

ARTHROGRYPOSIS MULTIPLEX

Multiple fixed joint deformities which are either local or generalized. It is neurogenic in 95% [loss of anterior horn cells] and in 5% it is myogenic.

Clinical.

Rare condition

In utero: oligohydramnios and paucity of fetal movement

At birth: Periarticular soft tissue structures become fibrotic causing teratological changes

Arthrogyposis is a clinical diagnosis, usually made at birth.

Distal joints are usually affected more severely

Normal facies and intelligence

No visceral abnormalities

Lack of skin creases

Initial Workup

Evaluation should include neurological studies

Enzymes tests: CPK [myopathies]

Muscle biopsy (at 3-4 months)

Differential diagnosis

Myelomeningocele, Moebius, Larsen's syndrome, Talipes equinovarus, Whistling face syndrome; [Freeman Sheldon]

Treatment

Shoulder Adduction and internal rotation
Shoulders rarely require treatment

Elbow When bilateral extension deformity; Restore flexion in one limb
A transfer of pectoralis major muscle to the biceps tendon

Wrists Wrist is in flexion and ulnar deviation deformity
Stretching exercises
Surgery: FCU to ECRB transfer rarely

Thumb Thumb in PAM deformity
1 web space release + skin graft
Adductor pollicis lengthening

Hips are usually flexed, externally rotated, and abducted
Teratological hip dislocation
Unilateral: Always needs open reduction
Bilateral: Leave it alone hip is high

Knee: Knee flexion contractures are common;

Treatment: Serial cast
Soft tissue releases + Posterior capsulotomy
Knee contractures should be corrected before hip reduction

Knee Extension: Stretching
Usually does not require surgery; Z plasty of the quadriceps

Spine 35% Scoliosis

Foot Rigid CTEV
Try first - serial manipulation
Casting in the newborn partially correct but deformity recurs
Soft tissue release
Talectomy

OVERGROWTH

Proteus syndrome - Overgrowth of feet and hands
Bizarre facial abnormalities
Scoliosis, genu valgum
Lipomas

Klippel-Treuaunay Syndrome - Overgrowth caused by underlying A-V malformations.
Cutaneous haemangioma and varicosities
Severely hypertrophied extremities.

Idiopathic Hemihypertrophy Caused by neurofibromatosis.
Often associated with renal abnormalities (Wilm's Tumour).
Leg length discrepancy