



2015 Annual Report



A cure is out there.
Help us find it.

Our Impact this year

2015 Year In Review

In 2015, our 3rd year, we continued to learn and grow as a 100% volunteer organization. With our mission to find a cure, raise awareness in the medical community, and support families affected by PCDH19 at the heart of every decision we make and every goal we set; we made an impact in all these important areas. Because of the generosity of our passionate and generous donors and fundraisers, we increased our revenue by 25%, which allowed us to award three research grants (including a second round of funding for the International PCDH19 Registry at Boston Children's Hospital and UCSF Benioff Children's Hospital, which we also funded in 2013-14). We were able to support families in need of therapeutic devices and medical equipment by providing grants through our patient assistance program. We continued to attend and exhibit at the Annual Child Neurology Society Conference and the Annual American Epilepsy Society Meeting. We also exhibited and educated Physician's and possible undiagnosed patients for our 2nd year at Epilepsy Awareness Day at Disneyland. It is important to note that we have continued to be a 100% volunteer based organization.

We officially launched our first Affiliate, PCDH19 Alliance UK, on Facebook. We have been fundraising together, sharing information and providing patient support to the UK since day 1. We even had UK representation on our Board of Directors and as more families in the UK are diagnosed, this seems like a natural course to take to better support our families in the UK. We couldn't be more excited that the UK is our first Affiliate Chapter: One Mission, One Heart, Two Continents.



Our Mission

Our mission is to improve the lives of children and families who are affected by PCDH19 Epilepsy. The Alliance focuses on raising and directing funds to scientific research with the goal of finding better, more effective treatments and, ultimately, a cure; providing information and support to affected families; and assisting the efforts of the medical community, so that no family suffers without a diagnosis and the most appropriate medical treatment.

PCDH19 Epilepsy

PCDH19 Epilepsy is a condition with a wide spectrum of severity in seizures, cognitive delays and other symptoms, which are all caused by a mutation of the PCDH19 gene on the X chromosome. Males with the mutation, who will be largely unaffected, will pass the mutation onto 100% of their daughters and none of their sons. Women with the mutation have a 50% chance of passing it to their daughters and will pass it to 50% of their sons. Recently, scientists have discovered some unaffected females and are studying to learn what is protecting them from the disorder. Mosaic males are also affected, but so far very few males have been diagnosed.

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Research Grants

Research gives us hope and is by far our largest program here at the Alliance. With the guidance of an esteemed international Scientific Advisory Board, we are vetting the most promising PCDH19 research on the planet today. Below are the three projects we funded for 2015-2016. We received several scientifically strong proposals, and wish we could have funded them all. For a rare and still underdiagnosed disease, the scientific and pharmaceutical community has an uncharacteristically strong interest in studying PCDH19 Epilepsy, and we are rising to the challenge to fund them.



Maria Passafaro PhD

Unraveling the Molecular Mechanisms of PCDH19 in Cultured Neurons and in PCDH19 KO Mouse Model



Paul Thomas PhD and Jozef

In Vivo Investigation of the Cellular Interference Model Using Unique PCDH19 Mouse Model and Brain Tissue from a PCDH19 Affected Female



Our goal is to fund research directly targeted toward understanding the expression of the PCDH19 gene and the function of the PCDH19 protein, finding therapeutic treatments, and a cure for PCDH19 Epilepsy.

We also hope that projects we fund will lead to additional research support from government or other funding agencies. We are pleased to be able to support many different types of projects, each critical for advancing all phases of PCDH19 research, from basic, to clinical, to treatment.

PCDH19 Gene Registry and Natural History

The PCDH19 Registry is organized by Boston Children's Hospital and the University of California, San Francisco, and is crucial to illuminating and understanding PCDH19 Epilepsy. Our first round of funding in 2013 made the registry possible, and this second round will allow continued growth and also add an important behavioral component.



Beth Sheidley, MS, CGC



Lacey Smith, MS, CGC



Annapurna Poduri, MD, MPH



Nilika Singhal, MD

Patient & Family Support

Patient Assistance

While investments in research today will lead to better treatments tomorrow, we also believe in supporting those living with PCDH19 today and their families through several programs: Patient Assistant Grants, Facebook PCDH19 Parent and Caregiver Community, Teddy Bear Program, and Travel Grants for our Biennial Professional and Family Symposium.

Patient Assistance Grants

We provided therapeutic devices and durable medical equipment not covered by insurance to children and families living with PCDH19.



“The PCDH19 Alliance Patient Assistance Grant was used to purchase the Proloquo2Go communication app for my daughter Freya. Since getting this app, Freya has not only become more confident asking for things with the symbols, her speech is improving as well. I am so glad the Alliance was able to give Freya a voice.”

Kristine Coultas,
Mother of 5 year old Freya
England

Facebook Support Group



We continued to grow our Closed Facebook family support group, and now have 240 members. Our group continues to grow while remaining a safe, non-judgmental home where families can go to celebrate, commiserate, vent, ask questions, find all kinds of support, and find the latest information about PCDH19 Epilepsy and the PCDH19Alliance. PCDH19 can be isolating, and this group is a lifeline and a home. Alliance volunteers administer the group, and must check in several times daily and also screen new member requests.

Providing Hope and Support— The Shepard Family

My daughter just started reading and I am over the moon excited. Alyssa is 7 years old and in the first grade so the fact that she is reading probably doesn't impress too many people. My husband and I weren't sure it would happen this year though so we are ecstatic. Alyssa has PCDH19, a rare and severe form of genetic epilepsy. What many people may not realize is that this type of epilepsy involves more than just seizures; it also comes with behavioral and cognitive deficits that can be severe. Even after years of therapy and billions of doctor visits, Alyssa struggles with short term memory and impulse control among other things. We used to think those challenges were the result of damage from seizures or trauma sustained before we adopted her but research funded by the Alliance has helped us understand how it is all related. That research also offers us hope on the bad days because we know there is cure out there and some of the best minds on the planet are working hard to find it.

PCDH19 impacts my family every day. We deal with meltdowns or confusion over simple instructions. Sometimes my boys miss out on what they desire because our schedules revolve around therapy and doctor visits. I'm job searching right now and the positions I can apply for are pretty limited considering the flexibility that raising her requires. I could write to you about all of the challenges because there are many of them. All of that fades when I hear her sounding out words in the backseat though. In spite of her diagnosis and her limitations and everything else stacked against her, my daughter just started reading.

- LeAnn Shepard



Education, Awareness, and Advocacy

Education

Our awareness and education program continued to grow and make an impact on the medical community and patients waiting for a diagnosis.

Volunteers from the Alliance exhibited at **Child Neurology Conference** in Washington DC for our 2nd year, where the goal was to inform and educate neurologists about PCDH19 Epilepsy, the symptoms and encourage them to test patients.

Awareness & Advocacy

This was our 3rd year exhibiting at the **American Epilepsy Society Conference**, where over 5000 epilepsy specific medical professionals and researchers from around the world convened to exchange ideas, discuss the latest research, and hold meetings.



We feel we reached a tipping point this year at these medical conferences, and **more than half of the medical professionals we approached knew about PCDH19 and many even had patients.** However, the problem still persists. For example, insurance companies aren't willing to pay for the genetic testing required to diagnose. This is a significant issue in identifying those with PCDH19 and delivering appropriate treatment.

We raised awareness among those with epilepsy, advocates and physicians at our 2nd year at **Epilepsy Awareness Day at Disneyland!** Thanks to the amazing Kathy Tenchka, for single-handedly setting up and running our booth again this year!



We continued to work with Marinus to help facilitate the first **PCDH19 drug trial.** The trial is ongoing, and it was a significant accomplishment to begin a drug trial for a rare disease that was identified just 7 years ago. Working together with researchers, families, pharmaceutical partners, and other epilepsy advocacy organizations, we are strong advocates for those with PCDH19 Epilepsy.

Several families and their friends laced up their running shoes and completed runs to raise awareness and much needed funds. We had Alliance runners in the US, Canada and the UK. We also held dinners, auctions, raffles, dine n' donate, cookies sales, and online fundraisers, raising awareness along the way.



The Numbers

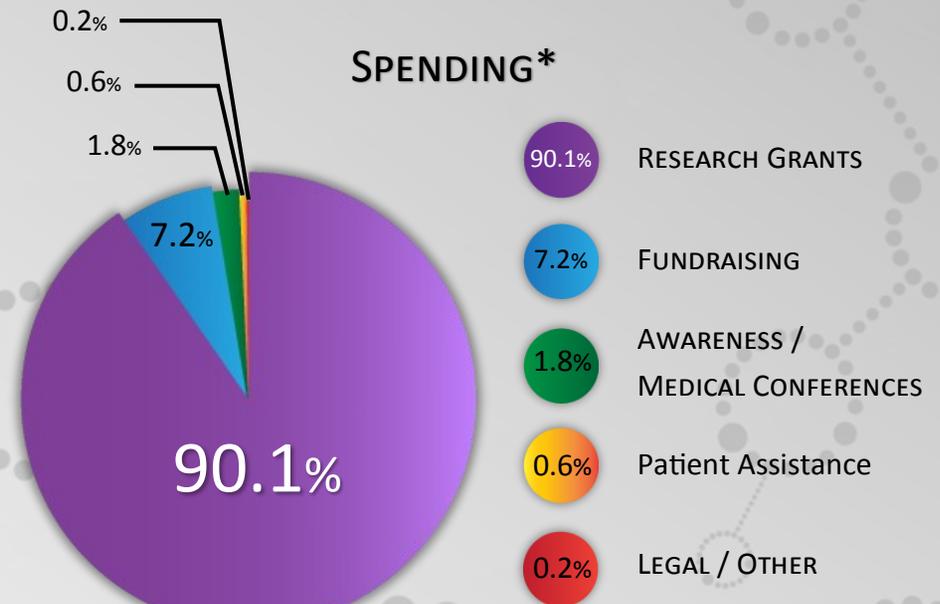
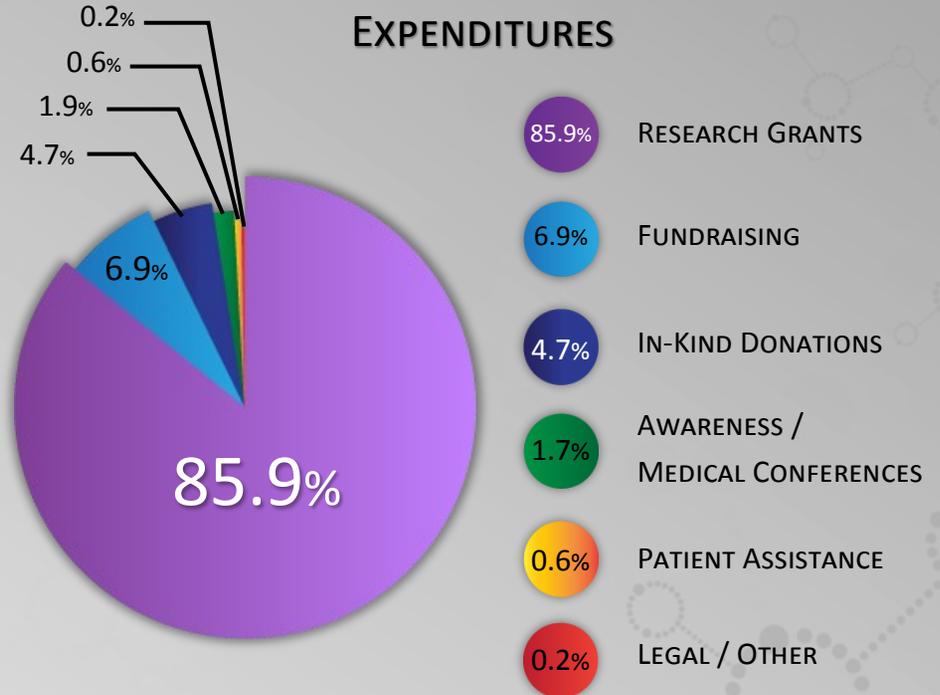
Raising and Investing Funds— Our Biggest Year Yet!

Most of our donations came from PCDH19 Families making personal donations and reaching out to family and friends to join our cause through fundraisers. Parents, Grandparents, Aunts, Uncles, siblings, friends and co-workers (motivated to provide a better future to our courageous children) hit the internet, and hit the streets to help us raise funds this year. We raised \$96,311 this year.

The Alliance would not be able to raise as much or invest as much directly into our programs without the support of our in-kind donors. In-kind donations were goods and services that were donated by generous supporters, such as all Alliance T-shirts, teddy bears, wristbands, wine for fundraisers, banners for conferences, almost all shipping costs (including shipping items to and from conferences), raffle items, gift baskets, printing services and much more.

With the guidance our Scientific Advisory Board, we awarded 3 research grants in 2015 and are excited to have the researchers update us all at our conference at UCSF in June 2016.

We continued to make a substantial impact with our Awareness, Education, and Advocacy programs by strategically leveraging funds and relying 100% on volunteers to travel, attend medical conferences and create awareness materials, forge relationships with industry partners and represent the PCDH19 patient population in larger groups and councils. Our volunteers pay their own flights, lodging and meals for working at medical conferences, regional events and Epilepsy Awareness Day at Disneyland. We cannot stress enough the return on investment and impact of the funds we raise, by being 100% volunteer run.



* net of in-kind donations \$5,445

Thank you

We are deeply grateful to all of our supporters.



Spirit of Progress Award: We recognized Joseph Sullivan MD for his dedication to diagnosing and treating PCDH19 Patients and for the immense time and commitment he has dedicated guiding the PCDH19 Alliance as Chair of our SAB.



Spirit of Service Award: We recognized Denise Fabio for being a committed volunteer and fundraiser from day one. Denise's daughter, Amanda, has PCDH19. Denise has dedicated her time and her heart to serving the Alliance and the entire PCDH19 Community.

Major Donors / Supporters

The Homewood Family Foundation	Kurt and Leslie Henkel
Karin and Bart Kilpatrick	David Dick
Julie Walters and Matt McManus	Robert Wells
Paras and Audrey Fancy	Bob and Kay McManus

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Volunteers / Fundraisers

Alison King	Karin and Bart Kilpatrick
Beth Bergstrom	Kate Ann
Bob Jessome	Kathy and Scott Tenchka
Coultas Family	Keigher Family
Daniel and Rachel Sexton	Kristen Sands
Denise Fabio	Kurt and Leslie Henkel
Gladys Rodriguez Chacon	LeAnn and Curt Shepard
Federico Cribiore	McManus Family
Haganey Family	Mikey's Ride (the whole team)
Heather Fryman	Sabrina Farrington
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