

Arthrogryposis

OVERVIEW

Arthrogryposis is a condition that can affect the spine and most joints such as the hips, knees, ankles, shoulders, elbows and hands. Arthrogryposis may also be referred to as amyoplasia or arthrogryposis multiplex congenita (AMC). Escobar syndrome is a form of arthrogryposis where there is skin webbing at the joints (pterygium).

The severity of the disease varies from person to person. Treatment varies according to the cause and severity of the condition and may include physical therapy, casting, surgery, or a combination of these options. In many cases, when treatment begins at an early age, children can gradually become stronger and experience improved joint mobility and function that lasts the rest of their lives.

DESCRIPTION

Arthrogryposis is a medical condition that is present in affected children from birth and occurs in about 1 in 10,000 live births. The word “arthrogryposis” is actually a catch-all term used to describe instances of joint contractures from birth. The condition is characterized by malformed or stiff joints, muscles, and tendons that result in arms, legs, hands, and/or feet having limited or no mobility. Scoliosis (a curvature of the spine) may be present in some children as well. The cognitive function of children with the condition is not affected. In fact, they are often extremely bright and communicative.

CAUSE

There is no one specific cause of arthrogryposis but the most common causes are genetics and intrauterine viruses. Genetic causes often only involve the hands and feet while other causes typically result in more generalized weakness and contractures.

DIAGNOSIS METHODS

Arthrogryposis tends to be found in its most severe form during newborn examinations. Given the various possible causes of arthrogryposis, proper diagnosis plays a very important role in determining treatment. Diagnosis methods may include MRI, muscle biopsies, blood tests, DNA testing, studies, and/or observations.

TREATMENT

Given that the condition is relatively rare, few surgeons and therapists are experienced enough to treat this disease effectively. Arthrogryposis requires very complex treatment and should only be undertaken by physicians, surgeons, and allied health professionals who are not only familiar with the disease but also have a high level of expertise in treating arthrogryptic patients.

I believe that if a child is to have surgery, there must be an expected outcome that can change the child's life. To achieve the best functional outcomes, I take into account the underlying muscle strength of the patient, outline realistic goals, assess the potential benefits of treatment, and partner with an exceptional team of experienced medical professionals to provide treatment. This ensures that children do not undergo multiple painful surgeries that make very little change in their condition.

Non-Surgical Treatments

- **Occupational/Physical Therapy**

The initial evaluation and treatment for a child is performed by an experienced therapist. In some cases it may be possible to correct arthrogryposis to some degree with therapy alone. For example, newborns are often most affected at birth with contractures that might include hyper-extended knees, flexed hips, or clubfeet. Therapists can often achieve significant correction in the first six months of life followed by work on activities of daily living activities and walking.

Therapy may include mobilization, casting, and splinting that would be ongoing in addition to any other treatments.

- **Casting / Splinting**

Casting and splinting is the primary treatment for wrists, hands and clubfeet and is often the only treatment needed. It can be performed by me and/or therapists and may be utilized at all ages.

- **Psychosocial and Emotional Therapy**

Any physical disability can have an impact on body image and all children who have multiple surgical procedures require a strong support system in order to thrive. Having your family work with experienced psychologists, social workers and child life specialists is essential for an overall happy child.

Surgical Treatments

The goal of surgical treatment for arthrogryposis is often to straighten curved bones and/or bent joints.

- **Soft Tissue Release**

Contractures are caused by shortened or abnormally tight soft tissue which prevents limbs from moving as they should. In soft tissue releases, contractures are relaxed by cutting the tight muscle, tendon, or ligament which allows the limb or extremity to move more freely. On some surgical procedures, I partner with a highly skilled (wound) plastic surgeon. We have been thrilled with the outstanding results of working in tandem on cases such as severe knee contractures with and without webbing (pterygium).

- **Tendon Transfer**

A tendon is a tough type of tissue which connects muscles to bones, keeps limbs in position, and plays an important role in the movement of body parts. Tendons may be moved from one part of the body to another to correct the positioning and/or enable movement of a limb.

- **Osteotomy**

Curved bones in many arthrogryptic children can be corrected through casting but surgery may be required in cases that are complex or involve older children. An osteotomy is a surgical procedure where a curved or deformed bone is broken and reset. Internal pins or plates and screws are utilized and at times external frames called “fixators” are used to hold/guide the bone into the correct position and proper alignment.

- **Frames & Devices (Fixators)**

External frames (ex: Taylor Spatial Frame, Ilizarov Frame, etc.) consist of adjustable stainless-steel discs and rods that are affixed to bone and held in place by pins and struts. The tension of the wires can be adjusted to allow force to be distributed through the top and bottom discs of the frame. This relieves stress from the osteotomy site while the bone re-aligns and tissues in the limb are slowly stretched.

- **Growth Modulation**

Growth plates (physeal plates) are bits of cartilage that are present in children and adolescents. The plates are located at the ends of the long bones (ex: femur, tibia, etc) and create new bone tissue which determines the length of mature bones. Growth modulation is a minimally invasive procedure that temporarily alters the growth plate to allow bones to be straightened. I often use small metal plates which are shaped like an '8' (8 plates) to gradually guide new bone growth during modulation.

CONCLUSION

Arthrogryposis is varied and challenging for the patient, family and caregiver. Parents might feel discouraged and the situation may seem hopeless but after treating many patients I know that we should never give up. While outcomes may vary and goals might change, my arthrogrypotic patients have been amongst the most personally rewarding patients to treat as I've been able to see them grow over time and reach new functional milestones with treatment.