

## ELLIS-VAN CREVELD SYNDROME

The Ellis-Van Creveld syndrome is found chiefly among the Amish, a group of individuals who live chiefly in Pennsylvania, Ohio and Indiana, USA. However, non-Amish individuals have been reported to have this condition. The disorder is characterized by abnormalities of the hands, short stature, and Ectodermal Dysplasia involving mainly the nails and teeth. Sweating is normal. Congenital heart problems may also be evident.

The abnormalities of the hands are chiefly an extra finger on the "pinkie" side of each hand. Rarely are there extra toes. The nails are thin, wrinkled or spoon-shaped. Congenitally missing teeth (hypodontia) are a consistent finding. Teeth which do erupt are usually small and conical in shape. About 25% of individuals with this syndrome are born with teeth; teeth present at birth or which erupt during the first month or so of life are called natal or neo-natal teeth. Natal or neo-natal teeth are usually small, malformed and are usually lost prematurely. When natal or neo-natal teeth fall out or are extracted by a dentist, they are not replaced by more baby teeth. Other abnormalities in the mouth include serrated (washboard-like) alveolar ridges.

Congenital heart disease is the most life-threatening feature of this syndrome and is found in about half of all patients with the disorder. The congenital heart problem is largely responsible for frequent neo-natal deaths in this condition.

Intelligence is usually normal. The inheritance pattern of the Ellis-van Creveld syndrome is autosomal recessive. That is, parents who have already had one child with the condition are at a 25% risk for having a similarly affected child with each succeeding pregnancy. Males and females are affected with equal frequency. An affected individual is at little risk for having an affected child unless he marries someone else carrying the gene. Unlike many of the other ectodermal dysplasia syndromes, this condition may be diagnosed prenatally by ultrasound and fetoscopy.

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### Supporting a normal lifestyle

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