Pulmonary agenesis and aplasia

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

What is Pulmonary agenesis and aplasia?

Pulmonary agenesis and aplasia are rare congenital conditions characterised by a complete or almost complete absence of lung development. It may affect one or both lungs.

How does Pulmonary Agenesis/Aplasia happen?

It is not clear why this condition occurs, but it is suspected that there is a disruption of the normal lung bud development that begins during the 6th week of gestation in the baby's development in utero.

How are chromosomes relevant to a CDH?

Pulmonary Agenesis/Aplasia is sometimes associated with other problems including chromosomal/genetic diseases. Therefore, it is recommended that we perform further tests during the prenatal and postnatal course.

Should I have more tests done?

The first step is to confirm the diagnosis and then to have a full multidisciplinary evaluation. Further tests are recommended including:

- Detailed fetal anatomy ultrasonography to confirm the diagnosis.
- Fetal Magnetic Resonance Imaging (MRI), if available, to also confirm the diagnosis.
- an **amniocentesis** to look for problems with the number of chromosomes and some of the problems within the chromosomes. This is done by removing a small amount of amniotic fluid surrounding the fetus.
- **Fetal echocardiography** - A specialised ultrasound of the heart of the baby during the pregnancy, which uses sound waves that "echo" off the structures of the fetus's heart.

What are the things to watch for during pregnancy?

During pregnancy, it is crucial that this pregnancy be followed by a specialist such as a Maternal-Fetal Medicine doctor in a center with a multidisciplinary team. Initially, the recommendation is to confirm the diagnosis and investigate other associated anomalies as described above. Then, regular fetal ultrasound follow-ups are necessary to evaluate the baby's heart function and to plan the delivery in a specialised tertiary center.

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

Babies with bilateral Pulmonary Agenesis/Aplasia (both lungs are absent) unfortunately don't survive after birth. This situation is extremely rare. In this event, parents may choose not to continue the pregnancy.

Babies with unilateral Pulmonary Agenesis/Aplasia (only one lung is not present) usually do well if there are no other anomalies or diseases. That is why it is important to perform the tests described above before the baby is born. Many of these babies will need to be born in a tertiary center specialised in congenital anomalies with a strong Neonatal Intensive Care Unit (NICU).

After birth, the diagnosis should be confirmed with x-rays of the chest and CT scans if available. In addition, an echocardiogram (ultrasound of the baby's heart) is also recommended to review the structures of the baby's heart and, if necessary, repeat genetic testing may be advised.

Will it happen again?

Pulmonary Agenesis/Aplasia is not associated with genetic diseases and, therefore, the chance of it happening again is extremely rare. So far, there has been no report of repeated occurrences of this disease.

What other questions should I ask?

- Does this look like a Pulmonary Agenesis/Aplasia?
- Is there another anomaly associated with this condition in my baby?
- Is it affecting one or both lungs?
- What prenatal tests do I need to do?
- How often will I have ultrasound examinations done?
- Is there any treatment during the pregnancy available?
- Where should I deliver?
- Where will the baby receive the best care after it is born?
- Can I meet the team of doctors that will be assisting my baby when it is born in advance?

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