Asphyxiating Thoracic Dysplasia

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

What is Asphyxiating Thoracic Dysplasia?

It is a genetic disease 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 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?

It is a defect in the genes: CEP120, CSPP1, IFT80, IFT140, IFT172, TTC21B, WDR19, WDR34, WDR60, and DYNC2H located in chromosome 15. It affects the function of the cilia of the cells and can affect other organs as the kidneys and liver.

Should I have more tests done?

Ultrasound scans should be repeated frequently to estimate the development of the lungs, X rays should also be performed at birth to evaluate other bones. Molecular analysis in amniotic fluid might 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.

What are the things to watch for during the pregnancy?

The size and development of the thorax and lungs, as a reasonable proportion of babies with Asphyxiating Thoracic Dysplasia can survive. The limb bones should also be evaluated

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

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, they can 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?

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?

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