

# 2020-2021 ANNUAL REPORT



## OUR MISSION

We empower individuals living with dup15q syndrome and other related rare diseases to reach their full potential by advancing breakthrough research and life-changing therapeutic treatments, supporting families affected by dup15q, and promoting advocacy.

[www.dup15q.org](http://www.dup15q.org)



# LETTER FROM BOARD CHAIR DAVID GIFFORD



Dear Dup15q Community,

It has been quite a couple of years. I'd like to run down some of the highlights.

The 2021 Family Conference was different to say the least, we went forward with an in-person plan as well as including remote attendance and speaker participation. The Moving Mountains Conference in Denver was a great success. There is no substitute for bringing our families together in person, but we acknowledged that many of our community and some of the speakers could not attend for obvious reasons. We will plan to take the same hybrid approach for our 2023 Family Conference slated for Nashville.

Our financial picture continues to improve. This mission is critical to advance breakthrough research and life-changing therapeutic treatments to support our families.

As we continue to thrive in the research arena, there are valuable updates at [dup15q.org](https://dup15q.org) regarding the latest information. There has been a great deal of focus on epilepsy in the rare disease community and dup15q is well represented in that work. In addition, we received a grant from the Chan-Zuckerberg Initiative (CZI) that has helped with the formation of the Commission for Neurodevelopmental Copy Number Variants (CNVs). The Commission is a new, one-of-a-kind collaboration formed with two other patient-led advocacy groups with complex copy number variants, aiming to investigate the challenges common to neurodevelopmental CNVs in order to accelerate therapeutic developments.

Regarding your board of directors and our incredible administration team, as I'm sure most are aware, we have a new Executive Director, Carrie Howell, who joined our team in 2021. I want to thank Vanessa Vogel-Farley for her service as our previous Executive Director. We are fortunate that Vanessa has agreed to stay on as part of our board, where she will continue to focus and drive the science and research efforts of dup15q syndrome. I also want to acknowledge Tessa Quinlan's past contributions, as she has decided to step back from being on the board. To say Tessa added to the board is an understatement, she has been our Family Conference committee chair for the past three conferences, which is quite an achievement. Thank you, Tessa.

Wrapping up, we will continue to strive to advance our Vision and envision a world where families, clinicians, and advocates enable Dup15q individuals to thrive.

Best regards,  
David Gifford



# LETTER FROM EXECUTIVE DIRECTOR CARRIE HOWELL



Dear Dup15q Community,

Many of you have reached out and asked how my first few months have been as the new Executive Director of Dup15q Alliance. I can unequivocally say that it has been an incredible few months. I have been in awe of the willingness of this team to work tirelessly to meet the needs of our community, raise awareness, advance the mission, educate our families, and fight for the future of research for those with dup15q syndrome.

We are excited as we look ahead to 2022 as we pursue our goals of:

- Finalizing our 3-year strategic plan
- Rolling out our updated mission and vision statement
- Continuing our work in advancing our research measures
- Offering more programming for families and
- Growing our fundraising efforts to support our future work

I want to personally thank each of you who have already spent time with me sharing your dup15q journey. To those who are our donors, we absolutely cannot grow beyond what our donors give no matter how many ideas we can conceive. Thank you for your part in making this work possible.

I look forward to meeting each of you whether via Zoom, at our biannual conference in 2023, or over a phone call. Thank you for being part of our community and allowing me to be part of your journey.

With Gratitude,

A handwritten signature in blue ink that reads "Carrie".

Carrie Howell  
Executive Director

# BOARD OF DIRECTORS - 2021

Our 100% volunteer Board of Directors is responsible for determining the organization's mission and purpose, ensuring adequate resources, and providing financial oversight. The board delegates responsibility for day-to-day operations to the Executive Director.



**Donna Bennett**  
Founder, Board Member Emeritus

**David Gifford**  
Board Chair

**Tony Marmo**  
Treasurer

**Joe Elassal**

**Elina Marchenko**

**Lisa Feehery**

**Mike Porath**

**Julia Jordanich**

**Tessa Quinlan**

**Steve Kadner**

**Carlos Romero**

**Anne Karch**

**Vanessa Vogel-Farley**

## STAFF

Executive Director through Sept 2021: Vanessa Vogel-Farley

Executive Director: Carrie Howell

Administrative Director: Naka King

Operations Manager: Amy Kotnik

15q Clinic Coordinator: Zoe Dannenberg

Support Services Director: Krissa Harris

Outreach and Education: Ember Burke



## PROFESSIONAL ADVISORY BOARD -2021

The Dup15q Alliance Advisory Board offers expertise on scientific developments, to provide insights on the needs of the dup15q syndrome population, and to assure that Dup15q Alliance's policies, research, grants, marketing, communications, and publications meet the highest standards of scientific rigor and accuracy. We are so thankful to the amazing individuals who serve the Dup15q community!

Jana Benesh, PhD: Clinical Strategy and Patient Advocacy Lead, Center for Rare Diseases, ICON

Dimitrios Arkilo, MD:  
Clinical Development at Acadia Pharmaceuticals Inc.

Agotino Battaglia, MD:  
DPed, DNeurol Calambrone, (Pisa), Italy

Stormy Chamberlain, PhD:  
University of Connecticut Health Center, Farmington, CT

Edwin H. Cook, Jr., MD: Department of Psychiatry  
University of Illinois at Chicago, Chicago, IL

Orrin Devinsky, MD: NYU School of Medicine

Scott Dindot, PhD: College of Medicine, Texas A&M University,  
College Station, TX

Elizabeth Jalazo, MD: Genetics and Metabolism, UNC School of  
Medicine

Shafali Spurling Jeste, MD: Neurological Institute Las Madrinas,  
Children's Hospital, Los Angeles, Keck School of Medicine of USC

Janine M. LaSalle, PhD: Medical Microbiology and Immunology  
UC Davis School of Medicine, Davis, CA

N. Carolyn Schanen, MD, PhD  
Nemours Biomedical Research, Alfred I. duPont Hospital for Children,  
Wilmington, Delaware

Sarah Spence, MD, PhD:  
Assistant Professor of Neurology, Harvard Medical School

Ron Thibert, DO, MSPH: Pediatric Epilepsy Program  
Massachusetts General Hospital, Boston, MA

# EDUCATION AND OUTREACH

## 2021 Conference



- 11th biannual International Family Conference
- Over 65 presentations
- Introduced online streaming
- Keynote speaker Temple Grandin
- Over 400 in-person attendees and 100 virtual attendees
- Collected 101 bio research samples, 49 participating families



### Conference Presentation Highlights

- Teen Life
- Adult Life
- How to Support Siblings
- Sibling Sessions
- Genetic Counseling
- Special Education
- Pediatric Neurology
- Seizure Devices
- Seizure Tracking
- Neuro Surgery
- Empowering Ability
- Special Needs Dentistry
- Gastroenterology
- Social Security
- Special Needs Trusts
- Sleep
- Sensory
- OT/Sensory Specialist
- Regression
- Research
- ABA/Life Skills Programming

## 2021 Big Give

On May 15th, 2021, our community came together as we went LIVE for our 4th annual BIG GIVE event: "Everything I know about Dup15q, I learned from my Dup15q Alliance Family." LIVE streams included dup15q doctors, researchers, and parents. It was a day of sharing stories, spreading awareness about dup15q syndrome, and answering questions.

MAY 15, 2021  
BIG GIVE

EVERYTHING I HAVE LEARNED  
ABOUT DUP15Q I LEARNED  
FROM MY DUP15Q FAMILY



### Highlights

- Upcoming Clinical Trials
- Dup15q Research Using Stem Cells
- Junior Believe Ambassadors
- Research Priorities of the 15q CRN
- Sibling Programming
- EEG Biomarker of dup15q syndrome
- Dup15q Clinics
- Sleep Patterns in dup15q



## Rare Disease Day

We aim to raise awareness through education for our community, researchers, clinicians, and policymakers to bring equitable access to diagnosis, treatment, health, and social care as we work towards a more inclusive society together.

Equity in practice means meeting peoples' specific needs and eliminating barriers that prevent their full participation in society including social opportunities, equitable access to health and social care, finding a diagnosis, and receiving proper treatments. Rare Disease Day is an annual event slated nationally for the end of February in conjunction with government advocacy efforts.

## International 15q Day

Together with Angelman Syndrome Foundation and the Foundation for Prader-Willi Research, we are raising awareness of the similarities between dup15q, Angelman, and Prader-Willi Syndromes.

These are all distinct neurodevelopmental disorders that are caused by changes within the specific q11 to q13 region of the long arm of chromosome 15 (called 15q). This region contains genes, like SNRPN and UBE3A, and others, which are very important for brain development. Unlike most other genes in the body, these genes behave differently depending on whether they are on the copy from the mom (maternal) or from the dad (paternal); this is a biological mechanism called 'genomic imprinting.' With our newborn screening initiative, families with loved ones with dup15q, Angelman, Prader-Willi Syndromes will find a diagnosis in weeks instead of years, avoiding a painful diagnostic odyssey. Diagnosing individuals earlier provides the best chance of treating the symptoms and improving their quality of life much sooner.



## Giving Tuesday

Dup15q Alliance, The Foundation for Angelman Syndrome Therapeutics (FAST), Angelman Syndrome Foundation (ASF), and the Foundation for Prader-Willi Research (FPWR) announced a collaborative initiative to fund the addition of chromosome 15 conditions to Early Check, a flexible framework amenable to screening for a variety of conditions. Led by RTI International, Early Check will support the infrastructure necessary to add dup15q, Angelman, and Prader-Willi Syndromes to the Newborn Screening Panel. In 2021, funds raised from our annual Giving Tuesday campaign were designated to this newborn screening initiative.





# FAMILY SUPPORT

## 2021 Highlights

- Launched Sibling Matters Program - distributed over 104 VIP sibling kits
- Raised awareness with over 90 new families joining the Alliance
- Created IEP Educational Resources and Help Ticket
- Provided blankets to dup15q individuals who were hospitalized
- Offered counseling to parents/caregivers at no cost
- Continued mailing of birthday cards through our Sunshine Committee
- Facilitated Dup15q Parent Support Facebook page
- Sponsored Regional Meetups
- Presented webinars on Financial Advocacy
- Hosted calls for Teen Life Resources
- Initiated calls for Adult Transitions
- Administered Clinical and Family Conference Travel Grants



### Our Vision:

We envision a world where families, clinicians, and advocates enable dup15q individuals to thrive.

# CLINICAL CARE AND RESEARCH



## 15q Clinical Research Network

Partnering with Angelman Syndrome Foundation, together we support 16 dup15q clinics in our 15q Clinical Research Network (CRN). Clinicians in our CRN are dedicated to developing standards of care and patient data management along with developing and maintaining a clinical research agenda.

This new combined network leverages and grows the existing infrastructure and provides greater depth of coverage geographically, allowing broader access to 15q clinical care. The combined 15q CRN is currently pulling together clinical expertise from across the globe and driving forward continued clinical research data collection. Additionally, we are ensuring the ongoing effort of attracting and educating the next generation of trainees in the field of genetic neurodevelopmental disabilities and 15q related disorders. Bringing together two patient-led foundations results in increasing the amount of passion, knowledge, accessibility and funding for drug and clinical breakthroughs. By bringing forth both Dup15q Alliance and Angelman Syndrome Foundation missions, we are ensuring that the patients and families come first.

## Highlights



Zoe Dannenberg joined the team in 2020 as the 15q Clinical Research Network Coordinator.

Zoe works closely with clinicians, industry partners and families to help strengthen the 15q clinic network and its impact on research. She is dedicated to assisting families with clinic visits and clinical trials, as well as answering questions about clinic services, procedures and any other concerns. Contact Zoe directly at [zoe.dannenberg@dup15q.org](mailto:zoe.dannenberg@dup15q.org) for further information regarding our CRN.



We are excited to announce the launch of a new program, the LADDER Database, designed to help advance research and clinical service for individuals living with dup15q or Angelman Syndrome. LADDER, Linking Angelman and Dup15q Data for Expanded Research, will allow information gathered about individuals with these disorders to be housed in one place, reducing the time families spend completing forms about their loved ones, while maximizing knowledge to help with interventions and future treatments. The LADDER Database will provide a wealth of new information to researchers, clinicians and families. The database will be maintained at RTI International, a non-profit research institute dedicated to improving the human condition. All the information about you and your family will be kept on a private network that has high security; only members of the research team can see it. Family members' names and contact information are never sold to anyone, and never given to anyone outside of the research team. LADDER will share de-identified information (which means it cannot be traced back to you or your family) with other researchers whose research studies have been reviewed and approved by an appointed data access committee.

# CLINICAL CARE AND RESEARCH

With your support, Dup15q Alliance provides funding for research fellowship grants. Closely reviewed and monitored, our goal is to improve clinical care and to discover treatments, such as behavioral and drug therapies, targeted specifically for dup15q syndrome.

## Dup15q Research Grants

**Dr. Ben Philpot** - This project will generate new mouse models for the study of how extra UBE3A copies confer risk for seizures. \$50,000 over two years.

**Dr. Gilles Trave** - This project studies the interaction between UBE3A and HERC2 and its impact on brain development and will build the first ever 3D structure of UBE3A with and without HERC2. \$100,000 annually - this project is jointly funded by the Dup15q Alliance and Angelman Syndrome Foundation

**Charlotte DiStefano** - This project will evaluate the use of a telehealth model of conducting remote assessments of development and clinical characteristics of children diagnosed with dup15q syndrome. \$25,000 annually.

**Vidya Saravanapandian** - This project focuses on understanding the mechanisms underlying EEG biomarkers in dup15q syndrome which will enable early diagnosis and prognosis, and aids in developing treatment-targets. \$25,000 annually.

We are pleased to share that the work of our grant recipients generated additional federal funding for research continuation.

## The Commission on Novel Technology in Neurodevelopmental Copy Number Variants (CNVs)

Dup15q Alliance has teamed up with Project 8P and Ring 14 (rare disorders with large chromosome anomalies) to develop the Commission for Neurodevelopmental CNVs. The Commission aims to investigate the challenges common to neurodevelopmental disorders involving copy number changes (duplications or deletions) involving multiple genes in order to accelerate therapeutic developments.

The Rare As One Grant through the Chan Zuckerberg Initiative (CZI) has supported the collaboration and development of the Commission. Knowing there needs to be new technology to support genetic disorders with multiple copy numbers and large anomalies.

By increasing dialogue about, research on, and funding for CNVs, along with providing tools and technologies that support understanding and treatment of these conditions, we hope to enable progress that accelerates scientific and medical breakthroughs with direct benefit for patient communities.





# CHEERS TO VOLUNTEERS

Volunteers are the life-blood of any organization, whether you volunteered to serve on a committee or support the Alliance with a fundraiser, we couldn't do what we do without your support. Thank you for your steadfast dedication.



## Fundraisers

Houston Half Marathon  
Chicago Marathon  
AJC Peach Tree Road Race  
Run for a Reason  
Walk for a Reason  
Donate Your Birthday  
Giving Tuesday  
Rare Disease Day  
Conference Fundraisers to support:  
Dup15q Birthday Party and Cupcakes  
Sensory Gifts  
Dup15q Backpacks and Blankets

## Volunteer Committees

Adult Life Committee  
Believe Committee  
Conference Committee  
Education/IEP  
Extended Support Network  
Governmental Advocacy  
International Committee  
Junior Believe Ambassadors  
State Representatives  
Sunshine Committee  
Thank You Committee  
Teen Life Committee

We are grateful to our families who generously choose Dup15q Alliance as a memorial donation in lieu of flowers when their loved one has passed. We hope that knowing loved ones will be remembered in all the work we do provides a measure of comfort.

*"There you are, bringing your passions, your talents, and your wisdom to your community. As gifts. Yes, there you are, asking nothing in return, but taking away so much: connection, camaraderie, and a better community for having you in it." - Anonymous*

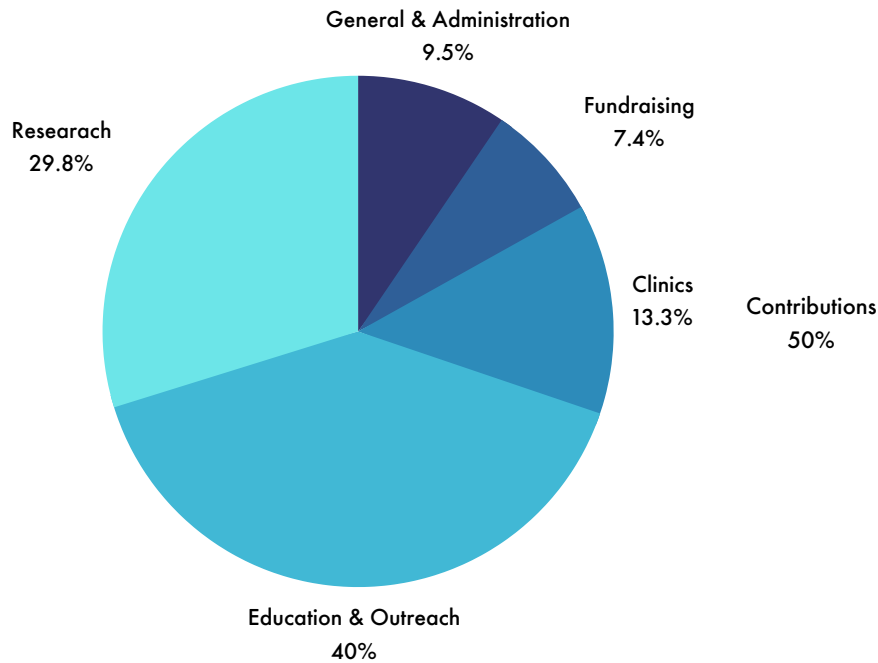
# PARTNERSHIPS

We recognize the importance of working together to achieve common goals through partnerships with other groups. Here are some of our partnerships and initiatives that are focused on improving the lives of those with rare disorders.

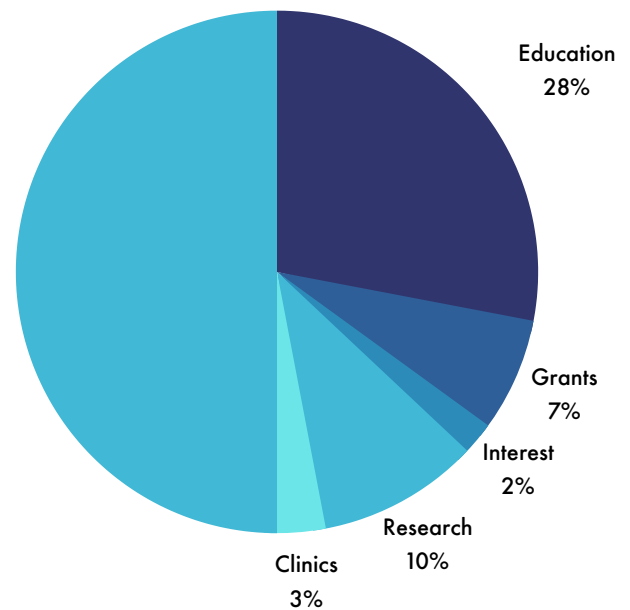


# FINANCIALS

## 2021 Actual Expenses\*

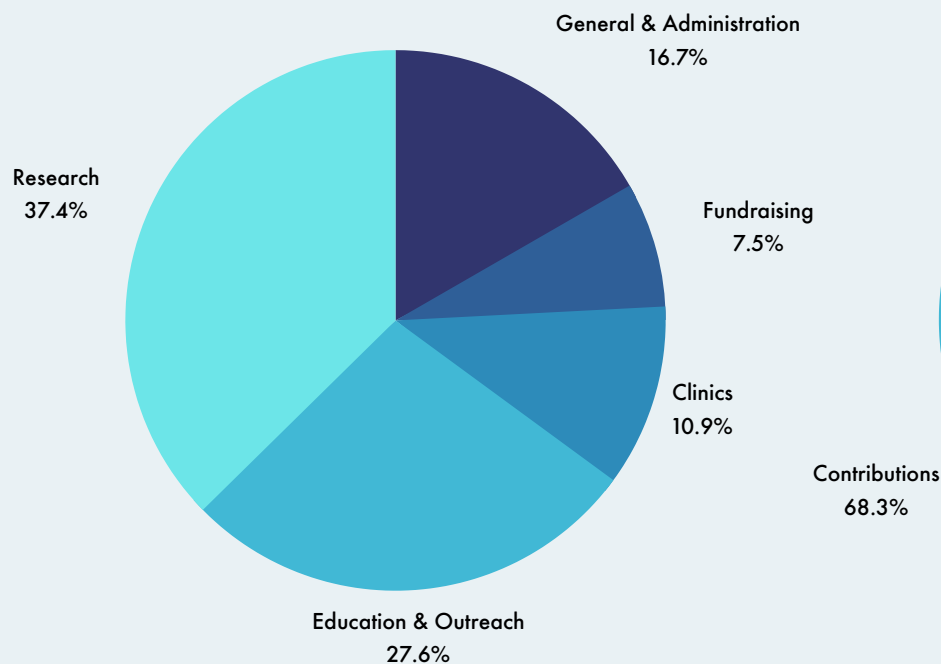


## 2021 Actual Income\*

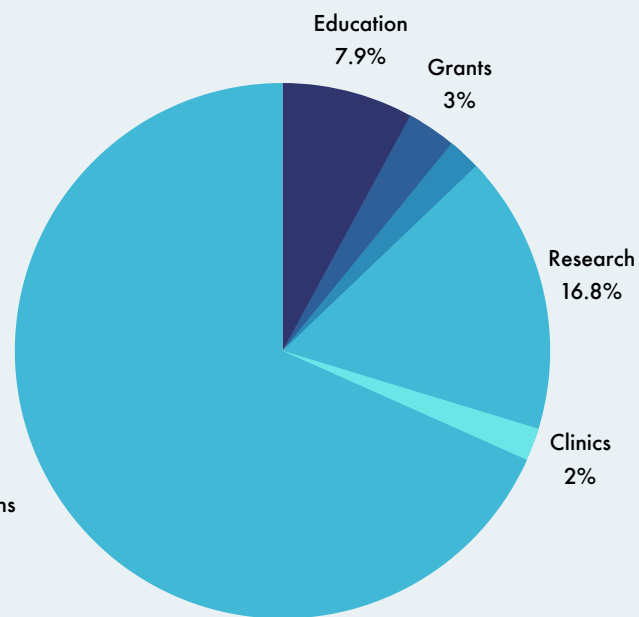


\* Financials have not been audited for 2021

## 2022 Budgeted Expenses



## 2022 Budgeted Revenue



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# SUPPORT

## WAYS TO DONATE:

### Check or Money Order

Make payable to Dup15q Alliance, mail to:

Dup15q Alliance

P.O. Box 1669

Highland Park, IL 60035 USA

### Online

To make one-time or monthly credit card donation visit:

[www.dup15q.org/donate](http://www.dup15q.org/donate)

### PayPal Giving

PayPal Giving Fund helps people support their favorite charities online. To donate via Paypal Giving Fund visit <https://www.paypal.com/fundraiser/charity/1398792>

**Check with your employer regarding matching donations from your company.**

Dup15q Alliance is a registered 501(c)(3) non-profit organization.

Donations are tax-deductible in the United States, as allowed by law. TIN: 20-0751232

