

Pitt Hopkins Syndrome

➤ **What is Pitt Hopkins Syndrome?**

- Pitt Hopkins Syndrome (PTHS) is a rare genetic neurological disorder that causes developmental delays, moderate to severe intellectual disability, behavioral differences, distinctive facial features, and breathing problems such as episodes of rapid breathing (hyperventilation) and breath holding. Other features may include symptoms of autism spectrum disorder, sleep disturbances, seizures, constipation, nearsightedness, and minor skeletal abnormalities.

➤ **What are the signs of Pitt Hopkins Syndrome?**

- Pitt-Hopkins Syndrome can have a variety of signs and symptoms. Specific symptoms present and their severity can vary from person to person. Early symptoms, which often become apparent in the first year of life, may include:
 - Very low muscle tone (hypotonia)
 - Significant developmental delays
 - Small head size in infancy (microcephaly)
 - Delayed walking and talking
- Additional signs and symptoms of PTHS may include:
 - **Behavioral differences.** Symptoms of autism spectrum disorder may be present. Other behavioral characteristics can include unusual behavior during feeding, aggressive outbursts, anxiety, hand biting and head banging, stereotypic hand or head movements.
 - **Distinctive facial features.** These may become more obvious with age and may include deeply-set eyes, a prominent nose, short philtrum, wide mouth, full lower lip, widely-spaced teeth, and/or a prominent chin.
 - **Breathing problems.** These may include episodes of rapid breathing (episodic hyperventilation) followed by episodes of struggling to breath or not breathing. Episodes of breathing problems may be triggered by anxiety, excitement, or tiredness. Failure to breathe may cause a bluish discoloration of the skin (cyanosis) or fainting.
 - **Seizures.**
 - **Sleep disturbances.** These may include problems falling asleep or staying asleep, and/or night terrors.

- **Eye or vision problems.** Nearsightedness (myopia) can be severe and may become apparent before 2 years of age. Other eye abnormalities may include misalignment (strabismus) and astigmatism.
 - **Gastrointestinal problems.** These may include early feeding issues (which often resolve), constipation (which can be severe), and reflux.
 - **Skeletal features.** Scoliosis, flat feet, clubfoot, small hands and feet, broad fingertips or tapered fingers, bent or curved fingers (clinodactyly), and overlapping toes.
- **What is Pitt Hopkins Syndrome a result of?**
 - Genetic mutation in the TCF4 gene, or by a loss (deletion) of the part of chromosome 18 that contains the TCF4 gene.
- **What are some complications of Pitt Hopkins Syndrome?**
 - Breathing
 - Apnea (pauses in breathing)
 - Hyperventilation (rapid breathing)
 - Digestion
 - Chronic (long-term) constipation
 - Drooling
 - Gastroesophageal reflux (GERD, also called acid reflux or heartburn)
 - Behavior Development
 - Medical Issues
 - Vision or hearing challenges
 - Pain
 - Seizures
 - Flat feet
 - Valgus positioning (when one or both knees bend inward toward one another)
 - Scoliosis
- **Resources/References**
 - [https://rarediseases.info.nih.gov/diseases/4372/pitt-hopkins-syndrome#:~:text=Pitt%2DHopkins%20syndrome%20\(PTHS\),hyperventilation%20and%20breath%2Dholding](https://rarediseases.info.nih.gov/diseases/4372/pitt-hopkins-syndrome#:~:text=Pitt%2DHopkins%20syndrome%20(PTHS),hyperventilation%20and%20breath%2Dholding).
 - <https://www.massgeneral.org/children/pitt-hopkins/general-pitt-hopkins-care-guidelines>