

# ARTHROGRYPOSIS

## OVERVIEW OF THE DIAGNOSIS

Arthrogryposis was first documented in 1841 by Adolph Wilhelm Otto in an article written in Latin. Arthro = joints, Grypo = curved, Multiplex = different forms, Congenita = present at birth.

The major cause of arthrogryposis is fetal akinesia or decreased fetal movements. Decreased movements may be due to fetal abnormalities (neurogenic, muscular, or connective tissue) or mechanical limitations to movement in the womb. Maternal disorders including infection, drug use, trauma or illness can also lead to decreased fetal movement. Sometimes nerve signals don't reach the muscles because of problems with the baby's central nervous system (CNS).

During early embryogenesis, joint development is almost always normal. As movement becomes limited or decreased, the joint becomes fixed. The frequency of arthrogryposis (Mankin, 2013) is about 1 in 3,000 live births in the United States. The causes of arthrogryposis are varied and not entirely understood, but are presumed to be multifactorial. In most cases, arthrogryposis multiplex congenita (AMC) is not a genetic condition. However, in approximately 30 percent of cases, a genetic cause can be identified. It is not uncommon in twins for one baby to have some form of arthrogryposis secondary to limited space in the womb.

In some cases, only a few joints are affected and the range of motion is nearly normal. In severe cases, many joints are involved, including the jaw and back. While there are many known forms, the most common is Amyoplasia (A = absent, Myo = muscle, Plasia = abnormal growth or development). No racial predilection has been described. Males are primarily affected in X-linked recessive disorders. Otherwise, males and females are equally affected. Arthrogryposis is detectable at birth or in utero using ultrasonography (see Picture 1).

## PHYSICAL PRESENTATION AND LIFE SPAN EXPECTATIONS

- Involved extremities are cylindrical in shape;
- Orthopedic limitations are usually symmetrical, and severity increases distally;
- Distal joints are affected more frequently than proximal joints;
- Joint rigidity and diminished range of motion may be present;
- The patient may have joint dislocation, especially the hips, and occasionally, the knees;

- Atrophy may be present, and muscles or muscle groups may be absent; and
- Sensation is usually intact, although deep tendon reflexes may be diminished or absent.

The individual's life span depends on the disease severity and associated malformations, but is usually normal, unless the nervous system and/or heart are involved. About 50 percent of patients with severe limb involvement and CNS dysfunction die in the first year of life. Scoliosis may compromise respiratory function which leads to early mortality.

## ORTHOPEDIC CONSIDERATIONS

Some joint limitations can be corrected and ambulation is possible. Interventions include a combination of surgical procedures and occupational and/or physical therapy for strengthening, range of motion, ambulation and ADL activities. Often, any recurrent joint limitations are addressed with splints

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PICTURE 1



PICTURE 2

and bracings. When indicated, night splints are prescribed. The parents are part of the team with home programs designed to assist with maintenance of range of motion and strength.

Most surgeries are performed early,

often before the child turns 2 years old. The most common surgeries are to the bone or tendon transfers. Due to the heightened risk during anesthesia, whenever possible or needed, two surgeries may be combined. When the individual is older, osteotomies may be performed when growth is completed.

When surgeries are not performed and joint rigidity and contractions occur, seating and wheeled mobility become the selected form for ambulation. A power wheelchair assessment may occur as young as 18 months.

As the child grows, the location and severity of orthopedic involvement can create unique seating challenges, particularly when surgical interventions are not provided (See Picture 2). For example, when there are fixed contractures and asymmetries in the hips, knees and ankles, creating an appropriate seating system that will support the individual and provide pressure redistribution requires multiple considerations (See Picture 3). One consideration is how to integrate this unique seating system into the mobility base.

### SKIN/SENSATION CONSIDERATIONS

Sensation is intact, so comfort is a major consideration within the seating system, contrary to what funding sources may consider in their coverage criteria. Often, bony prominences resulting from contractures need to be addressed. Skin webs



PICTURE 3

Picture 1: Ultrasound of a baby with arthrogryposis

Picture 2: Example of lower extremity involvement

Picture 3: Range limitations accommodated in seat and mobility base

(pterygia) may be present across fixed joints. Skin dimples are common where movement is limited. Secondary to muscle atrophy, muscle mass and circulation is decreased. There may be a history of skin breakdown in these areas depending on the ability of the individual to do a weight shift or redistribute pressure.

### BASIC SEATING AND MOBILITY CONSIDERATIONS

- Provide support;
- Accommodate/prevent orthopedic limitations;
- Increase functional mobility;
- Decrease fatigue;
- Anticipate change or growth;
- Protect skin integrity;
- Encourage independence in MRADLs;
- Enhance physiological function; and
- Consider future surgeries or other medical interventions.

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## ARTHROGRYPOSIS (CONTINUED FROM PAGE 41)

### SETTING GOALS

Most children born with arthrogyrosis have normal intelligence but can experience developmental delays related to decreased mobility and exploration of their environment. Providing mobility early can diminish learned helplessness and allow the individual to be as independent as possible. Some individuals may be able to roll or scoot, however it is not uncommon for young children to be carried or placed in strollers, limiting independent mobility opportunities. Commercially available strollers do not provide adequate postural support either. A thorough mat assessment and evaluation with trial equipment is needed to determine the best technology solution for each individual.

### THE TEAM SHOULD STRIVE TO:

- Encourage acceptance and exposure to mobility technologies;
- Educate the parents on product options, as well as the evaluation process;
- Encourage autonomy;
- Diminish learned helplessness by providing mobility options;
- Provide a safe, but less protective environment to promote independence; and
- Explore options and access to other technologies that will enhance independence and that can be operated from the mobility device.

### SUMMARY

While there are many levels of physical involvement seen in an individual with arthrogyrosis, the client we typically see for a seating and wheeled mobility assessment is the one who presents without surgical intervention. Considerations for accommodating contractures and decreased range of motion while providing postural support and access to mobility will continue to challenge the team's knowledge, creativity and problem solving abilities. The RTS/CRTS working together with the individual, their family, the therapist and equipment manufacturers will help provide the best outcomes for independence and function.

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