Normal Chondroectodermal Dysplasia (Ellis-Van Creveld Syndrome)

Patient Information Series – What you should know, what you should ask.

What is Chondroectodermal Dysplasia (Ellis-Van Creveld Syndrome)?

Ellis–van Creveld (EvC) syndrome is a rare disease of the bones with a prevalence of 1/60,000 approximately. It is characterized by short limbs, short ribs, six-fingered dwarfism, abnormal nails/teeth, and congenital heart defects (CHD).

How does Chondroectodermal Dysplasia happen?

EvC is a genetic disorder caused by a mutation in the chromosome 4, proximal to the gene involved in achondroplasia.
The name chondroectodermal dysplasia appears to be a generalized disorder affecting the maturation of cartilage ossification. Thus, the main features of the syndrome are a cartilage anomaly in the bones and abnormalities in the nails and teeth.

How is Chondroectodermal Dysplasia identified?

Prenatal ultrasound diagnosis of chondroectodermal dysplasia can be suspected from prenatal ultrasound findings of a narrow chest, six fingers in the hands and short limbs, which can be confirmed on different postnatal radiographic features.

Should I have more tests done?

In pregnancies known to be at risk of EvC syndrome, a targeted vaginal first-trimester ultrasound may help to identify fetuses with such anomaly since this syndrome is one of the severe skeletal abnormalities associated with an increased nuchal translucency during the first-trimester scan.
In case of potential recurrence, prenatal diagnosis can be feasible by including samples of the placenta during the early stages of pregnancy.

What are the things to watch for during pregnancy?

Chondroectodermal dysplasia is associated in a high proportion of cases with moderate narrowing of the chest that should be detected with a standard sonographic examination performed during the mid-trimester. This finding should prompt evaluation of the fetal long bones, heart, and all digits of the hands and feet.

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What does it mean for my baby after it is born?

The syndrome presents a 50% infant mortality rate, related to the severity of the chest narrowness and as a consequence of both heart and respiratory problems. Survivors will have a normal intelligence but short stature. The cardiac anomaly can be successfully corrected after birth. Dentition defects often require orthodontic procedures.

Besides, patients with EvC should be managed through a multidisciplinary approach. Particularly during the neonatal period, these patients require treatment for respiratory distress and heart failure. Management of bone deformities requires orthopedic follow-ups and oral manifestations need intense professional attention.
Prognosis is coupled to the respiratory problems during the first few months of life that are attributable to thoracic narrowness and heart defects. Treatment should be started as early as possible in order to attain the best possible clinical outcome.

Will it happen again?
There is a 25% risk of recurrence since the disease is inherited with a recessive pattern**.**

What other questions should I ask?

- Does this look like a case with severe chest narrowness?
- Is the heart affected?
- How big do the lungs seem to be?
- How often will I have ultrasound examinations done?
- When should surgery be done?
- Where should I deliver?
- Where will the baby receive the best care after it is born?
- Can I meet in advance the team of doctors that will be looking after my baby when it is born?

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