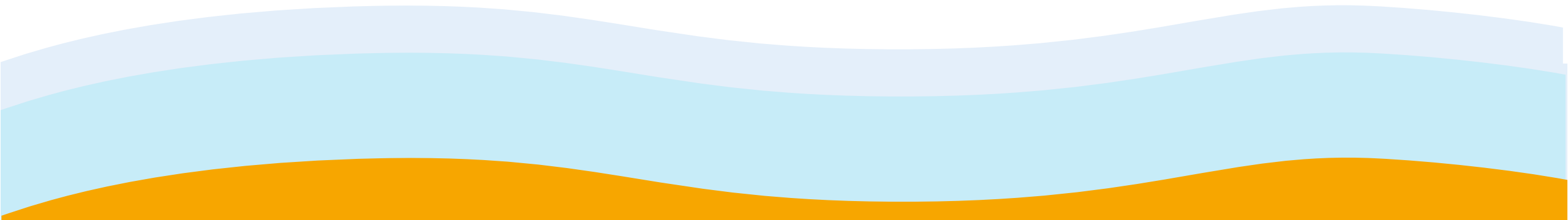


PCH2cure Online Consultation

Infections in PCH2

August 12, 2026



1. What's new at PCH2cure?
2. Specialist Subject: "Infections in PCH2"
3. Open Q&A Session

Status Update

- PCH2cure Foundation gGmbH: Fundraising from major donors and foundations in progress
- Translation of the Patient Brochure into Polish and Italian
- PCH2cure Research Symposium at the Karlsruhe Institute of Technology in October 2026
- Good progress on different studies: quality of life, restlessness in PCH2, MRI/PCH2 checkups. Initial results expected in fall.
- New publications:
 - Doctoral dissertation on PCH2A: [“Gastrointestinal Symptoms, Nutrition, and Growth in Pontocerebellar Hypoplasia Type 2A”](#) by **Alice Kuhn**.
 - [“Diagnostic Clues and Pitfalls in Pontocerebellar Hypoplasia Type 2A”](#) by **Herrmann et al.**

Storia naturale (decorso ed
evoluzione nel tempo)
dell'ipoplasia
pontocerebellare di tipo 2

Linee guida
per i genitori, familiari e caregiver



Historia naturalnego
przebiegu hipoplazji
mostowo-mózdkowej
typu 2

Przewodnik dla rodziców
i zainteresowanych osób





Infections in PCH2

Diagnostic and Therapeutic Challenges

Andrea Bevot, Neuropediatrics



**Klinik für Kinder-
und Jugendmedizin**
Universitätsklinikum
Tübingen

What is Known About Infections and PCH2?

1. Nothing!



What is Known

Despite advances in diagnosis, treatment, and care in recent years, patients with rare diseases worldwide have higher mortality rates and lower survival rates compared to the general population, with specific differences based on gender, age, and disease group.

Mazzucato M, Visonà Dalla Pozza L, Minichiello C, Toto E, Vianello A, Facchin P. Estimating mortality in rare diseases using a population-based registry, 2002 through 2019. Orphanet J Rare Dis. 2023 Nov 17;18(1):362. doi: 10.1186/s13023-023-02944-7. Erratum in: Orphanet J Rare Dis. 2024 Feb 6;19(1):43. doi: 10.1186/s13023-024-03051-x. PMID: 37978388; PMCID: PMC10655462.



What is Known

- Children and adolescents with severe neurological impairments are particularly affected by severe and recurrent lower respiratory tract infections (LRTI).
- Determining the indication and selecting antibiotic therapy for bacterial LRTI is often difficult, as there are no evidence-based treatment recommendations for this heterogeneous but vulnerable patient group.
- The frequent overuse of broad-spectrum or reserve antibiotics in this patient group increases the selection pressure for multidrug-resistant pathogens.

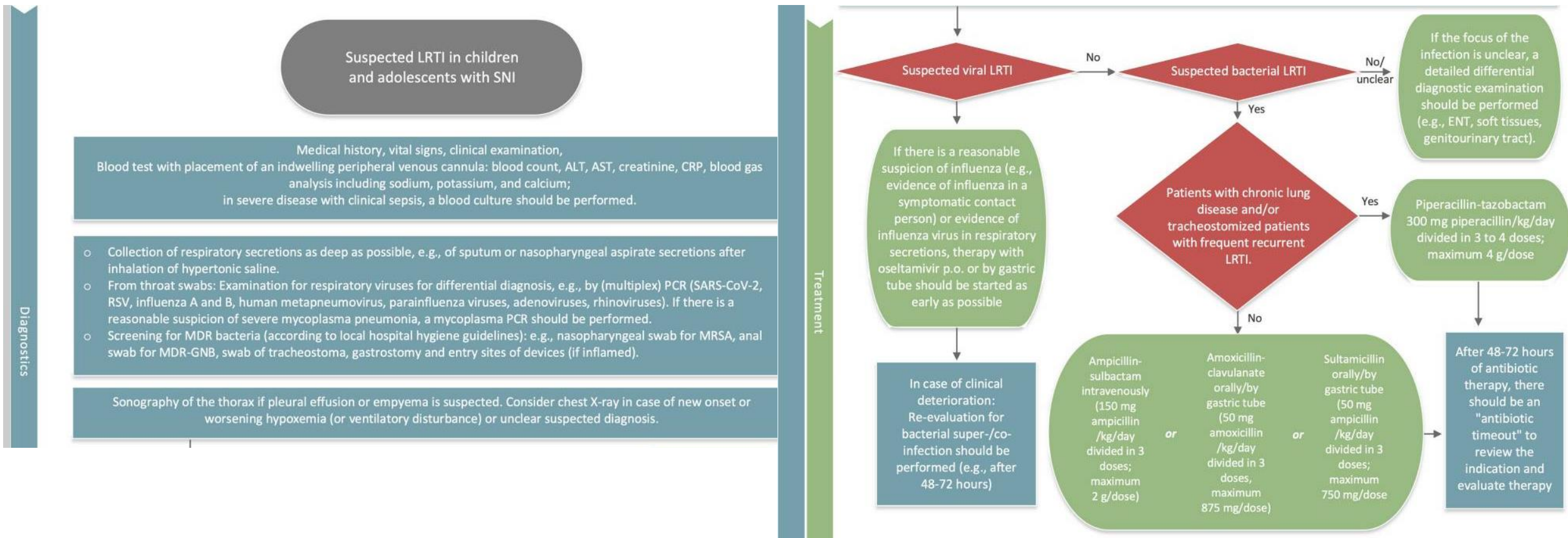
Eckerland M, Bock C, Olivier M, Pichlmaier L, Steindor M, Stehling F. Reducing the frequency of respiratory tract infections in severe neurological disorders by inhaled antibiotics: a retrospective data analysis. ERJ Open Res. 2019 Jul 22;5(3):00149-2018. doi: 10.1183/23120541.00149-2018. PMID: 31360698; PMCID: PMC6646963.



TABLE 1 Subject characteristics

Underlying disease	
Infantile cerebral palsy	8/20 (40)
Congenital myopathy	4/20 (20)
Spinal muscular atrophy	3/20 (15)
Duchenne muscular dystrophy	2/20 (10)
Neuronal ceroid lipofuscinosis	2/20 (10)
Tracheomalacia	1/20 (5)
Sex	
Female	14/20 (70)
Male	6/20 (30)
Age years	12.1±10.1 (1–37)
Tracheostomy	
Infantile cerebral palsy	9/20 (45)
Congenital myopathy	3/8 (37.5)
Spinal muscular atrophy	3/4 (75)
Duchenne muscular dystrophy	1/3 (33)
Noninvasive ventilation	2/2 (100)
Infantile cerebral palsy	5/20 (25)
Spinal muscular atrophy	2/8 (25)
Tracheomalacia	2/3 (66)
Mechanically assisted cough	1/1 (100)
Infantile cerebral palsy	15/20 (75)
Congenital myopathy	4/8 (50)
Spinal muscular atrophy	4/4 (100)
Duchenne muscular dystrophy	3/3 (100)
Neuronal ceroid lipofuscinosis	2/2 (100)

Data are presented as n/N (%) or mean±SD (range).



Eckerland M, Bock C, Olivier M, Pichlmaier L, Steindor M, Stehling F. Reducing the frequency of respiratory tract infections in severe neurological disorders by inhaled antibiotics: a retrospective data analysis. *ERJ Open Res.* 2019 Jul 22;5(3):00149-2018. doi: 10.1183/23120541.00149-2018. PMID: 31360698; PMCID: PMC6646963.



What's New in 2024

Inhaled antibiotic therapy offers a new approach to preventing respiratory tract infections in patients with severe neurological impairment who have chronic microbial colonization of the respiratory tract and recurrent respiratory tract infections.

<http://bit.ly/2HQRyz0>

Eckerland M, Bock C, Olivier M, Pichlmaier L, Steindor M, Stehling F. Reducing the frequency of respiratory tract infections in severe neurological disorders by inhaled antibiotics: a retrospective data analysis. ERJ Open Res. 2019 Jul 22;5(3):00149-2018. doi: 10.1183/23120541.00149-2018. PMID: 31360698; PMCID: PMC6646963.



Respiratory Tract Infections: Treatment Recommendations 2026

Update to the previous clinical recommendation from 2024:

For children with severe neurological impairment, first-time consideration in 2026 is given to (based on expert consensus and newly available data):

- Colonizing, multidrug-resistant pathogens
- Respiratory support
- Discharge planning
- Prophylactic Strategy

The new recommendation emphasizes comprehensive multidisciplinary care, antimicrobial stewardship and a stepwise risk stratification approach to the use of antibiotic therapy.

Ongoing treatment should include regular reevaluation, adjustment of antibiotic therapy, and optimization of supportive care.

Clinical Recommendations for the Inpatient Management of Lower Respiratory Tract Infections in Children and Adolescents with Severe Neurological Impairment in Germany – 2026 Update (in press)



What's New in 2026

- Careful Management of Secretions, Respiratory and Cough Support, Discharge Planning
- Decision-making regarding antibiotic therapy for infections caused by multidrug-resistant pathogens
- Recommendations for both non-pharmacological and pharmacological prophylactic strategies for LRTI

Clinical Recommendations for the Inpatient Management of Lower Respiratory Tract Infections in Children and Adolescents with Severe Neurological Impairment in Germany – 2026 Update (in press)



There Are Infections Other Than Respiratory Infections!

Common:

1. Urinary tract infections
2. Oral and dental infections
3. Gastrointestinal infections
4. Skin infections

All other infections common in childhood and adolescence also occur in children with PCH2!



Dental Infections / Tooth Decay

70 Children at Special Needs Schools in Java, Indonesia

- Odontogenic infections in children with intellectual disabilities are unfavorable
- Full spectrum of dental caries was observed
- predominantly severe forms

There is an association between odontogenic infections, the full spectrum of dental caries, and oral health-related quality of life (OHRQoL), as well as between odontogenic infections and the full spectrum of dental caries.

Sabila A, Riyanti E, Indriyanti R, Tjahajawati S, Rikmasari R, Rakhmatia YD. The Correlation Between Odontogenic Infection and Total Spectrum of Dental Caries with Oral Health Related Quality of Life in Children with Intellectual Disabilities. Clin Cosmet Investig Dent. 2024 Oct 30;16:431-439. doi: 10.2147/CCIDE.S486015. PMID: 39494164; PMCID: PMC11531729.



157 Children in a Hong Kong Institution, mean age 9.4 years

Endoscopy was performed on 8 of 15 seropositive children with a recent history of upper gastrointestinal bleeding. In all cases, a gastric infection with *H. pylori* was confirmed.

87 children (55.4%) were *H. pylori* seropositive, compared with 4 out of 50 children (8%, $P > 0.0002$) from an age-matched control group with a mean age of 7.2 ± 4.3 (SD) years.

Children with disabilities who receive long-term care in residential facilities in Hong Kong have been shown to be at increased risk for *H. pylori* infections.

Lewindon PJ, Lau D, Chan A, Tse P, Sullivan PB. *Helicobacter pylori* in an institution for disabled children in Hong Kong. *Dev Med Child Neurol*. 1997 Oct;39(10):682-5. doi: 10.1111/j.1469-8749.1997.tb07363.x. PMID: 9352730.



Children With SMID (n=18) And Previously Healthy Children (n=43) Under The Age of 16

Hospitalization for RSV-LRTI during five consecutive RSV seasons.

The clinical presentation and laboratory parameters of the SMID group were compared with those of the non-SMID group.

Results: Children with SMID were more likely to develop hypoxia following an RSV-LRTI.

Strategies for the treatment and prevention of RSV infections in children with SMID must be further improved.

Onoyama S, Hoshina T, Honjo S, Ihara K, Hara T. Respiratory syncytial virus infection in children with severe motor and intellectual disabilities. Eur J Clin Microbiol Infect Dis. 2013 Oct;32(10):1353-7. doi: 10.1007/s10096-013-1893-x. Epub 2013 May 17. PMID: 23681154.



217 Japanese Children With Severe Motor and Intellectual Disabilities (Ages 1-15), Hospitalized For LRTI

Comparison with and without systemic corticosteroid therapy:

- No improvement in treatment outcomes
- Conclusion: use with extreme caution

Nakamoto T, Hoshina T, Ishii M, Yamada G, Kusuvara K. Systemic corticosteroid as an adjunctive treatment for lower respiratory tract infection in children with severe motor and intellectual disabilities. J Infect Chemother. 2022 Mar;28(3):384-388. doi: 10.1016/j.jiac.2021.11.013. Epub 2021 Nov 23. PMID: 34823994.



Long-term Effects of COVID-19 on Children With Disabilities

Few reports on the topic. Hypotheses:

- Social factors (limited social contact)
- Behavioral factors (low visibility)
- Physiological factors
- Triage protocols were established very early on (functional ability, comorbidities, dependence in daily life). All protocols address people with chronic disabilities as a group.

Fry-Bowers EK. The Effects of the COVID-19 Pandemic on Children and Youth With Special Health Care Needs. J Pediatr Health Care. 2023 Mar-Apr;37(2):99-100. doi: 10.1016/j.pedhc.2022.12.003. Epub 2022 Dec 24. PMID: 36572587; PMCID: PMC9789749.

Dan, B. (2020), Long-term impact of COVID-19 on disabled children. Dev Med Child Neurol, 62: 1004-1004. <https://doi.org/10.1111/dmcn.14603>



Long-term Effects of COVID-19 on Children With Disabilities

To understand the effects of this pandemic and other phenomena like it, and to work effectively toward a better future, it is imperative to also consider carefully characterized **social determinants of health**, as well as the causes of the inequalities they create.



Prevention of Infections (Options, Individualized, As Needed)

1. Respiratory therapy (physical therapy, positioning, inhalation). Goal: maintaining chest elasticity (preventing stiffness), mobilizing secretions, stimulating coughing.
2. Secretion management (suctioning, reduction of saliva production with BTX, Glycopyrroniumbromide)
3. Mechanical cough-assist
4. Vaccinations (including pneumococcal)
5. Treatment of neuromyopathic scoliosis
6. Adequate hydration (not too much, not too little)
7. Bowel management (prevention of secondary bladder dysfunction)
8. Professional PEG care



Respiratory Infections: Treatment

1. See prevention
2. Early, targeted antibiotic therapy (run regular antibiotic susceptibility tests from sputum samples to allow guided therapy)
3. Early use of mechanical cough-assist and NIV
4. Early transfer to intensive care
5. Invasive ventilation before exhaustion (also to prevent rhabdomyolysis)
6. Antibiotic prophylaxis for frequent infections (primarily via inhalation; restrictive use, systemic antibiotics)



A Question From Parents

We experience frequent bouts of pneumonia (every month since March); tube feeding, the use of MucoClear, CoughAssist, suctioning, AirVo, Kürass – what else can help? What can be done to reduce the frequency of pneumonia? Thank you very much. Best regards.

Ideas:

1. *Inhalation with antibiotics, tailored to the antibiotic susceptibility test*
2. *Reducing saliva production*
3. *Systemic antibiotic administration if 1 in 2 treatments is unsuccessful*



Thanks For Your Attention!

Celebrating 2 Years of pch2cure.org

22.07.2026 | [News](#)

