Arthrogryposis

BIRTH DEFECT RESEARCH FOR CHILDREN

What is Arthrogryposis?

Arthrogryposis is a congenital (present at birth) condition characterized by the reduced mobility of many joints. The joints are fixed in various postures and lack muscle development and growth. There are many different types of arthrogryposis and the symptoms vary among affected children. In mild cases only a few joints are affected, and the range of motion may be almost normal.

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In the most severe cases, nearly every body joint may be affected including the jaw and back. The distal form of arthrogryposis involves the distal joints (joints away from the center of the body) and includes several subtypes which categorize the various combinations of anomalies. The most common form of arthrogryposis, which accounts for about 40% of the cases, is called amyoplasia. In amyoplasia the hands, wrists, elbows, shoulders, hips, feet, and knees are affected. The typical deformities are often severe and symmetrical. The limbs are usually fixed, shoulders are internally rotated, elbows are extended, wrists and digits are flexed, hips may be dislocated and slightly flexed, and the feet may have clubfoot deformity. Intelligence and sensation, however, are usually normal. Arthrogryposis may be associated with other conditions such as spina bifida, Trisomy 18, Amniotic Band Syndrome, and Poland's Syndrome.

How Many Children Have Arthrogryposis?

In the United States this condition occurs in one out of 3,000 live births.

How Do You Know If Your Child Has Arthrogryposis?

Arthrogryposis is detected at birth or through prenatal ultrasound. Prenatal ultrasound can detect decreased fetal movement and abnormal fetal position.

What Causes Arthrogryposis?

The causes of arthrogryposis are varied and not entirely understood. In many cases abnormal nerve, muscle, and connective tissue development is involved. Although most cases of arthrogryposis are non-genetic, genetics may be involved in some cases. Maternal exposure to environmental factors like drugs and alcohol may also play a part. The suspected cause of most cases is fetal akinesia (decreased fetal movements). Motion is essential for the normal development of joints. Lack of fetal movement causes extra connective tissue to develop around joints, fixing them in position. Akinesia may be due to fetal abnormalities or maternal disorders. The causes of akinesia include, but are not limited to, congenital muscular dystrophies, neurogenic abnormalities, maternal infection or illness, trauma, insufficient amniotic fluid, and an abnormally shaped uterus. Infants born to mothers with multiple sclerosis are at risk for arthrogryposis. There is no known prevention for arthrogryposis.

How Can You Help A Child With Arthrogryposis?

The goal of treatment is to improve the body's mobility function and to teach the child self-care. Treatment does not generally address the issue of a child's appearance. A multi-disciplinary team of specialists is recommended and should include a pediatrician, neurologist, orthopedic surgeon, physical and/or occupational therapist, geneticist, and counselor. Physical therapy (to improve muscle strength and joint range of motion) and bracing of the joints are usually attempted before surgical correction. In most cases it is important to mobilize the limbs early so that muscle atrophy is minimized. Long-term bracing is usually required. When necessary, surgery is performed to achieve better position or more range of motion. Multiple orthopedic procedures are usually required. In most cases it is beneficial to seek a doctor or therapist at a facility which treats a large number of arthrogryposis patients.

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What's In The Future For A Child With Arthrogryposis?

There is a wide variation in the degree of muscle and joint problems in arthrogryposis. Children will usually have normal life spans, but this depends on the severity of arthrogryposis and the associated malformations. In most cases the outlook is positive and physical therapy can greatly improve function. When other conditions are present such as central nervous system disorders, the outlook may be less favorable. In disorders with limb involvement and severe central nervous system dysfunction, about half of the children die in the first year of life. Children with scoliosis may experience respiratory problems. Most children will have normal intelligence and can lead productive, independent lives as adults. Children who develop good coping skills and receive support from family and friends contribute to better functional prognosis.

Fact Sheet by:

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