Arthrogryposis Syndromes:
Disorders with Congenital Contractures

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Arthrogryposis = “hooked/curved joints”
Physical finding and not a diagnosis
Multiple congenital joint contractures
Non-progressive limitation of movement
Heterogeneous group of disorders (150 entities)
Different etiologies, clinical courses, prognoses, genetics, and pathologic processes
Grouped from standpoint of treatment/management
# Hall Classification

## Classification of Conditions With Congenital Contractures

<table>
<thead>
<tr>
<th>Type of Disorder</th>
<th>Related Conditions</th>
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<tbody>
<tr>
<td>Limb involvement only</td>
<td>Amyoplasia (classic arthrogryposis)</td>
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<td>Distal arthrogryposis</td>
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<td>Poland anomaly</td>
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<td>Limb and other body areas</td>
<td>Diastrophic dysplasia</td>
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<td></td>
<td>Larsen’s syndrome</td>
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<td></td>
<td>Nail-patella syndrome</td>
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<td>Osteogenesis imperfecta</td>
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<td>Popliteal pterygium syndrome</td>
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<td>VATER association</td>
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<td>Limb and central nervous system</td>
<td>Fetal alcohol syndrome</td>
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<td></td>
<td>Myelomeningocele</td>
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<td>Spinal muscular atrophy</td>
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<td>Congenital muscular dystrophy</td>
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Etiology

Often unknown

Fetal Akinesia (decreased fetal movements)

1. fetal abnormalities (neurogenic, muscle, or connective tissue abnormalities, mechanical limitations to movement e.g. Oligohydramnios)
2. maternal disorders (infection, drugs, trauma, other maternal illnesses)
Frequency:

In the US: 1 in 3,000 live births.
Arthrogryposis Multiplex Congenita

- Amyoplasia
- Sporadic
- ~ 40%
- ~1:10,000 live births
- Risk of subsequent children ~ 5%
Prognosis:

- Life span is usually normal (related to the disease severity/associated malformations)
- ~20% of patients die in the 1st yr of life.
- Scoliosis may compromise respiratory function.
- Average of 5.7 orthopaedic procedures
- By age 5 yrs 85% ambulatory with normal mental development
- Complications of procedures
Evaluation:
- FHx
- Pregnancy history
- Delivery history
- Physical exam
- Multidisciplinary Team
Team

- Clinical geneticist
- Orthopedic surgeon
- Plastic surgeon
- Radiologist
- Neurologist
- Developmental pediatrician
- Pathologist
- Psychologist
- Physical and occupational therapists
- Social worker
- Educator
- Orthotist
- Rehabilitation engineer
Family history

- Affected children/family members (hyperextensibility, dislocated joints, dislocated hips, and clubfeet). Incidence of congenital contractures 2° and 3° relatives.
- Consanguinity
- Maternal age
- Intrafamilial variability (parent may be affected very mildly or may have had contractures early in infancy)
- Review previous miscarriages or stillbirths.
Pregnancy History

- Infants born to mothers affected with myotonic dystrophy, myasthenia gravis, or multiple sclerosis are at risk
- Maternal infections (rubella, rubeola, coxsackievirus, enterovirus, akabane)
- Maternal fever > 39°C, contractures due to abnormal nerve growth or migration.
- Teratogens
- Oligohydramnios
- Contractures, bleeding, trauma, hypoxia
Delivery history

- traumatic delivery in about 5-10% of cases.
- abnormal placenta, membranes, or cord insertion in case of amniotic bands or vascular compromise
- umbilical cord shortened or wrapped around a limb, leading to compression
- multiple births or twins
- death of one twin may lead to vascular compromise in the remaining twin
Physical Exam

- Best for establishing a diagnosis.
- The limbs are featureless and tubular.
- Normal skin creases are lacking.
- Deformities are usually symmetric, and severity increases distally, with hands and feet typically being the most deformed.
- 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.

Pain-free.

Firm, inelastic block to movement beyond a very limited range.

2/3 of patients: all four limbs are affected equally

1/3 of patients: lower-limb deformities predominate, and only on rare occasions do the upper extremities predominate.

Deformities tend to be more severe and more rigid distally.
Typical Baby with Arthrogryposis

Sometimes the face is long and the jaw large.

Wrist often bent up or out stiffly.

Hips often bent upward or outward stiffly; may be dislocated.

*Contractures* with 'webbing' of skin behind joints (at knees, hips, elbows, or shoulders).

Knees bent or straight, in a stiff position.

Mind completely normal.

Shoulders sometimes turned in.

Often arms are stiff at elbows and weak.

Hands and fingers often very weak.

Spine often curved but **trunk** strength usually normal.

Club foot common.
Upper Extremity

- Shoulders: adducted and internally rotated
- Elbow: more often extended than flexed
- Forearm: pronation
- Wrist: flexed severely, with ulnar deviation
- Fingers: flexed, clutching the thumb.
Lower Extremity

- Hips: flexed, abducted, and externally rotated, may be dislocated unilaterally or bilaterally.

- Knees: typically in extension (~50%), flexion (~10%) is possible
  - With no motion: articular surface deformity
    - Patellar elongation, flattening of femoral condyles and joint incongruity

- Feet: clubfeet are the rule, verical tallus deformity
Spine and CNS

- Scoliosis
- C-shape
- Normal intelligence
Clinical appearance of a child with amyoplasia.
Arthrogryposis multiplex congenita. The picture shows the classic limb position and fusiform limbs lacking flexion creases.
Arthrogryposis multiplex congenita at birth. Features include club feet, knee flexion deformity, and dislocated right hip. The articular surfaces are normal. Adaptive changes occur as a consequence of the fixed position. (From ref. 1, with permission.)
Investigations

Lab Studies:
- CPK
- IgM
- Viral titers (e.g., coxsackievirus, enterovirus, Akabane virus)
- Maternal antibodies to neurotransmitters in the infant may indicate myasthenia gravis.
- Cytogenetic studies
- Fibroblast chromosome study
- Nuclear DNA mutation analysis
- Mitochondrial mutation analysis
Imaging Studies:
- Photographs
- Radiographs
- Ultrasonography
- CT scan
- MRI

Other Tests:
- Skin biopsy
- Muscle biopsy
  - Distinguish myopathic from neuropathic conditions
- Electromyography (EMG)
- Nerve conduction tests

An autopsy should be performed to discover more about the following:
Management

- At birth:
  - Gentle stretching and ROM exercises
- Lightweight splinting
- Casting
- Soft tissue release
- Muscle transfer
- Osteotomy
Outcomes better if joint surgery is done early, before adaptive intraarticular changes.

Osteotomies are usually performed closer to the completion of growth.

Early motion, and avoidance of prolonged casting, may increase joint motion, improving function.

Many children require long-term bracing or other assistive devices.
Overview of the total patient

Overall goals:
- lower-limb alignment and stability for ambulation
- upper extremity motion for self care

Mobilization of joints by early and frequent range of motion exercises and splinting of the joint in a position of function with a removable orthotic

There are no studies clearly demonstrating that early mobilization improves these patient outcomes
Upper extremity
Shoulder

- Internal rotation rarely causes a problem
Elbow Deformities

- Range of motion exercises
- Early splinting
- Serial casting
Elbow Extension Contractures

1. One side to be treated at a time
2. Posterior capsulotomy and triceps tendon lengthening
3. Transfer of triceps, pectoralis, or latissimus dorsi
   - Improves active flexion if passive flexion $\geq 90^\circ$
   - Triceps to biceps transfer most common, good results in $\sim 80\%$
Wrist Deformities

Volar flexion and ulnar deviation

1. Splinting shortly after birth
2. Surgical
   • For fixed wrist contractures interfering with function
   • Release of:
     • Volar wrist capsule
     • Flexor Carpi Ulnaris tendon transfer to Extensor Carpi Radialis Brevis
     • Osteotomy of distal radius
     • Intracarpal extension osteotomy
     • Post-op splinting
       ….. to improve dorsiflexion
3. Arthrodesis
   • Near skeletal maturity in slight palmar flexion
Finger Deformities

Finger Flexion Contractures

1. Therapy and splinting
2. Surgery
   - Release of proximal intraphalangeal joint contractures not helpful for function
3. Arthrodesis
   - Intraphalangeal
   - At skeletal maturity
Thumb-in-Palm Deformity

1. Surgical
   - Z-plasty: release of adductor pollicis
   - Sublimis transfer
   - First metacarpal osteotomy
   - First metacarpophalangeal joint arthrodesis
   - Brachioradialis to thumb extensor transfer
Lower Extremity
Foot and Ankle Deformities

Clubfeet

1. Manipulation and serial casting (but generally resistant)
2. Surgical treatment at 6mo to 1 yr of age (before walking)
   - Aggressive soft tissue release, all tendons
   - Long term bracing, night bracing, ankle-foot orthosis
     - recurrence of up to 73% but more favored
     - taelectomy remains an option
Relapsed foot:

- **Talectomy**
  - may cause Tibiocalcaneal incongruity & loss of medial column
  - Progressive midfoot adduction if calcaneocuboid joint not fused
- **Decancellation of cuboid and/or talus**
  - Talus – maintains medial column and allows for easier triple arthrodesis later
- **Triple arthrodesis**
- **Gradual correction with circular frame external fixator**
  - wire transversely through distal tibial epiphysis and lock to tibial frame – prevent epiphyseal separation
- 95% can be made plantigrade with satisfactory outcome
**Vertical Talus Deformity**

1. In ~ 5%
2. Resistant to cast treatment
3. Surgical correction necessary
   - Anterior tibialis transfer to neck of talus
   - Permanent arch support necessary post-op
   - Subtalar fusion may be necessary in older patients
   - Triple arthrodesis may be necessary
4. Between 6 mo to 2 yrs
Knee Deformities

Flexion Contractures (~50%)

1. Mild: <15° - 20°, stretching and physiotherapy
2. 20° - 40°, surgery
   - Hamstring lengthening
   - Post-op splinting
3. Moderate: 40° - 50°, surgery
   - Z-plasty in popliteal fossa
   - Post-op serial cast changes
4. 60° – 80°, surgery
   - Gradual correction:
   - external fixation vs. femoral shortening
   - Post-op serial casting and chronic bracing

5. Sever: 80° - 90°, surgery
   - Soft tissue release and external fixator
   - Osteotomies for distal femoral extension
     - Internal fixation
     - Near skeletal maturity
   - Post-op splinting
Figure 1 Treatment of severe congenital knee flexion contracture. 

A. A 3-month-old infant with popliteal pterygium syndrome demonstrating 110° knee flexion contracture and popliteal web. 

B. At age 3 years, following daily stretching, serial casting, posterior capsular release, and Z-plasty of the skin has been performed. The residual knee flexion contracture measures 55°. 

C. Treatment involved application of a circular external fixator, distraction at the knee, and slow gradual extension of the knee flexion contracture. 

D. Full extension of the knee has been obtained. Range of motion and splinting were continued until age 6 years.
Extension Contractures

- Tight anterior capsule, hypoplasia of suprapatellar bursa, fibrosis of quadriceps

1. Percutaneous release of quadriceps tendon
2. V-Y plasty: quadriceps lengthening
3. Respond better to physical therapy and splinting
Considerable controversy
Closed reduction is rarely, if ever, successful
Operative reduction (improve function or decrease pain)
  - Studies to date have not found pain to be a problem with these hips
  - Operative procedures have potential to worsen function if they produce significant contractures.
Contractures $>45^\circ$ should undergo surgery

Flexion, external rotation and abduction contractures

1. Soft tissue release
2. Osteotomies

Unilateral dislocation

1. Bracing, traction, casting – rarely helpful alone
2. Open reduction (6mo-1yr)
   • Medial incision: best results but osteonecrosis
   • Anterior incision: stiffness
Bilateral Hip Dislocation

1. Controversial
2. Non-operative: functional ambulation without pain
3. Operative: improved quality and efficiency
   - Medial approach
   - Spica cast 8-12 weeks
4. Supple hip that is dislocated is preferred to a stiff reduced hip
Scoliosis

Uncommon at birth but 30-67% will develop it

Progressive, paralytic

Surgery

- progression, age, imbalance

25° - 40°

- brace treatment – not effective with progressive

>40°

- Spinal fusion with instrumentation
- Combined approach (ant/post)
- Treated same way as idiopathic scoliosis

Paralytic curves & lumbosacral obliquity >15°

- Fusion to pelvis recommended
Timing of Management

- **2-3 month**
  - Knee subluxation: closed reduction
- **4-5 month**
  - Knee subluxation: soft tissue release
  - Clubfoot deformity: surgical correction
- **9-12 month**
  - Hips dislocation(s): open reduction
  - Upper extremity splinting (may be from birth on)
- **3-4 years**
  - Upper extremity surgery
Outcomes

Overall function related to:
- Family support
- Patient personality
- Education
- Early effort to foster independence

Little correlation between physical deformity and function.
References

- Mihran O. Tachdjian, Pediatric Orthopedics
- Lovell and Winter, Pediatric Orthopaedics
- OKU: Pediatrics 2