



2016 Annual Report



A cure is out there.
Help us find it.

Our Impact this year

2016 Year In Review

In 2016, our 4th year, we continued to learn and grow as an organization. With our mission to find better treatments and eventually 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 progress in all these important areas. Thanks to the generosity of our donors and fundraisers, and the work of our dedicated volunteers we awarded two research grants, held a successful International PCDH19 Professional and Family Conference, and were able to send two families to the PCDH19 Conference for a life-changing experience.

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

We believe research brings us hope, and it is by far our largest program here at the Alliance. With the guidance of our esteemed International Scientific Advisory Board, we are funding cutting edge PCDH19 research and 2016 yielded two incredibly exciting projects. We received several scientifically strong proposals, and wish we could have funded them all. Below are the two projects we funded for 2016-2017. We are looking forward to hearing Dr. Pasca and Dr. Thomas speak at our 2018 Research Conference.

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.



Dr. Sergiu Pasca - Using IPS cells to understand PCDH19-related encephalopathy

"PCDH19 is a severe disease characterized by onset of epileptic seizures in infancy, intellectual disability and autism. The condition is caused by mutations in the PCDH19 gene present on X chromosome. PCDH19 codes for the protein protocadherin-19. Protocadherins are cell adhesion molecules involved in establishing connections between neurons. Our preliminary findings suggest that protocadherin-19 regulates the trafficking of GABA(A)Rs to synapses. GABA(A)Rs mediate fast inhibitory transmission in the brain. If GABA(A)R presence at synapses is affected, the correct balance between inhibition and stimulation of neurons is upset, possibly giving rise to epilepsy and disorders of neurodevelopment. However details of PCDH19's function in brain are largely unknown. We propose to investigate the role of PCDH19 in mammalian neurons and neuronal circuits, by working both with cultured neurons and with animal models in which PCDH19 has been mutated in a subset of brain cells. Our study will help us better understanding how the mutation gives rise to dysfunction at the synapse, in the neuron, and in neuronal circuits, in order to guide the future development of treatments for the disease."



Paul Thomas PhD - Identifying the pathological mechanism of PCDH19 Epilepsy

"Changes in the PCDH19 gene cause epilepsy and, in some cases, intellectual disability. An unusual and poorly understood feature of PCDH19-associated epilepsy is that it only affects girls. To investigate the underlying cause of PCDH19 associated epilepsy, we have developed Pcdh19 mouse models that mimic the genetic changes that cause epilepsy in girls and allow us to identify neurons in which the PCDH19 gene is active. We have recently found that PCDH19 is active in a subset of neurons that are responsible for "dampening down" electrical activity in the brain. We have also shown that changes in the Pcdh19 gene in female mice affect neuron connections. The aim of this project is to further investigate these preliminary results through detailed analysis of brain development and function in our mouse models. We will also begin to translate our research findings into a clinical context by looking for subtle changes in brain structure in affected girls. These experiments will lead to greater understanding of how changes in PCDH19 cause epilepsy in girls and facilitate the development of new treatments."

Patient & Family Support

Patient Assistance

Besides the emotional and physical impact that PCDH19 has, the financial strain on a family dealing with PCDH19 Epilepsy can be devastating in itself. If we can help a child or adult obtain necessary medical equipment, therapy, and related devices that would otherwise be out of reach, we consider it a privilege.

Every-other year we also offer a travel grant to PCDH19 families who would otherwise not be able to attend our PCDH19 Research and Family Conference. This year we were able to send 3 families to the conference, however one family could not make it. The Keigher family is one of those families.

Providing Hope and Support— The Keigher Family

Receiving the travel grant was such an incredible gift. We received our daughters diagnosis in 2014, 8 years after her seizures began. We live in a small community and we did not know anyone else dealing with anything like what our daughter was going through. Finding the Alliance was a Godsend for me. Finally, people who knew exactly what we were going through. Finally, we were no longer completely alone. Even our daughter's Neurologist had never heard of this specific epilepsy. I immediately began connecting with other family members and was learning from all of the other parents posts about their children's seizures, medications, and behaviors.

We have a large family. Norah is our youngest of 5 daughters. When we heard about the conference, we wanted to go, but there was just no way we could afford for all of us to go. It would be too long of a drive for Norah and flying would have been completely out of the question. When I received the phone call from Julie Walters saying that we had received the travel grant I was beside myself! Our family was able to fly out to California from Illinois.

My husband and I were able to meet other mothers and fathers doing all they can for their children. My daughter was able to meet other girls who suffer from seizures just like her...and are strong and beautiful and brave, just like her. My other children were able to meet other siblings who could relate to what they have been through...being put second while seizures are happening, worrying about their little sister, etc. We are so grateful to have been chosen to receive the travel grant. Thank you to the donors who made this incredible trip possible for us.



Facebook Support Group



We continued to grow our Closed Facebook family support group—from 240 members to 312 members (representing roughly 170 patients). 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 PCDH19 Alliance. 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.

Education, Awareness, and Advocacy

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

This was our 4th year Exhibiting at the **American Epilepsy Society Conference**, this year in beautiful Houston, TX, where over 5000 medical professionals and researchers from around the world convened to exchange ideas and learn the latest information about epilepsy.

This year we had 4 parent volunteers representing the Alliance (thank you Leslie Henkel, Le Shepard, and Nicole Meador) educating medical professionals about the importance of a diagnosis, meeting with corporate partners, attending meetings, and reaching out to young researchers. We do some of our most important work at this meeting each year, and this year was no exception.



We raised awareness among those with epilepsy and advocates and physicians at our 3rd 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!

Over 2 years in the making, we continued to work closely with our friends and partners at Ambry Genetics and UCSF to bring *The Color Violet*, a documentary about PCDH19 Epilepsy, Research, Genetics and Hope to a screen near you. The film, *The Color Violet*, was screened at the American Epilepsy Conference in December and we are working on wider release in the near future. We are excited to share it with the world.

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 and the UK. We also held dinners, auctions, raffles, dine n' donate, cookies sales, and online fundraisers, raising awareness along the way.



PCDH19 Professional & Family Conference

This was our 2nd Biennial PCDH19 Professional and Family Conference, and was the highlight of the year for the Alliance Community. Researchers and families from 3 continents and several countries came together to share the latest data, share stories and make friendships that will surely last a lifetime. Once again the conference was streamed live online and allowed viewers from around the world to ask questions and listen to the answers to their PCDH19-related questions.

A heartfelt thank you to Joseph Sullivan MD for volunteering his time each year to Chair this amazing and life-changing conference for the Alliance. Dr. Sullivan, who is also the Chair of our Scientific Advisory Board volunteers a significant amount of his time organizing, planning and then hosting the conference and because of his dedication, the dedication of Tym Peters at UCSF, our Alliance Volunteers, and our generous partners at Ambry Genetics, Upsher-Smith, Marinus, Novartis, and Lundbeck, The PCDH19 Alliance has created a highly regarded research and family conference.



The Numbers

Raising and Investing Funds

As an organization that serves and represents a rare disease with the number of patients diagnosed in the hundreds, we are making a relatively large impact. Most of our donations came from PCDH19 Families making personal donations and reaching out to family and friends to join our cause through various fundraisers. Parents, Grandparents, Aunts, Uncles, siblings, friends and co-workers (motivated to provide a better future to our courageous children) raised their voices, raised funds and raised hope this year!

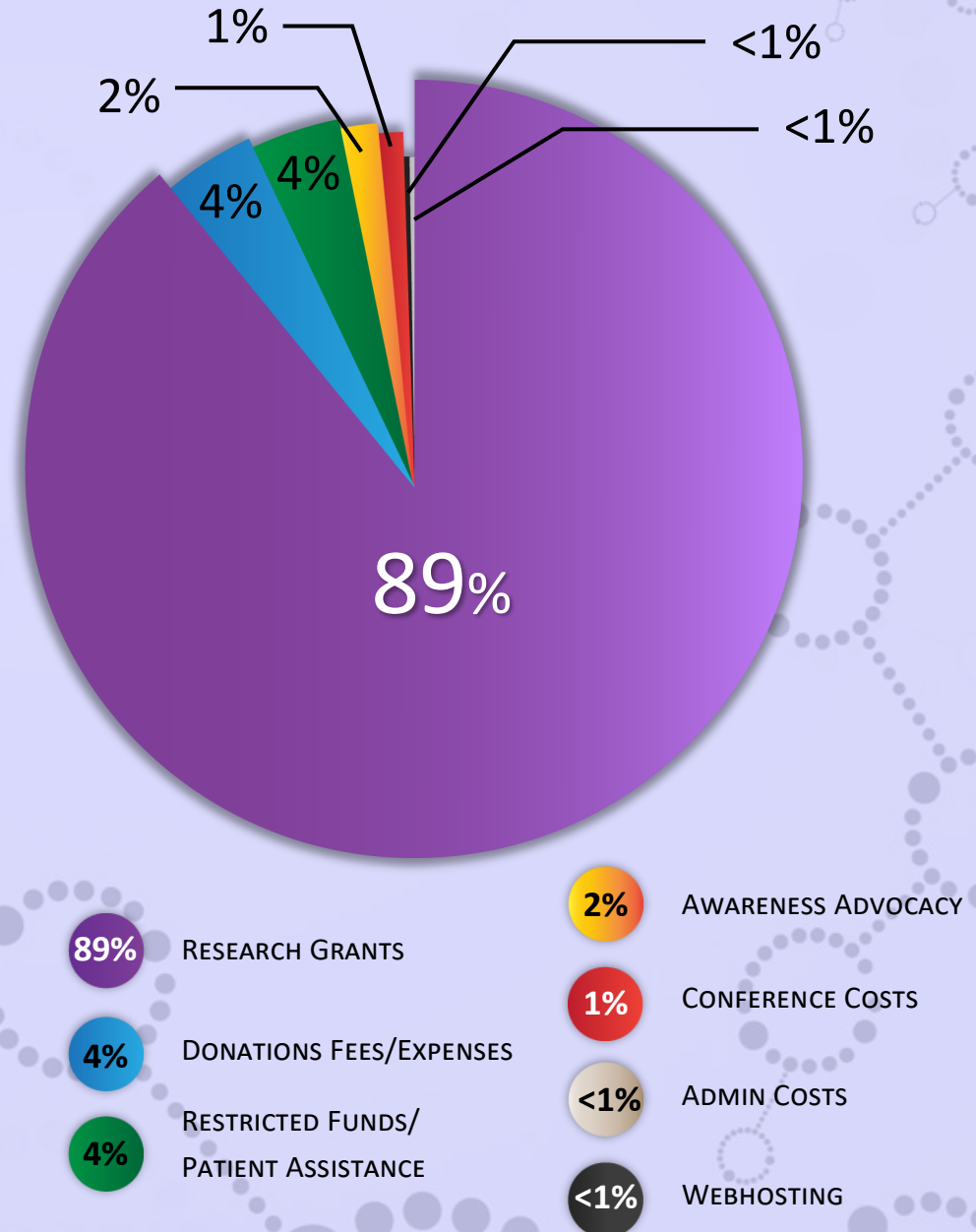
The Alliance would not be able to invest as much directly into our programs without the support of our in-kind donors. In-kind donations were goods and services that were donated by generous supporters, such as Production of "The Color Violet", Alliance T-shirts, teddy bears, wristbands, banners for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

With the guidance of our Scientific Advisory Board, we awarded 2 research grants in 2016 and are excited to have the researchers update us all at our Professional and Family Conference in June 2018.

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, create awareness and educational materials, forge relationships with industry partners and run our programs every day. 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.

We are inspired and grateful that you have joined us to make a real impact for individuals and families living with PCDH19 Epilepsy. We take stewardship of your hard earned funds very seriously. Here are the numbers:

EXPENDITURES



Thank you

We are deeply grateful to all of our supporters and we were honored to recognize Professor Jozef Gecz and April Haganey for special rewards.



Spirit of Progress Award

We recognized Professor Jozef Gecz with our Spirit of Progress Award for his unwavering dedication to understanding the mechanism of PCDH19 and finding an effective treatment. Since the PCDH19 gene was discovered in 2008, Professor Gecz has worked with an immense sense of urgency to find an effective treatment.



Spirit of Service Award

We recognized April Haganey for her service to the PCDH19 Alliance. While raising her own family, helping to run her family business, and dealing with the daily struggles of PCDH19, she has made an incredible impact in all areas of our mission from supporting others families, raising awareness, and fundraising.

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