

Thanatophoric Dysplasia (TD)

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

What is Thanatophoric Dysplasia (TD)?

TD is a rare disease affecting the growth and development of bones (skeletal dysplasia), occurring in 1 in 20,000 to 1 in 50,000 babies. It is mainly characterised by generalised by short limbs, a small thorax (rib cage), redundant skin folds, relative head enlargement, and a prominent forehead.

Thoracic growth restriction leads to underdevelopment of the lungs and respiratory insufficiency, resulting in early neonatal death in almost all cases.

How does TD happen?

TD is a genetic disorder in which a gene (a small part of DNA) that controls the development of bone and other tissues is affected, resulting in growth inhibition.

Which tests should I do during pregnancy?

TD can be suspected prenatally when the specific sonographic findings are seen on ultrasound and can be diagnosed later by genetic testing performed on fetal cells.

As some of the sonographic findings seen in TD are common to other skeletal dysplasias, a detailed ultrasound examination should be performed by an experienced physician, and the findings discussed with a geneticist. For a definitive diagnosis, you will be offered to undergo an invasive procedure (ultrasound-guided puncture) to obtain a sample of the placenta or amniotic fluid.

If the ultrasound findings are suggestive of TD, analysis of the sample can be targeted to the causative gene (FGR3). If the findings are indistinguishable from other skeletal dysplasias, a panel of different genes may be tested, or a more comprehensive DNA study may be performed such as whole exome sequencing.

What are the things to watch for during the pregnancy?

Termination of pregnancy may be an option because of low survival rates after birth. In some cases, the amount of amniotic fluid around the baby can be increased (polyhydramnios), which can lead to preterm labor.

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The size of the fetal head should be monitored since head circumference could be severely increased, which could interfere with safe vaginal delivery. Similarly, the position of the baby could be unfavorable for the normal progress of labor. If the pregnancy is managed expectantly, you will be given the opportunity to avoid monitoring for fetal distress during labor.

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

TD is considered a lethal condition. Survival after birth is extremely rare. Therefore, you will be offered the option of comfort care for the newborn, avoiding resuscitation efforts and extreme life support.

Will it happen again?

Parents with a baby affected with TD generally do not have an increased risk of their future baby developing the same condition.

What other questions should I ask?

- Can I receive genetic counseling?
- How often is the baby going to be scanned?
- Is there a risk of premature delivery?
- Is there a risk for me in case of a vaginal delivery?

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