Pitt Hopkins Syndrome (PTHS) is a rare neurodevelopmental disorder caused by a mutation of the TCF4 gene on the 18th chromosome. The TCF4 gene plays an essential role in the development of the nervous system and the brain.

Characteristics:
- Breathing problems: hyperventilation, apnea & breath-holding: 16%
- Lack of speech: nonverbal, pre-verbal/AAC users: 15%
- Gastrointestinal issues: low motility, gas & constipation: 15%
- Compromised gross motor: motor coordination (ataxia) or non-mobile: 15%
- Short attention span: sensory processing disorder: anxiety: 13%
- Developmental delay: seizures/epilepsy: 10%
- Vision impairment: strabismus or exotropia: myopia: 10%
- Compromised fine motor skills: feeding challenges: oral aversions & aspiration: 10%

- How they learn -
Give them time to process requests. Need repetition to build neuro pathways. Highly attracted to sensory rich environments (music, lights). Always presume competence.

Affects approximately between 1 in 34,000 and 1 in 41,000

Affects both genders and all races equally.

- Treatments -
There is currently no cure for Pitt Hopkins syndrome. However, treatment services can help people learn important skills. Services can include therapy to learn to walk, communicate and have improved fine motor skills. The PHRF is funding promising research to find treatments and eventual cure for PTHS, which you can read about at pitthopkins.org/research

You can help us by sharing this information & donating to research.

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