Diastrophic Dysplasia

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

What is Diastrophic Dysplasia?

Diastrophic dysplasia is a condition that is characterized by the presence of short arms and legs, clubbed feet, and hitchhiker thumbs (thumbs that are lower and deviated outwards). There may be problems with the spine, and cleft lip and palate. Babies born with this condition can have contractures in their legs and hips, early-onset arthritis, will be shorter than normal height, but have a normal intelligence. The incidence of this condition is 1:100,000 births.

How does Diastrophic Dysplasia occur?

This is a genetic condition, where both parents are often carriers of a faulty gene (The SLC26A2 gene). This gene codes for a protein that is essential for the development of cartilage, and its conversion to bone. This faulty protein interferes with the normal formation of cartilage, and as most cartilage is converted to bone in fetal life, it therefore also prevents bones from developing normally. A fault in this gene can also cause more dangerous types of skeletal dysplasia, which can result in stillbirth, or very early death following birth.

Is genetics relevant to Diastrophic Dysplasia?

Yes, this is an autosomal recessive condition, therefore both parents have to carry the faulty gene in order for the child to be affected. The parents will not express any symptoms if they are carriers but may have other members of the family with similar conditions. If you are aware of a history of skeletal dysplasias in your family, please mention this to your midwife or doctor.

Should I have more tests done?

Some women may choose to have further tests done to provide them with more information about the pregnancy, as well as future pregnancies. These can include:

- **Amniocentesis:** this can look for problems in the chromosomes and genetic makeup of your baby, in particular the SLC26A2 gene. 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 weeks and 14 weeks pregnant. This involves taking a small sample of the cells from the placenta, which will also look at the chromosomes and genes of your baby.

- A genetic consultation with a clinical geneticist can help you decide which genetic tests are the best choice for you, and how your future offspring may be affected.

- **Histological analysis** of your baby's skin and bone cells after birth is usually reserved for those babies who have signs of Diastrophic Dysplasia, but the genetic tests do not correlate. This involves taking a small sample of your baby's skin or bone cells and looking at them under a microscope.

What are the things to watch for during the pregnancy?

In most cases, Diastrophic Dysplasia is incidentally found during your routine 20-week scan. The appearance of short arms and legs, clubbed feet, and hitchhiker thumbs can sometimes be difficult to differentiate from other types of skeletal dysplasias. Regular and detailed ultrasound scans can help to look for other abnormalities, check the growth of the baby, look for the development of joint contractures, and increase in amniotic fluid volume.

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

Due to some babies having a small chest, there is unfortunately a 25% chance of the baby passing away in the first few months of life, due to difficulties in breathing. The babies who survive the initial months usually have a good prognosis. They will require regular physiotherapy and possibly surgery to correct the clubbed feet and contractures, in order to allow walking and normal range of movement. They may also need surgery later in life as they can develop early arthritis. Most babies will experience an inflammation of the outer ear early in life, which can lead to the ears appearing deformed, but should not affect the hearing. The upper spine is usually very curved, which usually resolves, but if not, may require surgery to avoid compressing the spinal cord. The children with this condition will have a normal level of intelligence, but a shorter than average height.

Will it happen again?

Because of the nature of inheritance, the chance of this happening again is 25% if both parents are carriers. A consultation with the 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?
- How big do the lungs seem to be?
- Are there normal movements of the joints?
- Does the amniotic fluid level appear increased?
- How often will I have ultrasound examinations?
- Where should I deliver?
- When should I deliver?
- 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?
- Can I meet with the Orthopedic doctors to talk about treatments for my baby after birth?

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