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Non-syndromic phocomelia: A rare case report signifying prenatal screening

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Abstract

Phocomelia is a rare congenital condition characterized by severe limb malformation, where the limbs are either partly or completely underdeveloped. Phocomelia can occur as a syndrome or as a limb-specific abnormality. The frequency of phocomelia ranges from 0.6 to 4.2 per 100,000 live births; hence, there are not many reports of this deformity. Genetic inheritance and the use of thalidomide are the two main etiological factors of phocomelia. Several symptoms and visceral abnormalities are associated with this condition. Ultrasonography is crucial for the early detection of phocomelia during the intrauterine stage. Presented here is a case of phocomelia in a 6-year-old boy who was diagnosed after birth, with no maternal history of thalidomide usage or family history of the same condition. This case is unique in that it involves a child born with phocomelia but no additional congenital defects observed in related syndromes. Because of that, we suggest this case may be isolated.

Keywords

Phocomelia, ultrasonography, congenital limb malformation, orthosis prosthesis, syndactyly, fibular deficiency

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Introduction

The term "phocomelia" is derived from the Greek words "fóke," meaning seal, and "melos," meaning limbs, describing the seal-like appearance of the affected limbs. The proximal part of the limb is either absent or conspicuously hypoplastic, while the hand and foot are typically normal or almost normal.[1] Phocomelia affects approximately 0.6 to 4.2 per 100,000 live births globally,[2] with a higher incidence observed in left-sided upper limbs. Unilateral defects are more common than bilateral ones. Out of 127 registered cases, there were 65 males for 53 females.[1]

Limbs develop from mesenchymal cells in the lateral mesoderm, which form limb buds covered by the ectoderm. An apical ectodermal ridge forms at the apex of each bud and guides longitudinal growth and tissue differentiation through interaction with the progress zone mesoderm. Phocomelia can result from apoptosis triggered by factors such as vascular insufficiency or medication toxicity (e.g., thalidomide), disrupting the connection between the apical ectodermal ridge and the progress zone. In its genetic form, it is inherited as an autosomal recessive condition linked to mutations on chromosome 8.[3]

This deformity typically impairs the musculoskeletal system's proper growth and development. It has occasionally been linked to thoracic dystrophy, craniofacial deformities, and genital malformations.[4]

This case concerns a 6-year-old male child diagnosed with isolated phocomelia.

Case presentation

A 6-year-old boy with a known case of phocomelia was diagnosed after birth. He presented to our pediatric orthopedics clinic a few months after birth for further evaluation.

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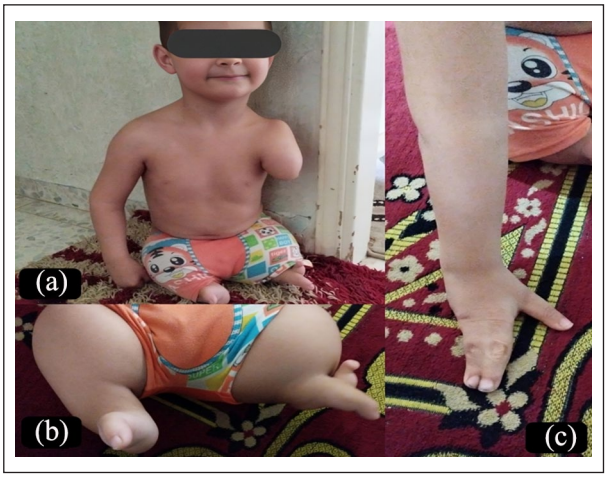

Figure 1. Gross images of the: malformed left upper limb (a), the malformation of both lower limbs (b), and the right index and middle finger syndactyly (c).

This child is the firstborn of a 21-year-old mother who had adequate antenatal care and monthly doctor visits per national guidelines. All booking investigations were normal. However, a detailed fetal ultrasound was not performed, leading to a missed prenatal diagnosis of phocomelia. The mother took only supplements and denied exposure to smoking, alcohol, vitamin A, illicit drugs, radiation, or local herbs. She reported no health issues or chronic diseases during pregnancy, resulting in an unremarkable prenatal history. There was no consanguinity between the parents and no family history of malformations or deformities. The baby's two younger siblings are healthy. Born at 40 weeks via cesarean section due to breech presentation, the baby weighed 2600 g. At birth, the baby had flipper-like deformities of the upper and lower limbs, but the neonatal history was otherwise uneventful.

Currently weighing about 14 kg, he is considered 100% disabled, moving by jumping due to limb deformities, and is unable to use his right hand because of syndactyly. Despite this, his language, cognitive, social-emotional, and behavioral development are normal for his age. He is otherwise healthy, with up-to-date immunizations.

Upon examination, the child was conscious, alert, and oriented. He had shortened, flipper-like upper and lower limbs on the left side. The left upper limb was deformed with only a stump, missing the elbow joint and the bones below it. The right upper limb was well-formed but had syndactyly of the right index and middle fingers (Figure 1). Both lower limbs were malformed: the left leg was absent, and the right leg was malformed with one digit on the right foot and two on the left (Figure 1). There were no spine deformities. Systemic, head and neck, chest, and abdominal examinations were normal, with no dysmorphic facial features or congenital defects in the genitalia. The child showed no growth or mental retardation.

After birth, a "skeletal survey"[5] showed short limb bones and a normal skull (Figure 2). A follow-up pelvic X-ray revealed proximal femur tapering on both sides with the absence of the head and neck, dysplastic hips, an underdeveloped acetabulum, right fibular deficiency, and absent bones below the left knee (Figure 2). Blood and renal function tests were normal, as was an abdominal ultrasound.

On follow-up, management is conservative, with plans for orthoses and prostheses for the upper and lower limbs. Surgery is recommended to separate the syndactylous index and middle fingers.

Discussion

Limb buds begin to develop on the 26th day of intrauterine life and are fully formed by the end of the 14th week. Deviations from this pattern can lead to deformities, including partial (meromelia) or total (amelia) absence of limbs.[6] Phocomelia, a type of meromelia, is a rare condition affecting newborns and can occur as a syndrome or a limb-specific abnormality.[2] Potential causes include thalidomide, substance use (alcohol or cocaine), X-ray exposure, and gestational diabetes.[7] Vascular insufficiency, specifically reduced blood flow through an abnormal subclavian artery, is also a possible cause, though no such abnormality was noted in this case.[8]

However, phocomelia can either be hereditary or caused by gene mutations.[9] Many different genes play crucial roles in the normal embryonic development of limbs. Among these, the Wnt7, Shh, and FGF genes are particularly significant. Specifically, the Wnt-7 gene controls the ventral-dorsal axis, determining the anterior and posterior surfaces of the limbs. By contrast, the Shh gene governs the anterior–posterior axis, influencing the placement of fingers and toes. Meanwhile, the FGF gene promotes limb development and elongation. Mutations in these genes can result in shortened limbs due to FGF mutations, improper positioning of fingers or toes from Shh mutations, and loss of limb surfaces caused by Wnt7 mutations.[10] Despite these genetic insights, genetic counseling was declined in this case, preventing the exclusion of sporadic causes of phocomelia.

It was reported that 9.9% of phocomelia cases are associated with other syndromes,[7] including Roberts syndrome, femur-fibular-ulnar syndrome, thrombocytopenia absent radius syndrome, and Holt-Oram syndrome.[10] In addition, 36.9% of cases had severe abnormalities[7] such as mental deficiency, craniofacial, heart, or kidney abnormalities, cryptorchidism, and a low platelet count.[11] The unique aspect of this case is the absence of any concomitant abnormalities or associated deformities, suggesting it is a non-syndromic phocomelia.

The diagnosis of phocomelia syndrome is often made through a fetal prenatal ultrasound during the second trimester, usually between weeks 18 and 22.[12] However, in our case, the mother did not undergo a detailed fetal ultrasound, resulting in the patient not being diagnosed with phocomelia

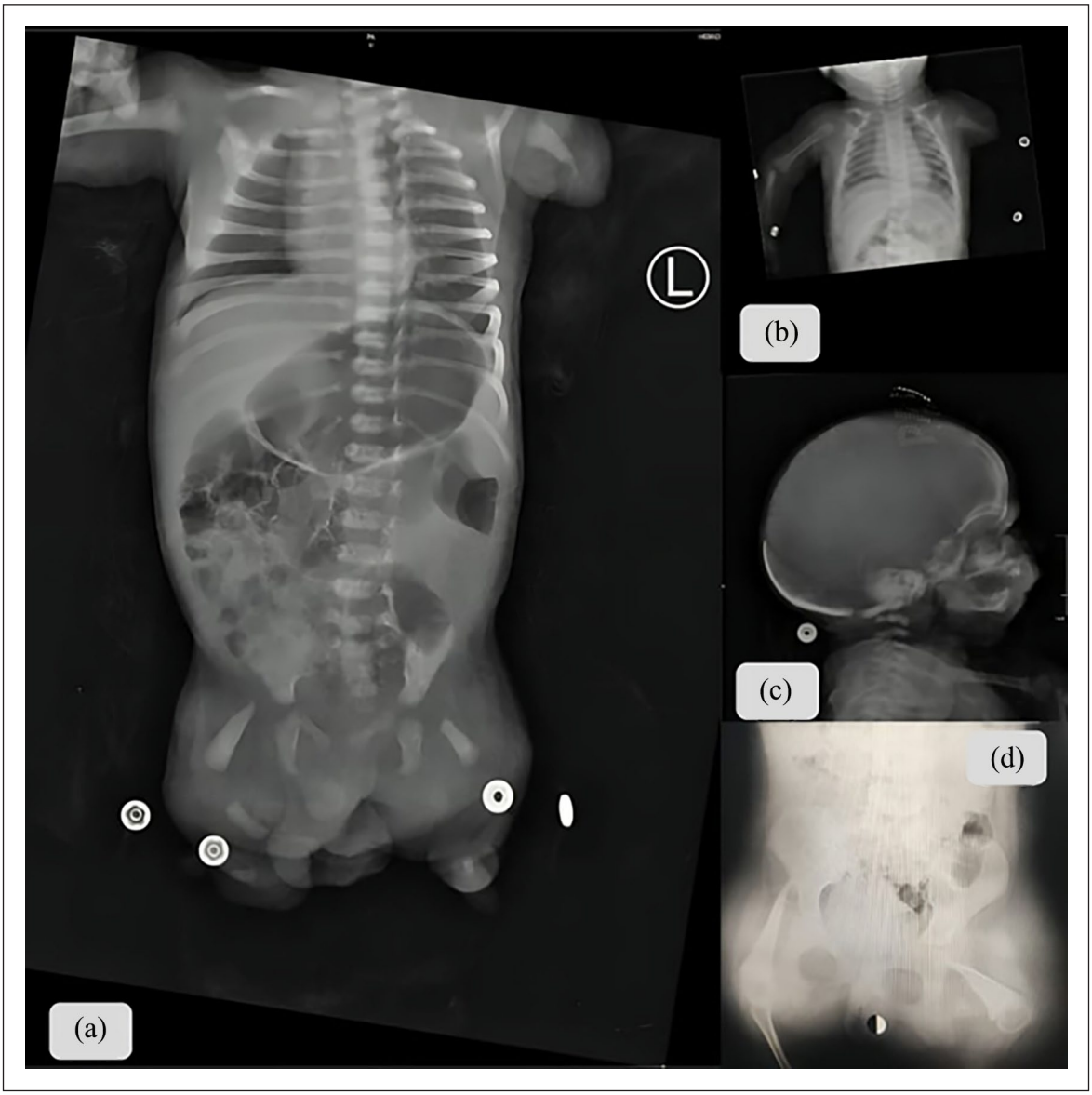

Figure 2. A skeletal survey of the upper extremities revealed a malformed rudimentary humerus was present on the left with an absence of elbow joint, forearm, hand, and fingers. On the right, the shoulder joint, humerus, elbow joint, radius, ulnar, and carpal bones were present, but the metacarpal bones could not be observed by the image. (b). In the lower extremities, both femora were grossly shortened, with only a small, somewhat triangular portion of bone in both thighs. A single bone was present in the right leg but absent in the left. The right showed a malformed ankle. However, the spine, clavicle, and ribs are normal (a). A lateral view of the skull revealed normal bone formation without any deformities (c). Pelvic X-ray showing both-sided proximal femur tapering end with absence of head and neck, dysplastic hip, and underdeveloped acetabulum; on the right side, there is an underdevelopment of the knee joint, an ankle joint, and an absent fibula (fibular deficiency). Left side: absent knee joint, no tibia or fibula, no ankle joint (d).

before birth. Our case illustrates the critical importance of prenatal ultrasound exams in identifying congenital defects and preventing experiences similar to ours.

The primary course of treatment is conservative. To help children become self-sufficient, various technologies have been developed. Surgery, however, is a modest intervention that may be recommended for limb shortening, insufficient thumb opposition, or unstable shoulders.[13] However, we attempt to identify and suggest a course of action for any abnormalities our patient may experience by utilizing speech, physical, occupational, and/or plastic therapy.

We are aware that raising a disabled child can be extremely challenging for certain individuals. The key to handling these situations is offering family counseling services, as parents typically feel guilty and accountable for their children's abnormalities.[7]

Conclusion

To sum up, phocomelia is a rare congenital limb anomaly characterized by shortened or missing extremities, which can occur in isolation or alongside other syndromic defects. This case appears to be sporadic, given the absence of consanguinity, family history of congenital anomalies, and teratogenic drug use. It underscores the importance of detailed prenatal ultrasound assessments, particularly for measuring extremities, to guide follow-up management and prepare parents psychologically and financially. With proper prosthetics and orthopedic rehabilitation, children with phocomelia can achieve normal lives.

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Author contributions

All authors contributed to the report, discussion, and conclusion of this work.

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Ethical approval

Our institution does not require ethical approval for reporting individual cases or case series.

Informed consent

Written informed consent was obtained from the patient's parents for their anonymized information to be published in this article

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