

THE ALLIANCE



The PCDH19 Alliance was founded by three parents of girls affected by PCDH19 Epilepsy in an effort to support ongoing research into this debilitating disorder. The Alliance is a 100% volunteer- run organization.

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.



HOW TO HELP

(For more info go to PCDH19info.org)

Like our Facebook page

Follow us on FB, and we'll introduce you to some of the brave children who are our heroes. We'll also keep you up to date on current research and upcoming events, such as our biannual symposium.

Donate

Visit our webpage to make a one-time or recurring donation. Your gift will help fund research and support affected families.

Volunteer

Donations can take other forms than funds! Are you willing to donate skills or time to support the Alliance in changing lives? Connect with us through our website at www.pcdh19info.org and we'll put you to work!

Find our Facebook community online at www.facebook.com/Pcdh19

WWW.PCDH19INFO.ORG



Providing hope through

- RESEARCH
- ADVOCACY
- SUPPORT

Research into PCDH19 Epilepsy will directly improve the lives of children who are living with life-threatening seizures, developmental delays, and behavioral problems that interfere with their health and safety on a daily basis.

Researchers now estimate 1 in 10 girls with seizure onset before age 5 have PCDH19 Epilepsy.

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Thank you to our sponsors:



WHAT IS PCDH19 EPILEPSY?

PCDH19 is a genetic epilepsy caused by a mutation of the PCDH19 gene on the X chromosome, affecting mostly females. As this gene was not isolated until 2008, researchers are still working tirelessly to understand the connection between the gene and the wide spectrum of symptoms for which PCDH19 epilepsy is known. Males with the mutation typically do not present with epilepsy unless mosaicism is present. All males carrying the mutation will pass it on to 100% of their daughters and none of their sons. Women with the mutation have a 50% chance of passing the mutation to both their daughters and sons. Scientists have discovered some unaffected females (carriers of the gene but not affected) and are studying to learn what is protecting them from the disorder.



ABOUT SEIZURES

Seizure Types

The most common seizure types in PCDH19 are generalized tonic-clonic, tonic, clonic, complex partial, atypical absence, atonic drop, and myoclonic seizures. The seizures may be accompanied by rapid oxygen desaturation and cyanosis. For some, seizures do not stop on their own and require hospitalization.

Age and Onset

- Most consistent feature: seizure clusters over a period of days or weeks, with variable seizure-free intervals in between
- Seizures are often drug resistant and clusters difficult to stop
- Average onset age: 9 months (average range: between 3 months and 3 years)
- At first the seizures usually occur in the presence of a fever, later with no apparent trigger
- Sometimes loss of hearing or developmental skills occurs after seizure clusters



COMMON SYMPTOMS

Cognitive/Intellectual Outcomes

It is estimated that about 70% of PCDH19 patients have intellectual disability ranging from mild to severe. The course of development usually follows one of three paths:

- Normal development and intellectual ability from birth without regression
- Normal development from infancy, but with regression after seizure onset
- Delayed development from birth, remains delayed through adulthood.

Ictal Apnea

Many individuals stop breathing during their seizures and have rapid and sometimes prolonged oxygen desaturation.

COMMON SYMPTOMS CONT'D.

Psychiatric

Common diagnoses include: Autism Spectrum Disorder or autistic features (estimated 60% affected), behavioral disorders, aggression, ADHD, anxiety, and OCD. In adolescents and adults, depression, bipolar, schizophrenia, postictal psychosis and other mental illness have been reported.

Other Symptoms

- Sleep disorders, difficulty falling and/or staying asleep
- Fine and gross motor skill deficiencies
- Language delay or non-verbal
- Sensory processing and integration issues
- Dysautonomia (malfunction of the autonomic nervous system)



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The PCDH19 Alliance is registered as a tax-exempt public charity by the Internal Revenue Service under Section 501(c)(3). Our tax identification number is 45-4020102

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