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Phocomelia: A Rare Congenital Anomaly in Term Neonate

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Abstract: ***Phocomelia is a congenital condition that involves malformations of human arms and legs which result in a flipper - like appendage. Usually the upper limbs are not fully formed and sections of the "hands and arms may be missing". Short arm bones, fused fingers, and missing thumbs will often occur. Legs and feet are also affected similarly to the arms and hands. Individuals with phocomelia will often lack thigh bones, and the hands or feet may be abnormally small or appear as stumps due to their close "attachment to the body". According to the National Organization for Rare Disorders (NORD), when phocomelia is transmitted (in its familial genetic form) it is seen as an autosomal recessive trait and the mutation is linked to chromosome 8.***

Keywords: Phocomelia, congenital anomalies, prosthetics, Thalidomide.

1. Case Report

A full - term male neonate weighing 2.5kg, 4th by birth order was delivered vaginally on December 13, 2022, at the District Hospital Hapur, to a 28 - year - old multigravida. After birth the child was immediately shifted to NICU for congenital limb anomaly.

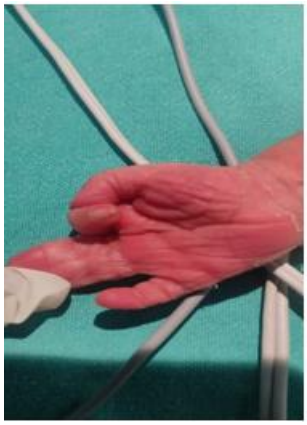
On examination there was -

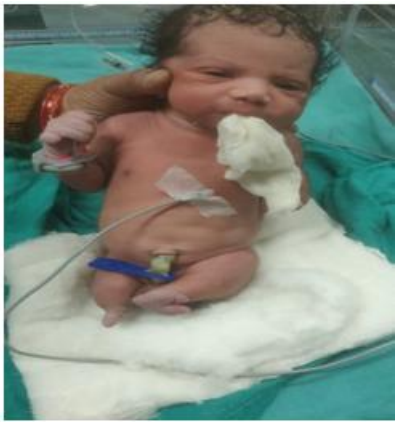
1) Absence of left upper limb distal to the humerus
2) Absence of both lower limbs distal to the femur
3) No craniofacial or thoraco - abdominal anomalies were detected

The child was kept in NICU for observation and further evaluation. Multidisciplinary consultation was initiated, including orthopedics and physiotherapy. The use of custom prosthesis was planned at 6 months of age, with the goal of early functional rehabilitation by 2 years of age. The mother had no history of consanguinity or teratogenic drug intake (including thalidomide). Family history revealed grandfather had a deformity in left foot since birth, grandmother had polio, father had history of intentional tremors, speech difficulty and limb abnormalities.

Antenatal USG (3rd trimester, dated 12 Nov 2022):

a) Left femur significantly shortened
b) non - visualization of bilateral tibia, fibula, and feet
c) Upper limb showed significant hypoplasia with non - visualization of distal forearm and hand





2. Discussion

Phocomelia is an extremely rare congenital limb deficiency disorder, characterized by the absence or underdevelopment of proximal limb segments, resulting in the attachment of hands or feet close to the trunk. Historically, it was most infamously associated with thalidomide exposure during pregnancy in the late 1950s and early 1960s, which led to over 10, 000 cases of birth defects globally [1]. However, with the cessation of thalidomide use in pregnant women, more recent cases have emerged in the absence of such exposure, indicating **alternative** genetic, vascular, or idiopathic causes [2]. In the present case, the neonate demonstrated classic features of phocomelia involving absence of the left upper limb distal to the humerus and bilateral lower limbs distal to the femur. Antenatal ultrasonography in the third trimester identified limb abnormalities, highlighting the value of prenatal imaging in early detection and counseling [3]. The mother's history was negative for drug intake, infections, or exposure to known teratogens, suggesting a possible sporadic genetic mutation or early intrauterine vascular disruption [4]. Phocomelia can present either as an isolated limb defect or in

association with other congenital anomalies, such as cardiac malformations, gastrointestinal atresias, choanal atresia, and genitourinary defects [5]. Our patient did not exhibit any associated anomalies, which is favorable for long - term outcomes. Early diagnosis allows for timely planning of prosthetic fitting and multidisciplinary rehabilitation. Management of phocomelia focuses on functional restoration through prosthetics, beginning as early as 6 months of age to enhance motor development and adaptation [6]. Physical and occupational therapy, along with family counseling, are crucial in ensuring holistic care. Studies have shown that early intervention significantly improves quality of life, independence, and psychosocial adjustment in affected children [7]. This case contributes to the limited literature on non - thalidomide - related phocomelia in India and underscores the need for further genetic studies, awareness programs, and guidelines for early intervention and long - term follow - up.

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