Isolated Bilateral Upper Limb Amelia – A Rare Case Report

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Learning Point of the Article:

The purpose of this case report is to enlighten the causes of Amelia and its presentations in current times.

Abstract

Introduction: Congenital upper limb amelia is one of the extremely rare conditions. It is defined as a complete absence of upper limbs. It may present as isolated or with other associated anomalies.

Case Report: We present a case of a 2-year-old male child with congenital complete absence of bilateral upper limb. This male child was born after four female children. With the advancement in modern-era prenatal diagnostic facilities and a better understanding of fetal-maternal drug pharmacology, such cases are rare entity.

Conclusion: Amelia is a very rare and challenging situation for clinicians. Regular prenatal checkup and knowledge of maternal and fetal drug interactions during pregnancy are key factors for prevention.

Keywords: Amelia, congenital limb deficiency, teratogenic drugs.

Introduction

Amelia is defined as a complete absence of a limb and is a rare congenital anomaly with incidence ranging from 0.053 to 0.095 in 10,000 live births [1-3]. It mostly affects the upper limb. Proximal part of the limb is absent with the distal extremity attached to the trunk. Sometimes, the limbs can be missing completely. It may also be associated with other malformations. The exact pathogenesis is still unclear, but mostly, it is believed that it occurs as a sporadic event [4]. It is found by some researchers that it may be related to some teratogens such as thalidomide, alcohol, maternal diabetes, and vascular compromise by amniotic bands [5-7].

Case Report

We present a case of a 2-year-old male child who came to the outpatient department with the absence of both upper limbs. On examination, no dysmorphic features were noted aside from bilateral absent upper limbs from the shoulder (Fig. 1 and 2). The child was otherwise healthy and had normal intellectual abilities. The child was delivered by normal vaginal delivery at home with the help of a local village dai. On further history, the child's mother mentioned that she has four daughters, all born uneventfully by normal vaginal deliveries. Patient's parent had consulted some osteopath in their village and took some treatment in the form of tablets and powder in the desire to have a male baby. She took these medicines from the time of conception and continued for 3 months during pregnancy. Due to lack of resources and awareness, she was unable to consult any health-care provider for antenatal checkup. She delivered a full-term

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Author's Photo Gallery


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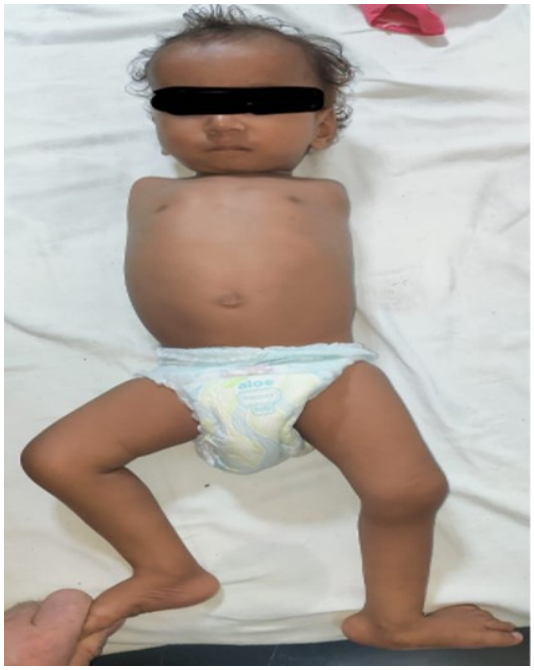

Figure 1: Clinical photograph of child showing complete absence of a bilateral upper limb.

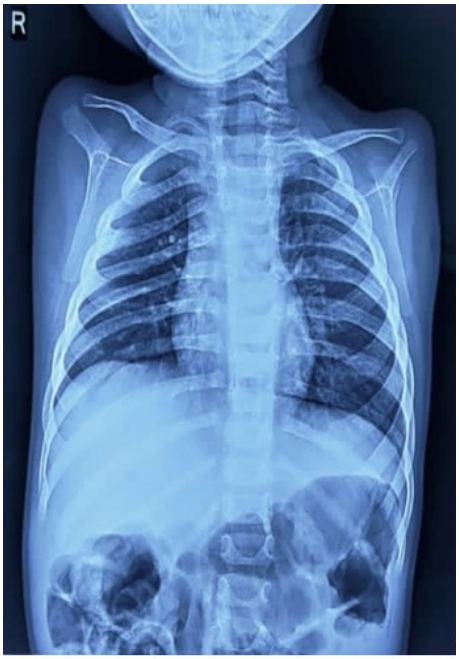

Figure 2: X-ray photograph showing complete agenesis of bilateral upper limbs without any bony growth.

male baby with absent bilateral upper limbs.

Discussion

Amelia is a rare congenital entity. The incidence of upper limb amelia is approximately 7/1,000,000 live births [3]. Children with amelia with other associated malformations usually have a bad prognosis and die within 1 year. Amelia has been associated with early-onset scoliosis.

Powers et al. [8] performed a retrospective study to determine the incidence of scoliosis in patients with upper limb skeletal abnormalities and concluded that patients with amelia had a higher incidence of idiopathic scoliosis (unilateral amelia = 50% incidence/bilateral = 100%) [8]. Froster et al. [9] suggested that the combination of brachial amelia, forebrain defect, and facial cleft may represent a new syndrome. Urinary tract abnormalities are known to be associated with limb defects [10].

Several causes have been proposed such as consumption of alcohol and teratogenic drugs such as thalidomide, vascular compromise by amniotic bands, maternal diabetes, and autosomal recessive mutations [5-7].

This case report describes the case of amelia in a couple with four healthy daughters without any obvious historical event. Although the exact cause is not known in this patient, one big possibility could be the effect of taking unknown unsafe drugs during pregnancy in the desire to have a male child, which is prevalent in developing countries. People consume toxic and harmful drugs in the hope of having a male child. One more important point is, still in developing countries, osteopaths are doing unethical practices and due to lack of education and knowledge, especially in rural areas, people consume unknown and unsafe drugs which lead to such conditions.

Conclusion

Amelia is one of the very rare and challenging conditions affecting limb development. Other associated anomalies in these patients should be identified. Regular prenatal checkup and knowledge of maternal and fetal drug interactions during pregnancy are key factors for prevention.

Clinical Message

Bilateral upper limb amelia is very rare congenital anomaly which may be associated with other anomalies. Several factors are involved but to highlight one important factor is taking some unsafe drugs such as thalidomide during pregnancy in desire of having a male child. This practise is still prevalent in developing countries wherein such teratogenic drugs are prescribed by osteopaths and due to lack of knowledge, it is consumed during pregnancy which leads to congenital anomalies

Declaration of patient consent: The authors certify that they have obtained all appropriate patient consent forms. In the form, the patient has given the consent for his/ her images and other clinical information to be reported in the journal. The patient understands that his/ her names and initials will not be published and due efforts will be made to conceal their identity, but anonymity cannot be guaranteed.

Conflict of interest: Nil **Source of support:** None

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