THE DILEMMA OF
THE CONGENITAL SHORT LIMB

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OBJECTIVES

• Congenital Longitudinal Deficiencies
  1. Congenital femoral deficiency (Proximal femoral focal deficiency)
  2. Fibula Hemimelia
  3. Tibial Hemimelia

• Management Dilemma:
  – Management Goals
  – Reconstruction & Lengthening
  – Amputation & Prosthesis
MANAGEMENT GOALS

• Function
• Appearance
• Convenience
• Quality of life
• Efficiency
  – Number of interventions
• Expense
  – Interventions
  – Prostheses
GENERAL CONSIDERATIONS

- Magnitude of length discrepancy
  - Current length predicts length at maturity
  - Multiplier Method
- Integrity of hip joint
- Integrity of the knee joint
- Integrity of the ankle joint
- Size and shape of the foot
- Deformity of bone
- Contralateral lower limb
“Congenital Femoral Deficiency”

- Spectrum of conditions/severity
  - Congenital short femur without an ossification defect
  - Apparent or true discontinuity between femoral neck and shaft (PFFD)
    - Hip dysplasia
    - Deformity
    - Knee joint

- Etiology unknown
CONGENITAL SHORT FEMUR

- Short thigh
- Femur: Anterolateral bow
- Genu valgum
- Valgus- external rotation contracture of knee
- ACL deficiency
- Short hamstrings
- Fibular hemimelia +/-
CLASSIFICATION OF PFFD

• Aitken: ABCD

• Gillespie-Torode: ABC

• Paley CFD: 1,2,3

• Hamanishi: I-V with 10 subgroups

• Fixsen, Lloyd-Roberts: I, II, III
PFFD: AITKEN CLASSIFICATION

<table>
<thead>
<tr>
<th>Type</th>
<th>Femoral Head</th>
<th>Acetabulum</th>
<th>Femoral Segment</th>
<th>Relationship Among Components of Femur and Acetabulum at Skeletal Maturity</th>
</tr>
</thead>
<tbody>
<tr>
<td>A</td>
<td>Present</td>
<td>Normal</td>
<td>Short</td>
<td>Bony connection between components of femur</td>
</tr>
<tr>
<td></td>
<td></td>
<td></td>
<td></td>
<td>Femoral head in acetabulum</td>
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<tr>
<td></td>
<td></td>
<td></td>
<td></td>
<td>Subtrochanteric varus angulation, often with pseudarthrosis</td>
</tr>
<tr>
<td>B</td>
<td>Present</td>
<td>Adequate or moderately dysplastic</td>
<td>Short, usually proximal bony tuft</td>
<td>No osseous connection between head and shaft</td>
</tr>
<tr>
<td></td>
<td></td>
<td></td>
<td></td>
<td>Femoral head in acetabulum</td>
</tr>
<tr>
<td>C</td>
<td>Absent or represented by ossicle</td>
<td>Severely dysplastic</td>
<td>Short, usually proximally tapered</td>
<td>May be osseous connection between shaft and proximal ossicle</td>
</tr>
<tr>
<td></td>
<td></td>
<td></td>
<td></td>
<td>No articular relation between femur and acetabulum</td>
</tr>
<tr>
<td>D</td>
<td>Absent</td>
<td>Obturator foramen enlarged, Pelvis squared in bilateral cases</td>
<td>Short, deformed</td>
<td>None</td>
</tr>
</tbody>
</table>

- **Femoral head**: Present
- **Acetabulum**: Normal to moderate dysplasia
- **Femoral head**: Absent
- **Acetabulum**: Severe dysplasia to absent
RIGHT: AITKIN D; LEFT: AITKIN B
PFFD: GILLESPIE-TORODE CLASSIFICATION

Group A

- Foot opposite midpoint of contralateral tibia
- LLD $\leq$ 20%
- Femur $>$ 60%
  - Limb lengthening
- Femur $\leq$ 50%
  - Rotationplasty
PFFD: GILLESPIE-TORODE CLASSIFICATION

Group B (Aitken A,B,C)

• Foot opposite contralateral knee or above

• LLD: 40%

• Options: Prosthesis +
  – Knee fusion
  – Amputation
  – Rotationplasty
Group C (Aitken D)

- Subtotal absence of femur
- Options: Prosthesis +
  - Prosthetic management
  - May have to retain foot to improve suspension
  - Brown or Steel procedure (fuse femur to pelvis, knee becomes hip)
PFFD: PHYSICAL EXAM

- Hip: flexed, abducted, externally rotated
- Femur: Apex anterior bow
- Distal femur: externally rotated
- Short thigh
- Genu valgum
- Knee flexion contracture
- Knee: ligament deficiency
- Fibular hemimelia: 45% -75%
  - Short tibia
  - Equinovalgus deformity
  - Absent lateral rays
- 15% bilateral
PFFD: GAIT

• Usually walk at expected age

• Walk on knee of long side

OR

• Walk with knee flexed on long side
• Equinus on short side
**MANAGEMENT OPTIONS**

<table>
<thead>
<tr>
<th>RECONSTRUCTIVE</th>
<th>AMPUTATION/PROSTHESIS</th>
</tr>
</thead>
<tbody>
<tr>
<td>• Hip joint stabilization</td>
<td>• Extension prosthesis</td>
</tr>
<tr>
<td>• Contracture releases</td>
<td>• Amputation + prosthesis</td>
</tr>
<tr>
<td>• Pseudarthrosis stabilization</td>
<td>• Rotationplasty + prosthesis</td>
</tr>
<tr>
<td>• Deformity correction</td>
<td></td>
</tr>
<tr>
<td>• Knee joint</td>
<td></td>
</tr>
<tr>
<td>• Ankle joint</td>
<td></td>
</tr>
<tr>
<td>• Lengthening/s: up to 3</td>
<td></td>
</tr>
<tr>
<td>• Contralateral epiphyseodesis</td>
<td></td>
</tr>
<tr>
<td>• Bracing: AFO</td>
<td></td>
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</tbody>
</table>
PFFD: LIMB LENGTHENING

Final discrepancy: 17-20cm or > 50% of other side

Stage 1

1. Contracture release
   - Flexion contracture release (Rectus femoris & psoas)
   - Abduction-external rotation contracture release
   - Release contracted IT band

2. Proximal femoral osteotomy & fixation
   - Repair/resect pseudarthrosis if present
   - Correct apex anterior bow
   - Correct proximal femoral varus: NSA of 135°
   - Correct external rotation deformity

3. Stabilize hip
   - Correct acetabular dysplasia: Salter, Pemberton, Dega

Stage 2

- Can be corrected in 3 or fewer lengthenings +/- contralateral epiphysseodesis
- Aim 15% - 20% (5 cm to 8 cm) gain at each lengthening
STAGE 1: PREPARATION FOR LENGTHENING
Complications of Lengthening

- Pin site infections
- Incomplete corticotomy
- Premature consolidation
- Poor regenerate formation
- Neurovascular injury
  - Intraoperative
  - Excessive/rapid distraction
  - Impingement against wires/pins

- Joint contractures
  - Hip
  - Knee
- Hip instability
- Knee instability
- Fracture
PFFD - PROSTHESIS

Final discrepancy >20cm or <50% of Other Side

- Distal end of residual limb to be at least 5 cm above contralateral knee at skeletal maturity (allow proper placement of prosthetic knee)
- Consider thigh reduction to facilitate prosthetic fitting
  - Surgical reduction or liposuction
- Make the limb a more efficient lever arm for the prosthesis
- Provide a more cosmetic prosthesis
- Optimal age for surgery 2½ to 3 yrs
PFFD - PROSTHESIS
Final discrepancy >20cm or <50% of Other Side

1. Equinus prosthesis
2. Symes amputation + prosthesis
3. Syme’s amputation + knee arthrodesis + prosthesis (take 1 or both growth plates)
4. Syme’s amputation + femoral-pelvic arthrodesis + prosthesis
5. Rotationplasty+ knee arthrodesis + prosthesis (ankle becomes knee)
6. Rotationplasty + femoro-pelvic arthrodesis + prosthesis (knee becomes hip)
PFFD: VAN NES ROTATIONPLASTY

• Advantages
  – Control knee with gastrocnemius
  – Better gait kinematics
  – More energy efficient

• Disadvantages
  – Cosmetic concerns
  – Tendency to derotate with growth (or failure to achieve sufficient rotation at surgery)
  – Rotational control problems if hip unstable
PFFD: VAN NES ROTATIONPLASTY

Knee Arthrodesis & Rotation

Tibial Segment Resection & Rotation

[Diagram of knee arthrodesis and tibial segment resection with rotational correction.]
BROWN PROCEDURE
ILIO-FEMORAL FUSION & ROTATIONPLASTY
Fibular Hemimelia

- Most common cong. limb deficiency
- Incidence: 1 in 40,000 live births
- Spectrum of abnormalities
- Post-axial hypoplasia of the limb
  - Congenital femoral deficiency
  - Genu valgum
  - Femoral external torsion
  - Knee instability: - ACL
  - Tibial shortening
  - Anterior bow +/- dimple
  - Hypoplasia or absent Fibula
- Foot abnormalities
  - Rigid valgus foot
  - Frequent tarsal coalition
  - Lateral rays missing
FIBULAR HEMIMELIA: 
RADIOGRAPHIC FEATURES

- Leg length discrepancy
- Hypoplastic or absent fibula
- Shallow femoral intercondylar notch
- Small tibial spines
- Hypoplastic lateral femoral condyle
- Mild to severe femoral shortening
- Ankle valgus
- Ball and socket ankle
- Tarsal coalitions
FIBULA HEMIMELIA: CLASSIFICATION: ACHTERMAN & KALAMCHI
FIBULA HEMIMELIA

BIRCH FUNCTIONAL CLASSIFICATION

Type I: Functional foot
IA 0 to 5% inequality
IB 6% to 10% inequality
IC 11% to 30% inequality
ID > 30% inequality

Type II: Nonfunctional foot
IIA Functional upper extremity
IIB Nonfunctional upper extremity
MANAGEMENT GOALS

- Function
- Appearance
- Convenience
- Quality of life
- Efficiency
  - Number of interventions
- Expense
  - Interventions
  - Prostheses

Strategies

A. Reconstructive
  - Ankle & foot
  - Lengthening
  - Deformity correction

B. Amputation & prosthesis
  - Syme’s or Boyd +
  - Corrective osteotomy
  - Bilateral fibular deficiency
  - Preserve feet is upper limbs are non-functional
## FIBULAR HEMIMELIA: TREATMENT
### BIRCH RECOMMENDATIONS

### “Functional” foot vs. “Nonfunctional” foot

<table>
<thead>
<tr>
<th>Classification</th>
<th>Treatment</th>
</tr>
</thead>
<tbody>
<tr>
<td>Type I: Functional foot</td>
<td></td>
</tr>
<tr>
<td>IA 0 to 5% inequality</td>
<td>Orthosis/epiphysiodesis</td>
</tr>
<tr>
<td>IB 6% to 10% inequality</td>
<td>Epiphysiodesis ± lengthening</td>
</tr>
<tr>
<td>IC 11% to 30% inequality</td>
<td>1 to 2 lengthenings (or amputation)</td>
</tr>
<tr>
<td>ID &gt; 30% inequality</td>
<td>&gt; 2 lengthenings versus amputation</td>
</tr>
<tr>
<td>Type II: Nonfunctional foot</td>
<td></td>
</tr>
<tr>
<td>IIA Functional upper extremity</td>
<td>Early amputation</td>
</tr>
<tr>
<td>IIB Nonfunctional upper extremity</td>
<td>Consider salvage</td>
</tr>
</tbody>
</table>
FIBULA HEMIMELIA: CLASSIFICATION:
PALEY CLASSIFICATION

Type I - Stable
LDTA = 85 - 90°
ADTA = 80 - 85°

Type II - Dynamic Valgus

Type III - Fixed Equino-valgus
a) ankle type
ADTA ≥ 90°

Type III - Fixed Equino-valgus
b) subtalar type
ADTA ≤ 90°

Type III - Fixed Equino-valgus
c) combined ankle and subtalar type
ADTA ≥ 90°
FIBULAR HEMIMELIA: RECONSTRUCTION

- Resection of fibular anlage
- Tendo-Achilles lengthening
  - To correct equinus
- Supramalleolar osteotomy
  - To correct valgus and procurvatum
- Application of frame
  - To correct anterior bow
  - Lengthening
FIBULAR HEMIMELIA: RECONSTRUCTION LENGTHENING

- Age 18-24 months: Ankle reconstruction + lengthening 5 cm

- Age 7-10: 2nd Lengthening 5 cm

- Age 12-14: 3rd Lengthening 5 cm &/or Epiphysiodesis 5 cm

- Genu valgum
  - Distal medial femoral physeal hemi epiphyseodesis
  - Guided growth
Predicted 14 cm LLD
TIBIAL HEMIMELIA

• Longitudinal preaxial deficiency of the tibia
  – Hypoplasia
  – Partial absence
  – Complete absence

• One in million children

• Autosomal dominant; mainly sporadic
TIBIAL HEMIMELIA: CLINICAL FEATURES

- Short or absent tibia
- Rigid equinovarus supinated foot
- Preaxial polydactyly
- Relatively long fibula
- Prominent ends of fibula
- 60-75% associated congenital anomalies
  - Musculoskeletal
  - Other organ systems
TIBIAL HEMIMELIA: JONES CLASSIFICATION

Radiographic Description

1a
Tibia not seen
Hypoplastic lower femoral epiphysis

1b
Tibia not seen
Normal lower femoral epiphysis

2
Distal tibia not seen

3
Proximal tibia not seen

4
Diastasis
JONES TYPE 1 TIBIAL HEMIMELIA (R)

- 1y 3m male
- h/o coarctation of the aorta
- R knee contracture 80°
- No quads fxn
- L knee full ext with quads fn
- Equinovarus feet

Tibia not seen
Hypoplastic lower femoral epiphysis
JONES TYPE 2 TIBIAL HEMIMELIA (L)

Distal tibia not seen
Jone’s Type 4 Tibial Hemimelia

Type 4 (congenital diastasis of ankle)

Rx:
Lengthening and realignment through supramalleolar osteotomy (position the foot plantigrade)
TIBIAL HEMIMELIA
TREATMENT OPTIONS

• Knee disarticulation

• Centralization of fibula under femur

• Transfer/synostose fibula to proximal tibia (if good quads & active knee extension)

• Syme amputation
TIBIAL HEMIMELIA MRI FOR DECISION MAKING

• Helpful in assessing the presence of
  – cartilaginous anlage in the proximal tibia,
  – presence of the patella,
  – quadriceps and patellar tendons

• Helpful for deciding
  – Level of amputation (Syme or knee)
  – Feasibility of proximal reconstruction
TIBIAL HEMIMELIA

Bottom line: if you have a knee, you can lengthen. If not, then amputate.
Amputation Strategy: Level

Through knee Disarticulation
- In the absence of a knee joint or in the absence of active quadriceps function (Jones Types 1 & 3)

Below Knee amputation
- Short proximal tibial fragment can become a useful below-knee amputation stump through natural growth (if proximal growth plate is preserved) or by surgical lengthening with innovative modern prosthetic fitting.

Syme or Boyd Amputation
- If knee joint and active quadriceps function is present and there is
  - Severe shortening of the extremity, &/or
  - Severe deformity of foot and ankle
  - Severe ankle joint instability
TREATMENT PRINCIPLES

1. Preserve length
2. Preserve important growth plates
3. Perform disarticulation, rather than transosseous amputation, whenever possible
4. Preserve the knee joint whenever possible
5. Stabilize and normalize the proximal portion of the limb
6. Be prepared to deal with other issues in addition to limb deficiency
Bilateral Absent Tibia

- 1. 3 year old boy
- Born with flexion contracture of both knees
- 80 degree flexion contracture both knees
- No active quadriceps
- Knee walks
- Varus feet
- X-Ray: Bilateral Type 1a tibial hemimelia
Bilateral through knee disarticulation
TYPE 2 TIBIAL HEMIMELIA
FIBULA TO TIBIA SYNOSTOSIS: 1 BONE LEG

Type 2 (proximal tibia):
Rx: fibular to tibia synostosis – one bone leg
6 month old girl with Left Jones Type 2 Tibial Hemimelia
At age 2 yrs:
Fibular to Proximal tibial synostosis
10 yrs old 8.5 cm LLD
Expected LLD = 10.2 cm

High riding fibular head
10 year old Tibial Hemimelia
8.5 cm LLD (Exp: 10.2 cm)

Fibular head descended
10 yr old Tibial Hemimelia: 8.5 cm LLD
5.5 cm lengthening
TYPE 4 TIBIAL HEMIMELIA
LENGTHENING & REALIGNMENT THROUGH
SUPRAMALLEOLAR OSTEOTOMY

Type 4 (congenital diastasis of ankle)

Rx:
Lengthening and realignment through supramalleolar osteotomy (position the foot plantigrade)
1 Month Old
6 YRS 4 MTHS OLD: Bil. Tibial Hemimelia
6 YRS 4 MTHS OLD

RIGHT

LEFT
Thank You