


# 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

Paul Karch

Families of children with dup15q, children who love the speed and jerk of roller coasters but not the loud music or waiting in line of most amusement parks, smile and scream as they enjoy a special session at the indoor amusement park at Mall of America.

A parent of a child newly diagnosed with dup15q finds detailed information and a sympathetic, understanding ear of a parent who has been through the same thing in facing a challenging and little known condition.

A caregiver asks for ideas in a Facebook conversation on how to respond to a medical, social or educational challenge in the life of a person with dup15q. An enthusiastic and skilled music therapist shares ideas and hope with a roomful of caregivers who face similar challenges and who understand when

children run around, hide behind curtains or lie on the floor of the conference room.

A woman with dup15q, in a manner that would be startling to and considered inappropriate by most, climbs into a restaurant booth of people not her family, reaches for a drink that is not hers and is met with a welcome laugh rather than fear and rejection.

A child with dup15q visits a group of specialty physicians who have seen many children with dup15q and are familiar with what does and does not work in treating people with dup15q.

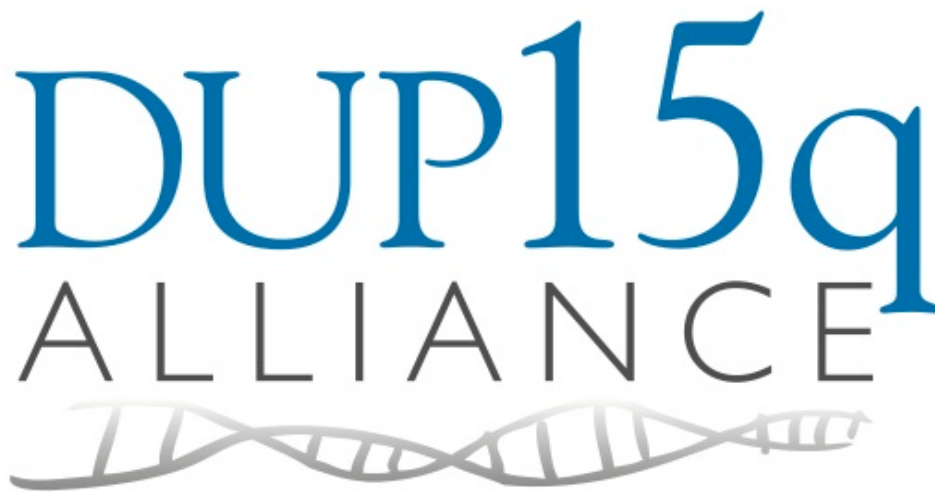
A group of scientists, including geneticists, biochemists, neurologists and others, meets to learn about the latest bench and clinical research into dup15q and to consider next steps in research and development which will lead to greater understanding of and possible new treatments for dup15q.

These scenes all took place in 2013 as generous donors, volunteer family leaders and a staff of two worked together to realize the mission of Dup15q

Alliance to provide family support and promote awareness, research and targeted treatments for chromosome 15q duplication syndrome. The depth and breadth of Alliance activities continued to grow in 2013 as we expanded our membership, held our largest conference and scientific meetings ever and opened new dup15q clinics, bringing the total to nine around the country. You will find further details of all this activity in this Annual Report.

I was honored to become chair of the Alliance Board this year and enjoyed working with a dedicated and enthusiastic group of families to provide the great support and services which were unknown, as was even the specific identity of dup15q, when our daughter Rachel was born 27 years ago.

I would like to thank our professional advisory board, dup15q researchers, the Board of Directors, our dedicated staff, other family volunteers and our generous fund-raisers and donors for this exciting progress. We look forward to continued growth and service in 2014.



## Board of Directors

In 2013, the board filled one of the open board seats. On March 17th, Fernando Gomez was elected with a term ending in June 2014. In addition to his role as director, Fernando will also head up the International Committee of Dup15q Alliance, a job he has already had years of practice doing!

In October, the Dup15q Alliance Board of Directors voted to expand the Professional Advisory Board by two members. Shafali Spurling Jeste, MD & Scott Dindot, PhD were asked to join the PAB and both accepted. Both have a long history of work with chromosome 15q duplications. Dr. Jeste heads up our Los Angeles dup15q clinic and Dr. Dindot created a dup15q mouse model. We look forward to working with these talented scientists.

Also in October, the Board voted Paul Karch as Chair of the Dup15q Alliance Board of Directors. Paul has been on the board for more than two years and is well quipped to lead Dup15q Alliance. We look forward to his insightful guidance as Dup15q Alliance continues to expand and grow.

### Officers:

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

### Board Members:

Donna Bennett (PA) - Co-Founder  
 Guy Calvert (CA)  
 Fernando Gomez (NJ)  
 Rylie McHam (OH)  
 Linda Meagher (TX)  
 Mike Porath (CA)  
 Dana Tilton (GA)

### 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

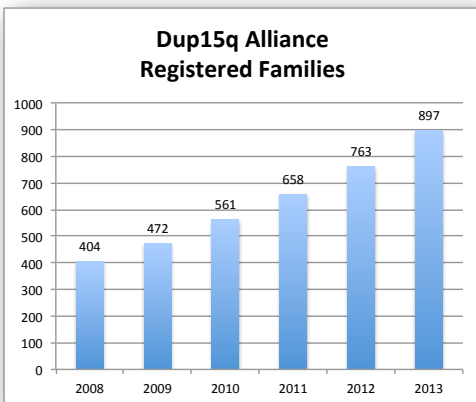
Sarah Spence, MD, PhD  
 Children's Hospital Boston

Ron Thibert, DO, MsPH  
 Massachusetts General Hospital

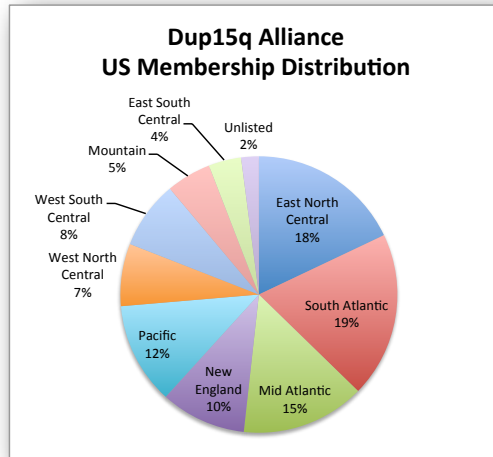
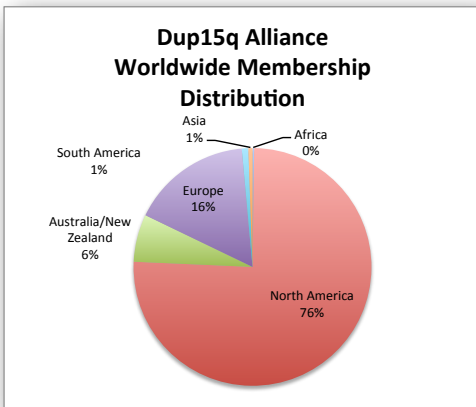
# Dup15q Alliance Membership

Dup15q Alliance serves members around the world. While the organization is incorporated in the United States, international families affected by chromosome 15q duplication syndrome 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, 2013, Dup15q Alliance had 897 registered families, continuing its steep growth trend with a 18% increase in members during 2013.



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 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, 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

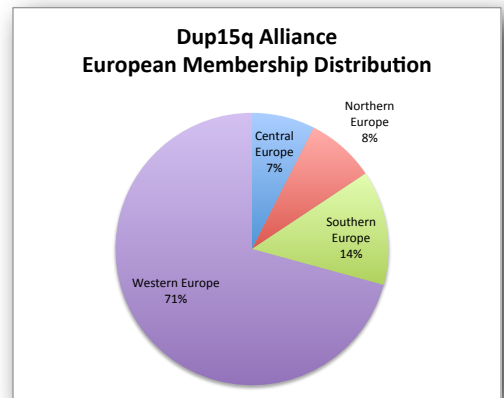
Outside of the United States, more than fifty-five percent of the remaining Alliance membership is located in Europe, primarily in Western Europe. The chart below illustrates the Dup15q Alliance's membership distribution by European Geographical Division. The European divisions include:

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

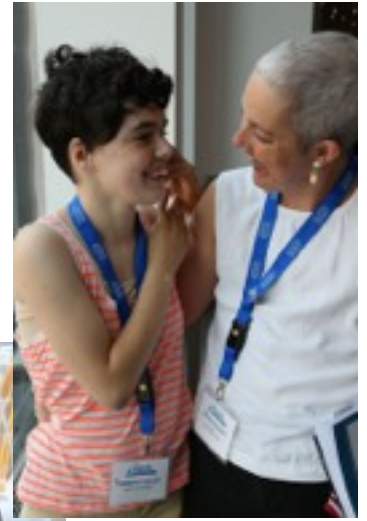
**Northern Europe:** Denmark, Norway, Finland and Sweden

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

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



## Seventh International Dup15q Alliance Conference

*July 25-27, 2013 in Bloomington, Minnesota*

*Photo credit to Rick Guidotti, [www.positiveexposure.com](http://www.positiveexposure.com)*

Our 7th International Conference was held at the Radisson Blu Hotel at the Mall of America in Bloomington, Minnesota. Our largest to date, a total of 409 people attended the conference. Ninety-five dup15q families were represented, including 62 individuals with dup15q syndrome. More international families travelled to the conference than ever before, as well. Nine countries other than the United States were represented by 36 family members.

The dup15q parade which opens each conference was once again a family favorite. Special events were held at the Nickelodeon Universe Amusement Park and the Aquarium. Families had a wonderful time on the rides at the amusement park which was opened for two hours just for Dup15q Alliance conference attendees.

A variety of sessions on a wide range of topics were offered at the conference. Keynote speaker Chantal Sicile-Kira, a

dup15q parent and award-winning author, speaker and leader in the field of adolescence and transition to adulthood, presented several inspirational sessions about raising a child with special needs. Other speakers included Sheila Merzer, a licensed psychologist and expert on autism spectrum disorders, Lori Guzman, an attorney specializing in special needs trusts, and David Geslak, an autism fitness specialist.

In addition to the main speakers, presentations were given by researchers and physicians on the latest science of dup15q syndrome. Practical workshops on potty training, planning for life after high school, assistive technology, managing anxiety and epilepsy, and music therapy were also held. Another conference favorite also returned - breakout sessions for women, men, extended families and parenting siblings.



This conference also taught us that growth often comes hand-in-hand with increasing complexity. Several families affected by edge or microduplications of chromosome 15q attended the conference. With symptoms often different from individuals with core 15q duplications, more questions were raised than answered. We hope as science moves forward, we have a better understanding of how all this fits into the bigger puzzle of dup15q.

Reach for the Stars was great success thanks to all of the volunteers who made it happen. We salute them and look forward to 2015 in Orlando, Florida!



# Dup15q Alliance Events

*Dup15q Alliance events bring together families, raise funds and increase awareness of chromosome 15q duplication syndrome.*

## Regional Gatherings

Traditionally, Dup15q Alliance regional gatherings tend to be less frequent during a conference year. However, in 2013 multiple regional gatherings were held across the country!

As always, those who attended appeared to have a wonderful time connecting and sharing their experiences with other dup15q families. Region two held a gathering in New York at the Syracuse zoo. Five families enjoyed a picnic and a fun day at the zoo.

Over Labor Day seven families from region seven gathered in Goldendale, Washington at the Maryhill state park. It was described as a fabulous day!

In May, three families from region eight gathered in Visalia, California at the Damkos' home for a barbeque and swimming. The families had a wonderful day and several other families in California met up in August at Disneyland.

Region 10 gathered at the home of Dana and Luke Tilton in Tyrone, Georgia. It was held on a beautiful March day and six families attended. Everyone had a great time eating, talking and the kids enjoyed the playground.

The regional gatherings are a special time for dup15q families to connect with other families who truly understand and appreciate the joys and challenges associated with raising or caring for an individual affected by dup15q syndrome. We are currently planning gatherings for 2014, so if you are interested in hosting or attending a gathering please contact your regional representative!

## Paint the Town Blue

In December 2013, the Ward family held their first Dup15q Alliance fundraiser in Hokes Bluff, AL.

After their son's diagnosis, they were frustrated with the lack of information from their geneticists. They began researching and found Dup15q Alliance's website. Tracy writes, "Without this tool and the Facebook page, I would have been lost. It was my one source to learn, ask questions, vent and not feel alone. My family and I got together and decided to have a fundraiser to support that organization that had helped me to understand my son."

What began as an idea for a 5k run and 1k fun run, soon became a day long event. In addition to the run, they hosted a dinner event and silent



auction and called it "Paint the Town Blue".

The event was a huge success both in terms of awareness of dup15q syndrome and fundraising. Thank you to the Ward family for this fantastic event!



Dup15q families gather at Maryhill State Park in Goldendale, Washington above.

## More News & Events from 2013

### Nonsolo15 Annual Gathering

Nonsolo15, a support group in Italy for families affected by dup15q syndrome, held their annual gathering in Tuscany in September 2013. Thirteen families participated in the holiday at the beach and the main topic of conversation was the report of Dup15q Alliance's International Conference that the Piredda family attended in July. The spoke of Dup15q Alliance's projects and the importance of the Registry. They translated some of Dup15q Alliance's information into Italian for posting on their website.

Dup15q syndrome is a disorder without borders. Dup15q Alliance welcomes the opportunity to work with organizations in other countries who work to fulfill the same mission.

### Dup15q Alliance Joins NORD

In 2013, Dup15q Alliance became a member of the National Organization for Rare Disorders (NORD). NORD is a unique federation of voluntary health organizations dedicated to helping people with rare "orphan" diseases and assisting the organizations that serve them. NORD is committed to the identification, treatment, and cure of rare disorders through programs of education, advocacy, research, and service.

Dup15q syndrome is a rare disorder, as defined by one that affects less than 200,000 Americans. Membership in NORD helps Dup15q Alliance to join forces with other small non-profits to encourage education, advocacy and research for rare disorders. It is also a referral source for Dup15q Alliance membership. If individuals inquire about support for dup15q syndrome they are told about Dup15q Alliance.

### Seven Additional Dup15q Clinics Open Around the United States

2013 was a great year for dup15q clinics. Seven clinics opened, in addition to the ones in Miami and Boston that opened in 2012. The new locations are Los Angeles, New York City, Seattle, Memphis, St. Paul, San Francisco and Lewisburg, PA.

Dup15q clinics provide comprehensive evaluation and treatment of all facets of dup15q syndrome. Patients are first seen by a neurologist and then referred to whichever specialists they need. Arrangements can be made for testing to be done in a short period of time for families who travel longer distances to visit the clinic.



For more specific information on each clinic please visit our website - [www.dup15q.org](http://www.dup15q.org).



Above: dup15q families at the Rosamond Gifford Zoo in Syracuse, New York.

NORD also sponsors such events as Rare Disease Day. Dup15q Alliance celebrates Rare Disease Day with "wear blue for dup15q" and the end of our \$15,000 in 15 Days for Dup15q fundraiser.





# 2013 Scientific Meeting

September 19-20, 2013

*MIND Institute, University of California Davis*

Over seventy-five researchers studying the genetics, biochemistry and clinical characteristics of dup15q syndrome gathered at the UC Davis MIND Institute for the annual Dup15q Alliance Scientific Meeting. Even with all that scientific firepower, it was clear that the families of Dup15q Alliance are the most critical resource to accelerating the breadth and speed of understanding dup15q syndrome.

Families contribute vital information to the Dup15q Alliance Registry, providing a collective look into the lives of those with dup15q syndrome. This information is used by clinicians and researchers to better understand dup15q and push science forward.

Because dup15q syndrome was first identified through genetic testing, the bench (or laboratory) science is ahead of the clinical science, but it is slowed by the lack of extensive and detailed data on the clinical characteristics of

people with dup15q syndrome. One of the keys to further progress is better clinical data, which can be learned through the Registry and dup15q clinics.

During the two-day science meeting, presentations were given on sleep and dup15q syndrome, genetic links to autism, the dup15q clinics, the gene UBE3a, and studying dup15q using animal models and stem cells. There were also sessions on the genetics of chromosome 15q duplications, which grows more and more complex as scientists dig deeper.

The overall goal of both bench science and clinical research is the development of targeted treatments which could make a real difference in the lives of people with dup15q syndrome and their families. We learned clinical research is already yielding some useful information, such as a sense of what

anti-epileptic drugs are effective for those with dup15q syndrome.

Exciting treatments based on understanding other rare genetic disorders provide a basis of hope that we will find more successful treatments for dup15q syndrome, even while understanding that dup15q is more complex than most and it will take many years to develop targeted treatments. Dup15q Alliance supports and encourages basic research by providing opportunities, such as this science meeting, for researchers to share findings, methods and possible paths for future inquiry.

We are pleased to report that there is a great deal of interesting and important work being done on chromosome 15q duplications. While much of it is difficult for non-specialists to fully grasp, rest assured it points to a very hopeful future.

## Research Funding

Another first for Dup15q Alliance occurred in 2013. After careful consideration, the Board decided to directly fund a scientific research project. In the past, we have offered our non-financial support and have matched families with studies, but this was the first time that Dup15q Alliance has provided scientists directly with funding for a project.

What was it, you ask? Dup15q Alliance funded the creation of a

dup15q mouse model at Texas A&M University. These mouse models will help scientists study the causes of dup15q symptoms with the hope of creating targeted treatments.

Our pilot project was so highly regarded that Simons Foundation granted Texas A&M University an additional \$85,000 to further the project under the direction of Dr. Scott Dindot.



# Financial Status

Tom Doyle

As we enter our 20th year Dup15q Alliance is well established as an organization. In just the last decade, we have grown from about 300 families to almost 1,000. We fulfill our mission by providing family support through The Mirror, biennial conferences, parent contacts, regional representatives, an informative website, social media, and a First 100 Days handbook. In addition to family support, we have funded research, created a searchable registry, provided annual scientific meetings, organized and promoted clinics in nine cities

throughout the United States and developed relationships with other organizations around the world in dup15q and related communities.

We have gone from an all-volunteer organization to one that now pays a small stipend to our Executive Director and an Executive Assistant, who manage the organization. Imagine the number of phone calls, emails, letters, meetings, grant proposals, donation responses and other organizational tasks that have multiplied many times over the past decade.

Deep down, we don't want our organization to get bigger, as that

means more families are receiving a dup15q diagnosis, but we know the reality is that there are many, many more individuals who will be diagnosed and referred to the Alliance as doctors and geneticists help direct families to the Alliance. We are growing exponentially and we need to prepare for that.

We have many dreams of how Dup15q Alliance can help families in the next 20 years and are trying to position the organization to do that financially. Your support and financial gifts make a tremendous difference to all of our children. We look forward to the next 20 years and to many new, exciting and helpful advances.

## 2 0 1 3 I N C O M E S T A T E M E N T

### Income

Conference	\$ 39,739.75
Fundraising	137,294.31
Grants/Memorials	11,345.00
Interest	374.75
Store	7331.99
	Total \$ 196,085.80

### Expenses

Independent Contractors	\$ 19,200.00
Administration	8,960.00
Bank Charge	99.00
Conference	89,623.44
Education and Outreach	208.69
Fundraising	3,283.30
General Administration	6,406.27
Legal/Professional Fees	721.14
<i>The Mirror</i>	3,087.94
PR and Marketing	8,129.31
Registry	37,676.84
Research Grants	-
Research Roundtable	10,963.29
Store	3,981.55
Supplies/Computers	0.00
Website	8,016.89
	Total \$ 200,357.66

## 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 3

### Assets

Checking/Savings Accounts	\$ 45,335.63
Money Market Account	307,194.46
True Family Restricted	6,045.55
	Total Assets \$ 358,575.64

### Liabilities and Equity

Opening Balance Equity	\$ 362,847.50
Net Income/Loss	(4,271.86)
	Total Liabilities and Equity \$ 358,575.64



2 0 1 4  
B U D G E T

**Income**

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

**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		12,000.00
<b>Total</b>	<b>\$</b>	<b>288,100.00</b>

## 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  
 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/IDEAS/305990303151](https://www.facebook.com/pages/IDEAS/305990303151)  
**Twitter:** [www.twitter.com/IDEASdup15q](https://www.twitter.com/IDEASdup15q)  
**BigTent:** [www.bigtent.com/groups/dup15q](http://www.bigtent.com/groups/dup15q)

