THE ALLIANCE

The PCDH19 Alliance was started by three parents of girls affected by PCDH19 Female 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 Female 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

There are several ways to contribute to the PCDH19 Alliance. Please consider donating to the Alliance through our website in order to help fund ongoing research into the gene PCDH19 and potential treatments.

We also have a variety of PCDH19 Alliance merchandise available for purchase. Please visit our website and click on the "How to help" tab for lots of great ideas.

If you would like to get more involved, you can also host an event to help raise money for the Alliance. We offer guidelines and suggestions for how to organize a fundraiser in support of the Alliance.

Have some extra time? We are always looking for volunteers to help in various capacities.

If you or someone you love has been diagnosed with a PCDH19 gene mutation, please join our mailing list through our website in order to keep in touch with important updates, potential clinical trials, and research opportunities.

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

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PCDH19 alliance™

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SUPPORTING RESEARCH, AWARENESS, FAMILIES, AND HOPE

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

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GeneDx DNA DIAGNOSTIC EXPERTS
Support for PCDH19 Alliance and printing of this brochure provided by GeneDx
www.genedx.com

WHAT IS PCDH19 FEMALE EPILEPSY?

PCDH19 is a condition with a wide spectrum of severity in seizures, cognitive abilities and outcomes, and other symptoms, all of which can be seen in females who carry the gene by a mutation of the PCDH19 gene on the x chromosome. Males with the mutation, who will be largely unaffected, will pass the mutation on to 100% of their daughters and none of their sons. Women with the mutation have a 50% chance of passing it to their daughters and sons. Recently, scientists have discovered some unaffected females and are studying to learn what is protecting them from the disorder.

ABOUT SEIZURES

Seizure Types

The most common seizure types for 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.

COMMON SYMPTOMS

Age and Onset of Seizure Clusters

- *Most consistent feature: seizure clusters which can last for days or weeks with variable periods of seizure freedom in between*
- *Seizures are often drug resistant.*
- *Seizures are often difficult to control.*
- *Onset of the initial seizures usually occurs between 3 months and 3 years of age, with an average age of onset of 9 months.*
- *At first, the seizures usually occur in the setting of a fever, later they come with no known trigger.*
- *Sometimes loss of hearing and loss of skills are experienced after seizure clusters*

Cognitive / Intellectual

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

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

Ictal Apnea

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

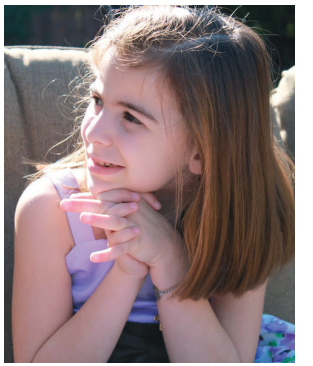
COMMON SYMPTOMS CONT.

Psychiatric

Autism Spectrum Disorder or autistic features (estimated at 60%), behavioral problems, aggression, ADHD, anxiety, and OCD are common. In adolescents and adults, depression, bipolar, schizophrenia, psychosis and other mental illness have been reported.

Other Symptoms

- *Sleep disturbances, trouble falling and/or staying asleep*
- *Hypotonia—decreased muscle tone*
- *Fine and gross motor deficits*
- *Language delay or non-verbal*
- *Sensory integration issues*
- *Dysautonomia—malfunction of the autonomic nervous system*
- *Delayed tooth eruption seems common*



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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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