

Arthrogryposis

(Amyoplasia or Arthrogryposis Multiplex Congenita [AMC])

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OVERVIEW

The word “arthrogryposis” is actually a catch-all term used to describe instances of joint contractures that are present at birth.

Treatment varies according to the cause and severity of the condition but when treatment begins at an early age, children can gradually become stronger and experience improved joint mobility and function that can last the rest of their lives.

DESCRIPTION

Arthrogryposis is a rare medical condition that only occurs in about 1 in 10,000 live births.

The condition is characterized by malformed or stiff joints, muscles, and tendons that result in arms, legs, hands, and / or feet having limited to no mobility.

The cognitive function of children with the condition is not affected. In fact, they are often extremely bright and communicative.

CAUSES

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

Arthrogryposis may require very complex treatment and should therefore only be undertaken by health professionals who are not just familiar with the disease but 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. This ensures that children do not undergo multiple painful surgeries that result in very little change in their condition.

To achieve the best functional outcomes, I:

- Assess the patient's underlying muscle strength
- Outline realistic goals
- Assess the potential benefits of treatment
- Partner with an exceptional team of experienced medical professionals to provide treatment

Non-surgical Treatment

Occupational / Physical Therapy

In some cases it may be possible to correct arthrogryposis to some degree with therapy alone. Therapy may include mobilization, casting, and splinting that would be ongoing in addition to any other treatments.

Casting / Splinting

The primary treatment for wrists, hands and clubfeet and 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 children who have multiple surgical procedures need a strong support system. Having your family work with experienced psychologists, social workers and child life specialists is essential for an overall happy child and family.

Surgical Treatment

The goal of surgical treatment for arthrogryposis is often to straighten curved bones and / or bent joints. While many of these issues can be corrected through casting, surgery may be required in cases that are complex or involve older children.

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*).

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.

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

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 are adjusted to distribute force 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 are bits of cartilage at the ends of children's long bones which create new bone tissue. Growth modulation is a minimally invasive procedure that temporarily alters the growth plate allowing bones to be straightened as they grow.

CONCLUSION

Arthrogryposis is varied and can be 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.

CASE STUDIES

Axel: Arthrogryposis, Clubfeet, & Dislocated Hips and Knees

At the time of Axel's birth, it was apparent to both Axel's parents and doctors that there was something very wrong with his legs. His pediatrician had never seen a case like his before. The family hoped to begin treatment immediately and I began treating Axel when he was three days old.

<http://www.davidsfeldmanmd.com/patient-education/case-studies/axel-arthrogryposis-clubfeet-dislocated-knees-hips>

Douglas: Arthrogryposis, Bilateral Clubfeet & Ankle Varus

I first met Douglas when his parents brought him in at the age of 1 week old for a consultation on his bilateral clubfeet. However, during my initial examination of Douglas I found a combination of symptoms that indicated he actually had arthrogryposis with clubfeet.

<http://www.davidsfeldmanmd.com/patient-education/case-studies/douglas-arthrogryposis-bilateral-clubfeet-ankle-varus>

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