

Arthrogryposis

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

What is Arthrogryposis?

Arthrogryposis is the presence of two or more joint contractures. This means that the affected joints don't move much and may even remain in one position. The muscles around these joints are thin, weak, stiff or even missing. Most contractures occur in the arms and the legs. The incidence of Arthrogryposis is 1/3,000 to 1/5,000 live births.

How does Arthrogryposis occur?

Movement is essential for joint, muscle, and tendon development and growth. When there is lack of fetal movement (fetal akinesia), extra tissue might develop around the joint, causing stiffness and immobility. The longer the joint does not move, the worse the contracture will be. This reduction of movements, in turn, can be caused by muscle/neurological problems, limitation of space (for example in twin pregnancies), or in maternal illness, but, in most cases, the reason is unclear.

Is genetics relevant to Arthrogryposis?

Some cases of arthrogryposis are caused by a mistake in the baby's genetic information, however, in other cases, the baby might inherit the syndrome from one or both of the parents. In case of movement problems or intellectual disability in your family, be sure to mention this to your physician.

Should I have more tests done?

Many women will choose to have more tests done to know more about the condition of the baby. Tests to ask about include:

- 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, the waters surrounding the fetus. With this sample of fluid, other more specific genetic tests can be done to look for particular alterations in the DNA of the baby.
- An **MRI scan** can be useful to provide more information about fetal movements, the fetal brain, and to detect whether other malformations are present in the baby.
- A genetics consultation, with a specialist in genetics, to help you decide which genetic testing is most appropriate for your family, and to determine the genetic basis of your baby's condition.

What are the things to watch for during the pregnancy?

Determining the cause of congenital contractures is not always easy, especially in the prenatal period. Most specialists will recommend regular ultrasound examinations in order to observe the grade of joint contractures and presence of other anomalies, particularly in the brain, face, abdominal wall and lungs. Frequent ultrasound scans can also monitor any worsening condition of the baby, such as a decrease or increase of amniotic fluid.

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

There is no absolute cure for this condition; however, quality of life can be greatly improved depending on the severity of the syndrome and each individual patient's needs.

The main goal of treatment for arthrogryposis is to help your child's joints move as normally as possible. This means improving their flexibility, their strength and the way the bones line up. For your child's lower body, the focus is on working with their feet and legs so that they may be able to stand and walk. For their upper body, the focus is on working with their hands and arms so they may be able to do things on their own.

There are a variety of orthopaedic surgery options that may be needed for a child with arthrogryposis. Some are simple, such as an Achilles tendon release, some are moderate, including elbow and knee surgeries, and some are very involved, including limb lengthening and spinal surgery.

Will it happen again?

If no other genetic reason is found to explain the arthrogryposis, the risk of this happening again is 3-5%. If there is a genetic reason, this will determine the risk, and a consultation with a geneticist may be helpful in sorting this out.

What other questions should I ask?

- How many joints are involved?
- Are there other malformations in the baby's body?
- Is the brain normal?
- How big do the lungs seem to be?
- How often will I have ultrasound examinations done?
- 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 of the delivery?

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