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ELLIS-VAN CREVELD SYNDROME

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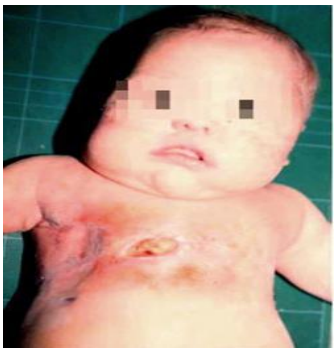
Abstract:

Ellis–van Creveld Syndrome[1] (also called chondroectodermal dysplasia or mesoectodermal dysplasia) is a rare genetic disorder of the skeletal dysplasia type.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.

Key Words: Ellis - Van Creveld Syndrome & Genetic Disorder

Introduction:



Richard W.B. Ellis of Edinburgh and Simon van Creveld of Amsterdam first described Ellis–van Creveld (EVC) syndrome. They met in a train compartment while traveling to a pediatrics conference in England in the late 1930s and discovered that each had a patient with the syndrome. In 1940, Ellis and van Creveld (Ellis and van Creveld, 1940) formally described the syndrome that would bear their names, although they termed it chondroectodermal dysplasia.

Meaning:

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.

Causes:

Ellis-Van Creveld syndrome is associated with abnormalities (mutations) in two genes on the number 4 chromosome called EVC and EVC2. These gene mutations result in the production of abnormally small EVC and EVC2 proteins. Some affected individuals do not have mutations in these genes, so it is likely that other unknown genes are also responsible for EVC.

Pathophysiology:

- ✓ Pathophysiology is unknown; however, recent identification of the *EVC* gene should lead to a better understanding.
- ✓ Histopathologic examination of fetuses with Ellis–van Creveld syndrome revealed that the cartilage of long bones showed chondrocyte disorganization in the physeal growth zone. Variable chondrocyte disorganization was seen in the central physeal growth zone of the vertebrae.

Symptoms:

Individuals with Ellis-Van Creveld syndrome typically have arms and legs that are abnormally short while the head and trunk are normal. Extra fingers (polydactyly) are present in all patients with this condition and both hands are usually affected. Ectodermal abnormalities include abnormal development of hair, nails and teeth.

Diagnosis:

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.

Treatment:

It is often necessary to treat respiratory distress shortly after birth that results from a narrow chest and/or heart failure. Natal teeth should be removed because they can interfere with feeding. The treatment of Ellis-Van Creveld syndrome is directed toward the specific symptoms that are apparent in each individual. Such treatment may require the coordinated efforts of a team of medical professionals, such as pediatricians, surgeons, cardiologists, dentists, pulmonologists, orthopedists, urologists, physical and occupational therapists and/or other health care professionals. Genetic counseling is recommended for affected individuals and their families.

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