Paediatric Lower Limb Deficiencies

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Outline

- Congenital vs. Acquired
- Limb Morphogenesis
- Terminology and Classification
- Transverse Deficiencies
- Longitudinal Deficiencies
- Acquired Amputations

Congenital & Acquired deficiencies

Aetiology

Congenital :

- Genetic
- Vascular
- Intrauterine amputation
- Maternal factors

Acquired:

- Meningococcal
- Burns
- Trauma
- Vascular malformations
- Tumour

Congenital

- No sense of loss
- Nothing new to adjust to
- Prosthesis as an aid
- Family adjustment issues

Acquired

- Profound sense of loss
- Period of readjustment
- How well they adjust affects acceptance of prosthetic limbs

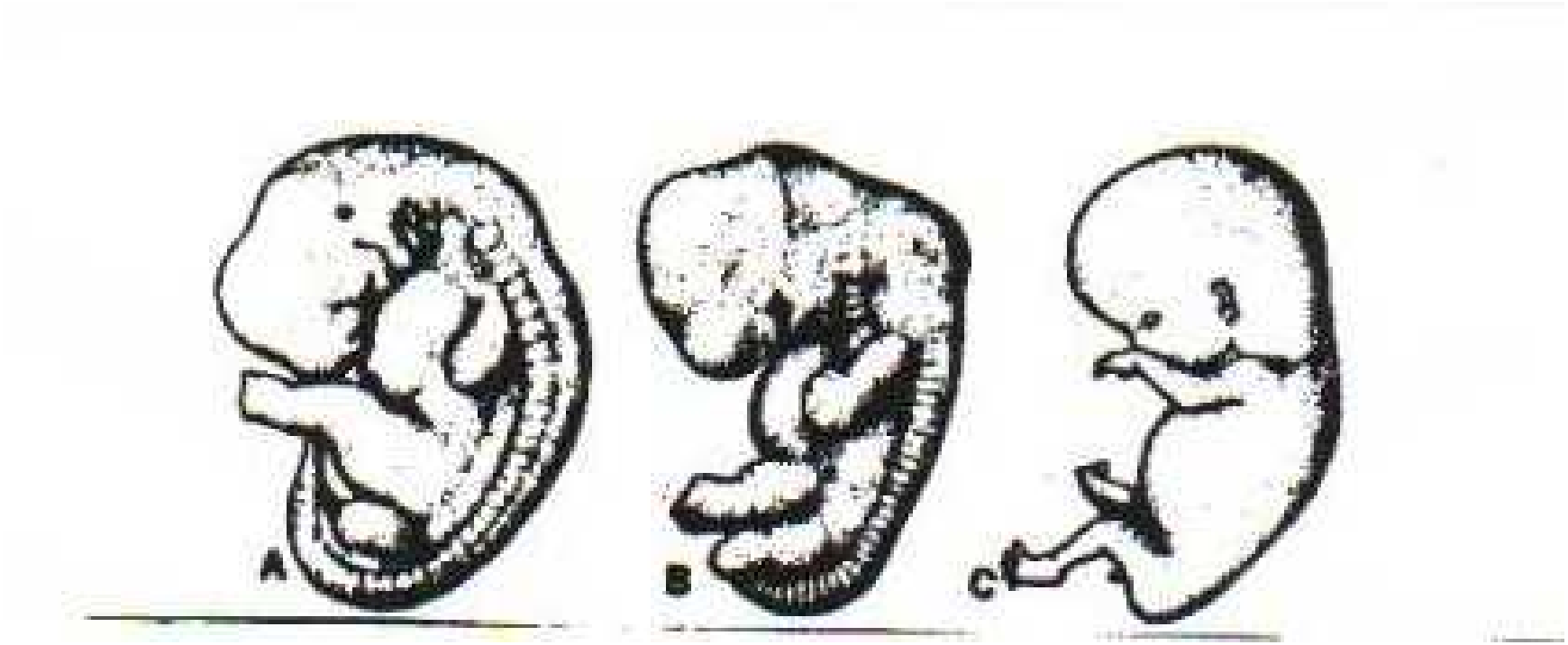
Limb Bud development

- Key genes involved in growth and patterning of the limb buds
- Formation involves numerous genes, the actions of which are interlinked
- Limb bud development begins 4th week

Limb Morphogenesis

- Thickening of lateral plate mesoderm signals the overlying ectoderm to thicken and form a ridge
- Apical ectodermal ridge (AER) controls proximal-distal limb
- Limb develops in a proximal-distal direction

Limb Morphogenesis



Congenital Limb Deficiencies

- About 1 : 5-10,000 births
- May have complex genetics - important for geneticist to see family.
- In most cases cause unknown, low recurrence risk

Congenital Limb Deficiencies

- Most defects occur in period of limb morphogenesis
- Weeks 4-8 of gestation most critical time
- Sensitive period peaks 5^{th} and 6^{th} weeks

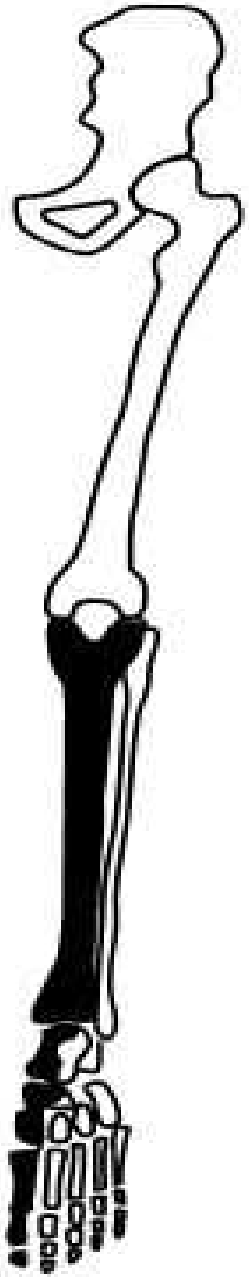
Upper and Lower Limb buds rotated at 7 weeks but digits not separated


Upper limb total deficiency

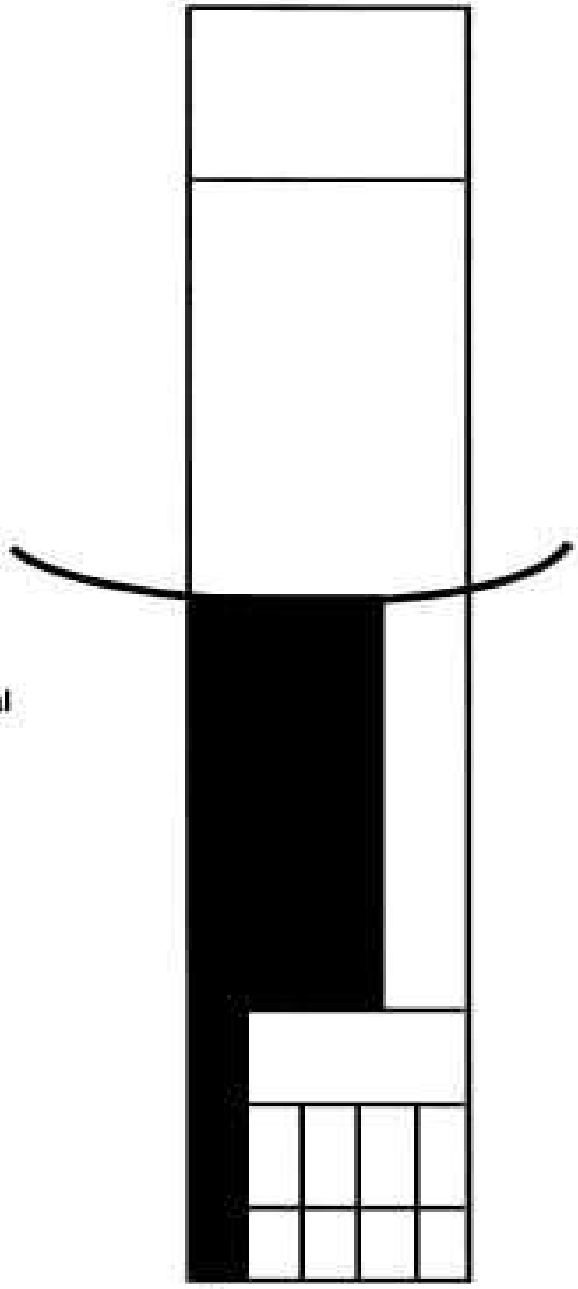
Terminology and Classification

- ISO Classification 1989 is the accepted international standard
  - Transverse
    - limb developed normally to a particular level beyond which no skeletal elements exist
  - Longitudinal
    - Reduction or absence of an element/s within the long axis. There may be normal distal skeletal elements. Name the bones affected
  - Partial / Total

Standard 8548-1:1989 http://www.iso.org



Longitudinal
Tibia Total
Tarsus Partial
Ray 1 Total



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Further Terminology

Amelia: complete absence of the limbs

Hemimelia: absence of some portion of the limb

Adactyly: absence of fingers

Achiera: absence of a hand

Apodia: absence of a foot

- Paediatric deficiencies are often mixed and need to be considered in very functional terms :
  - Shortening
  - Unstable
  - Terminal loss
- Conversion Amputation is never applicable in the Upper Limb

Transverse Deficiencies

- The limb has developed normally to a particular level beyond which no skeletal elements exist, although there may be digital buds

- Aetiology: Vascular disruption, Failure of formation, Constriction/ Amnionic Bands

Vascular Disruption

Constriction Rings/
Amnionic Bands

Longitudinal Deficiencies

- Proximal Focal Femoral Deficiency (PFFD)
- Fibula Deficiency
- Tibial Deficiency
- Femur Fibula Ulna Syndrome
- Partial foot (lateral ray deficiency)

PFFD

- Profoundly short femur with bulbous thigh segment lying in external rotation & flexion
- flexed knee with cruciate insufficiency
- foot at level of opposite knee or just below

- most unilateral
- > 60% associated absence of fibula / other skeletal abnormality


PFFD

Proximal Femoral Focal Deficiency (PFFD)

Type A

- defect between femoral head & shaft with spontaneous restoration during growth

Type B

- persistent discontinuity between hip joint & femur

Type C

- femoral head never ossifies / dysplastic acetabulum

Type D

- complete absence of the femoral head and acetabulum

PFFD Management options

- Lengthening of femur
- Surgical procedures to provide hip stability & bony continuity
- Syme amputation / removal of foot + fusion of knee joint & prosthesis
- Van Nes rotationplasty
- non standard prostheses


HOSPITAL

Longitudinal Deficiency of Fibula

- Shortening and anterior bowing of tibia
- absence of lateral metatarsal rays
- equinvalgus foot deformity
- cruciate ligament deficiency

Fibula Deficiency Management Options

- Extension prosthesis
- leg lengthening +/- ankle stabilisation
- conversion amputation through ankle & prosthetic restoration with supracondylar suspension for knee stability

Improving ankle stability and leg length discrepancy

Bilateral Longitudinal Fibula deficiency and complete deficiency of the 5th ray of the foot

Longitudinal Deficiency of Tibia

- Complete or partial

In complete absence:

- Short and relatively functionless leg
- Gross knee and ankle instability
- Equinovarus foot deformity
- No potential for development

Longitudinal Deficiency of Tibia

Management:

- Through knee amputation
- Ankle disarticulation
- Centralisation of fibula / reconstruction

“Conversion” amputations

- Aim for a weight bearing stump
- Enables better prosthesis use

Joint disarticulation:

- Less risk of bony overgrowth as bones grow
- Maximises the residuals growth potential as leaves both growth plates intact

Acquired Amputations

- Lawn mower
- motor vehicle
- farm machinery
- burns
- vascular catheterisations
- Landmines
- Tumours

Tumours

- May require amputation or various strategies for limb salvage
- The Van Nes Rotationplasty: distal femoral tumour

Van Nes Rotationplasty

- Tumour removed while the neurovascular bundle and distal portion of the tibia and foot are maintained
- Tibia and foot are rotated 180 degrees, attached to the remaining proximal femur
- The ankle is at the hight of the contralateral knee
- Benefits: functional “knee” joint
- Disadvantage: appearance of the limb




Questions?