CONGENITAL UPPER LIMB DEFORMITIES

SWANSON’S CLASSIFICATION

1. Failure of Part formation
   Transverse absence: Congenital amputation
   Longitudinal absence: Radial Club hand
                        Ulnar Club hand
                        Cleft hand

2. Failure of Part Differentiation
   Radioulnar synostosis
   Camptodactyly
   Trigger thumb
   Syndactyly

3. Duplication
   Polydactyly: Pre-axial, Post-axial

4. Overgrowth
   Macrodactyly

5. Undergrowth
   Thumb hypoplasia (Blauth)

6. Miscellaneous
   Constriction ring syndrome
   Congenital dislocation of radial head
   Madelung’s deformity

FACTS

Congenital anomalies affect 1% to 2% of newborns
Approximately 10% of those children have upper-extremity
Genetics
   Trisomy of 21, 18, 13 Polydactyly, Syndactyly
   Apert’s syndrome AD

Environmental factors:
   Drugs: Talidomide, Dilantin, Warfarin
**ASSESSMENT**

Initial visit: an emotion filled event.
Parents: Underlying guilt.
Parents have difficult for families to accept.
Risk for Future Pregnancies: Genetic counselling

**TRANSVERSE AMPUTATION**

Part distal is missing.
Rare
Eg: Absence hand at wrist

**INTERCALARY AMPUTATION**

Segment in between is absent.
More common with Talidomide Intake during pregnancy

**RADIAL CLUB HAND**

1: 30,000

Bilateral in 60%

**Clinical**

Absence of radius and radial rays
Bowing of the ulna

Associated
- Anal atresia
- Tracheo- oesopharyngeal fistula
- Radial bone absence
- Renal anomalies
- Cardiac anomalies
**Treatment**

Use of serial casts
Realignment and stabilization: Lower end of ulna is transferred radially.
Tendon transfers - Ulnar side tendons transferred radial side
Correction of the ulnar bowing – osteotomy

**RADIO-ULNAR SYNOSTOSES**

AD;

Bilateral in 60%

Site: Proximal 1/3 of forearm

Deformity: Forearm pronation

Rx: Leave it alone.

When bilateral: rotational osteotomy in one forearm to a functional position

**TRIGGER THUMB**

Often bilateral, with fixed flexion of thumb

If present >1 yrs Rarely recover

Treatment: - Surgery

Release A1 pulley [base of the tendon sheath]
SYNDACTYLY

Syndactyly: Syn = together in Greek

Mnemonic 5,15,50,30:[Thumb and Index 5%;
Ring and little: 30%]

More common in white males, AD
Present bilaterally in 50%

Types: I Isolated
II Complex  Trisomy 13, 14, 21

Apart syndrome [clinodactyly; facies]
Holt Oram Syndrome [ASD, Radial club hand]
Poland’s [Absence of Pectoralis major]

Treatment

Surgery: At 18 months old.

Full-thickness skin grafting is almost always required for soft tissue coverage.

Technique

Mark the skin flaps  Z plasty
Dorsal butterfly flap, to make the web
Both sides of the same digit : never separated on the same day (>6 wks)
POLYDACTYLY

POST AXIAL POLYDACTYLY

Post axial [next to the little finger]
Is a common hand anomaly
10 times more in Blacks
AD
Nonsyndromal

PRE AXIAL POLYDACTYLY

Preaxial [next to the thumb]
Is common duplication in whites
AR [17 chromosome]
Syndromal

Treatment
Excise the rudimentary finger

MACRODACTYLY

- Hamartomatous enlargement of soft tissue
- Associated with neurofibromatosis

When hemi-hypertrophy, suspect
   Wilm’s tumor
   Adrenal carcinoma
   Hepatoblastoma

Treatment
   Debulking,
   Osteotomy
   Epiphysiodesis
CONSTRICTION RING SYNDROME [CRS]

Also called: Streeter’s syndrome
1 in 15,000 live births
The etiology of CRS is unclear
It is not genetic
CRS frequently affects both arms and all four limbs
Commonly associated with clubfoot
Surgical treatment: Z-plasty of constriction

CONGENITAL DISLOCATION OF THE ELBOW

Incidence: 0.16%

D/D
Old Monteggia
Multiple endochondromatosis or exostosis
Radio-ulnar synostosis

Clinical: Family history; Bilateral anomaly;
Loss of supination

Radiological: Hypoplasia of the Capitellum
Dome shaped Head

Treatment: If painful, excision after maturity
MADELUNG’S SYNDROME

Epiphyseal arrest on ulnar and volar half of the distal radius

Joint is directed ulnarward and volarward

AD variable expressivity; Females (4:1)

Wrist motion: Extension and supination are limited

X ray may show: Classical deformity

Inferior Radio-ulnar dislocation

**Treatment**  
Corrective osteotomy

Physeal resection and fat graft

SPRENGEL’S SYNDROME

Failure of the normal caudal migration of the scapula

Associated with Congenital scoliosis

Klippel-Feil syndrome

The superomedial scapula is connected to the proximal cervical spinous process through an omovertebral connection (bone, cartilage) in 30%

**Treatment:** Surgical correction
TORTICOLLES

Associated with other “Molding disorders”

Hip dysplasia
Metatarsus adductus
Plagiocephaly

Treatment
Stretching exercise 90% good results
Surgery if necessary at one year (Distal or proximal release or both)

Look for
Upper cranial anomaly
Ophthalmologic disorder
Posterior fossa brain tumor

KLIPPEL-FEIL SYNDROME

Low hair line
Limited ROM of the neck
Can be associated with Sprengle’s shoulder,
Renal or cardiac anomaly

Avoid contact sports
3. LOWER LIMB CONGENITAL DEFORMITIES

INFANTILE COXA VARA

Clinical

Trendlenburg gait
Adduction is more than abduction
Supratrochanteric shortening

X ray

Coxa Var a <110°
Hilgenreiner epiphyseal angle > 60°

Treatment  Corrective Valgus osteotomy

PROXIMAL FEMORAL DEFICIENCY

Can be associated with absence of fibula. tarsal coalition is common

Type A  [congenital short femur]
Gap between proximal and distal femur, which ossifies by maturity
Femur is 80% short

Type B
The femoral head and acetabulum are hypoplastic; The femoral gap does not unite

Type C
The Femoral head is absent
The Acetabulum is present but dysplastic

Type D
The femoral head and acetabulum: absent
Treatment

Type I  Ilizarov lengthening of the femur
Type II  Valgus osteotomy and fusion
Type III and IV
  Extended prosthesis
  Syme’s and extended prosthesis
  Knee fusion and Syme’s [Brown]
  Van Ness procedure: Informed consent and Psych evaluation
**ABSENCE OF FIBULA**

Absence of Fibula more common than tibia

Can be associated

- PFFD,
- ACL absence,
- Toe ray absence
- Tarsal coalition
- Tibial bowing: Anteromedial bow

Treatment: Syme’s amputation

**POSTEROMEDIAL BOWING OF TIBIA**

Postural

Posteromedial bowing of tibia

Is usually benign

Observe for shortening of the limb

Treatment: Regresses with stretching exercises

**ANTEROLATERAL BOWING OF TIBIA**

Rare but serious condition

**Treatment**

- Protective brace
- Bone graft and fixation of tibia
- Sometimes repeat operation
- May need disarticulation of the ankle
CONGENITAL VERTICAL TALUS

Autosomal dominant

Associated with: trisomy of 13

Talus remain in plantar flexion both in dorsiflexion and plantarflexion.

**Treatment:** Soft release and plaster cast