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Case Report

Radiology Section

Bilateral Upper Limb Congenital Anomalies in a Neonate: A Case Report

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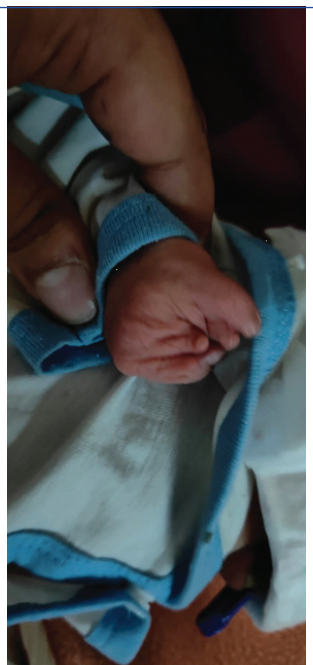
ABSTRACT

Congenital anomalies are diverse in nature and can affect multiple organ systems, posing significant diagnostic and management challenges. Among these, nubbins, characterised by the absence or underdevelopment of distal terminal phalanges, present a rare clinical finding. The present case report discusses a neonate born via normal vaginal delivery who demonstrated immediate cries upon birth. The patient's clinical presentation led to a series of diagnostic investigations. The absence of distal terminal phalanges in both hands was concerning; however, radiographs of the forearms showed normal structural findings. The detection of mild Developmental Dysplasia of the Hip (DDH) on hip Ultrasonography (USG) necessitated regular monitoring with USG and radiographs of both hips to ensure proper hip joint development. The USG of the spine revealed no anomalies, which was reassuring. The most significant finding came from the Echocardiogram (ECHO), which identified a small ostium secundum Atrial Septal Defect (ASD) with mild tricuspid regurgitation and pulmonary hypertension. The present case underscores the importance of ongoing research and collaboration in advancing our understanding and management of rare congenital anomalies.

Keywords: Atrial septal defect, Developmental dysplasia of the hip, Nubbins, Symbrachydactyly

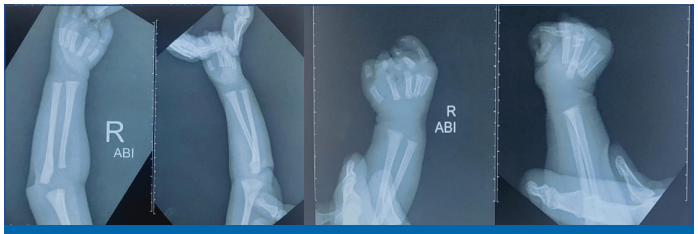
CASE REPORT

A single live-term baby was born via normal vaginal delivery, displaying immediate cries upon birth. The infant was delivered at a gestational age of 39 weeks to a 28-year-old female with an obstetric history of G4P2L2A1. The birth weight was recorded at 3.14 kg, with a length of 50 cm and a head circumference of 35 cm. The mother has a known history of Rheumatic Heart Disease (RHD) and is on penicillin prophylaxis. Antenatal USG scans were normal, and there is no family history of congenital anomalies or genetic syndromes. Clinical examination revealed the absence of distal terminal phalanges in both hands [Table/Fig-1].

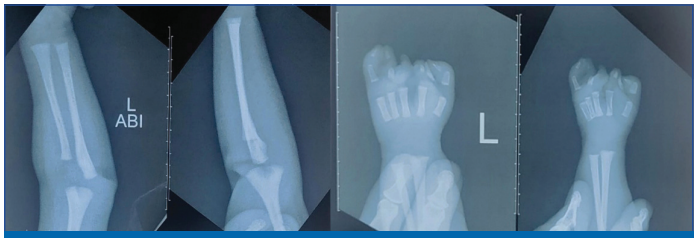


[Table/Fig-1]: Clinical picture of the baby showing nubbins in both hands.

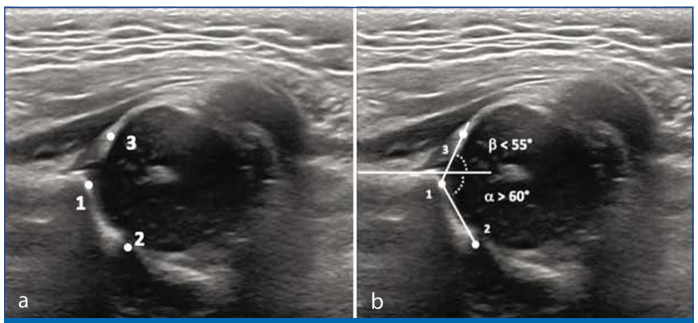
Radiographic evaluation of the forearms and spine revealed normal findings, while both hands exhibited an absence of distal phalanges [Table/Fig-2,3]. Ultrasound (USG) indicated mild DDH (Graf type 1) [Table/Fig-4a,b], with a normal spine [Table/Fig-5]. Echocardiogram (ECHO) identified a small ostium secundum ASD with a left-to-right shunt, mild tricuspid regurgitation and mild pulmonary hypertension [Table/Fig-6a,b]. Based on these findings,



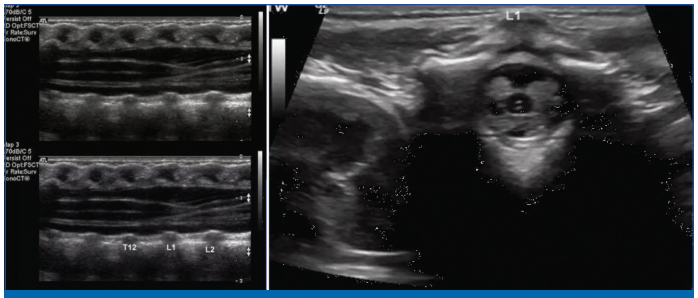
[Table/Fig-2]: Radiographs of right forearm and hand shows no anomalies in forearm and symbrachydactyly (no terminal phalanges in all fingers of right hand).



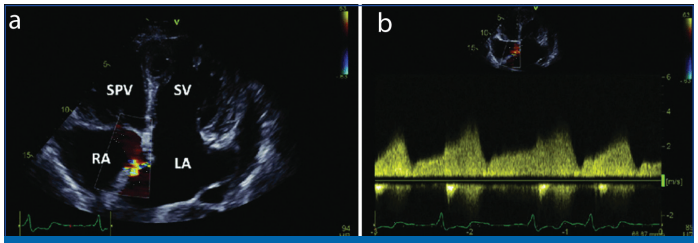
[Table/Fig-3]: Radiographs of left forearm and hand show no anomalies in forearm and symbrachydactyly (no terminal phalanges in all fingers of left hand).



[Table/Fig-4]: Ultrasound showing Graf type 1 hip: a) Landmarks on standard imaging plane 1-bony acetabular rim, 2-triradiate cartilage, 3-labrum; b) Measurement of alpha angle (α) and beta angle (β).



[Table/Fig-5]: Ultrasonography (USG) of the present case showing normal spine.



[Table/Fig-6]: a,b) Small ostium secundum Atrial Septal Defect (ASD) with a left-to-right shunt.

the diagnosis includes congenital absence of distal phalanges in the hands (nubbins or symbrachydactyly), developmental hip dysplasia grade 1, and a small ASD with associated mild cardiac anomalies. The parents were counselled regarding genetic counselling, but it could not be pursued due to financial constraints. The patient did not return for further follow-up, and the authors were unable to contact them.

DISCUSSION

This is a case of a newborn with multiple congenital anomalies: absence of distal terminal phalanges in both hands (symbrachydactyly), DDH, and a small ostium secundum ASD. The combination of these anomalies, along with the mother's history of RHD and the absence of family history or antenatal indicators, creates a unique and complex clinical scenario.

The absence of distal terminal phalanges, commonly referred to as nubbins, is an infrequent congenital anomaly that necessitates thorough evaluation [1]. The mild DDH observed highlights the need for early detection and intervention to prevent long term complications such as hip dysplasia or osteoarthritis [1,2].

The combination of symbrachydactyly, DDH, and ASD raises the possibility of a genetic or syndromic condition, although no specific syndrome can be definitively diagnosed without genetic testing. Conditions like Holt-Oram syndrome, which involves congenital heart defects and limb anomalies, as well as, other limb-heart syndromes, might be considered [3]. However, the absence of forearm involvement and the specific nature of the anomalies in this case do not perfectly align with known syndromes.

The maternal history of RHD and ongoing penicillin prophylaxis is notable. RHD itself is not typically associated with congenital anomalies, but the medication and underlying maternal condition could potentially influence foetal development. However, there is no direct evidence linking penicillin prophylaxis to the congenital anomalies observed in the present case infant.

Symbrachydactyly represents a specific subset of congenital hand differences with various morphological presentations, complicating diagnosis and treatment planning. With an incidence of about 0.6 per 10,000 live births, it predominantly affects males and is usually unilateral, more frequently involving the left side [4].

The diagnosis of mild DDH necessitates monitoring and possible early intervention to optimise outcomes. DDH, characterised by the abnormal development of the hip joint, can lead to instability if untreated, underlining the importance of vigilance in neonates presenting with multiple anomalies [5].

Atrial septal defect, a common congenital heart defect, can lead to significant complications, if not managed appropriately. The presence of a small ASD in the current case, along with mild tricuspid regurgitation and pulmonary hypertension, underscores the necessity for comprehensive cardiac evaluation in infants with congenital anomalies [6].

According to current recommendations for a small ASD with mild symptoms, regular follow-up with a paediatric cardiologist and medical management with medications to manage symptoms have been prescribed. If the ASD does not close on its own or if symptoms worsen, interventional procedures such as transcatheter closure will be considered [4-6].

The management of the present case involves a multidisciplinary approach. The mild DDH (Graf type 1) generally responds well to conservative management with a Pavlik harness. Close monitoring is essential to ensure proper hip development. The small ASD with mild associated cardiac anomalies requires regular follow-up with paediatric cardiology to monitor for potential spontaneous closure or the need for intervention.

Despite financial constraints precluding immediate genetic testing, it remains important to counsel the family about the possibility of genetic factors and discuss the potential for future testing, although this could not be done due to the parents' financial difficulties.

The primary challenge in the present case was the inability to perform genetic counselling and testing because of these financial constraints. Genetic testing could provide valuable insights into the aetiology of congenital anomalies and guide future family planning. Additionally, the lack of follow-up limits the understanding of the long-term outcomes and the effectiveness of the initial management strategies.

CONCLUSION(S)

The combination of absent distal terminal phalanges, mild DDH, and ASD in this neonate requires coordinated care from various specialists, including Neonatology, Orthopaedics and Cardiology. Early detection, thorough evaluation, and ongoing monitoring are crucial for optimising outcomes and ensuring the holistic well-being of affected infants. Comprehensive and collaborative medical care is vital for neonates with complex congenital anomalies. Timely diagnosis and intervention can significantly improve outcomes.

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