Congenital Amputation of Limbs: Meromelia

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Abstract: Meromelia is a partial absence of a limb or absence of part of the limb so that hands or feet are attached to the body like stumps. Such shrunken and deformed extremities are caused mainly by genetic factors, but some teratogenic (or environmental) factors have been identified, such as the use of thalidomide which was used in between 1957-62 for morning-sickness. The antenatal period of the case presented here was medically unsupervised but uneventful. The baby had bilateral upper limb Meromelia. Except for the young age of mother, there was no other obvious risk factor in this case. The preterm stillborn female baby of 1.6 kg was born of a non consanguineous marriage to a primigravida via vaginal mode at District Head Quarter Hospital. Ultrasound head, abdomen and 2D echocardiography were normal.

Keywords: Amelia, Limb defect, Limb reduction, Micromelia, Phocomelia

I. Introduction

Upper and lower limb reduction defects occur when a part of or the entire upper limb or lower limb of a fetus fails to form completely during pregnancy. The defect is referred to as a “limb reduction” because a limb is reduced from its normal size or is missing. Limb anomalies can either be minor or major. The minor limb anomalies are relatively common and can usually be corrected surgically but the major ones are mostly caused by genetic factors or environmental factors. The following are the common types of limb anomalies [1]:

- **Amelia**: complete absence of one or more limb
- **Peromelia**: Absence or deformity of the terminal part of a limb or limbs
- **Hemimelia**: Absence of the distal portion of one limb (form of peromelia)
- **Meromelia**: partial absence of a limb or absence of part of the limb
- **Acheira**: Absence of one or both hands
- **Apodia**: Absence of one or both feet
- **Acheiropodia**: Absence of hands and feet
- **Adactyly**: Absence of fingers or toes
- **Phocomelia**: A malformation in which the proximal portions of the extremities are poorly developed or absent. Hands and feet are directly attached to the trunk
- **Ectromelia**: Hypoplasia of the long bones of the limbs
- **Oligodactyly**: Subnormal number of fingers or toes
- **Polydactyly**: Condition of having supernumerary fingers or toes; **Preaxial**: Extra digit on radial or tibial side; **Postaxial**: Extra digit on ulnar or fibular side
- **Brachydactyly**: Abnormal shortness of the fingers or toes
- **Ectrodactyly**: Absence of all or part of a digit (split hand, lobster claw)
- **Sirenomelia**: Anomaly in which the lower extremities are fused
- **Syndactyly**: A fusion of two or more fingers or toes
- **Clinodactyly**: Permanent medial or lateral deflection of one or more fingers
- **Camptodactyly**: Permanent flexion of fingers or toes
- **Micromelia**: Shortness of the segment of the extremities
- **Phocomelia**: long bones are absent and the rudimentary hands and feet are attached to trunk by small irregular bones.
- **Talipes**: Cubfoot; **Valgus**: Heel and foot are turned outward; **Varus**: Heel and foot are turned inward

II. Case Presentation

19 years old primigravida in the 24th week of pregnancy came for regular antenatal check up to District Head Quarter Hospital, Koraput, Odisha, India. On examination: Average body built with weight- 50 kgs. Pulse Rate- 86/min; Respiration rate- 44/min, Pallor +, Pedal edema– not present. Heart and chest– NAD. Per
abdomen: height of uterus - 24 weeks; Cephalic presentation. Liquor not increased. Foetal movement - Felt. Rest of the systemic examination was normal. On investigation: Hb% - 10 mg/dl. FBS: 80 mg/dl. VDRL - Non reactive, Blood group: ‘O’+ve, Urine R/M - NAD. She was advised sonography for routine checkup.

Ultrasound of head, abdomen and 2D echocardiography were normal. All the fetal biometry like B.P.D., H.C., A.C., F. L. was normal for the gestational age except for the bilateral absence of both radius and ulna (Fig. 1).

The amniotic fluid was echogenic, Amniotic fluid index was 16 cm. Placenta: anterior, high up with grade-II maturation. The feet of the fetus were normal along with the tibia and fibula. The movements of fetal limbs were seen. The fetal hands were clenched and directly attached to the humerus. No other anomaly detected.

**Fig. 1.** Showing the fetal hand being attached to humerus with the absence of radius and ulna

The pregnancy was terminated after taking the patient’s consent and proper counseling. The newborn female baby showed classical features of bilateral upper limb meromelia (Fig. 2). There was bilateral terminal transverse limb deficiency below the elbow joint. The condition of the patient was stable after abortion and she was discharged after proper advice.

**Fig. 2.** Newborn with bilateral upper limb meromelia (bilateral terminal transverse limb deficiency below the elbow joint)

### III. Discussion

The overall prevalence of missing or incomplete limbs at birth is 7.9/10,000 live births [2]. Most are due to primary intrauterine growth inhibition or disruptions secondary to intrauterine destruction of normal embryonic tissues. The upper extremities are more commonly affected. Amelia and Meromelia may either present as an isolated defect or associated with other malformations; and the diagnosis is mainly clinical [3]. The antenatal period of the case presented here was medically unsupervised but uneventful. Except for the young age of mother, there was no other obvious risk factor in this case.

Congenital limb deficiencies have many causes and often occur as a component of various congenital syndromes. Teratogenic agents (e.g., thalidomide, vitamin A) are known causes of hypoplastic/absent limbs. The most common cause of congenital limb amputations are vascular disruption defects, such as amniotic band-related limb deficiency, in which loose strands of amnion entangle or fuse with fetal tissue. The International
Society for Prosthetics and Orthotics (ISPO) classification describes limb deficiencies as either Longitudinal (more common) or Transverse [4].

Longitudinal deficiencies involve specific maldevelopments (e.g., complete or partial absence of the radius, fibula, or tibia). Radial ray deficiency is the most common upper-limb deficiency, and hypoplasia of the fibula is the most common lower-limb deficiency. In transverse deficiencies, all elements beyond a certain level are absent, and the limb resembles an amputation stump. Amniotic bands are the most common cause; the degree of deficiency varies based on the location of the band, and typically, there are no other defects or anomalies.

Etiological factors include genetic, teratogens (infamous-thalidomide use), vascular disruptions and ischemia, chemicals and radiation exposure [5]. Children with limb anomalies have associated malformations like craniofacial, gastrointestinal, heart, kidney and nervous system [6]. The present case was unique that the baby had only isolated limb meromelia with no other congenital anomalies.

IV. Conclusion

When a genetic syndrome is suspected, evaluation should also include a thorough assessment for other physical, chromosomal and genetic abnormalities by a clinical geneticist. Typically, x-rays are done to determine which bones are involved. Treatment consists mainly of prosthetic devices, which are most valuable for lower-limb deficiencies and for completely or almost completely absent upper limbs. If any activity in an arm or hand exists, no matter how great the malformation, functioning capacity must be thoroughly assessed before prosthesis or surgical procedure is recommended. Therapeutic amputation of any limb or portion of a limb should be considered only after evaluating the functional and psychological implications of the loss and when amputation is essential for fitting prosthesis.

Upper-limb prosthesis should be designed to serve as many needs as possible so that the number of devices is kept to a minimum. Children use prosthesis most successfully when it is fitted early and becomes an integral part of their body and body image during the developmental years. Devices used during infancy should be as simple and durable as possible; e.g., a hook rather than a bioelectric arm. With effective orthopedic and ancillary support, most children with congenital amputations lead normal lives.

References

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