Asphyxiating Thoracic Dysplasia

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

What is Asphyxiating Thoracic Dysplasia?

Asphyxiating Thoracic Dysplasia is a rare genetic skeletal dysplasia characterized by the presence of a very small thorax and reduced lung development. There are three types: type 1) lethal due to severe lung hypoplasia; types II and III where the thorax is less affected, and the lung function is maintained. In types II and III the newborn might survive but can also present with renal and hepatic complications later in life. The classification of the different types is based on the prenatal evaluation of the fetal thorax and lungs.

How does Asphyxiating Thoracic Dysplasia happen?

Asphyxiating Thoracic Dysplasia is a ciliopathy (a condition which affects the function of the cilia of the cells) which mostly arises from defects in the genes CEP120, CSPP1, IFT80, IFT140, IFT172, TTC21B, WDR19, WDR34, WDR60, and DYNC2H located in chromosome 15. It can affect other organs such as the kidneys and liver.

Should I have any more tests?

Evaluation should be carried out by an expert in Fetal Medicine. Ultrasound scans should be repeated frequently to estimate the development of the lungs. Molecular analysis in amniotic fluid might also help to confirm the diagnosis, but not in all cases, as not all genes related to Asphyxiating Thoracic Dysplasia have been described. The severity of the condition is determined by the ultrasound appearance of the thorax, lungs, and limbs. At birth, X-rays should also be performed to evaluate the bones.

What are the things to watch for during the pregnancy?

During the pregnancy, regular assessment of the size and development of the thorax and lungs will be performed as a reasonable proportion of babies with Asphyxiating Thoracic Dysplasia can survive. Limb bones should also be evaluated.

What is the prognosis? What treatment will my child require?

Babies with Type I Asphyxiating Thoracic Dysplasia will develop severe pulmonary hypertension and lung hypoplasia.
Babies with types II and III show less severe signs of lung hypoplasia and therefore, they are more likely to survive, but the renal and liver function should be evaluated periodically. X-ray evaluation of all limbs should be evaluated and their growth monitored. There have been reports of successful surgical procedures using titanium struts to expand the thorax in severely affected newborns.

Will it happen again?

This condition is inherited in an autosomal recessive pattern which means that if both parents are carriers of the affected gene the condition can happen again (25% risk).

What other questions should I ask?

- Is Asphyxiating Thoracic Dysplasia isolated or associated with other anomalies?
- What type is it?
- Should I have other tests?
- How is the development of the lung and thorax?
- Is surgery after pregnancy available?
- Where should I deliver?
- Where is the best place for the baby to be born?
- Can I meet in advance the team of doctors that will be looking after my baby when it is born?

Last updated on December 3rd 2023