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CASE REPORT OPEN ACCESS

Brachydactyly type B: A rare case

Juan Pablo Dominguez, Waiz A. Wasey, Sharefi Saleh

ABSTRACT

Introduction: Brachydactyly is the term used to describe disproportionately short fingers and/or toes. The abnormality may be isolated or a part of an underlying syndrome. There are many types of isolated brachydactyly, of which type B is a very rare one. It is characterized by underdevelopment of the fingers with the complete absence of fingernails with the thumb intact. However, some cases may show flattening or splitting of the thumb. ROR2 gene mutations are responsible for the condition. Very few cases of such nature has been reported.

Case Report: We report a case of a 35-year-old male who presented to the emergency room with an injury to the right hand. On physical examination both his hands had underdeveloped fingers with absent fingernails. His thumbs were intact and functional. He had no facial dysmorphism or abnormalities in his toes. The patient acknowledged a strong family history of similar features.

Conclusion: Brachydactyly is the shortening of fingers with or without nails. The condition is diagnosed by clinical features, radiological studies, family history and genetic testing. Type B brachydactyly has an autosomal dominant inheritance pattern. The strong family history, typical clinical features and the X-ray of the hands helped us to label the patient as having brachydactyly type B.

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Keywords: Brachydactyly type B, Short fingers, Absent fingernails, Underdeveloped fingers

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INTRODUCTION

Brachydactyly is the term used to describe disproportionately short fingers and/or toes. The abnormality may be isolated or a part of an underlying syndrome. There are many types of isolated brachydactyly, type B being very rare. It is characterized by underdevelopment of the fingers with complete absence of fingernails. The thumb is generally intact, however, some cases may show flattening or splitting of the thumb. Very few cases of such nature have been reported. Brachydactyly is diagnosed from its clinical features, radiological studies, strong family history and genetic studies. The ROR2 gene mutations have been shown to be responsible for the abnormality. Brachydactyly type B presents with an autosomal dominant inheritance pattern.

CASE REPORT

A 35-year-old male walked into the emergency room with a bleeding right hand. The patient was working in the garden when one of the tools slipped and he injured his hand. While attending to his wounds, unusual abnormalities were seen in both of his hands. The four digits, from index to little finger, were all short with

no fingernails, except for the thumbs; which were well developed (Figure 1). On further inquiry, the patient stated that his hands have been like this since childhood, and his father and only brother have similar shortened fingers. On detailed physical examination his fingers were short with either absent or hypoplastic distal phalanges. The thumbs were of normal size and fingernails present. No flattening or splitting of the thumb phalanges was seen or felt. The proximal interphalangeal joints were functional. He had good muscle strength and neurological sensations were intact. Further examination of the patient revealed no dysmorphic facial features or toe abnormalities. No coloboma of iris or cataracts were seen on eye examination. No murmurs or abnormal heart sounds were heard on auscultation of the chest. His stature was within normal limits and his gait was normal. The patient was not interested in undergoing any further testing, as he was already aware of his condition. He only wanted treatment for the current injury. An old X-ray image was provided by the patient on request (Figure 1).

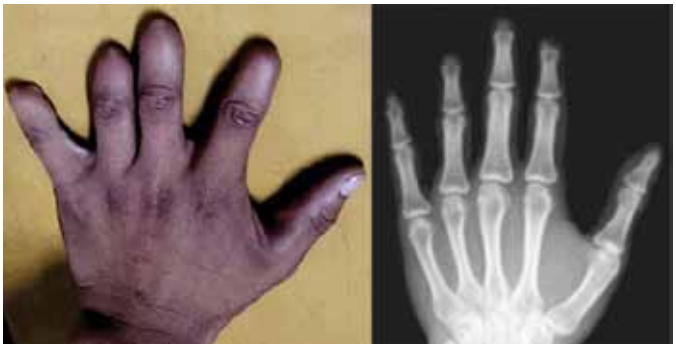

Figure 1: An image of patient's left hand showing shortening of the fingers with no fingernails and intact thumb, with a comparing X-ray.

DISCUSSION

Brachydactyly is the term used for shortening of fingers and/or toes. This abnormality maybe isolated or a part of an underlying syndrome. There are various types of isolated Brachydactyly; the common ones are A3 and D [1]. Brachydactyly type B, which we reported, is one of the rare subtypes of this condition. It involves the underdevelopment of fingers with the absence of fingernails, with the exception of the thumbs having normal morphology in most cases. There have been reported cases documenting flattened or split thumb bones. The majority of cases have shown an autosomal dominant inheritance pattern, and is caused by mutations in the ROR2 gene. [2] The toes may or may not be involved. In our case, the toes were not affected.

Being a congenital disorder, the shortening maybe noticed at birth. However, in some cases it may become apparent as the body grows. Brachydactyly is not associated with pain or other symptoms, unless another underlying syndrome is associated with it. Some of the syndromes associated are Robinow syndrome, Rubinstein-Taybi syndrome and Pan syndrome [3]. Individuals affected with brachydactyly may have difficulty in using their hands to complete daily tasks or may have gait abnormalities when the toes are involved.

The diagnosis of brachydactyly involves careful clinical examination, X-rays and family history. Mild cases may be diagnosed with X-rays only. When other signs and symptoms are present with brachydactyly, then extensive investigation with full skeletal X-rays maybe warranted to rule out congenital syndromes [4].

Isolated brachydactyly are of various subtypes. These subtypes are differentiated based on the number of fingers affected or the type of shortening involved. Brachydactyly type A is shortening of the middle phalanges only. It is further divided into subtypes depending on which digits are involved. Type A1 presents with absent or malformation of the middle phalanges of all digits [5]. The toes are also affected, with the proximal phalanges of the thumbs and big toes being short. Type A2 in which malformation of the middle phalanx of the index and second toe are involved [6]. Type A3 is the shortening of the middle phalanx of the little finger only. A radial deviation of the distal phalanx maybe noted. This subtype is common with a frequency ranging from 3–21% [7]. Type A4 involves shortening of the middle phalanx of the second and fifth digits. The feet also show absence or shortening of middle phalanx of the lateral four toes. Sometimes the fourth digit of the hand may be involved with a radial deviation [8]. Type A5 presents with abnormalities in the middle phalanges, along with nail dysplasias. A duplication of the distal phalanx of the thumb has also been reported. Temtamy and Aglan argue that this should be a part of the Type B brachydactyly [9]. Brachydactyly type C is characterized by shortening of the middle phalanx of the index, middle and little fingers; and hyperphalangy of the index and middle fingers. The first metacarpal is short and the ring finger is the longest. The toes may or may not be affected. The pattern of inheritance of this subtype is also autosomal dominant [10]. Type D is the subtype in which only the thumbs are affected. It maybe be unilateral or symmetrical. This subtype is common and ranges from 0.4–4%. Type E brachydactyly is shortening of the metacarpals, the terminal phalanges maybe normal or short. Individuals with this condition may also have a short stature [1].

Those affected with isolated brachydactyly may not require any treatment apart from physical and/or occupational therapy and rehab when needed. These individuals have a normal life span. However, when the brachydactyly is a part of a syndrome, generally presenting with other signs, a thorough examination and extensive investigations maybe needed to diagnose and appropriately treat the affected person. [11]

CONCLUSION

Brachydactyly is a rare congenital deformity, and type B is one of the rarest subtypes. Due to its rarity, we felt the need to report this case. The clinical features along with the radiological evidence pointing to the underdevelopment of the fingers from index to the little one, with intact thumbs, confirmed our diagnosis of brachydactyly. The patient did not present with other malformations or complains requiring any further evaluation. A complete thorough physical examination is warranted for patients who report other symptoms, as brachydactyly may be associated with other syndromes. The patient was treated for his wounds and referred to physical therapy, to help improve his fine motor skills, to prevent similar accidents in the future.

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Author Contributions

Juan Pablo Dominguez – Substantial contributions to conception and design, Acquisition of data, Analysis and interpretation of data, Drafting the article, Revising it critically for important intellectual content, Final approval of the version to be published
Waiz A. Wasey – Substantial contributions to conception and design, Acquisition of data, Analysis and interpretation of data, Drafting the article, Revising it critically for important intellectual content, Final approval of the version to be published
Sharefi Saleh – Acquisition of data, Drafting the article, Revising it critically for important intellectual content, Final approval of the version to be published

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Conflict of Interest

Authors declare no conflict of interest.

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SUGGESTED READING

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