Arthrogryposis: When A "Non-Progressive" Diagnosis Progresses

Kay Koch, OTR/L, ATP and The van Halem Group
Stephanie Tanguay, OTR/ATP Motion Concepts

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

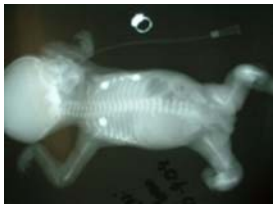
- Involved extremities are cylindrical in shape
- Deformities are usually symmetrical, and severity increases distally
- Distal joints are affected more frequently than proximal joints.
- Joint rigidity and diminished ROM (Range of Motion) may be present.
- The patient may have joint dislocation, especially 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