Some information about epilepsy caused by mutations in the PCDH19 gene:

PCDH19 is a protocadherin gene. Protocadherins are calcium-dependent cell adhesion proteins. The formal name of this gene is: Protocadherin 19. This protein is important for communication between nerve cells.

What medical conditions are associated with mutations in this gene?

- Early Infantile Epileptic Encephalopathy Type 9 (EIEE9) – previously known as epilepsy and mental retardation restricted to females (EFMR)

What are the some of the features of PCDH19 epilepsy (EIEE9)?

- Delayed development, variable severity, from birth in some patients
- Developmental regression in about 50% of patients
- Normal development in some patients
- Seizures, convulsive
- Seizures, tonic-clonic
- Seizures, partial
- Seizures, absence
- Seizures, atonic
- Seizures, myoclonic
- Status epilepticus
- Intellectual disability is variable
- Seizure onset at a mean of 14 months (range 6 to 36 months)
- Some patients stop having seizures at a mean of 12 years
- Carrier males are unaffected except for psychiatric/behavioral abnormalities
- Mutations in this gene are inherited in an X-lined manner and may occur de novo (not found in either parent)

Who can I contact for more information about PCDH19 epilepsy?

Please contact these groups for more information about PCDH19 epilepsy (listed in alphabetical order):

- Cute Syndrome Foundation
- PCDH19 Alliance

If you know another group that can be added to this list, please email EGI@CUREepilepsy.org.
References


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