Ellis-van Creveld syndrome

Description

Ellis-van Creveld syndrome is an inherited disorder of bone growth that results in very short stature (dwarfism). People with this condition have particularly short forearms and lower legs and a narrow chest with short ribs. Ellis-van Creveld syndrome is also characterized by the presence of extra fingers and toes (polydactyly), malformed fingernails and toenails, and dental abnormalities. More than half of affected individuals are born with a heart defect, which can cause serious or life-threatening health problems.

The features of Ellis-van Creveld syndrome overlap with those of another, milder condition called Weyers acrofacial dysostosis. Like Ellis-van Creveld syndrome, Weyers acrofacial dysostosis involves tooth and nail abnormalities, although affected individuals have less pronounced short stature and typically do not have heart defects. The two conditions are caused by mutations in the same genes.

Frequency

In most parts of the world, Ellis-van Creveld syndrome occurs in 1 in 60,000 to 200,000 newborns. It is difficult to estimate the exact prevalence because the disorder is very rare in the general population. This condition is much more common in the Old Order Amish population of Lancaster County, Pennsylvania, and in the Indigenous (native) population of Western Australia.

Causes

Ellis-van Creveld syndrome can be caused by mutations in the *EVC* or *EVC2* gene. Little is known about the function of these genes, although they appear to play important roles in cell-to-cell signaling during development. In particular, the proteins produced from the *EVC* and *EVC2* genes are thought to help regulate the Sonic Hedgehog signaling pathway. This pathway plays roles in cell growth, cell specialization, and the normal shaping (patterning) of many parts of the body.

The mutations that cause Ellis-van Creveld syndrome result in the production of an abnormally small, nonfunctional version of the EVC or EVC2 protein. It is unclear how the defective proteins lead to the specific signs and symptoms of this condition. Studies suggest that they prevent normal Sonic Hedgehog signaling in the developing embryo,

disrupting the formation and growth of the bones, teeth, and other parts of the body.

Together, mutations in the *EVC* and *EVC2* genes account for more than half of all cases of Ellis-van Creveld syndrome. The cause of the remaining cases is unknown.

Learn more about the genes associated with Ellis-van Creveld syndrome

- EVC
- EVC2

Inheritance

This condition is inherited in an autosomal recessive pattern, which means both copies of the gene in each cell have mutations. The parents of an individual with an autosomal recessive condition each carry one copy of the mutated gene, but they typically do not show signs and symptoms of the condition.

Other Names for This Condition

- Chondroectodermal dysplasia
- Ellis-van Creveld dysplasia

Additional Information & Resources

Genetic Testing Information

- Genetic Testing Registry: Ellis-van Creveld syndrome (https://www.ncbi.nlm.nih.gov/gtr/conditions/C0013903/)

Genetic and Rare Diseases Information Center

- Ellis Van Creveld syndrome (https://rarediseases.info.nih.gov/diseases/1301/index)

Patient Support and Advocacy Resources

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

Clinical Trials

- ClinicalTrials.gov (https://clinicaltrials.gov/search?cond=%22Ellis-van Creveld syndrome%22)

Catalog of Genes and Diseases from OMIM

- ELLIS-VAN CREVELD SYNDROME; EVC (https://omim.org/entry/225500)

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?term=%28Ellis-Van+Creveld+Syndrome%5BMAJR%5D%29+AND+%28Ellis-van+Creveld+syndrome%5BTIAB%5D%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+1800+days%22%5Bdp%5D)

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