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Brachydactyly type B: rare case report

Braquidactilia tipo B: a propósito de un caso poco frecuente

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Key words:

Foot, brachydactyly, genetic, malformation.

Abstract

Brachydactyly is a genetic condition characterized by shortening of the fingers or toes that can occur in isolation or as a part of a syndrome. This article report the clinical case of a 62-year-old woman who presented subungual hematoma in both first toes and isolated brachydactyly affecting the second, third and fourth toes of the left foot. The objective of this article is to highlight a rare clinical finding and its possible treatments.

Palabras clave:

Pie, braquidactilia, genética, malformación.

Resumen

La braquidactilia es una condición genética caracterizada por el acortamiento de los dedos de las manos o de los pies que pueden ocurrir de forma aislada o formando parte de un síndrome. El presente artículo muestra el caso clínico de una mujer de 62 años, que presentaba hematoma subungueal en ambos primeros dedos del pie y braquidactilia aislada afectando al segundo, tercer y cuarto dedo del pie izquierdo. El objetivo de este artículo es poner en evidencia un hallazgo clínico infrecuente y sus posibles tratamientos.

Introduction

Brachydactyly (BD) is a limb malformation that affects the length of the fingers and toes, characterized by dysostosis, a defective ossification of fetal cartilage, which may occur in isolation or in combination with various bone malformations as part of a syndrome[1,2].

Malformations of the hands and feet that may appear occur during blastogenesis, which happens from week 4 to week 8 after conception or much later once the limb has been formed[3,4]. Under normal conditions, during embryonic development, the formation of the lower limb occurs on week 6 of development, when the condensed mesoderm chondrifies to form a hyaline cartilage model of the bones of the lower limb[5].

Different anomalies such as symphalangism, syndactyly, or digital deviation are characteristic of presenting BD, which can be recognized at birth or in childhood after a failure of the ossification of digital components. Different types of BD can be distinguished based on anatomical criteria and radiological patterns[3]. Most types are rare except for type A3 BD and type D BD, which have a prevalence of around 2 %[6,7] (Table I). This article focuses on type B BD (BBD; OMIM # 113000)[11].

The distinctive features of DBD are hypoplasia or absence of distal phalanges and nail plates, as well as shortening of the middle pha-

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Table I. Classification of Brachydactyly[8-10].

Type	Main Characteristics	OMIM®
A1	Short middle phalanges. Femoral head dysplasia	112500
A2	Short middle phalanges of the 2nd finger of the hand and foot	112600
A3	Radial deviation of the terminal phalanx of the 2nd finger. Shortening, flexion, and curvature of the 5th finger	112700
A4	Short middle phalanges of 2nd and 5th fingers. Absence of four lateral fingers	112800
A5	Absence of middle phalanges. Nail dysplasia. Duplication of the distal phalanx of the 1st finger	112900
B	Short middle phalanges. Absent or shortened distal phalanges. Nail dystrophy of the 2nd and 5th fingers. Partial widening or duplication of the distal phalanx of the 1st finger. Syndactyly	113000
C	Short and deformed proximal and distal phalanges of the 2nd and 3rd fingers	113100
D	Short and wide distal phalanx of the 1st finger of the hand and foot	113200
E	Short metacarpals and metatarsals. Short stature. Short phalanges, especially the 1st finger of the hand and foot, and middle phalanx of the 5th finger. Cone-shaped epiphyses in the hand	113300
Brachymetatarsus IV	Shortening of the 4th toe and 4th metatarsal	113475
Sugarman's Brachydactyly	Pronounced shortening of the proximal phalanx	272150
Kinner Deformity	Curvature of the 5th finger distal phalanx	128000

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langes, producing an appearance of amputations from the 2nd to the 5th phalange[3,12,13].

DBD is an autosomal dominant disorder caused by heterozygous mutations in the ROR2 gene, which encodes a receptor tyrosine kinase located on chromosome 9 at 9q22[8,14,15]. This article aims to present this rare case to the podiatric community, its possible treatments, and have bibliographic references available.

Case report

We present the case of a 62-year-old Caucasian woman who came to the clinic due to the presence of subungual hematoma on both big toes. Upon physical examination, hypoplasia of the 2nd, 3rd, and 4th toes of her left foot was found along with micronychia, without presenting keratosis at plantar level (Figure 1).

Subsequently, a weight-bearing X-ray was performed in the anteroposterior projection to complement the examination. Exceptionally, the absence of middle and distal phalanges of the 2nd, 3rd, and 4th toes of the left foot was found (Figures 2 and 3). A difference in the metatarsal formula was observed between both feet, showing a minus index in the left foot and a plus-minus index in the right one. Findings were consistent with isolated BBD.

The patient had no family history of BD or genetic disorder. Regarding her medical history, she had hypertension and hypothyroidism, both pharmacologically treated. Conservative treatment was chosen since DBD was not an aesthetic issue for the patient, recommending the use of flexible, wide-toe footwear.

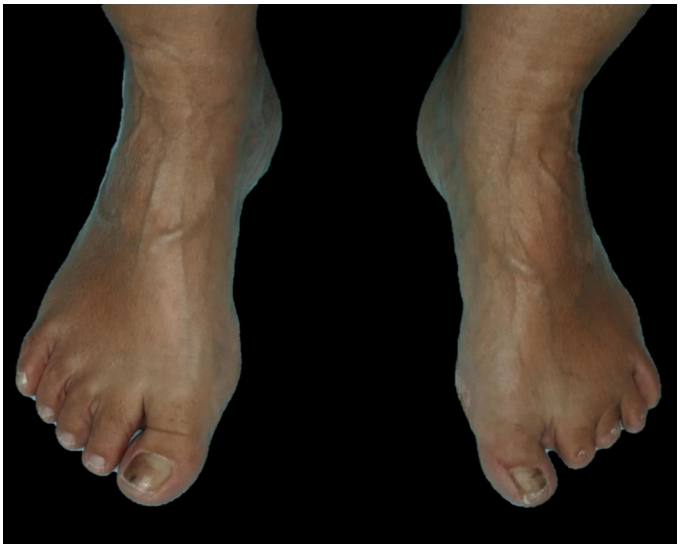

Figure 1. Patient standing, showing the absence of middle and distal phalanges of the left foot 2nd, 3rd, and 4th toes and micronychia affecting the 2nd, 3rd, and 4th toes, along with subungual hematoma on both big toes.

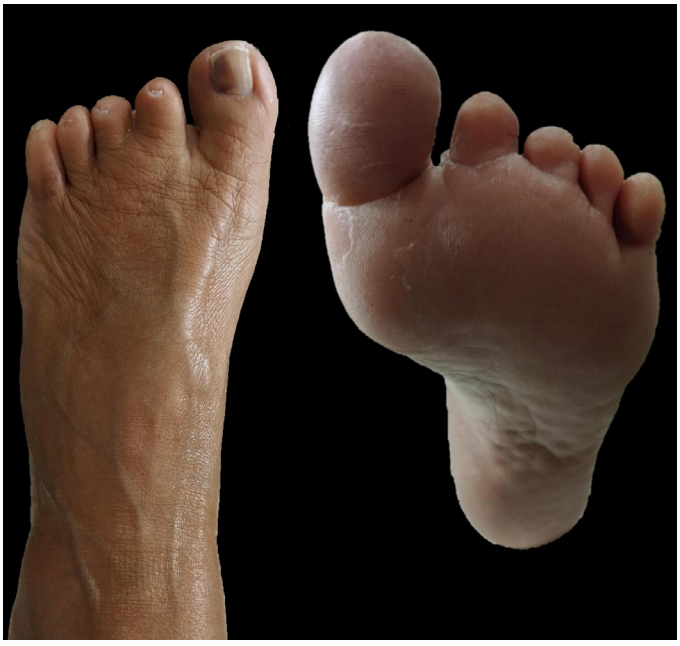

Figure 2. Dorsal and plantar view of the left foot.

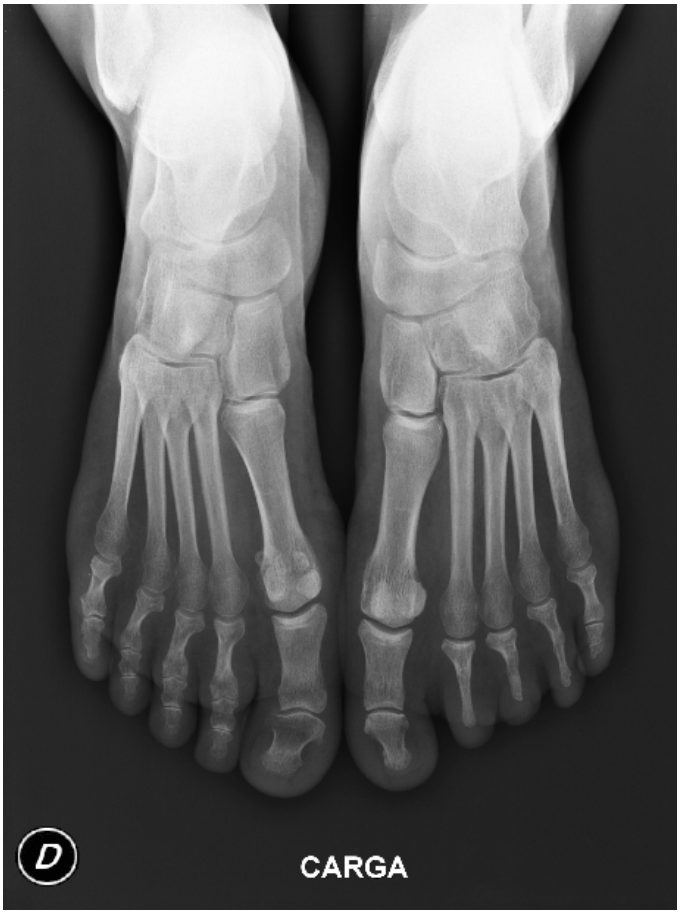

Figure 3. Weight-bearing X-ray, showing the absence of middle and distal phalanges of the left foot 2nd, 3rd, and 4th toes and bipartite sesamoids in the right foot.

Discussion

The prevalence of DBD is extremely rare and may occur as an isolated trait or as part of a complex malformation of a syndrome[14]. The phenotypes of DBD include facial defects and deformities of the fingers, including the nails, all of which are part of a syndrome. In this case report, the patient presented unilateral DBD with involvement of the 2nd, 3rd, and 4th middle and distal phalanges. The review article by Temany et al. indicated that surgery is only recommended if BD affects the function of the limb or for aesthetic reasons, but it is generally not necessary[9]. In our case report, no plantar keratosis was observed, which would suggest a biomechanical alteration, with a different metatarsal formula in each foot.

In 2010, Poerink et al. presented a case report similar to ours: a teenager from the Middle East with DBD and anonychia, whose phenotypes are part of Cooks syndrome[10]. Records of isolated DBD exist; AbuHaweeleh et al. presented the case of an 8-month-old infant of Asian origin who presented with absence of the middle and distal phalanges of the left foot 3rd toe, which did not require any surgical treatment[16]. Medical literature on DBD is limited vs other malformations such as brachymetatarsia, whose incidence is 0.02-0.05%, for which various surgical treatments are mentioned to lengthen the metatarsals, restoring the metatarsal parabola[17]. However, the feasibility of lengthening the phalanx is possible through the use of mini-fixators. In a study conducted in 2008, Kim JY et al. managed to lengthen distal phalanges to correct nail deformities, achieving a gradual elongation of 0.125 mm/day, with a mean lengthening of 9.8 mm. Fixation was performed with 3 transversely placed Kirschner wires in the distal phalanx and 1 Kirschner wire fixed to the head of the middle phalanx. Subsequently, a CK mini-fixator was placed, and an osteotomy was performed using an electric saw with a #15 blade between the nearest and distal pin[18]. Another treatment option was proposed by Lundborg G et al. through an osteotomy in the middle phalanx, followed by the insertion of Kessler distractors, initiating a progressive daily lengthening of 0.5-1 mm, allowing the phalanx to lengthen by 15 mm. After removing the device, a spongy cortical bone graft was applied from the iliac crest, with subsequent fixation using Kirschner wires[19]. Another example of phalanx elongation is that of Jiang Y et al., who achieved the elongation of the distal phalanx of the 1st toe using external fixators in patients with DBD, lengthening the toe by a mean 4 mm and also improving the nail aesthetic appearance. The nail plate was removed at the eponychium edge using a lifter, followed by a transverse incision in the nail bed near the lunula, and the nail bed and periosteum were lifted to expose the distal phalanx. Two 1.6 mm surgical screws were inserted into the distal phalanx, 1 at the proximal base to the osteotomy and the other distal to it. The osteotomy was performed with a mini-saw on the diaphysis. An external mini-fixator (Double Medical, Xiamen, China) was then placed[20].

In conclusion, BD remains an uncommon finding that can have surgical and/or conservative treatment depending on the aesthetic complexities that may impact the patient's life.

Conflicts of interest

None declared.

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