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Case report

Symbrachydactyly: a rare congenital anomaly – A case report

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Abstract

Introduction: Symbrachydactyly is a rare unilateral congenital hand anomaly with an incidence of approximately 1 in 32,000 babies born each year. It usually presents with shortened or absent central digits, sparing border digits. It can occur following an insult to the apical ectodermal ridge mainly affecting the vascular supply of the upper limb. According to the extent of skeletal involvement of the hand, there are variety of clinical presentations. This paper presents a case of symbrachydactyly involving the right upper limb with absent central digits, highlighting the presentation, classification, etiology, and possibilities of effective management.

Case report: A 38-year-old mother of 2 children presented in her 3rd pregnancy, following an uncomplicated pregnancy period. She was not exposed to any chemicals or radiation during pregnancy. A male neonate was born following an uncomplicated normal vaginal delivery. The neonate was born with absent fingers on the right hand, which were not identified prenatally. He had a normal left hand. The parents were counseled and the baby was referred to a pediatric plastic surgeon after a multidisciplinary meeting.

Conclusion: Symbrachydactyly, despite its rarity, can significantly affect the child's quality of life. In this case, the neonate had absent middle fingers with two nubbins instead of border digits on the right hand, which can be classified under the peromelic type according to the Blauth classification. Prevention is challenging due to uncertain etiology. The child's quality of life is limited by the degree of skeletal involvement. Depending on the degree of involvement, surgical or non-surgical methods and rehabilitation can improve the function of the hand.

Keywords

Case report, symbrachydactyly, hand anomalies, apical ectodermal ridge, finger nubbins, Blauth classification.

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Introduction

Symbrachydactyly is a rare unilateral congenital hand anomaly, with shortened or absent central digits with sparing of border digits [1-7]. It is nonhereditary and can occur following an insult to the apical ectodermal ridge (AER), mainly involving the vascular supply of the upper limb [2]. The presentation can vary with the degree of skeletal involvement, which can be identified clinically and radiographically [3]. Management is focused on improving the appearance and function, considering the expectations of the parents [2]. The etiology is unknown, but vascular dysgenesis during fetal development is a leading hypothesis [5]. This case report elaborates on the presentation, classification, etiology, and management of a case of symbrachydactyly involving the right upper limb with absent central digits.

Case presentation

A 38-year-old mother of 2 children presented in her 3rd pregnancy at term, for a vaginal delivery. It was an uncomplicated pregnancy, with no infections or medical comorbidities. She was not exposed to any chemicals or radiation during the pregnancy. She has 2 healthy living children delivered via normal vaginal delivery. There is no family history of congenital anomalies. No anomalies were detected by ultrasound scan during pregnancy.

A male neonate was delivered via a normal vaginal delivery at term. On neonatal examination, the baby was weighing 3.25 kg. He was born with absent central fingers on the right hand (**Fig. 1**) and with a normal left hand, suggestive of symbrachydactyly. On genital examination, the right side testis was absent. The anus was patent.

Based on the presentation, investigations were done to exclude other anomalies, including a 2D echocardiogram (which revealed an ostium secondum, atrial septal defect), and an ultrasound of abdomen and kidney, ureter, and bladder (which was normal). Poland syndrome was considered as a differential diagnosis, which was excluded due to the absence of the typical alterations associated with symbrachydactyly. A referral to a pediatric plastic surgeon was done after having a multidisciplinary meeting with a neonatologist, paediatric radiologist, psychiatrist, anaesthetist, physiotherapist, and geneticist.

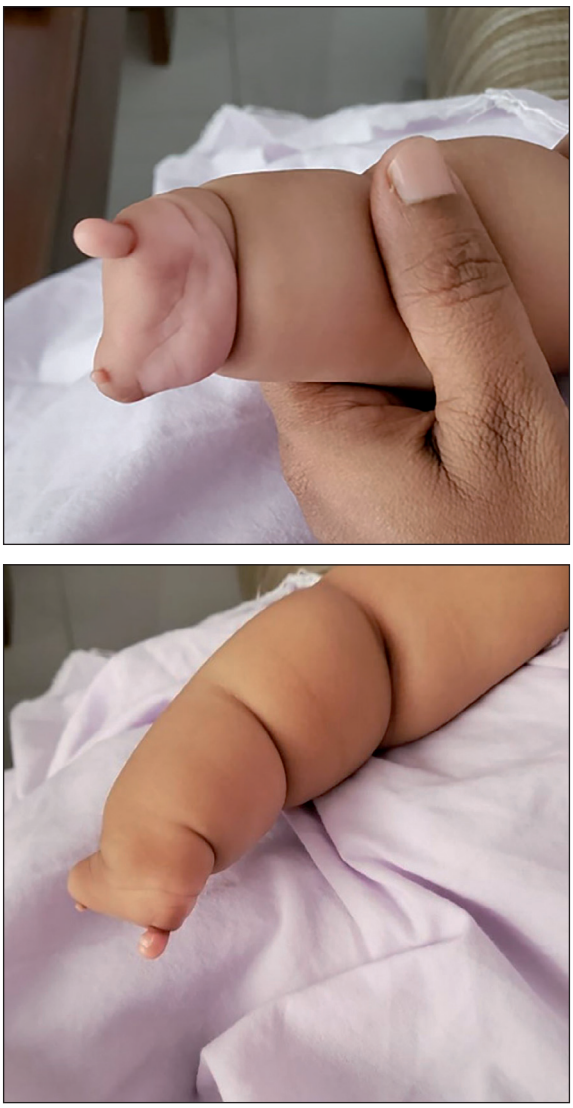

Figure 1. Symbrachydactyly of the right hand with absent central digits and two nubbins instead of border digits.

Discussion

Symbrachydactyly is a rare congenital anomaly with unilateral involvement, shortened or absent central digits with sparing border digits of the upper limb [1]. In the Oberg, Manske, and Tonkin (OMT)

classification adopted by the International Federation of Societies for Surgery of the Hand (IFSSH), it is included under the abnormal axis formation of the whole upper limb, considering the association with Poland syndrome or the hand plate [4]. In this case, we mainly focus on the involvement of the hand plate.

Symbrachydactly is classified according to the degree of involvement. Blauth classification includes 4 phenotypes: short fingers – brachydactylous type; cleft hand – oligodactylous type; absence of digits 2 to 5, with the thumb present – monodactylous type; adactyly with rudimentary nubbins – peromelic type. The short-finger (brachydactylous) symbrachydactly is characterized by the presence of a thumb and four short digits that might have missing distal phalanges. The oligodactylous symbrachydactly is characterized by the presence of a thumb and a fifth digit, referred to as "an atypical cleft hand", and central aplasia of digits. The monodactylous symbrachydactly is characterized by the presence of a thumb and four partially formed digits, referred to as "remnants" or "nubbins". The peromelic symbrachydactyly is characterized by the absence of the thumb and no fingers at the metacarpal level. In this case, the baby had absent central fingers with two nubbins instead of border digits on the right hand, which can be classified under the peromelic type.

Yamauchi and Tanabu included 7 types based on morphology and skeletal involvement described by radiological findings, but Foucher's modification of Blauth classification aided surgical management [3].

Limb development occurs in the proximal to distal directed by the AER [2]. Disruption of AER can result in symbrachydactyly, and nubbins might be a result of a regenerative ability following an insult [2]. Embryonic flow disruption in the 5th or 6th week affecting the subclavian artery, vertebral artery, and branches is the main hypothesis [3]. Several internal and external factors are considered possible, as the exact cause is unknown. Internal factors include intrauterine compression of the embryo, cervical rib, and soft tissue edema. Exposure to external factors (such as chemicals, medication, hypoxia, and hyperthermia compromising vascular supply) are also suggested in the literature, but none of the causes are approved to have significant correlation [3].

A prenatal ultrasound scan can detect symbrachydactyly, which was not detected in our case. After birth, hand anomalies can be detected clinically, and radiography of the hand can identify skeletal involvement to decide on management accordingly. The main aim of the treatment is to improve the function and appearance as permitted by the extent of the involvement and expectations of the parents [1]. Non-surgical methods can use orthotics and prostheses to improve function. In unilateral involvement, occupational therapy can be used to improve the activities of daily living by using the unaffected limb. Hand reconstruction by surgery can improve the function to a certain degree [2].

Conclusion

Symbrachydactyly, despite its rarity, can significantly affect the quality of life of the child. In this case, the baby has absent central fingers with two nubbins instead of border digits on the right hand, which can be classified under the peromelic type, according to the Blauth classification [3]. Prevention is challenging due to uncertain etiology. The quality of life of the child is limited by the degree of skeletal involvement. Depending on the extent of involvement, surgical or non-surgical methods and rehabilitation can improve the function of the hand [2].

Informed written consent

Informed written consent was obtained from the parents to publish the case and photographs.

Declaration of interest

The Authors declare that there is no conflict of interest.

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