# Rehabilitation Management Involving a child with Multiple Congenital Limb Deficiency

Brenda Reagan, OTR/L, MS
Jared McNeill, CPO
Cindy Dodds, PT, PhD

1

## Learning Objectives

Following this learning session, participants will:

1. Be introduced to role of a quality of life model and International Classification of Functioning, Disability and Health framework in rehabilitation
2. Review congenital deficit causes, types, and prosthetic considerations
3. Learn therapeutic activities and environment adaptions to improve function and participation across for a 3-year-old with rare disability
4. Discuss rehabilitation experiences involving other pediatric cases with similar congenital limb deficiencies

2

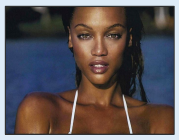
Article 23 of the United Nation's Rights of the Child asserted that:

- **"a child with mental or physical disabilities is entitled to enjoy a full and decent life, in conditions that ensure dignity, promote self-reliance and facilitate the child's active participation in the community."**

- http://www.un.org/esa/socdev/unyin/documents/children_disability_rights.pdf

3

## Models?

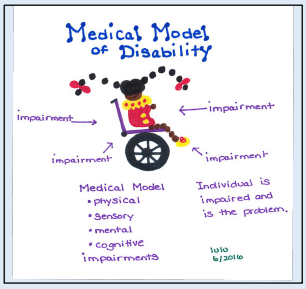




4

## Custodial Model

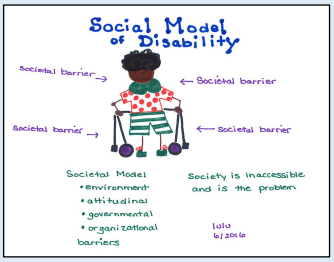
5

## Medical Model




6

## Social Model

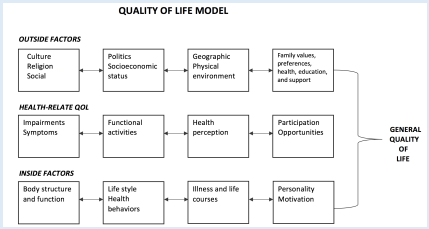



7

## Quality of Life Model

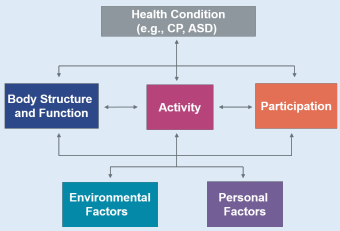
8

## Quality of Life Model



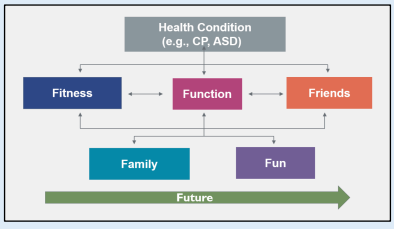

9

## International Classification of Functioning, Disability and Health




10

## F-Words: International Classification of Functioning, Disability and Health




11


12

# Prosthetic Management of Children with Congenital Multiple Limb Deficiencies

13

## Limb deficiency statistics

- CDC estimates
  - 4 out of every 10,000 babies will have upper limb reductions
  - 2 out of every 10,000 babies will have lower limb reductions.
  - Some of these babies will have both upper and lower limb reduction defects.
  - 0.8% of all amputations

Content source: Division of Birth Defects and Developmental Disabilities, NCBDDD, Centers for Disease Control and Prevention

14

## Congenital Limb Deficiency

- Congenital limb defects occur when a portion or the entire upper or lower limb fails to form normally when the baby is developing in the uterus.

15

## Congenital deficiency causes

- Constriction band syndrome (Streeter's Dysplasia),
  - amniotic bands result in complete or nearly complete antenatal amputation.
- Genetic
  - as in some cases of longitudinal deficiency of the radius or ulna.
- Injury or developmental failure of the limb during the first six weeks of pregnancy.
  - Due to anoxia, drugs, irradiation, chemicals, certain viral infections or an accident during the early part of the pregnancy.
- In the majority of cases, the cause is simply not identifiable

Cummings and Kapp, 1992

16

## Assessment

- **Level of amputations: To define amputation levels and how this relates to functional outcomes.**
- Ideal outcomes: Determining expected functional independence.
- The role of the rehabilitation team: Individual roles of the team members in accomplishing the rehabilitation of the amputee patient.
- Prosthetic candidacy: Based on comorbidities, compliance, energy expenditure, K-levels, and objective measures and subjective assessments.

17

## Congenital Limb Deficiency

- The most common of these defects are:
  - No limb at all
  - Part of the limb doesn't separate, often seen in fingers or toes
  - Duplication, often seen as extra fingers or toes
  - The limb is much larger than the normal limb (overgrowth)
  - The limb is much smaller than the normal limb (undergrowth)

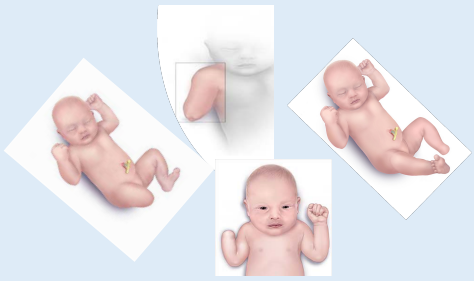
18

## Types of deficiencies

- **Transverse deficiencies**
  - occur when the limb does not develop past a certain point.
    **Amelia** - absence of limb
    **Acheiria** - missing a hand
    **Apodia** - missing a foot
- **Longitudinal deficiencies**
  - occur when a bone is missing or does not develop normally.
    (Usually the foot or hand is present although it may not have developed normally.)
    **Hemimelia** absence of a portion or half of a limb
    **Proximal Femoral Focal Deficiency**

19

## Transverse deficiencies



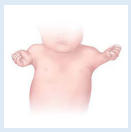
20

## Types of deficiencies

- **Transverse deficiencies**
  - occur when the limb does not develop past a certain point.
    **Amelia** - absence of limb
    **Acheiria** - missing a hand
    **Apodia** - missing a foot
- **Longitudinal deficiencies**
  - occur when a bone is missing or does not develop normally.
    (Usually the foot or hand is present although it may not have developed normally.)
    **Hemimelia** absence of a portion or half of a limb
    **Proximal Femoral Focal Deficiency**

21

## Longitudinal deficiencies



TMC, 1998

22

## Assessment

- Level of amputations: To define amputation levels and how this relates to functional outcomes.
- **Ideal outcomes: Determining expected functional independence.**
- The role of the rehabilitation team: Individual roles of the team members in accomplishing the rehabilitation of the amputee patient.
- Prosthetic candidacy: Based on comorbidities, compliance, energy expenditure, K-levels, and objective measures and subjective assessments.

23

## Treatment for congenital limb defects

- There are no standardized treatment protocols for congenital limb defects. Treatment options may include:
  - prosthetics
  - orthotics
  - surgery
  - physical and occupational therapy

24

## Treatment goals

- promoting normal development
- discovering sense of independence
- encouraging self-care
- improving cosmetic appearance

Potential emotional and social issues because of physical appearance

- adaptation

Limitations with certain movements, sports, or activities

25

## Treatment for congenital limb defects

- Specific treatment based on:
  - the child's age, overall health, and medical history
  - the extent of the condition
  - the type of condition
  - the child's tolerance for specific medications, procedures, or therapies
  - parent's opinion or preference
  - expectations for the course of the condition

26

## Assessment

- Level of amputations: To define amputation levels and how this relates to functional outcomes.
- Ideal outcomes: Determining expected functional independence.
- **The role of the rehabilitation team: Individual roles of the team members in accomplishing the rehabilitation of the amputee patient.**
- Prosthetic candidacy: Based on comorbidities, compliance, energy expenditure, K-levels, and objective measures and subjective assessments.

27

## Congenital deficiency factors

- Complexity of multiple limb loss leads to confusion
- Early rehab should involve
  - Patient/family education
  - Discuss expectations

28

## Congenital deficiency factors

- Teamwork is fundamental to achieving rehabilitative success
  - Surgeon
  - Physiatrist
  - Physical Therapist
  - Occupational Therapist
  - Orthotist/Prosthetist
  - Nurse
  - Case Manager
  - Social Worker
  - Behavioral Health Specialist
  - Dietician
  - Assistive Technology Specialist

29

## Prosthetic considerations

- **Staging**. Children are changing, growing and dynamic; hence, prosthetic designs should be staged based upon the child's developmental readiness. For example, a prosthetic component that may be too complex for the child today may be exactly what he or she needs two years from now.
- **Age at Fitting**
- **Growth**

Cummings and Kapp, 1992

30

## Prosthetic considerations

- **Staging**
- **Age at Fitting**. The child with lower limb congenital limb absence or early amputation is considered ready for lower extremity prosthetic fitting when he or she begins pulling up to stand. This usually occurs between nine and 12 months of age. Independent ambulation will begin between 15 and 22 months. Initially, all children walk with a wide-based gait with hips and knees flexed. Normal heel-to-toe gait patterns do not usually begin until age five.
- **Growth**

Cummings and Kapp, 1992

31

## Child milestones

- 4 – 5 Months
  - Head control
- 9 Months
  - Sits without support, crawls,
- 12 Months
  - Walks with or without support
- 18 Months
  - Walks independently
- 2 Years
  - Runs and jumps
- 3 Years
  - Climbs well
- 4 Years
  - Rides a tricycle
- 5 Years
  - Jumps, hops, and skips

32

## Prosthetic considerations

- **Staging**
- **Age at Fitting**
- **Growth.** Children grow both longitudinally and circumferentially. Bony alignment is changing also. For example, a newborn child's knee will generally exhibit genu varum. This condition usually straightens out by the first or second year, moves into genu-valgum by the third year, then resolves spontaneously thereafter.

Cummings and Kapp, 1992

33

## Prosthetic considerations

- Socket liners.
- "Slip" or "triple-wall" sockets.
- More socks.
- Distal pads.
- Flexible sockets have been used successfully in many pediatric centers.
- Frequent follow-up.
- Modular systems.
- Growth-oriented suspension system.
- Growth-oriented modifications/alignment.
- Activity Level.
- Maximize prosthetic performance.
- Protect from injury.
- Reinforce the prosthesis.
- Minimize weight.

34

## Assessment

- Level of amputations: To define amputation levels and how this relates to functional outcomes.
- Ideal outcomes: Determining expected functional independence.
- The role of the rehabilitation team: Individual roles of the team members in accomplishing the rehabilitation of the amputee patient.
- **Prosthetic candidacy: Based on comorbidities, compliance, energy expenditure, K-levels, and objective measures and subjective assessments.**

35

## LE Assessment

| Level of Amputation | Increased Energy Expenditure Above Normal (%) |
|---|---|
| Transtibial | 20–25 |
| Bilateral transtibial | 41 |
| Transfemoral | 60–70 |
| Transtibial/transfemoral | 118 |
| Bilateral transfemoral | >200 |

Energy requirements for types of amputation
Data from Cuccurullo SJ. Physical medicine and rehabilitation board review. New York: Demos Medical Publishing; 2004; with permission.

36

## K Levels

Level 0
Does not have the ability or potential to ambulate or transfer safely with or without assistance and a prosthesis does not enhance their quality of life or mobility.

Level 1
Has the ability or potential to use a prosthesis for transfers or ambulation on level surfaces at fixed cadence. Typical of the limited and unlimited household ambulator.

Level 2
Has the ability or potential for ambulation with the ability to traverse low level environmental barriers such as curbs, stairs or uneven surfaces. Typical of the limited community ambulator.

Level 3
Has the ability or potential for ambulation with variable cadence. Typical of the community ambulator who has the ability to traverse most environmental barriers and may have vocational, therapeutic, or exercise activity that demands prosthetic utilization beyond simple locomotion.

Level 4
Has the ability or potential for prosthetic ambulation that exceeds basic ambulation skills, exhibiting high impact, stress, or energy levels. Typical of the prosthetic demands of the child, active adult, or athlete.

37

## K Levels

Outcome measures commonly used to establish K levels include the following:

- Amputee Mobility Predictor (AMP) - **most frequently used outcome measure**
- Patient Assessment Validation Evaluation Test (PAVET)
- Prosthesis Evaluation Questionnaire (PEQ)
- Timed Up and Go (TUG)
- Timed Walk Tests
- Distance Walk Tests

38

## UE Assessment

- Upper Extremity Functional Index (UEFI) - a patient-reported outcome measure (PROM) for quantifying UE function
- Atkins Prosthetic Functional Adaptation Rating Scale
  - 100%: Wearing all day, using well in bilateral tasks, incorporating well in body scheme
  - 75%: Wearing all day, using in gross and fine motor tasks
  - 50%: Wearing all day, primarily for cosmetic reasons, using in gross motor tasks
  - 0%: Not wearing or using the prosthesis; unilaterally independent

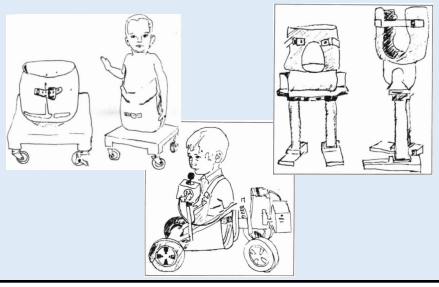
39

## What about tetra-amelia syndrome?

- congenital disorder characterized by the absence of arms and legs
- Tetra-amelia syndrome has been reported in only a few families worldwide.

40

Past Ideas



41

Past Ideas

42

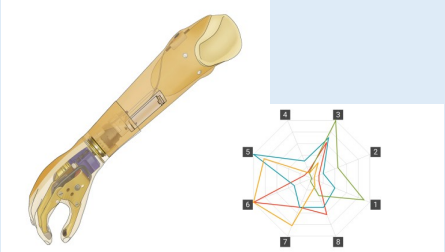
Modern Ideas

43

What can we do better for the person with multiple limb amputations?

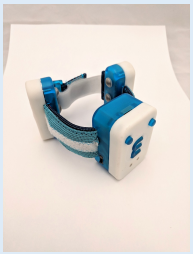
44

Myoelectric Prostheses



45

Myoelectric Prostheses

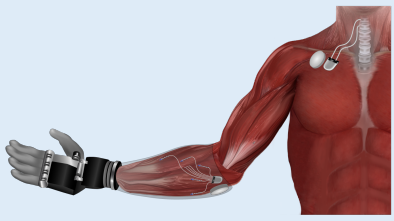


46

Myoelectric Prostheses

47

Sensory feedback

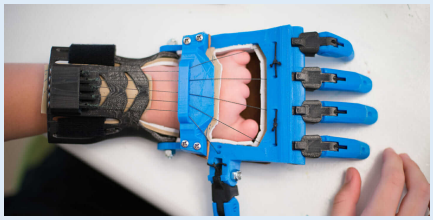


48



49

3D Printing



50

3D Printing

51

FORREST'S ADVENTURES IN THERAPY

Progression of activities from when he was 3 months old until 2 years old

52

Forrest was born full term on 9/10/2017. He was first seen by OT on 12/13/2017 when he was 3 months old.

**Birth History**: Forrest was delivered via C-Section at MUSC and spent 1.5 days in the NNICU for observation.

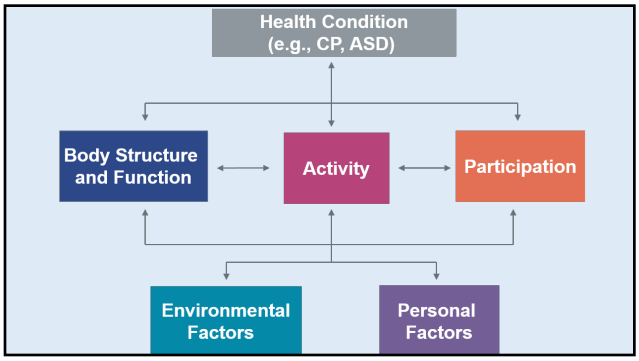
**Diagnosis**: He is diagnosed with Amelia, he has lower extremity bilateral residual limbs. His left upper extremity (LUE) is deficient above the elbow and his right upper extremity (RUE) is typically formed from his shoulder to his wrist. His fingers are potentially fused (no individual isolated fingers at this time). Recent X-rays reveal no heads of femurs bilaterally.

53

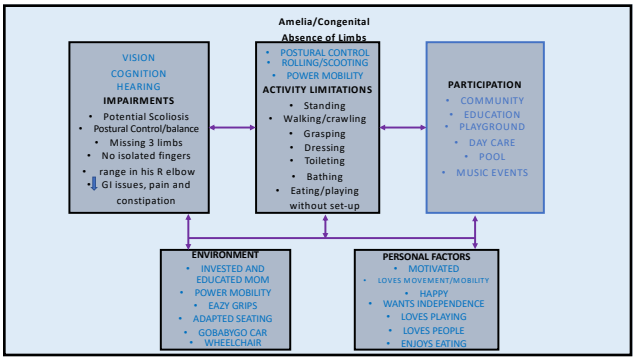
***The Team:***

Forrest and his mom, Alex
Early Interventionist, Laura Corn
Orthotist/Prosthetist, Jared McNeill
Occupational Therapist, Brenda Reagan
Speech Therapist – Stacie Devries
Physical Therapist – Denise Fredericks

54




55




56

**2019 ICD-10-CM Diagnosis Code Q73.0 Congenital absence of unspecified limb(s)**

**2016 2017 2018 2019 BillabA Exempt**

•Q73.0 is a billable/specific ICD-10-CM code that can be used to indicate a diagnosis for reimbursement purposes.
•The 2019 edition of ICD-10-CM Q73.0 became effective on October 1, 2018.
•This is the American ICD-10-CM version of Q73.0 - other international versions of ICD-10 Q73.0 may differ.

•Approximate synonyms: Amelia, congenital absence of limbs

57

NEW GOALS: Within 6 months, Forrest will
1. Get from sitting to lying on floor for play without the use of furniture for support.
2. Be able to stop 90% of the time in power wheelchair when seeing obstacles without verbal cueing from mom.
3. Get into sitting from lying with moderate assist using furniture for support.
4. Getting on and off a bench/step surface with minimal assist.
5. Balancing in sitting on a scooter board or mobile surface with 4-inch weight shift from center of gravity

58

References


- Classification of Functioning, Disability and Health. 2001; http://www.who.int/classifications/icf/en/.
- Rosenbaum P, Gorter JW. The 'F-words' in childhood disability: I swear this is how we should think! *Child Care Health Dev.* 2012;38(4):457-463.
- Patrick, D. L. and Y. P. Chiang (2000). "Measurement of health outcomes in treatment effectiveness evaluations: conceptual and methodological challenges." Med Care **38**(9 Suppl): II14-25.
- Centers for Disease Control and Prevention. Facts about Upper and Lower Limb Reduction Defects Online. Available: http://www.cdc.gov/ncbddd/birthdefects/ul-limbreductiondefects.html
- Cummings, Don, Kapp, Susan (1992) Lower-Limb Pediatric Prosthetics: General Considerations and Philosophy Journal of Prosthetics and Orthotics 4(4) 196-206
- Rossi, Roger., Alexander, Michael., and Cuccurullo, Sara. Pediatric Limb Deficiencies, Physical Medicine and Rehabilitation Board Review Online. Available: http://www.ncbi.nlm.nih.gov/.
- Texas Medical Center Newsletter. (1998) 20(5) Online. Available: http://www.tmc.edu/tmcnews/03_15_98/page_06.html.
- Ernst Marquardt, M.D Chapter 36A - Atlas of Limb Prosthetics: Surgical, Prosthetic, and Rehabilitation Principles. Special Considerations: The Multiple-Limb-Deficient Child


59

Any Questions

60