

Ellis-Van Creveld syndrome is a rare genetic disorder characterized by short limb dwarfism, additional fingers and/or toes (polydactyly), abnormal development of fingernails and, in over half of the cases, congenital heart defects. Motor development and intelligence are normal. This disorder is inherited as an autosomal recessive condition. Ellis-Van Creveld syndrome occurs in many ethnic groups throughout the world and affects males and females in equal numbers. This condition has been reported in approximately 150 individuals. It is more common in the Old Order Amish population of Lancaster County, Pennsylvania and in the native population of Western Australia. Ellis-Van-Creveld syndrome is diagnosed by the observation of short stature, slow growth, skeletal abnormalities determined by imaging techniques and sometimes teeth present at birth (natal teeth). Molecular genetic testing for the EVC and EVC2 genes is available on a research basis only. Prenatal diagnosis is possible by ultrasound.