Hypochondrogenesis

Description

Hypochondrogenesis is a rare, severe disorder of bone growth. This condition is characterized by a small body, short limbs, and abnormal bone formation (ossification) in the spine and pelvis.

Affected infants have short arms and legs, a small chest with short ribs, and underdeveloped lungs. Bones in the skull develop normally, but the bones of the spine (vertebrae) and pelvis do not harden (ossify) properly. The face appears flat and oval-shaped, with widely spaced eyes, a small chin, and, in some cases, an opening in the roof of the mouth called a cleft palate. Individuals with hypochondrogenesis have an enlarged abdomen and may have a condition called hydrops fetalis in which excess fluid builds up in the body before birth.

As a result of these serious health problems, some affected fetuses do not survive to term. Infants born with hypochondrogenesis usually die at birth or shortly thereafter from respiratory failure. Babies who live past the newborn period are usually reclassified as having spondyloepiphyseal dysplasia congenita, a related but milder disorder that similarly affects bone development.

Frequency

Hypochondrogenesis and achondrogenesis, type 2 (a similar skeletal disorder) together affect 1 in 40,000 to 60,000 newborns.

Causes

Hypochondrogenesis is one of the most severe conditions in a spectrum of disorders caused by mutations in the *COL2A1* gene. This gene provides instructions for making a protein that forms type II collagen. This type of collagen is found mostly in the clear gel that fills the eyeball (the vitreous) and in cartilage. Cartilage is a tough, flexible tissue that makes up much of the skeleton during early development. Most cartilage is later converted to bone, except for the cartilage that continues to cover and protect the ends of bones and is present in the nose and external ears. Type II collagen is essential for the normal development of bones and other connective tissues that form the body's supportive framework. Mutations in the *COL2A1* gene interfere with the assembly of type II collagen molecules, which prevents bones and other connective tissues from

developing properly.

Learn more about the gene associated with Hypochondrogenesis

- COL2A1

Inheritance

Hypochondrogenesis is considered an autosomal dominant disorder because one copy of the altered gene in each cell is sufficient to cause the condition. It is caused by new mutations in the *COL2A1* gene and occurs in people with no history of the disorder in their family. This condition is not passed on to the next generation because affected individuals do not live long enough to have children.

Other Names for This Condition

- Achondrogenesis type II/hypochondrogenesis

Additional Information & Resources

Genetic Testing Information

- Genetic Testing Registry: Achondrogenesis type II (https://www.ncbi.nlm.nih.gov/gtr/conditions/C0220685/)
- Genetic Testing Registry: Hypochondrogenesis (https://www.ncbi.nlm.nih.gov/gtr/conditions/C0542428/)

Patient Support and Advocacy Resources

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

Clinical Trials

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

Catalog of Genes and Diseases from OMIM

- ACHONDROGENESIS, TYPE II; ACG2 (https://omim.org/entry/200610)

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?term=%28hypochondrogenesis%5BTIA

B%5D%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+3600+days%22%5Bdp%5D)

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