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Case Report

Atypical cleft hand with complex syndactyly: An unusual form of hand oligodactyly

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ABSTRACT

Hand oligodactyly refers to a developmental defect which results in the presence of less than 5 digits on a hand. It presents as a component of 4 main categories of congenital upper limb malformations. Herein, we present a 50-year-old man with an unusual form of hand oligodactyly which is characterized by atypical cleft hand accompanied by complex syndactyly between the thumb and the index finger. Accurate characterization of hand oligodacticity can sometimes be challenging due to unusual phenotypic appearances accompanying the abnormality. Radiological evaluation is of great importance for correct identification and classification of such complex hand anomalies, and the treatment should be highly individualized in these patients.

Introduction

Hand oligodactyly refers to a developmental defect which results in the presence of less than 5 digits on a hand. It presents as a component of 4 main categories of congenital upper limb malformations. These are symbrachydactyly, central deficiency (typical cleft hand), ulnar deficiency, and radial deficiency, in order of prevalence. Hand oligodactyly may also be the result of amniotic band syndrome which causes disruption of the growth of a digit or amputation of a formed digit [1].

Symbrachydactyly, which actually means short and webbed fingers, presents as 1 of the 4 main clinical types which are in the continuity of each other in terms of clinical severity. Cleft hand refers to a hand anomaly characterized by a V-shaped slit between the index and the long fingers. Ulnar and radial deficiencies are characterized by the ulnar or radial longitudinal deficiency of the upper limb which affects the structural development of the affected side of the forearm and hand. Amniotic band syndrome may display a variable clinical presentation ranging from a slight banding around a limb to multiple amputations [2]. Herein, we present a 50-year-old man with an unusual form of hand oligodactyly which is characterized by atypical cleft hand accompanied by complex syndactyly between the thumb and the index finger.

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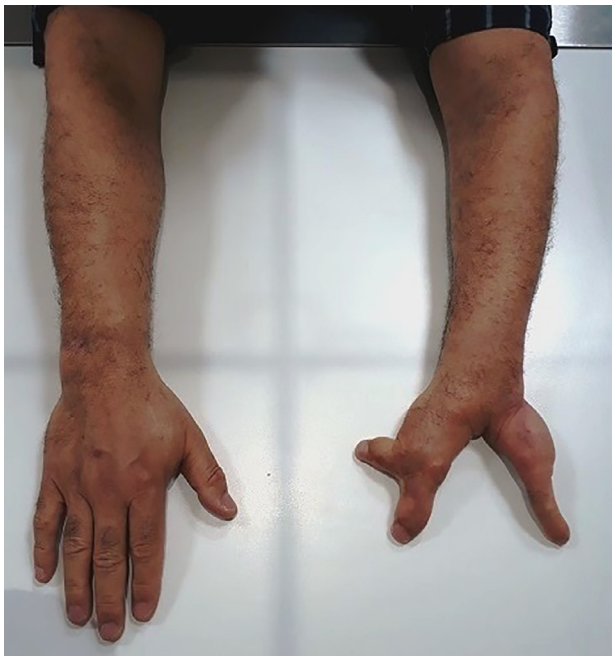

Fig. 1 – Clinical photograph of both forearms and hands demonstrates oligodactyly of the left hand and a normal right hand. Note the skin and nails of the present fingers of the left hand are intact.

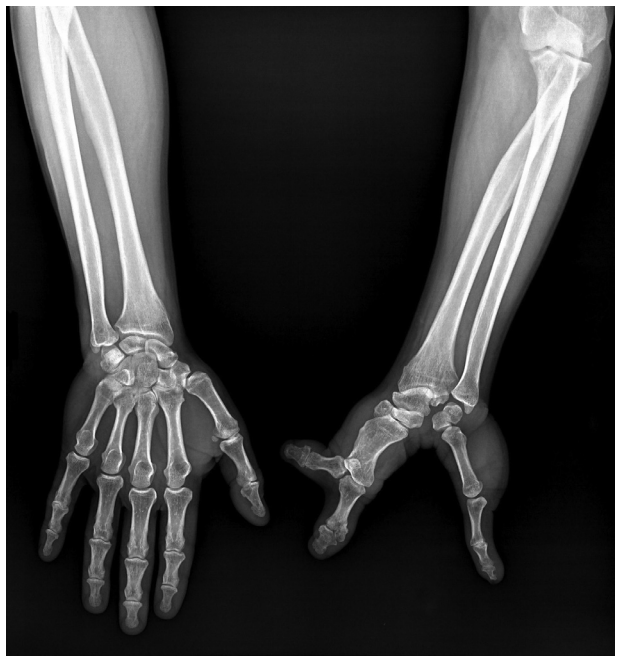

Fig. 2 – Posteroanterior radiograph of both forearms shows that both ulnas and radiuses as well as the right hand bones were of normal size and shape. There is oligodactyly of the left hand.

Case report

A 50-year-old male with a left hand disability admitted to the hospital for compulsory health screening to qualify for disability benefits from the state social insurance agency. He had no health complaints other than the skeletal disorder involving his left hand. He is the second male child born to non-consanguineous healthy parents. His brother is completely healthy. There is no history of any kind of congenital skeletal abnormalities in the extended family. No other significant health problem is present in the history of his childhood. Oligodactyly of the left hand and a normal right hand was noted in the inspection. The skin and nails of the present fingers of the left hand were intact (Fig. 1). Using his left hand, he was unable to perform most of the manual activities but was capable of grip. The patient stated that he was offered surgery several times during childhood. However, neither he nor his family requested surgical treatment on the grounds that there was no promise of complete recovery.

Posteroanterior radiograph of both forearms showed that both ulnas and radiuses, as well as the right hand bones, were of normal size and shape. However, there was oligodactyly of the left hand (Fig. 2). Posteroanterior radiograph of the left hand showed the absence of the third and fourth fingers as well as the respective metatarsals. There was complex syndactyly between the thumb and the index finger where the proximal phalanges of both fingers were articulating with a common bulky metatarsal. A delta-shaped small middle phalanx was present in the structure of the index finger. The fifth finger and metatarsal were present and were of normal size and shape. The carpal bones were deficient and the present carpals were of abnormal shape. Trapezium and trapezoid were separated and well developed. Although the form of scaphoid was atypical, its development was normal. Capitate was absent and lunate was coalesced with the distal radius. Hamate, triquetrum and pisiform seemed coalesced with each other. No ulnar variance was present (Fig. 3).

Subsequently, the patient underwent genetic testing for limb malformations, and no causative genetic alterations were identified. Based on the characteristic radiographic findings and genetic test results, the patient was diagnosed as having an atypical cleft hand accompanied by complex syndactyly between the thumb and the index finger.

Discussion

The typical cleft hand is 1 of the 4 main categories of congenital upper limb malformations which is characterized by the variable absence of the index, middle and ring fingers, and central carpus. A typical appearance of cleft without ectodermal elements, including fingernails, is present in patients with cleft hand. In mild forms of the disorder, a deep web space may be the only finding. However, in cases of severe cleft hand, complex syndactyly, deletion of 1 or more fingers or bizarre bone and joint structures may be observed. At first glance, the phenotype of the hand oligodactyly we currently present looks very similar to that of the cleft hand. However, in contrast to the cleft hand, ectodermal elements of the present case are all preserved. Further, our case differs from the cleft

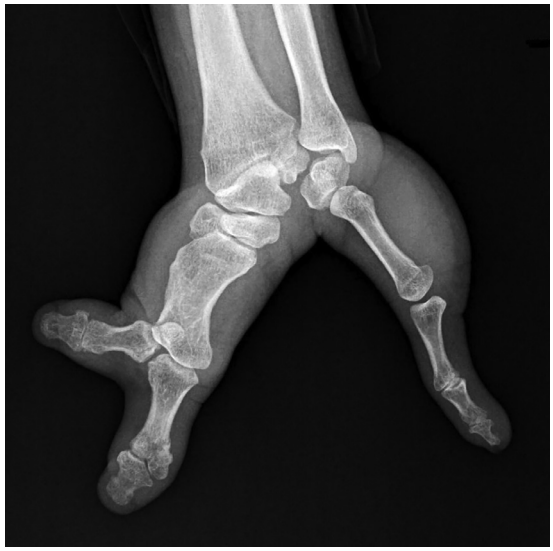

Fig. 3 – Posteroanterior radiograph of the left hand shows the absence of the third and fourth fingers as well as the respective metatarsals. There is complex syndactyly between the thumb and the index finger where the proximal phalanges of both fingers are articulating with a common bulky metatarsal. A delta-shaped small middle phalanx is present in the structure of the index finger. The fifth finger and metatarsal are present and of normal size and shape. The carpal bones are deficient and the present carpals are of abnormal shape.

hand in that the inheritance of the cleft hand is typically familial, and it is characterized by bilateral hand and foot involvement in the majority of the cases [1,2].

Symbrachydactyly is an other upper limb malformation category which is almost always a noninherited sporadic anomaly that involves only 1 limb. It has a spectrum of presentation which comprises 4 main clinical types: short finger type, oligodactylous type, monodactylous type, and the peromelic type. Preservation of ectodermal elements, including the distal skin and fingernails, is a characteristic feature of all types of the disorder. Short finger type represents the mildest form. As the involvement progresses, the index finger is suppressed, followed by the middle, the ring, and the small fingers, respectively. The oligodactylous type is also referred to as "atypical cleft hand" owing to its similarity to the typical cleft hand. In an atypical cleft hand, the central parts, including metacarpals, phalanges, and musculotendinous elements are characteristically absent, and the border digits are hypoplastic. The thumb is mostly preserved in patients with atypical cleft hand [3,4]. The present case is consistent with the atypical cleft hand in terms of its sporadic nature, its unilaterality, the preservation of the ectodermal units and the deficiency of the central parts. However, the complex syndactyly involving the thumb and index finger of our patient is an unusual phenotypical feature accompanying the atypical cleft hand. Moreover, different from the atypical cleft hand, our patient does not have any forearm hypoplasia which usually accompanies symbrachydactyly.

Radiological evaluation is of great importance for correct characterization and classification of the hand anomalies. Currently, the classification proposed by the International Federation of Hand Surgery Associations is used to classify congenital hand and upper extremity anomalies [5]. The phenotype of the case we currently present seems to fit the 1B category, which includes the hand malformations with failure of axis formation/differentiation-hand plate, according to this classification. Treatment of hand malformations should be planned during childhood, and the choice of a specific treatment must be planned with realistic functional goals and a reasonable expectation of achieving these goals after treatment. For children with unilateral hand deficiency, quality of life is usually very close to normal, but often the most difficult thing for the child and the family is the psychological distress of appearing different from others [3]. Nonsurgical treatments such as prosthetic and orthotic applications, occupational therapy and psychotherapy can help children with unilateral malformations to gain mastery of daily living activities and to increase self-confidence [6]. Surgical treatments must be planned according to the specific aspect of the disorder including syndactyly, web contracture, brachydactyly, digit instability, and lack of opposition, and should be highly individualized [7].

Supplementary materials

Supplementary material associated with this article can be found, in the online version, at doi:10.1016/j.radcr.2019.09.004.

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