Ellis van Creveld Syndrome

Overview

Ellis van Creveld syndrome (EVC) is a rare form of skeletal dysplasia that effects approximately 1 in 150,000 people, though it is more common in certain ethnic and racial groups, such as the American Amish. Common features include short stature, extra fingers and toes (polydactyly), and knock-knees (genu valgum). EVC is the result of a mutation in either the EVC or EVC2 genes and may be detected using radiological techniques, physical exams and genetic testing. Treatment of symptoms may include monitoring and surgery by doctors who specialize in skeletal dysplasia.

Heredity and genetics

Ellis van Creveld syndrome can be caused by a mutation in either the *EVC* or *EVC2* genes. Although the function of the genes is relatively unknown, they seem to be important for cell-to-cell communication during development. People demonstrate the same features regardless if the mutation is in *EVC* or *EVC2*.

EVC is inherited in an autosomal recessive pattern. This means that someone would need two copies of the "bad" gene in order to have the disease. If two parents are both carriers (have one mutated copy), they have a 25 percent chance of their offspring having the disease.

Diagnosis and testing

A diagnosis of Ellis van Creveld syndrome can be made using clinical assessments, radiological imaging and genetic testing.

Clinical features:

- Short stature
    - Adult height range: 3-foot-7 to 5-foot-3
- Teeth irregularities
- Polydactyly
- Finger and toenail underdevelopment
- Genu valgum

Radiographic features:

- Polydactyly with or without fusion of metacarpals and/or phalanges
- Shortening of the tubular bones
- Acetabulum abnormalities during infancy that resolve with age
- Short ribs during infancy

Orthopaedic problems and treatment

Growth monitoring

The Little People of America does not have EVC-specific growth charts available on its website. There are such growth charts that have been generated from growth studies of EVC patients, but the studies have limited patient numbers and include patients from other countries. They may not be a true reflection of growth in EVC patients in the United States, but they are useful as a guide.

Polydactyly

Polydactyly (extra digits) is a common orthopaedic issue seen in Ellis van Creveld syndrome. This most commonly affects the fingers, though there can be extra toes in some cases. Surgery is usually necessary to remove the extra digit.

Leg alignment

Most children with EVC have genu valgum (knock-knees) that will progress as the child gets older. Treatment varies according to the severity of the deformity and may include clinical monitoring using a series of X-rays called an extremity alignment series or surgery when the child experiences significant pain walking.

Other common problems and treatment

Congenital Heart Defects

More than half of people with EVC will have some sort of heart defect at birth. This is most commonly atrial septal defect, but single atrium, ventricular septal defect or atrioventricular septal defect may occur. Heart problems are life-threatening during infancy and should be followed by a cardiologist.

Teeth

Teeth may be present at birth and have an increased risk of cavities. The teeth may also be weak, missing or poorly formed. Other deformities of the lips and gum may be present. A pediatric dentist should be seen during childhood to manage teeth concerns, and a referral to an orthodontist may be necessary for management via surgery or prosthetics.

Resources

Ellis van Creveld General Information

Pauli, R.M. Legare, J. Ellis van Creveld Natural History. Rev. 2009.
http://www.lpaonline.org/natural-histories-of-dwarfism-types

Little People of America

http://www.lpaonline.org/

Adaptive Products

http://www.lpaonline.org/adaptive-products-