EVC2 Gene

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EvC ciliary complex subunit 2

genes

1. Normal Function

The *EVC2* gene provides instructions for making a protein whose function is unknown. However, it appears to be important for normal growth and development, particularly the development of bones and teeth. The EVC2 protein is found in primary cilia, which are microscopic, finger-like projections that stick out from the surface of cells and are involved in signaling pathways that transmit information between cells. In particular, the EVC2 protein is thought to help regulate a signaling pathway known as Sonic Hedgehog, which plays roles in cell growth, cell specialization, and the normal shaping (patterning) of many parts of the body.

EVC2 and another gene, *EVC*, are located very close together on chromosome 4. Researchers believe that the two genes may have related functions and that their activity may be coordinated.

2. Health Conditions Related to Genetic Changes

2.1 Ellis-van Creveld Syndrome

More than 30 mutations in the *EVC2* gene have been found to cause Ellis-van Creveld syndrome, an inherited disorder characterized by dwarfism, abnormal nails and teeth, and heart defects. The mutations that cause this condition occur in both copies of the *EVC2* gene in each cell. Most of the genetic changes lead to the production of an abnormally small, nonfunctional version of the EVC2 protein. Although it is unclear how the loss of this protein's function underlies the signs and symptoms of Ellis-van Creveld syndrome, researchers believe that it may prevent normal Sonic Hedgehog signaling in the developing embryo. Problems with this signaling pathway may ultimately lead to the abnormal bone growth and heart defects seen with this condition.

2.2 Weyers Acrofacial Dysostosis

At least three mutations in the *EVC2* gene have been found to cause Weyers acrofacial dysostosis, a condition that affects the development of the teeth, nails, and bones. Its signs and symptoms are similar to, but typically milder than, those of Ellis-van Creveld syndrome.

The mutations that cause Weyers acrofacial dysostosis occur in one copy of the *EVC2* gene in each cell. Each mutation changes a single protein building block (amino acid) near the end of the EVC2 protein. These mutations probably result in the production of an abnormally short, malfunctioning version of the protein. It is unclear how the defective protein leads to the specific features of Weyers acrofacial dysostosis. Studies suggest that it interferes with Sonic Hedgehog signaling in the developing embryo, disrupting the normal formation and growth of the teeth, nails, and bones.

3. Other Names for This Gene

- Ellis van Creveld syndrome 2
- Ellis van Creveld syndrome 2 (limbin)
- LBN
- LBN_HUMAN
- limbin

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