

## Case Report

## Congenital Amputation Syndrome Affecting All Four Limbs -A Rare Case Report

Dr Dhan Raj Bagri<sup>1</sup>, Dr R K Gupta<sup>2</sup>, Dr Mahesh Kadel<sup>3</sup>

<sup>1</sup>(Js Pediatrics), Ex Senior Registrar, SPMCHI, J K LON Hospital, SMS Medical College, Jaipur, Rajasthan

<sup>2</sup>Professor and Unit Head, SPMCHI, J K LON Hospital, SMS Medical College, Jaipur, Rajasthan

<sup>3</sup>(GYNAE &OBST), JS (GYNAE), DMHS, Government of Rajasthan, Jaipur

**\*Corresponding author**

Dr Dhan Raj Bagri

Email: [meena.drdhanraj6@gmail.com](mailto:meena.drdhanraj6@gmail.com)

**Abstract:** Intrauterine amputation of the extremities is an uncommon congenital deformity which is characterized by the absence of one or more distal limb portions. This deformity is caused mainly by genetic factors, but some teratogenic (or environmental) factors have been identified, such as the use of thalidomide for morning-sickness. The primigravida mother had recurrent episodes of allergic rhinitis and asthma and was prescribed fluticasone nasal spray and intermittent inhalation therapy. Her elder sister was affected from polio in childhood with residual deformity. Antenatal history regarding radiation exposure, febrile illness and rash were negative. HIV and VDRL were negative. The antenatal ultrasound was done in all three trimesters and revealed no obvious anomaly. The Borderline preterm baby was delivered in government hospital by LSCS due to premature rupture of membranes after appropriate antenatal corticosteroids.

**Keywords:** Amelia, Congenital amputation, Limb defect, Limb reduction

**INTRODUCTION:**

Intrauterine amputation of the extremities is an uncommon congenital deformity which is characterized by the absence of one or more distal limb portions. The term 'amputation' suggests separation, by mechanical force, of a limb already formed rather than a failure of development, but such a view of the pathogenesis of this abnormality is far from universally accepted [1].

Thomas Bartholin of Copenhagen (1616-1680) was the first to mention a congenitally deficient extremity. The idea that amniotic bands, by interfering with the blood supply, cause ischaemic necrosis of an extremity and eventual separation, should be credited to Montgomery (1832; 1833).

**CASE REPORT:**

The 22 years old primigravida mother had recurrent episode of allergic rhinitis and asthma and was prescribed Fluticasone nasal spray and intermittent

inhalation therapy with antenatal protocol treatment. She conceived after active married life of 9 months and there is no history of any abortion or abortifacient drug use; neither any drug use for conception. She tested negative for HIV and VDRL. Her BP was normal throughout pregnancy and no radiation exposure could be documented or reported.

She was anaemic with a haemoglobin level of 9.4 gm%, other tests were normal. USG for foetal well-being was done three times but no deformity was reported. The patient presented at 34 weeks with leaking PV since three days and was given antibiotics and antenatal steroids with a plan of LSCS. The baby cried immediately after birth and on examination his both upper limbs found 'amputated' at elbow and lower limbs were also in same condition below knee. The child weighed 2.25 kg and no other malformation was noted.



**Fig 1: Congenital Amputation**



**Fig 2: Baby gram**

**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].

Congenital limb deficiencies have many causes and often occur as a component of various congenital syndromes. Congenital amputation syndrome affecting all four limbs is a rare anomaly. 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 all four limbs amputation and didn't required resuscitation.

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.

#### CONCLUSIONS:

Antenatal ultrasounds should be crosschecked with more sophisticated imaging techniques in suspected cases. Other physical, chromosomal and genetic abnormalities should be assessed by a clinical geneticist if a genetic syndrome is suspected. X-rays were done to determine which bones are involved. Diagnosis is mainly clinical. Management of this condition requires a team based approach. Psychological trauma to the parents should be taken care of with appropriate counselling in a trustworthy environment. The team should include a paediatrician, psychologist, orthopaedician and physiotherapist.

Use of prosthesis and training of parents and child may serve as important management plans. 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

1. Aitken GT, Frantz CH. Congenital amputation of the forearm. *Annals of surgery*. 1955 Apr; 141(4):519.
2. Boyadjiev SA, Kim SD, Hata A, Haldeman-Englert C, Zackai EH, Naydenov C, Hamamoto S, Schekman RW, Kim J. Cranio-lenticulo-sutural dysplasia associated with defects in collagen secretion. *Clinical genetics*. 2011 Aug 1;80(2):169-76.
3. Gupta P, Kumar A. Amelia-Meromelia Sequence with Atrial Septal Defect—A Rare Occurrence. *Fetal and pediatric pathology*. 2014 Apr 1;33(2):92-7.
4. Day HJ. The ISO/ISPO classification of congenital limb deficiency. *Prosthet Orthot Int*. 1991; 15:67-9.
5. Moore KL, Persaud TVN, Torchia MG. *Textbook of Essentials of Embryology and Birth Defects. The Musculoskeletal System*. 7<sup>th</sup> ed. Philadelphia: John F. Kennedy; 2008: 248.
6. Chawla D, Marwah P. Meromelia in a Newborn—A Rare Congenital Anomaly. *The Indian Journal of Pediatrics*. 2014 Dec 1; 81(12):1422.