can continue to spread awareness of dup15q syndrome in the medical community; and that the database will continue to provide information for many years to come!

## Communication Study at UCLA

Children with 15q11-q13 duplications are at high risk for neurodevelopmental disabilities, particularly autism spectrum disorder (ASD) and intellectual disability (ID). Dr. Shafali Jeste and Dr. Connie Kasari began a study at UCLA's Center for Autism Research and Treatment to comprehensively and systematically characterize cognitive and social-communication function in children with dup15q syndrome, using standardized behavioral testing, play-based assessments, and EEG. The overarching goal is to determine if there are specific areas of strength or impairment that may serve as targets for behavioral intervention.

We also will be comparing the data from this study to data from age-matched children with "idiopathic" or non-genetic ASD. This pilot study is currently funded by Dup15q Alliance, with the infrastructure for behavioral and EEG testing funded by the National Institutes of Health.



# Financial Status

## Tom Doyle

2014 was a great year for fundraising for Dup15q Alliance. Thanks to the generous support of our family, friends, and businesses, we set a record by raising over \$325,000 for the year. This couldn't have happened without dedicated people working behind the scenes putting on family fundraisers, contributing to our \$15K in 15 days, and participating in so many more of our fundraising activities. It also couldn't happen without dedicated people working out of their homes for our organization, particularly Kadi and

Kim, who managed most of the Dup15q Alliance's major fundraising efforts.

We have learned through our 20 years that we are a family and that all of our members are critical to our success and our ability to fulfill our mission. We could not have developed the Registry, grown the number of dup15q clinics, built mouse models, or put on conferences without the goodwill of so many of our family members.

As a board member, I want to personally thank you for your time, talent, and treasure that you have so willingly shared with other Dup15q

Alliance families through the years. It is gratifying to see the love and concern that everyone models for our group. We are pleased that we have been able to grow our resources so that we will be able to respond to the needs of tomorrow. We know that conferences, family assistance, clinics, and research will be critical down the road. We are working very hard to manage and wisely steward our resources.

Please contact me at [tom.doyle@dup15q.org](mailto:tom.doyle@dup15q.org) if you have any questions or concerns about Dup15q Alliance's Financial Report.

### 2014 INCOME STATEMENT

#### Income

Conference	\$	3,500.00
Fundraising		314,526.60
Grants/Memorials		13,776.00
Store		<u>2,629.54</u>
Total	\$	334,742.50

#### Expenses

Independent Contractors	\$	39,850.00
Administration		18,435.00
Bank Charge		340.22
Conference		1,282.90
Education and Outreach		664.44
Fundraising		28,811.15
General Administration		12,215.17
Legal/Professional Fees		1,693.14
<i>The Mirror</i>		3,393.30
PR and Marketing		12,483.47
Registry		13,636.25
Priority Projects		93,145.62
Research Roundtable		10,773.51
Store		1,957.50
Website		<u>12,643.35</u>
Total	\$	251,325.02

### BALANCE SHEET AS OF DECEMBER 31, 2014

#### Assets

Checking/Savings Accounts	\$	128,442.75
Money Market Account		307,501.80
True Family Restricted		<u>6,048.57</u>
Total Assets	\$	441,993.12

#### Liabilities and Equity

Opening Balance Equity	\$	358,575.64
Net Income/Loss		<u>83,417.48</u>
Total Liabilities and Equity	\$	441,993.12

## 2015 BUDGET

**Income**

Fundraising	\$	200,000.00
Grants/Memorials		5,500.00
Interest		300.00
Store		<u>4,000.00</u>
Total	\$	209,800.00

**Expenses**

Independent Contractors	\$	80,000.00
Administration		7,000.00
Bank Charges		100.00
Education and Outreach		3,000.00
Fundraising		20,000.00
General Administration		7,000.00
Legal/Professional Fees		1,500.00
<i>The Mirror</i>		4,000.00
PR and Marketing		20,000.00
Registry		15,000.00
Priority Projects		100,000.00
Research Grants		5,000.00
Research Roundtable		10,000.00
Store		2,500.00
Supplies		1,000.00
Website		<u>12,000.00</u>
Total	\$	288,100.00

## 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*, as well as through periodic emails and mailings. The newsletter is available on Dup15q Alliance's 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 platforms has increased awareness of the Alliance's mission as well as significantly assisting in fundraising efforts.

## Contact Information

**Dup15q Alliance**

**P.O. Box 674, Fayetteville, NY 13066 USA**

**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)