

Digital Consultation: “Meet the expert”

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

Questions we have already received

Is gut innervation being studied? Seems like many with PCH2A have dysmotility symptoms. Treatment focusing on vagal nerve “issues”?

- Yes, patients with PCH2A have dysmotility symptoms. We’re trying to start studying gut innervation in PCH2A, but to do this, we’re in need of full-thickness biopsies of the gut. Full-thickness biopsies can be taken only during abdominal surgery.
- One of our study patients had a vagus-nerve-stimulator. Parents reported a positive effect on epilepsy but not on gastrointestinal symptoms.

Q & A session

Questions we have already received

Also wondering if research has looked at the great variability in symptoms - why are some people able to live with relatively minor symptoms (no seizures, able to eat orally, no respiratory concerns, minor effects of dyskinesia, etc) while others are greatly affected (multiple seizure types, ventilation required, tube fed, hospitalization required for storming, etc) with an extremely short life?)

- A clinical variability can also be found in other neurogenetic conditions despite identical pathogenic variants, and might be caused by other modifying genetic alterations, which are not yet identified (and are not evaluated in routine genetic diagnostics). Epigenetic and environmental conditions might also play a disease-modifying role.

Q & A session

Questions we have already received

My child is 9, it eats peptamen + Fleur Alpine (porridge as food thickener), but she digests this food for 3,5 hours, how can I speed up food digestion (motilium and trimebutine dont help)?

- It is difficult for us to answer such specific questions for individual cases
- Peptamen is tube food, so we suppose Fleur Alpine is administered orally and Peptamen via tube
- Average residence time in the stomach for food is 1-3 hours; we assume the digestion time mentioned refers to the stomach residue
- The name of the active ingredient of Motilium is Domperidone (a prokinetic medication, meaning it drives digestion forward): in our study 7 children received Domperidone as a treatment for reflux and vomiting; in 4 out of 7 patients, the medication was stopped in the further course
- MCP (Metoclopramide): usually not a first line medication for children due to adverse events including extrapyramidal motoric disorder
- Trimebutine is an antispasmodic medication usually used in irritable bowel syndrome; as far as we know it is not available in Germany, so we have no experience in our German patients with PCH2A
- Input from other parents: What we usually do in case of stomach residue is to change frequency, amount of nutrition, and try out another formula, before changing medication (one parameter change at a time)
- If intestinal motility disorder is present in patients with PCH2A, other therapeutic options could include Prucalopride (used in PCH2A with limited experience)

Q & A session

New questions

A 16-year-old PCH2 patient with the CASK mutation presents with less symptoms than PCH2 children without CASK mutation. The patient is able to eat by herself, shows no digestive symptoms, has fine motor control and walks freely. Is there any potential to use her data, i.e. MRI, health records to compare with other PCH2 patients?

- Yes, there is a CASK research group based in UK, who can be contacted:
<https://caskresearch.org/cask-gene-disorders/>

Q & A session

New questions

What can be done about very thick mucus and problems with coughing/ vomiting?

- Usage of a cough assist
- Inhalation of NaCl
- In case of hypersalivation (present in many patients due to dysphagia): Glycopyrronium or Tiotropium as drug options or botox injections in the salivary glands
- Rule out any additional diseases, such as cystic fibrosis
- In our study, 6 patients with PCH2A required trachostoma due to respiratory problems

Q & A session

New questions

Will it be possible to donate in different currencies via the pch2cure.org website?

- Yes, in the near future we will implement the possibility to donate in a wider range of currencies. So far it is only possible to donate via PayPal and SEPA Lastschrift.