

# PCDH19 related epilepsy

## Patient journey



Insieme per la Ricerca  
PCDH19 ETS



### First symptom

**Timeline:** 3-60 months old (median age 8-10 months).

### Clinical Presentation / Symptoms

Focal seizures occurring mainly in clusters, sometimes over a period of days or weeks.  
High sensitivity to fever or illness which are frequent seizure triggers.

### Identify Patient Needs

- Parents need reassurance (the first seizure may have been traumatic for parents).
- Parents need basic information about epilepsy, seizures, and clusters.
- Parents need to know what to do if the seizure happens again.
- If the seizure was fever-related, parents need to know how to proceed during fever episodes in a safe way and/or in case of vaccination (if fever occurs).

### Ideal Outcome / support

- The worries of the parents are taken seriously and reassurance is offered.
- Parents receive instructions to handle a seizure, if it happens again, and trigger factors.
- Patient is referred to a child neurologist.

### Diagnosis

**Timeline:** After the first seizures up to 2 years.

### Clinical Presentation / Symptoms

Child will often present with more seizures in clusters and may have different seizure types like focal or generalised tonic-clonic seizures with or without trigger factors. Other seizure types such as tonic, atonic, myoclonic and atypical absence may also occur. Fever-related seizures, long clusters of seizures, and status epilepticus are possible.

Development is different from child to child, but many have developmental delay and cognitive impairment. Intellectual disability (from mild to severe), autistic traits, speech, motor and behaviour problems, hypotonia and psychiatric disorders are also seen.

### Identify Patient Needs

- Parents need a clear explanation of the diagnosis and prognosis with psychological support.
- Parents need to be offered a genetic test.
- Parents need to understand that development varies from child to child and with the age.
- Parents need to know whether there are ongoing clinical trials for PCDH19 related epilepsy in their countries.
- Parents need to know what social help is available from their government.

### Ideal Outcome / support

- Family is explained that a prognosis is difficult to give.
- Genetic counselling is provided for explaining the cause and reoccurrence.
- Parents are offered a professional support for coping with diagnosis and are referred to local parent support groups/patient associations.
- Parents receive clear instructions about emergency protocol and about ongoing clinical trials.
- Parents are informed that education and rehabilitation are extremely important for child's development and should be very closely monitored.
- Parents receive defined rehabilitation pathway (developmental assessments, speech therapy, postural rehabilitation, behavioural therapy).
- Parents receive information about social help available in their country.

### Treatment

**Timeline:** Lifelong.

### Clinical Presentation / symptoms

PCDH19 related epilepsy is drug resistant, although in some patients, seizures tend to decrease after puberty.  
Treatment should aim at best possible seizure control with minimal side effects.  
Special attention is necessary for trigger factors and seizure preventive measures.  
Paramedic treatment may be needed to optimize physical and cognitive development.

### Identify Patient Needs

- Parents need support and respite care from professional team with multidisciplinary approach.
- Parents need information about the used medication and their side effects, and access to potential trials/new treatment options.
- Parents need advice about dealing with trigger factors.
- Parents need advice about non-epileptic symptoms.
- Parents need education/school advice.

### Ideal Outcome / support

- Regular counselling with professionals is offered.
- Up to date information is available to families at any time.
- Parents receive help finding a school/daycare and respite care.
- Family/epilepsy nurse is offered to educate family for managing seizures and trigger factors.
- Defined rehabilitation plan (developmental assessments, speech therapy, postural rehabilitation, behavioural therapy).

### Follow-up

**Timeline:** Up to 16-18 years old.

### Clinical Presentation / symptoms:

Other issues may occur, like Intellectual disability, motor and speech impairment, and behavioural and psychiatric issues.

### Identify Patient Needs

- Parents need evidence-based advice and expertise when these additional symptoms occur.
- Parents need emotional support.
- Parents need evidence of enabling needs (psychomotricity, speech therapy, behavioural therapy).

### Ideal Outcome / support

- Monitoring of non-epileptic symptoms and offering treatment where possible.
- Developing standards for quality of life of adult patients.
- Availability of home care and/or institutional care of excellent quality.
- Defined rehabilitation pathway (developmental assessments, speech therapy, postural rehabilitation, behavioural therapy).
- Assistance programs in context of education (e. g. schools).

### Adult follow-up

**Timeline:** 16-18 years old and up.

### Clinical Presentation / symptoms:

Convulsive seizures may be present. Temperature-sensitivity usually decreases. Comorbidities continue, in particular behavioural and psychiatric issues. Late-onset psychosis may appear. Lack of coordination between paediatric and adult neurologists may entail an insufficient support for patient and family.

### Identify Patient Needs

- Parents need advice and support in transition to adulthood.
- Parents need adequate adult management with multidisciplinary team.
- Parents need continuous monitoring of seizures and psychiatric and behavioural issues.

### Ideal Outcome / support

- A process for transition from paediatric to adult physician should be put in place.
- Defined rehabilitation path for skill maintenance.
- Occupational therapy.
- Taking timely actions for possible aggravation of the behavioural, social and cognitive issue.