Campomelic Dysplasia

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

What is Campomelic dysplasia?

Campomelic dysplasia is a rare abnormality that can affect the baby in the uterus. It belongs to a category of diseases that affect the bones of the developing baby and are called skeletal dysplasias. In this specific condition there is defect of the cartilage cells, called “chondrocytes” of the bones, so that the bones become short, poorly ossified and bowed. The bones mainly affected are the femur, (thigh bone) the tibia (lower leg bone), many facial bones and the ribs, resulting to a narrow, barrel-shaped thorax. Another common finding is the presence of ambiguous or indistinct genitalia or even male-to-female sex reversal.

How does Campomelic dysplasia happen?

Campomelic dysplasia is a rare condition and happens in about 1 in 200,000 babies. It is caused by a genetic abnormality (faulty gene). That means that there is an error in the message of the DNA in the cells. Each message of the DNA is called a gene. Campomelic dysplasia occurs due to a mistake (called mutation) in a gene known as SOX9. Usually, the error happens after conception, so the parents are healthy. This is called a “de novo” or sporadic mutation.

Should I have more tests done?

Campomelic dysplasia may be diagnosed before birth by testing DNA taken from the baby. This can be obtained by an invasive procedure called amniocentesis after the 15th week of pregnancy. With amniocentesis we collect cells from the amniotic fluid around the baby and we examine the DNA inside them. The suspicion for the condition comes from ultrasound findings at the routine anatomy scan of the second trimester or ultrasound findings at the growth scan of the third trimester. Some cases may be diagnosed after birth.

What are the things to watch for during the pregnancy?

Campomelic dysplasia can be suspected through ultrasound in pregnancy, however, other skeletal dysplasias can have similar sonographic features. Thus, the diagnosis must be confirmed via DNA examination or/and radiographic evaluation after birth. Some women will also accumulate extra amniotic fluid around the baby. This condition is called polyhydramnios. It can stretch the uterus too much and cause early labor before the due date. It is also possible that the baby might die during the pregnancy before birth.

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

Unfortunately, babies with Campomelic dysplasia generally die shortly after birth, due to severe respiratory issues, such as defective structure of the larynx and trachea airway compromise or cervical spine instability causing neural damage. For babies who survive further, intense support will be needed by a team of specialists, such as respiratory support, physiotherapy and surgery cleft palate, clubfeet and cervical spine instability repair. Intellect may be variable. Kyphoscoliosis (abnormal curvature in the spine), short stature and

hearing loss are common issues. No prenatal treatment exists, and pregnancy termination is an option. If pregnancy continues, follow-up and obstetric care should be as usual. Delivery should take place in a tertiary maternity center.

Will it happen again?

In most cases the underlying cause is a de novo faulty gene, so there is no increased risk of recurrence. In the rare case of inherited form, the recurrence risk is estimated to be 25%. This could be detected early in the next pregnancy with an invasive procedure, as mentioned above.
.

What other questions should I ask?

- Does this look like a severe case of Campomelic dysplasia?
- Are there extra signs apart from the affected long bones?
- Is there a way to be sure of the diagnosis?
- How should the pregnancy be followed-up?
- Is there a treatment available during the pregnancy?
- Where and when should I deliver?
- What care will the baby receive after it is born?
- Can I meet the genetic specialist?
- Can I meet the team of doctors that will be assisting my baby when it is born in advance?

Last updated May 2022