

Pitt Hopkins Syndrome (PTHS) is a rare genetic disorder affecting a specific gene on chromosome 18. Most kids with PTHS have some physiological and behavioural differences that can make it difficult to assess how sick they actually are.

1. Poor temp control:



People with PTHS have dysautonomia, so temp dysregulation & cold extremities are common.

4. Seizures:

Just under half of people with PTHS have seizures, sometimes triggered by abnormal breathing (#2)

2. Breathing anomalies:

A pattern of hyperventilation and/or apnoea can occur regularly or sporadically. Cyanosis - and disturbance to acid/base status - can occur quickly, and may trigger seizures. See #4

3. Pain tolerance:

People with PTHS have a high threshold to pain and discomfort, and pain symptoms or signs may be undetectable until the painful stimulus is severe: take care not to miss injuries or painful illnesses

Top tips for triaging & treating



kids with Pitts Hopkins Syndrome

5. Ask what's normal for this individual child

Assessing levels of alertness, responsiveness, tone (hypotonia is common) etc. can all be difficult if you don't know the individual child at baseline. Ask parents: they know their child best!

8. Bowel & urinary complications

People with PTHS can develop severe faecal loading despite opening bowels regularly. Urinary retention and persistent UTIs are also common.

6. Co-morbidities common

Be aware of GORD, GIT dysmotility, constipation, autism and mitochondrial dysfunction... and take a careful drug history!

9. Act fast if unwell

The dysautonomia and mitochondrial dysfunction seen in this condition means that rapid physiological decompensation can occur when the person with PTHS is unwell.

7. Explain and reassure

Kids with PTHS often have sensory processing difficulties and can be very wary of new sensations: BP cuffs and sats probes may be terrifying. Take time to explain and reassure.

10. Optimise communication strategies

PTHS covers a vast spectrum of abilities, but speech & language development may lag behind understanding, so children with PTHS often understand more than they can express.

Although Pitt Hopkins Syndrome is very rare, there's a thriving international community of parents whose children have this condition. They'll probably know more than you do, and also know from whom you should be seeking advice. Keep calm, and utilise parental power!

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