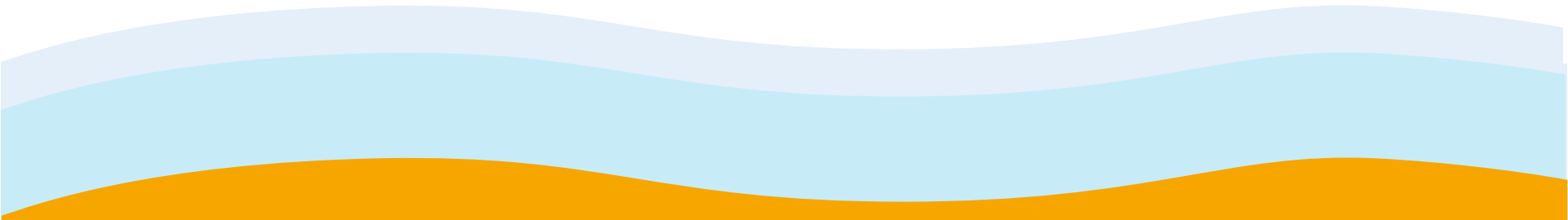


Online consultation hour: “Meet the expert”

Nutrition and gastrointestinal problems in PCH2

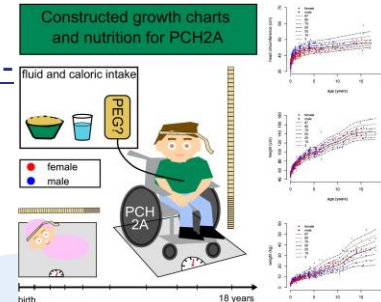
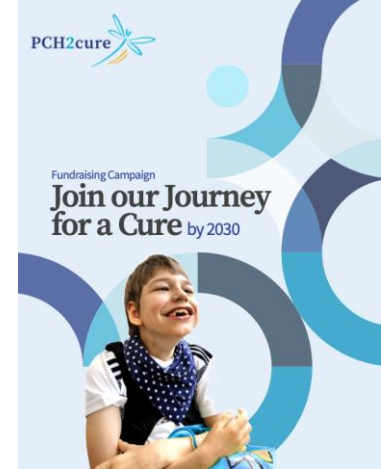
Nov 10th, 2025



1. News from PCH2cure
2. Nutrition and gastrointestinal problems in PCH2
3. Q&A session

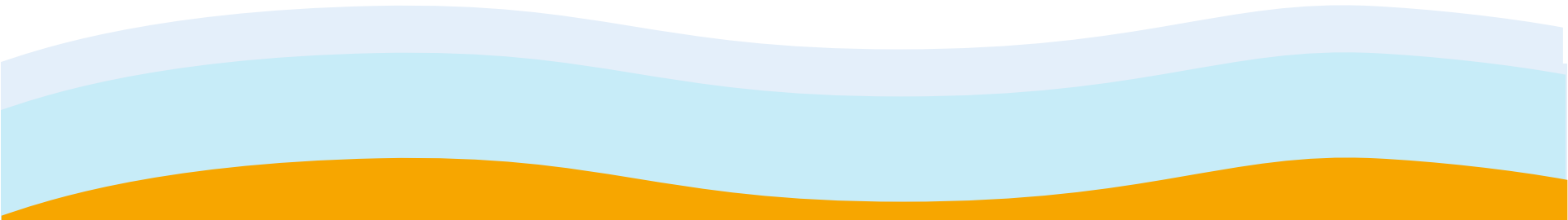
Status Update Q4/2025

- PCH2cure Foundation gGmbH: Fundraising brochure and Scientific Advisory Board to be completed
- Internal PCH2cure project-meeting in September
- New „Life Hacks“ and additional content on PCH2cure.org
- New publication:
 - Constructed growth charts and nutrition for pontocerebellar hypoplasia type 2A (Kuhn et al. 2025)
→ <https://www.pch2cure.org/en/research/disease-specific-percentiles-can-help-to-monitor-the-individual-development-of-a-child-with-pch2a/>



Nutrition and gastrointestinal problems in PCH2

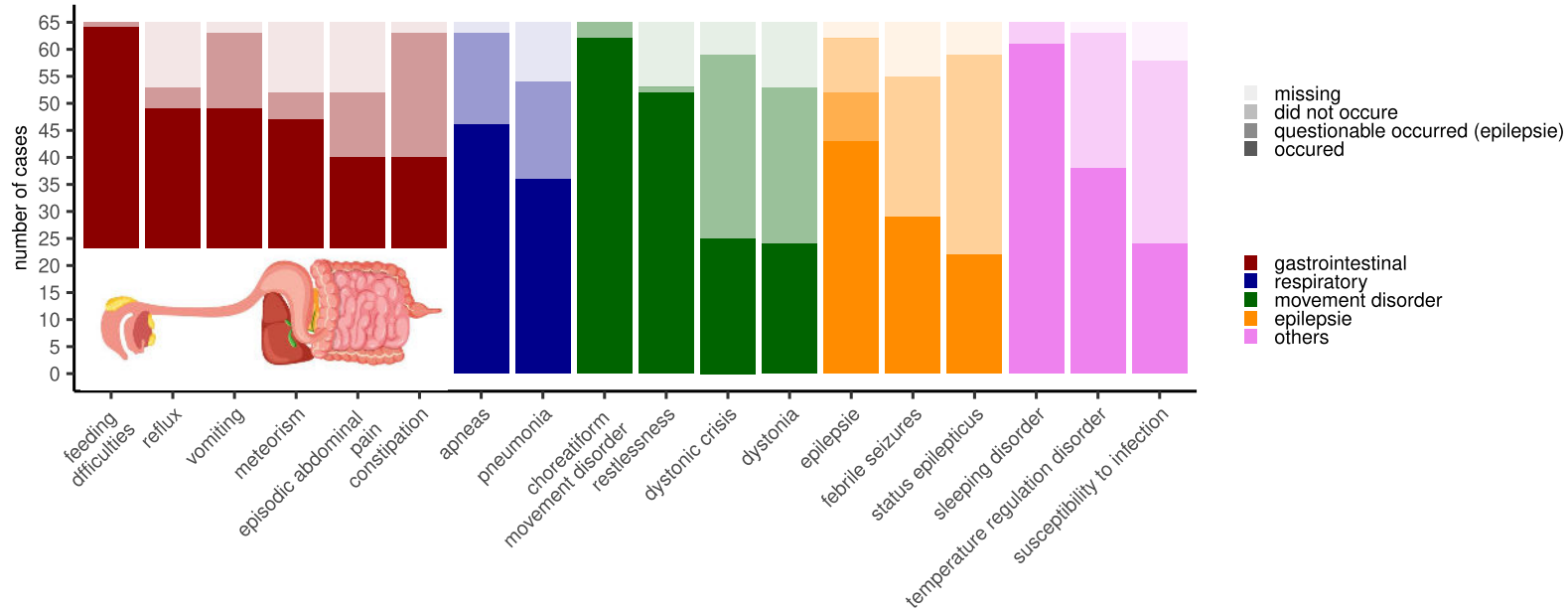
Alice Kuhn (Freiburg), PD Dr. Wibke Janzarik (Freiburg), Dr. Andrea Bevot
(Tübingen)



1. Data from the Natural History Study on PCH2A: Overview
2. „Feeding difficulties“
3. Tube feeding
4. Reflux and vomiting
5. „Flatulence“ und cramp-like abdominal pain
6. Constipation and laxatives
7. Prepared questions
8. Open question and answer session



1. Data from the Natural History Study on PCH2A: Overview





2. „Feeding difficulties“ = dysphagia

What exactly is it?

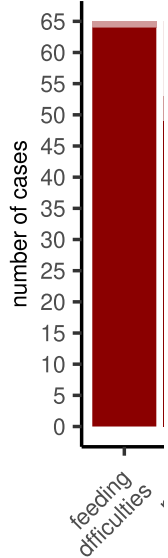
Choking, coughing while drinking/eating, uncoordinated swallowing, meals lasting longer than 30 minutes, tube feeding

When does this occur in children with PCH2?

Usually in the first months of life, every day, improves in individual cases, but does not disappear

Why do children with PCH2 have a swallowing disorder?

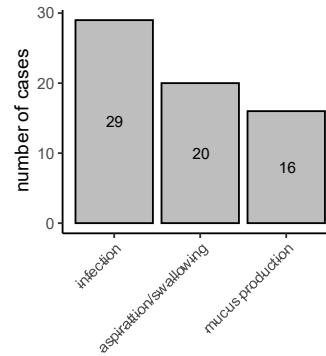
Chewing is difficult, coordination of the many functions involved in swallowing is impaired



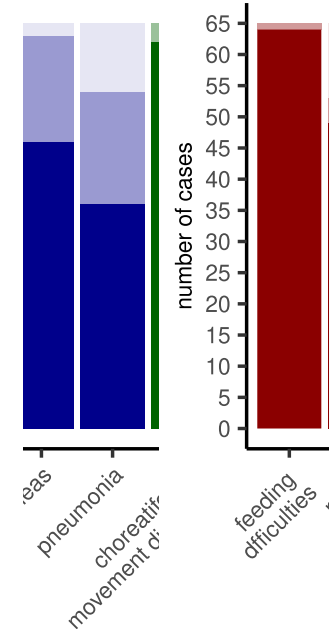
2. „Feeding difficulties“ = dysphagia

Is this a problem?

*Risk of aspiration → can lead to pneumonia,
risk of food refusal and development of feeding disorders,
strain on the family situation*



Causes of pneumonia (n=36)





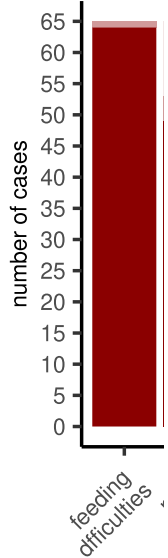
2. „Feeding difficulties“ = dysphagia

What can be done?

physiotherapy, logopaedics, consistency optimization, diagnostics, tube feeding

What should be considered?

- *trained logopaedics (does not have to be Castillo-Morales)*
- *physiotherapy to train head control*
- *consistency optimization: what works can vary from child to child*
- *use of eating and drinking aids (e.g., bottles, sippy cups)*
- *positioning: in the case of reflux, slightly elevated rather than completely seated*

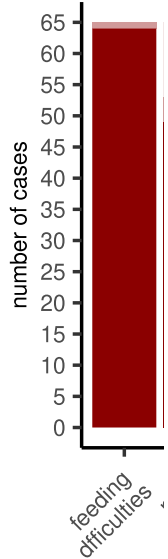


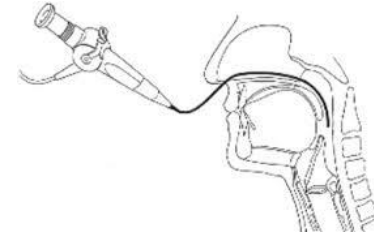


2. „Feeding difficulties“ = dysphagia

When should further diagnostics be considered?

1. *Does your child frequently suffer from severe respiratory infections or pneumonia?*
2. *Does your child struggle to breathe or cough when eating or drinking?*
3. *Do you feel that food gets stuck in your child's throat?*
4. *Does a meal take longer than 30 minutes?*
5. *Is the food adapted to make chewing or swallowing easier?*
6. *Are there any dental problems?*
7. *Does your child lose a lot of saliva?*
8. *Does your child's voice sound different during or after eating?*
9. *Does your child's voice sound increasingly hoarse throughout the day?*

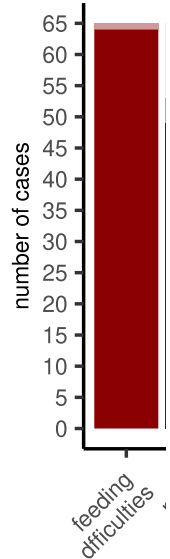




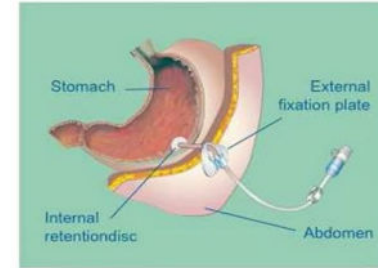
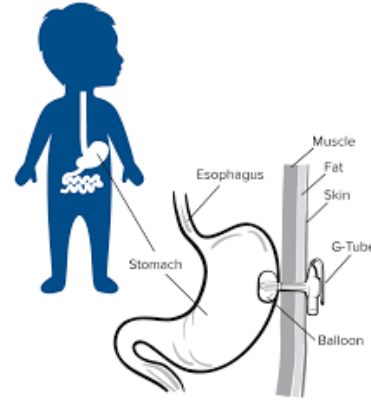
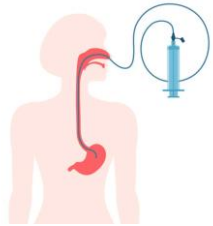
2. „Feeding difficulties“ = dysphagia

What types of diagnostics are available?

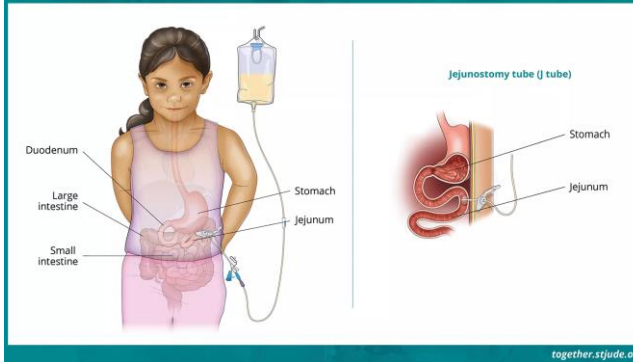
	<i>FEES = Fiberoptic endoscopic evaluation of swallowing: the swallowing process is “filmed.”</i>	<i>Video fluoroscopy: Contrast medium is swallowed and X-rayed from the side.</i>	<i>chest X-ray</i>
	• <i>no anaesthesia or sedation necessary</i> • <i>you can try out different consistencies and positions</i> • <i>even if the child cries or shows no reaction, you still get an impression</i>	• <i>no anesthesia or sedation required</i> • <i>non-invasive</i>	• <i>acute event</i> • <i>non-invasive</i>
	• <i>unpleasant</i> • <i>child must be restrained</i>	• <i>radiation exposure</i> • <i>contrast agents taste disgusting</i>	• <i>not a good diagnostic tool for repeated aspirations</i>



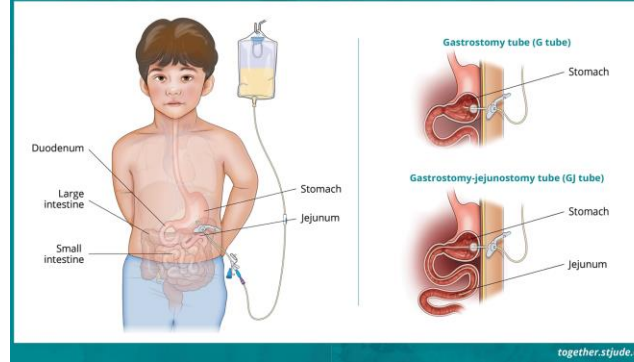
3. Feeding tube



Jejunostomy Tube (J tube)



Gastrostomy Tube (G tube) and Gastrostomy-jejunostomy Tube (GJ tube)





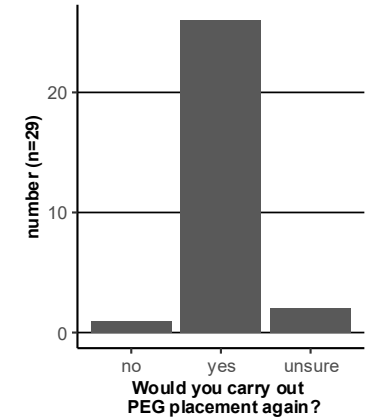
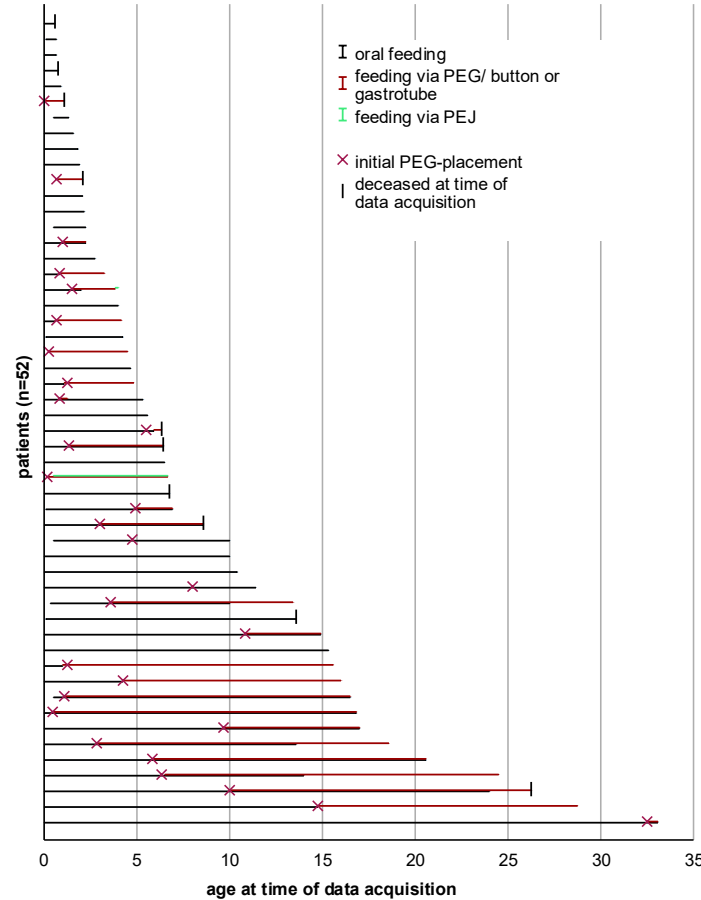
3. Feeding tube

PEG (n=29)

- Easier food and fluid intake → relief during mealtimes (11)
- Easier administration of medication (6)
- Life-sustaining (4)
- Fewer gastrointestinal problems (3)
- Fewer infections (2)



- Endoscopic placement of PEG not the right choice (1)
- Initially feels like a step backwards (1)
- Post-operative complications (1)
- Oral feeding no longer possible (1)
- Life extension from a certain point onwards (1)
- More GER (1)



PEG does not exclude oral food intake!

3. Feeding tube

Tube feeding

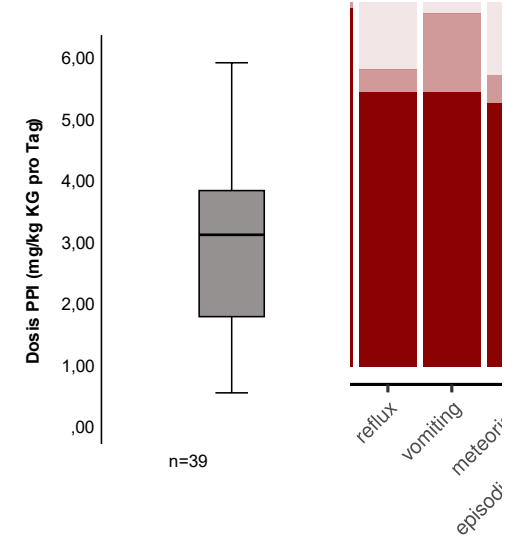
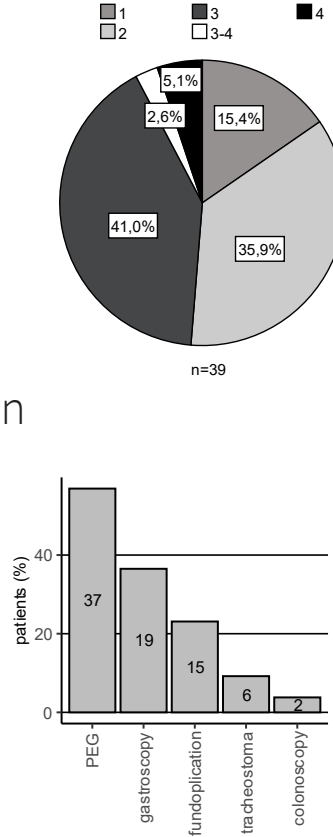
- Commercial tube feed
 - Isocaloric or high-calorie
- Homemade tube feed
- Medication: consider whether they can be dissolved or crushed





4. Reflux and vomiting

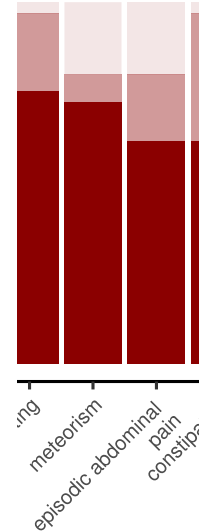
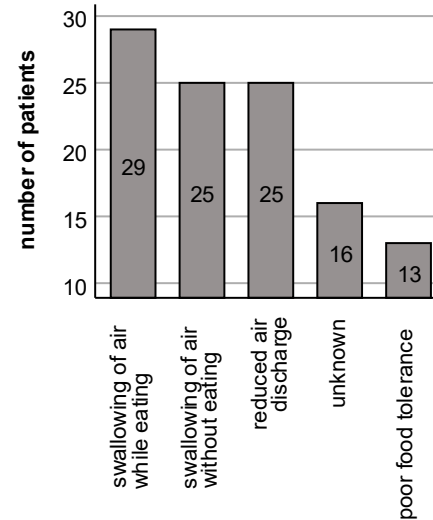
- Reflux as a common problem
- Vomiting tends to improve with age
- Treatment: high-dose PPI, fundoplication
- Fundoplication (n=11)
 - Less reflux and vomiting (8)
 - Improvement of QoL (6)
 - Less infections (2)





5. “Flatulence” and cramp-like abdominal pain

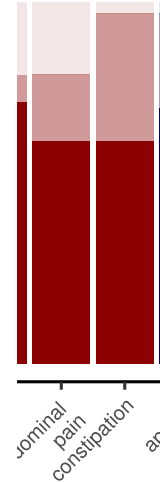
- Frequent, high level of suffering
- No changes in the intestine found so far
- Measures:
 - Defoaming agent: e.g., simethicone
 - Buscopan
 - Regular laxatives
 - Choose foods individually
 - Check for intolerances





6. Constipation and laxatives

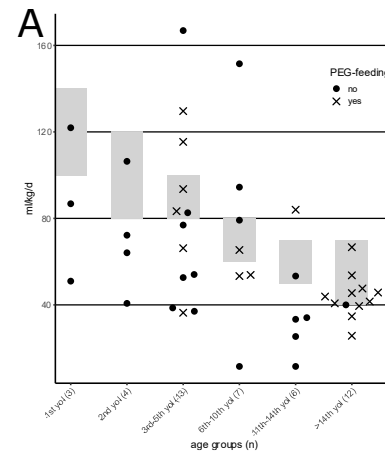
- Not classic constipation but uncoordinated defecation
- Frequent rectal evacuation
 - Difficult to ingest such large amounts orally
 - Bowel movements often only after rectal stimulation



7. Prepared questions

Which foods slow down bowel activity?

- *This can vary from person to person*
- *Large quantities of cow's milk should be avoided, as they often lead to constipation*
- *Low fluid intake: a common problem in children with severe multiple disabilities*
- *High-fat foods*
- *Foods with a high tannin content (unripe bananas, black tea)*
- *Foods with a high sugar content*
- *High saturated fat content (especially animal products)*



7. Prepared questions

Can food in puree form be digested faster than chunky food (our daughter eats pasta and bread, for example), or is there no difference?

- *The smaller the food is before it is eaten, the faster it can be digested*
- *The “speed” of digestion varies greatly from person to person and depends heavily on the food*
- *It depends on how the child is “feeling”*

7. Prepared questions

Several small meals or a few large ones—what is the medical opinion on this for our children?

- *In cases of recurrent vomiting and severe reflux, generally speaking, many small portions are better, BUT*
- *it must suit you, your child, and your everyday routine.*
- *Caloric intake should be based on the age-appropriate norm—if in doubt, consult a nutritionist or have a blood test done: restless children tend to have higher caloric requirements, while calm children tend to have slightly lower requirements.*

Thank you for your attention!

Platzhalter für Adresse

Gefördert von

PCH-Familie e.V.
PONTocerebelläre Hypoplasie



Eva Luise
und Horst Köhler
Stiftung



8. Open question and answer session

Are there any further questions?

8. More Questions

- How long does mashed food take to digest in children with PCH?
- What should the interval between meals be? - for 1.5-year-old child and 18-year-old
- How much food should be eaten at once for 1.5-year-old child?
- My younger daughter doesn't have an appetite at all. She can go up to 6 hours between meals and feel fine. Could this mean that her food doesn't have enough time to digest and she doesn't feel hungry?