# Achondrogenesis

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

## What is Achondrogenesis?

Achondrogenesis is a group of rare disorders affecting a baby's bones. Features include severe shortening of the limbs, short ribs, abnormal spine, and other skeletal abnormalities. These abnormalities are so severe that most babies with this condition will be stillborn, or pass away shortly after birth, due to breathing difficulties. There are 3 subtypes of this condition (Types IA, IB, and II), each caused by a different faulty gene and with different characteristics.

## How does Achondrogenesis occur?

This is a genetic condition that is inherited through the parents' genes or occurs as a new mutation (faulty gene) in the baby. Type IA is caused by a fault on the TRIP11 gene, which is required for the body's cells to manage certain cartilage proteins. Type IB is caused by a problem on the SLC26A2 gene, which helps with normal development of cartilage and its conversion to bone. Type II is related to a fault on the COL2A1 gene, which codes for a very important protein in cartilage. All of these forms can lead to abnormal development of cartilage and bones.

## Is genetics relevant to Achondrogenesis?

Yes. Types IA and IB are autosomal recessive conditions. This means that both parents carry the mutated (faulty) gene; when the child inherits 2 copies of the faulty gene (one from each parent), it will be affected with achondrogenesis. The parents will not have any symptoms themselves if they are carriers but may have other members of the family with similar conditions. If anyone in your family has had a bone or cartilage problem associated with short limbs, stillbirth or infant loss, please mention this to your midwife or doctor. Type II is an autosomal dominant condition; however, it is usually caused by a new mutation in the gene in the baby (and not inherited from its parents).

## Should I have more tests done?

Some women may choose to have further tests done to get a definite diagnosis. This may help you to decide whether you wish to continue with the pregnancy. It may also be helpful for planning future pregnancies. These tests include:

- **Amniocentesis:** this can look for problems in the chromosomes and genetic make-up of your baby, in particular the genes mentioned above. This involves taking a small sample of the fluid around your baby by passing a thin needle into your womb. If you have a known family history of skeletal dysplasia, or you have had a previously affected child, you may be offered a **chorionic villus sampling** between 11 and 14 weeks of pregnancy.

- This involves taking a small sample of the cells from the placenta, which will also look at the chromosomes and genes of your baby.
- If you opt for genetic testing of the baby, a consultation with a clinical geneticist can help you decide which genetic tests are the best for you, and how your future pregnancies may be affected.
- **Post-mortem:** If your baby passes away in the womb or shortly after birth, you may wish to have further tests to confirm the diagnosis. This can involve X-rays of your baby's bones, detailed studies of the organs, and histological analysis of your baby's skin and bone cells to look for the characteristic features.

## What are the things to watch for during the pregnancy?

In most cases, achondrogenesis is diagnosed during your routine 20-week scan, or less commonly, at your 12-week scan. As the baby's chest is very narrow, it affects their swallowing of fluid in the womb, causing an excessive build-up of amniotic fluid around the baby (polyhydramnios). This can cause your tummy to be more distended and uncomfortable and can trigger early labor. If you are becoming increasingly uncomfortable, your Fetal Medicine specialist may suggest draining some excess amniotic fluid to ease your symptoms. Your baby may develop a condition called hydrops fetalis, which involves swelling of their body and heart failure, due to the excessive pressure on the heart in a very small chest. This could lead to stillbirth. You will require regular detailed scans to monitor your baby's well-being.

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

Sadly, babies with achondrogenesis will either pass away in the womb or shortly after birth. Therefore, babies born alive will be made comfortable (palliative care), but will not routinely receive more active resuscitation. Your doctors will arrange for you to meet with the Neonatal doctors before giving birth to discuss exactly what this will involve.

## Will it happen again?

In Achondrogenesis types IA and IB, there is a 25% (1 in 4) chance in each future pregnancy that it will be affected by this condition. In type II, the chance of this happening again is usually <1%. Occasionally, however, the faulty genes may reside in your reproductive cells (egg or sperm), but not in the cells of the rest of your body, giving an increased chance of recurrence. A consultation with a geneticist before planning another pregnancy is advisable, so that you can be fully informed of the risks.

**What other questions should I ask?**

- What other abnormalities are there in my baby's body?
- Does the amniotic fluid level appear increased?
- Has my baby developed fetal hydrops?
- How often will I have ultrasound examinations?
- When should I deliver my baby?
- Can I meet with the genetic doctors to talk about my future pregnancies?
- Can I meet with the Neonatal doctors who will look after my baby when it is born?

*Last updated June 2022*