



Case Report

Isolated Upper Limb Phocomelia - Case Study of a Rare Congenital Disorder

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ABSTRACT

Phocomelia is a very rare congenital disorder where the limbs are characteristically absent or hypo plastic with a normal or nearly normal looking hand and foot. Even though, it is well known to be produced by Thalidomide, so many other causes are peeping into the scene presently and in some cases, the cause is more or less obscure. This article deals with the study of a case of phocomelia motivated by its rare features.

Key words: Phocomelia, congenital limb defects, synostosis.

INTRODUCTION

The term Phocomelia was first coined by Etienne Geoffroy Saint in 1836. Phocomelia is a very rare congenital malformation that presents with absence or severe hypoplasia of the long tubular bones, especially of the limbs. An extreme case results in the absence of the upper bones of both the arms and legs, so that the hands and feet appear attached directly to the body as stumps resembling flippers of a seal and hence the name. It may present as complete or incomplete forms. A complete form is the one in which both the proximal and distal bones of limb are absent and the incomplete form occurs with the absence of either the proximal or the distal bones. Abnormalities vary greatly, and they may be represented by partial [meromelia] or complete absence [Amelia] of one or more of the extremities. Sometimes all segments of the extremities

are present but abnormally short [micromelia].^[1]

Limb defects are caused by genetic factors, such as chromosomal abnormalities associated with trisomy 18, mutant genes, environmental factors, such as teratogens, a combination of genetic and environmental factors [multifactorial inheritance] vascular disruption and ischemia, as in limb reduction defects.^[2]

CASE STUDY

The present case is a 13 year old boy weighing 34kg with a height of 140 cm presenting to pediatric OPD with complaint of right sided limb defect since birth. The boy was born as a first child of his parents who had two children. He was born of LSCS with the indication of cord around the neck at full term. He was handed over to parents immediately after birth. A corrective surgery

for the limb defect was attempted 10 days after birth but with less promising results. He attained his milestones normally. The marriage of the parents was non-consanguineous and there was no history of any specific drug intake, radiation exposure or infections in the antenatal period. His sibling is normal and there is no history of limb defects in the family in both maternal and paternal lines.



Figure 1: left upper limb showing the deformity.

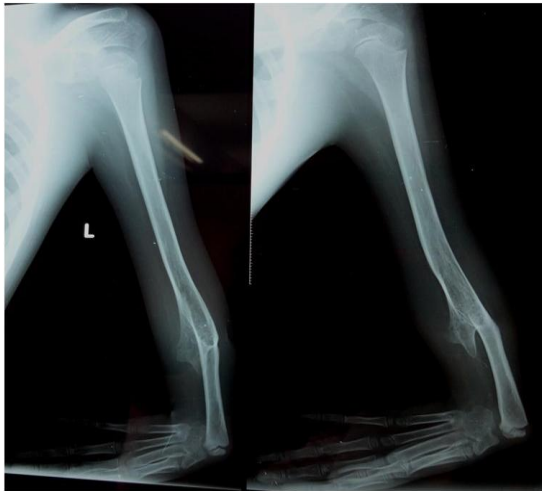


Figure 2: X-ray of the affected limb.

On general examination, he showed normal cranio-facial features. The spine was normal. The right upper limb measured 30cm from shoulder to elbow joint, 24 cm

from elbow joint to wrist joint and 16 cm from wrist joint to tip of the middle finger. The left upper limb showed the deformity. The shoulder is elevated with decreased shoulder width on the same side. The spine of the scapula as well as its medial border appears to be prominent. The length from point of the shoulder to wrist was 34.2 cm and appeared as a single segment. There was no recognizable elbow joint on inspection. On palpation, an irregular hard bone-like mass can be felt posteromedially. A linear scar is seen posteriorly which was said to be resulted from the attempted corrective surgery. On lateral aspect, at this level presents a dimple in the skin. The wrist is represented by a constriction and from that point, the length up to the tip of middle finger was recorded to be 14.2 cm. The fingers were abnormally short with the little finger to be the shortest. The palmar creases are sparse and not at all prominent. All the movements of shoulder joint are possible but to a limited range. No other movements are exhibited by the limb as a whole either at elbow or at wrist region. The limb muscles are atrophied and wasted. The child exhibited normal intelligence and memory and he had no other visible abnormalities. Nothing abnormally detected in Systemic examinations clinically.

Radiography: X-Ray of the affected side is taken and evaluated. Elevation of scapula with elevated lateral end of clavicle is noted. The upper end of humerus appears to be normal, taking part in shoulder joint formation. No definite elbow joint can be defined. The forearm portion presents a single bone which is more lateral in position and exhibit cortical continuity with lower end of humerus through synostosis. A Rudimentary Ulna is seen fused with the lower end of humerus. An attempt for the formation of upper end of ulna is exhibited. The proximal rows of carpals are absent. The distal row appears to be fused together-

congenital bony ankylosis is seen. The fourth and fifth metacarpals are fused through syndesmosis. Rest of the metacarpals and phalanges were well formed and normal in appearance.

DISCUSSION

The first case of phocomelia was reported in Germany in 1956. Limb malformations occur in approximately 6 per 10,000 live births, with 3.4 per 10,000 affecting the upper limb.^[1]

The limb buds appear as the outpunching from the antero-lateral body wall at about the 26th day of gestation. Initially there will be a mesenchymal core derived from the somatic layer of lateral plate mesoderm, covered by a layer of cuboidal ectoderm. At the distal border of the limb, Apical Ectodermal Ridge is formed. This induces the adjacent mesenchyme to proliferate. The development of the limb proceeds proximodistally. The lower limb also develops similarly but with some delay. The upper limbs are formed fully by 12 weeks and the lower limbs by 14 weeks. The muscles and bones also will be developed subsequently. When this normal pattern is disturbed because of various factors, it may lead to malformed appendages.^[1]

As per NORD (National Organization of rare diseases), phocomelia is an autosomal recessive trait linked to chromosome 8, when it is familial.^[3] Symptoms of phocomelia syndrome include underdeveloped limbs and absent limb bones. Short arm bones, fused fingers, missing thumb will often occur. Legs and feet may also get similarly affected and pelvic bones may be totally absent.

Phocomelia syndromes present with multiple malformations including skeletal, genitourinary such as renal agenesis, gastrointestinal system, ocular abnormalities such as cloudy corneas, craniofacial

abnormalities including silver blonde hair, extensive hemangiomas and hypoplastic nasal cartilage.^[4] Sometimes, it will be accompanied by other defects such as anotia, duodenal stenosis and cardiac defects.^[5] Prominent widely set eyes (hypertelorism), an underdeveloped nose with thin nostrils, malformed ears, cleft lip with or without cleft palate, and small jaws (micrognathia) may also occur. The testes of males may fail to descend (cryptorchidism).

Phocomelia was seen with thalidomide embryopathy or could be a part of some pseudo-thalidomide syndromes, which could be familial. Roberts syndrome,^[6] DK Phocomelia syndrome,^[7] Odontotrichomelic tetramelic ectodermal dysplasia,^[8] congenital hemidysplasia with ichthyosiform erythroderma and limb defects (CHILD) syndrome,^[9] Syndrome of spleno-gonadal fusion^[10] are some of the syndromes which present with associated phocomelia.

CONCLUSION

In this case phocomelia is confined to only one upper limb with no other congenital defect. It does not seem to be familial. No history of any radiations or teratogenic drug intake. The cause here seems to be obscure. However, proper prosthesis and other orthopedic rehabilitation had to be provided so that the quality of life can be improved. Isolated single limb phocomelia has a better prognosis compared to other varieties.

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