

Pallister-Hall syndrome

Description

Pallister-Hall syndrome is a disorder that affects the development of many parts of the body. Most people with this condition have extra fingers and/or toes (polydactyly), and the skin between some fingers or toes may be fused (cutaneous syndactyly). An abnormal growth in the brain called a hypothalamic hamartoma is characteristic of this disorder. In many cases, these growths do not cause any health problems; however, some hypothalamic hamartomas lead to seizures or hormone abnormalities that can be life-threatening in infancy. Other features of Pallister-Hall syndrome include a malformation of the airway called a bifid epiglottis, an obstruction of the anal opening (imperforate anus), and kidney abnormalities. Although the signs and symptoms of this disorder vary from mild to severe, only a small percentage of affected people have serious complications.

Frequency

This condition is very rare; its prevalence is unknown.

Causes

Mutations in the *GLI3* gene cause Pallister-Hall syndrome. This gene provides instructions for making a protein that controls gene expression, which is a process that regulates whether genes are turned on or off in particular cells. By interacting with certain genes at specific times during development, the GLI3 protein plays a role in the normal shaping (patterning) of many organs and tissues before birth.

Mutations that cause Pallister-Hall syndrome typically lead to the production of an abnormally short version of the GLI3 protein. Unlike the normal GLI3 protein, which can turn target genes on or off, the short protein can only turn off (repress) target genes. Researchers are working to determine how this change in the protein's function affects early development. It is uncertain how *GLI3* mutations can cause polydactyly, hypothalamic hamartoma, and the other features of Pallister-Hall syndrome.

[Learn more about the gene associated with Pallister-Hall syndrome](#)

- [GLI3](#)

Inheritance

This condition is inherited in an autosomal dominant pattern, which means one copy of the altered gene in each cell is sufficient to cause the disorder. In some cases, an affected person inherits a mutation in the *GLI3* gene from one affected parent. Other cases result from new mutations in the gene and occur in people with no history of the disorder in their family.

Other Names for This Condition

- Hall-Pallister syndrome
- PHS

Additional Information & Resources

Genetic Testing Information

- Genetic Testing Registry: Pallister-Hall syndrome (<https://www.ncbi.nlm.nih.gov/gtr/conditions/C0265220/>)

Genetic and Rare Diseases Information Center

- Pallister-Hall syndrome (<https://rarediseases.info.nih.gov/diseases/7305/index>)

Patient Support and Advocacy Resources

- National Organization for Rare Disorders (NORD) (<https://rarediseases.org/>)

Clinical Trials

- ClinicalTrials.gov ([https://clinicaltrials.gov/search?cond=%22Pallister-Hall syndrome %22](https://clinicaltrials.gov/search?cond=%22Pallister-Hall%20syndrome%22))

Catalog of Genes and Diseases from OMIM

- PALLISTER-HALL SYNDROME; PHS (<https://omim.org/entry/146510>)

Scientific Articles on PubMed

- PubMed (<https://pubmed.ncbi.nlm.nih.gov/?term=%28%28pallister-hall+syndrome%5BTIAB%5D%29+OR+%28hall-pallister+syndrome%5BTIAB%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+3600+days%22%5Bdp%5D>)

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