

## Arthrogryposis: When A “Non-Progressive” Diagnosis Progresses

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## Arthrogryposis Multiplex Congenita

- \* • Arthro = joints
- \* • Grypo = curved
- \* • Multiplex = different forms
- \* • Congenita = present at birth

## Review of the diagnosis

- \* 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.
- \* The most common form is Amyoplasia.
  - \* • A = absent
  - \* • Myo = muscle
  - \* • Plasia = abnormal growth or development

## Review of the diagnosis

- \* The major cause of arthrogryposis is fetal akinesia (ie, decreased fetal movements) due to fetal abnormalities (eg, neurogenic, muscle, or connective tissue abnormalities; mechanical limitations to movement) or maternal disorders (eg, infection, drugs, trauma, other maternal illnesses)
- \* During early embryogenesis, joint development is almost always normal. This results in fixation of the joint, limiting movement and further aggravating the joint contracture
- \* The frequency is about 1 in 3,000 live births in the United States

## Review of the diagnosis

- \* **Race**  
No racial predilection has been described.
- \* **Sex**  
Males are primarily affected in X-linked recessive disorders; otherwise, males and females are equally affected.
- \* **Age**  
Arthrogryposis is detectable at birth or in utero using ultrasonography.

## Physical Presentation

- \* 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% of cases, a genetic cause can be identified.

## Physical Presentation

- \* Involved extremities are cylindrical in shape
- \* Deformities are usually symmetrical, and severity increases distally
- \* Distal joints are affected more frequently than proximal joints.
- \* Joint rigidity and diminished ROM ( 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.
- \* Sensation is usually intact, although deep tendon reflexes may be diminished or absent.

## Life Span

- \* The 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% of patients with severe limb involvement and CNS dysfunction die in the first year of life.
- \* Scoliosis may compromise respiratory function.

## Orthopedic Considerations

- \* If possible most procedures are performed early, before two years of age
- \* Two surgeries may be combined to reduce risks with surgery
- \* Often night splints are used to correct
- \* Recurrent deformities addressed with splints, braces
- \* PT and or OT recommended for strengthen and range of motion and ADL activities
- \* Some deformities can be corrected and ambulation is possible



## Seating and Mobility Challenges

- \* Positioning for support and maximum function
- \* Customizing seating to accommodate body size and limb length
- \* Conservation of energy
- \* Home, classroom and other environmental access
- \* Growth and changes in positioning needs

## Seating and Mobility Challenges

- \* Non-operative management of deformities
- \* Casts or splints after operative management of deformities
- \* Integration of other assistive technologies
- \* Access to alternate controls for driving powered mobility
- \* Normal sensation, so comfort may be a challenge
- \* Provide mobility that can be self-initiated

## 5 Step Assessment Process

- \* Data Gathering
- \* Observation
- \* Mat Assessment
- \* Skin considerations
- \* Trial and simulation / Overall goals

## Data Gathering

- \* History
- \* Family/ Child goals
- \* Transportation
- \* Home access
- \* Current equipment- if any

## Observation

- \* Current positioning
- \* Current equipment likes and dislikes
- \* Transfer ability

## Mat assessment

- \* ROM
- \* MMT
- \* Support needed to maintain functional seated position
- \* Measurements

## Skin considerations

- \* Present history or past history of breakdown
- \* Bony prominences
- \* Sensation
- \* Ability to redistribute pressures

## Overall Mobility Goals

- \* Increase independence/ positive conscious experiences
- \* Increase access to environment/peers
- \* Increase functional mobility
- \* Decrease fatigue



## Other Goals

- \* Educate the parents
- \* Encourage autonomy
- \* Work to diminish learned helplessness
- \* Safety, but less protective

## Will

- \* DOB: 8/4/2004
- \* Arthrogryposis
- \* Lives in a rural environment
- \* First equipment was a dependent mobility system : Convaid

