PCH2cure
Connecting Families and Science

Digital Consultation: “Meet the expert” about Epilepsy

Q&A

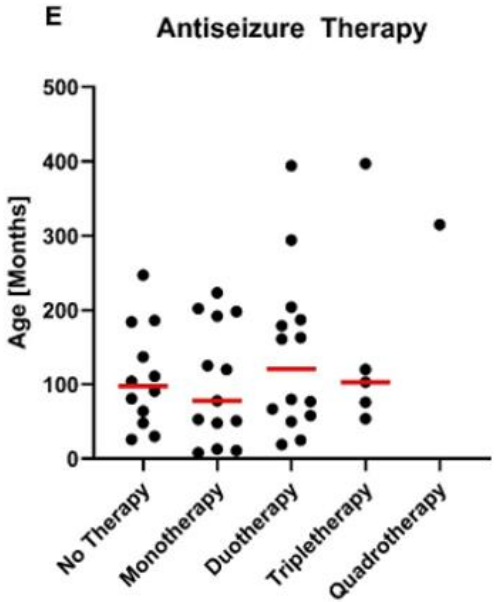
Disclaimer

The PCH2 treatment options listed here are based on data from the 2023 Natural History Study on PCH2 and personal experience from parents. The answers make no claim to be exhaustive and represent neither a specific recommendation nor an endorsement of the drugs or PCH2 treatment options mentioned. It is a compilation of measures that have been tried in the past and is intended for informative purposes only. PCH2cure assumes no liability in this respect.

Q&A Session

Are other children diagnosed with PCH type 2 also often resistant to antiepileptic drugs?

- Some children with PCH2 do not require medication.
- Similarly, there are children who do well on monotherapy; these tend to be younger.
- However, many children require **two or more medications**, which means that their epilepsy is difficult to treat.
- Over time, **changes in medication are often necessary, but there are also periods of stability**.



Q&A Session

Do you think the SEEG method can be helpful for children diagnosed with PCH type 2?

SEEG = Stereoelectroencephalography – what is it? EEG recording using deep electrodes that are temporarily implanted via a minimally invasive neurosurgical procedure.

- It is typically performed as part of the evaluation for epilepsy surgery to determine whether a patient with drug-resistant epilepsy is a candidate for the procedure.
- More commonly used, for example, after strokes followed by epilepsy

→ Usually not recommended for patients with PCH2A, as there is often more than one circumscribed epileptogenic region.

Q&A Session

My child is currently taking four antiepileptic drugs (Etil, Keppra, Losmorid, and Frisium). However, focal seizures still occur upon waking, due to focal epilepsy with foci in both hemispheres. Do you consider this treatment appropriate, or would you recommend an adjustment?

- Anticonvulsant medications: Keppra (active ingredient: levetiracetam), Losmorid (active ingredient: lacosamide), and Frisium (active ingredient: clobazam)
- Medication for low blood pressure: Etil (active ingredient: etilefrine)
- General considerations regarding this issue:
  1. Key question regarding treatment goal: Are the seizures distressing or dangerous?
  2. Has the dosage already been maximized (e.g., LEV up to 50 mg/kg, LCM up to 10 mg/kg)?
  3. Optimize dosing intervals?
  4. Consider a different dosing schedule or additional doses?

Q&A Session

How can you tell the difference between epileptic seizures and dystonic attacks in everyday life?

- During a dystonic attack:
  - the children are bent into a C-shape and
  - make sweeping, alternating movements that vary greatly
  - the child is awake and whining
- During an epileptic seizure:
  - the child makes repetitive, monotonous movements
  - the child does not respond

→ It can be difficult to tell the difference in individual cases, especially at the beginning.

→ Recommendation: It is best to film the child during a dystonic episode or an epileptic seizure and show the video to the neurologist.

Q&A Session

When do epileptic seizures occur most frequently? (Time of day? Situations?)

- Varies—but **may recur** in individual children
- Possible triggering factors, e.g., **fever/ infection, lack of sleep, falling asleep/ waking up**
- Not relevant for PCH2A: flashes of light, falling blood alcohol levels