U.S. National Library of Medicine
National Center for Biotechnology Information

NLM Citation: National Center for Biotechnology Information (US). Genes and Disease [Internet]. Bethesda (MD): National Center for Biotechnology Information (US); 1998-. Ellis-van Creveld syndrome.
Bookshelf URL: https://www.ncbi.nlm.nih.gov/books/

Ellis-van Creveld syndrome

Ellis-van Creveld syndrome. The search for a molecular basis for the disease is on-going. Image credit: Clement D. Erhardt, Jr., Baltimore, MD, USA.

Ellis-van Creveld syndrome, also known as "chondroectodermal dysplasia," is a rare genetic disorder characterized by short-limb dwarfism, polydactyly (additional fingers or toes), malformation of the bones of the wrist, dystrophy of the fingernails, partial hare-lip, cardiac malformation, and often prenatal eruption of the teeth.

The gene causing Ellis-van Creveld syndrome, EVC, has been mapped to the short arm of chromosome 4. As yet, the function of a healthy EVC gene is not known; this is one of the most important questions that must be answered about the disease, since it would give an indication as to the molecular mechanism of the disease.

Ellis-van Creveld syndrome is often seen among the Old Order Amish community in Lancaster County, Pennsylvania. Because this group of people is small and isolated, it affords a rare opportunity to observe the passage of this particular disorder from generation to generation. A pattern of inheritance can be observed that has indicated the disease is autosomal-recessive (i.e. a mutated gene form both parents is required before the effects of the disease to become apparent).

Related diseases

See other Muscle and Bone Diseases

See other Skin and Connective Tissue Diseases