Greig cephalopolysyndactyly syndrome

Description

Greig cephalopolysyndactyly syndrome is a disorder that affects development of the limbs, head, and face. The features of this syndrome are highly variable, ranging from very mild to severe. People with this condition typically have one or more extra fingers or toes (polydactyly) or an abnormally wide thumb or big toe (hallux). The skin between the fingers and toes may be fused (cutaneous syndactyly). This disorder is also characterized by widely spaced eyes (ocular hypertelorism), an abnormally large head size (macrocephaly), and a high, prominent forehead. Rarely, affected individuals may have more serious medical problems including seizures, delayed development, and intellectual disability.

Frequency

This condition is very rare; its prevalence is unknown.

Causes

Mutations in the *GLI3* gene cause Greig cephalopolysyndactyly 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.

Different genetic changes involving the *GLI3* gene can cause Greig cephalopolysyndactyly syndrome. In some cases, the condition results from a chromosomal abnormality—such as a large deletion or rearrangement of genetic material—in the region of chromosome 7 that contains the *GLI3* gene. In other cases, a mutation in the *GLI3* gene itself is responsible for the disorder. Each of these genetic changes prevents one copy of the gene in each cell from producing any functional protein. It is unclear how a reduced amount of this protein disrupts early development and causes the characteristic features of Greig cephalopolysyndactyly syndrome.

Learn more about the gene and chromosome associated with Greig cephalopolysyndactyly syndrome

- GLI3

- chromosome 7

Inheritance

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

Other Names for This Condition

- Cephalopolysyndactyly syndrome
- GCPS

Additional Information & Resources

Genetic Testing Information

- Genetic Testing Registry: Greig cephalopolysyndactyly syndrome (https://www.ncbi.nlm.nih.gov/gtr/conditions/C0265306/)

Genetic and Rare Diseases Information Center

- Greig cephalopolysyndactyly syndrome (https://rarediseases.info.nih.gov/diseases/6550/index)

Patient Support and Advocacy Resources

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

Clinical Trials

- ClinicalTrials.gov (https://clinicaltrials.gov/search?cond=%22Greig cephalopolysyndactyly syndrome%22)

Catalog of Genes and Diseases from OMIM

- GREIG CEPHALOPOLYSYNDACTYLY SYNDROME; GCPS (https://omim.org/entry/175700)

Scientific Articles on PubMed

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

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