Ellis-van Creveld syndrome

Ellis-van Creveld syndrome is a rare genetic disorder that affects bone growth.

Causes

Ellis-van Creveld is passed down through families (inherited). It is caused by defects in 1 of 2 Ellis-van Creveld syndrome genes (*EVC* and *EVC2*). These genes are positioned next to each other on the same chromosome.

The severity of the disease varies from person to person. The highest rate of the condition is seen among the Old Order Amish population of Lancaster County, Pennsylvania. It is fairly rare in the general population.

Symptoms

Symptoms may include:

- Cleft lip or palate
- Epispadias or undescended testicle (cryptorchidism)
- Extra fingers (polydactyly)
- Limited range of motion
- Nail problems, including missing or deformed nails
- Short arms and legs, especially forearm and lower leg
- Short height, between 3.5 to 5 feet (1 to 1.5 meters) tall
- Sparse, absent, or fine textured hair
- Tooth abnormalities, such as peg teeth, widely-spaced teeth
- Teeth present at birth (natal teeth)
- Delayed or missing teeth

Exams and Tests

Signs of this condition include:

- Growth hormone deficiency
- Heart defects, such as a hole in the heart (atrial septal defect), occur in about half of all cases

Tests include:

- Chest x-ray
- Echocardiogram
- Genetic testing may be performed for genetic variants in one of the two EVC genes
- Skeletal x-ray
- Ultrasound
- Urinalysis

Treatment

Treatment depends on which body system is affected and the severity of the problem. The condition itself is not treatable, but many of the complications can be treated.

Support Groups

More information and support for people with EVC and their families can be found at:

- National Foundation for Ectodermal Dysplasias (NFED) -- rarediseases.org/organizations/national-foundation-for-ectodermal-dysplasias-nfed/ [https://rarediseases.org/organizations/national-foundation-for-ectodermal-dysplasias-nfed/]
- Little People of America, Inc -- rarediseases.org/organizations/little-people-of-america-inc/ [https://rarediseases.org/organizations/little-people-of-america-inc/]

Outlook (Prognosis)

Many babies with this condition die in early infancy. Most often this is due to a small chest or heart defect. Stillbirth is common.

The outcome depends on which body system is involved and to what extent that body system is involved. Like many genetic conditions involving bones or the physical structure, intelligence is normal.

Possible Complications

Complications may include:

- Bone abnormalities
- Breathing difficulty
- Congenital heart disease (CHD) especially atrial septal defect (ASD)
- Kidney disease

When to Contact a Medical Professional

Contact your provider if your child has symptoms of this syndrome. If you have a family history of EVC syndrome and your child has any symptoms, visit your provider.

Genetic counseling can help families understand the condition and how to care for the person.

Prevention

Genetic counseling is recommended for prospective parents from a high-risk group, or who have a family history of EVC syndrome.

Alternative Names

Chondroectodermal dysplasia; EVC

References

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