Cantú syndrome

Cantú syndrome is a rare condition characterized by excess hair growth (hypertrichosis), a distinctive facial appearance, heart defects, and several other abnormalities. The features of the disorder vary among affected individuals.

People with Cantú syndrome have thick scalp hair that extends onto the forehead and grows down onto the cheeks in front of the ears. They also have increased body hair, especially on the back, arms, and legs. Most affected individuals have a large head (macrocephaly) and distinctive facial features that are described as "coarse." These include a broad nasal bridge, skin folds covering the inner corner of the eyes (epicanthal folds), and a wide mouth with full lips. As affected individuals get older, the face lengthens, the chin becomes more prominent, and the eyes become deep-set.

Many infants with Cantú syndrome are born with a heart defect such as an enlarged heart (cardiomegaly) or patent ductus arteriosus (PDA). The ductus arteriosus is a connection between two major arteries, the aorta and the pulmonary artery. This connection is open during fetal development and normally closes shortly after birth. However, the ductus arteriosus remains open, or patent, in babies with PDA. Other heart problems have also been found in people with Cantú syndrome, including an abnormal buildup of fluid around the heart (pericardial effusion) and high blood pressure in the blood vessels that carry blood from the heart to the lungs (pulmonary hypertension).

Additional features of this condition include distinctive skeletal abnormalities, a large body size (macrosomia) at birth, a reduced amount of fat under the skin (subcutaneous fat) beginning in childhood, deep horizontal creases in the palms of the hands and soles of the feet, and an increased susceptibility to respiratory infections. Other signs and symptoms that have been reported include abnormal swelling in the body's tissues (lymphedema), side-to-side curvature of the spine (scoliosis), and reduced bone density (osteopenia). Some affected children have weak muscle tone (hypotonia) that delays the development of motor skills such as sitting, standing, and walking. Most have mildly delayed speech, and some affected children have mild intellectual disability or learning problems.

Frequency

Cantú syndrome is a rare condition. About three dozen affected individuals have been reported in the medical literature.

Genetic Changes

Cantú syndrome results from mutations in the *ABCC9* gene. This gene provides instructions for making one part (subunit) of a channel that transports charged potassium atoms (potassium ions) across cell membranes. Mutations in the *ABCC9* gene alter the structure of the potassium channel, which causes the channel to open when it should be closed. It is unknown how this problem with potassium channel function leads to excess hair growth, heart defects, and the other features of Cantú syndrome.

Inheritance Pattern

This condition is inherited in an autosomal dominant pattern, which means one copy of the altered *ABCC9* gene in each cell is sufficient to cause the disorder.

Most cases result from new mutations in the gene and occur in people with no history of the disorder in their family. In a few reported cases, an affected person has inherited the mutation from one affected parent.

Other Names for This Condition

- Cantu syndrome
- hypertrichosis-osteochondrodysplasia-cardiomegaly syndrome
- hypertrichotic osteochondrodysplasia

Diagnosis & Management

Genetic Testing

- Genetic Testing Registry: Hypertrichotic osteochondrodysplasia
  https://www.ncbi.nlm.nih.gov/gtr/conditions/C0795905/

Other Diagnosis and Management Resources

- GeneReview: Cantú syndrome
  https://www.ncbi.nlm.nih.gov/books/NBK246980

General Information from MedlinePlus

- Diagnostic Tests
  https://medlineplus.gov/diagnostictests.html
- Drug Therapy
  https://medlineplus.gov/drugtherapy.html
- Genetic Counseling
  https://medlineplus.gov/geneticcounseling.html

- Palliative Care
  https://medlineplus.gov/palliativecare.html
- Surgery and Rehabilitation
  https://medlineplus.gov/surgeryandrehabilitation.html

Additional Information & Resources

MedlinePlus

- Encyclopedia: Patent Ductus Arteriosus
  https://medlineplus.gov/ency/article/001560.htm
- Health Topic: Bone Diseases
  https://medlineplus.gov/bonediseases.html
- Health Topic: Congenital Heart Defects
  https://medlineplus.gov/congenitalheartdefects.html

Genetic and Rare Diseases Information Center

- Cantu syndrome
  https://rarediseases.info.nih.gov/diseases/8585/cantu-syndrome

Additional NIH Resources

- National Heart Lung and Blood Institute: Patent Ductus Arteriosus
  https://www.nhlbi.nih.gov/health/health-topics/topics/pda/
- National Heart Lung and Blood Institute: What is Pulmonary Hypertension?
  https://www.nhlbi.nih.gov/health/health-topics/topics/pah/

Educational Resources

- Disease InfoSearch: Hypertrichotic osteochondrodysplasia
  http://www.diseaseinfosearch.org/Hypertrichotic+osteochondrodysplasia/3618
- MalaCards: hypertrichotic osteochondrodysplasia
  http://www.malacards.org/card/hypertrichotic_osteochondrodysplasia
- Merck Manual Consumer Version: Overview of Heart Defects
  http://www.merckmanuals.com/home/children-s-health-issues/birth-defects-of-the-heart/overview-of-heart-defects
- Orphanet: Hypertrichotic osteochondrodysplasia, Cantu type
  http://www.orpha.net/consor/cgi-bin/OC_Exp.php?Lng=EN&Expert=1517

Patient Support and Advocacy Resources

- American Heart Association
  http://www.heart.org
- International Skeletal Dysplasia Registry, UCLA
  http://ortho.ucla.edu/isdr
- Washington University in St. Louis: Cantu Syndrome Interest Group
  http://cantu.wustl.edu/

GeneReviews

- Cantú syndrome
  https://www.ncbi.nlm.nih.gov/books/NBK246980

Scientific Articles on PubMed

- PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?term=%28cantu+syndrome%5BTIAB%5D%29+AND+english%5Bla%5D+AND+human%5Bmh%5D

OMIM

- CANTU SYNDROME
  http://omim.org/entry/239850

MedGen

- Hypertrichotic osteochondrodysplasia
  https://www.ncbi.nlm.nih.gov/medgen/208647

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