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Type-E Brachydactylous Family: Autosomal Dominant Inheritance with Variable Expression and Non-Penetrance

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Abstract

The word brachydactyly is used to describe the hands and feet with shortened digits. Autosomal dominant brachydactyly Type-E (BDE) is a congenital limb malformation characterized by shortened digits in hands and feet predominantly as a result of shortened metacarpals and metatarsals . A 45 yrs old female presented with shortened 4th toe bilaterally with no other complaints.The 4th metatarsals were shortened bilaterally, rest other metatarsals, tarsals and phalanges appeared normal on X-ray examination.On family history there was positive history of similar brachydactyly of 4th toe bilaterally in her paternal grandfather and unilateral brachydactyly of 4th toe in her son. These features match approximately with isolated Type-E Brachydactyly. Type-E brachydactyly always inherited as an autosomal dominant trait.In present study when pedigree was plotted it was found that though the brachydactylous trait inherited as autosomal dominant , IInd generation showed nonpenetrance of the concerned trait while same was appeared in the IIIrd generation and the same trait showed variable expression in IVth generation which is rarely observed. Isolated brachydactyly has excellent prognosis and the role of surgical correction is limited only for cosmetic purpose in the present case.

Keywords: Brachydactyly; Toes; Autosomal dominant; Non-penetrance; Variable expression.

Introduction

The word brachydactyly is used to describe the hands and feet associated with shortened digits or toes. (Brachy meaning "short" and dactylous meaning "digit") The digits themselves may be shorter than normal or they may appear small because of shortening of metacarpal and metatarsal bones in the hands or feet.[1] Most of the patients with brachydactyly do not require any specific treatment. When use of the hands is impaired, physiotherapy may improve their function. Corrective surgery can also be used to lengthen the hand or foot bones in some severe forms of brachydactyly. If brachydactyly is associated with other medical problems, such as hypertension, specific treatments for these problems may be indicated.[1,2] The various types of isolated brachydactyly are rare, except for types A3 and D, which are common, prevalence being around[3] 2%. In isolated brachydactyly, the inheritance is mostly autosomal dominant. In usual pedigree showing an autosomal dominant inheritance an affected person has affected either parent but with few exceptions those involve nonpenetrance of concerned trait. Though the trait was inherited as autosomal dominant not all characters are going to present in next generation and that is governed by many genetic and environmental factors leading to variable expression of that concerned trait. Isolated brachydactyly generally has an excellent prognosis.

Case report

A 45 yrs old female patient presented withshortened 4th toe bilaterally without functional deformity since birth. On

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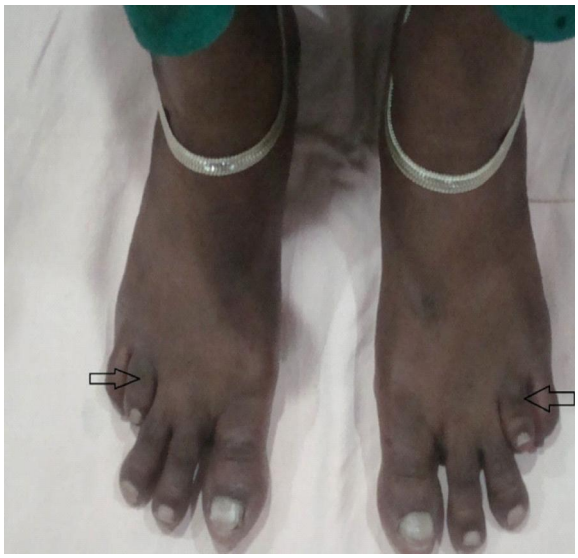
Fig 1a: Show bilateral 4th toe brachydactyly



examination it was found that the 4th toes were shortened bilaterally. No evidence of any lesions of nails. Both right and left hands were absolutely normal. Her height was 5ý2ýý. Rest general and systemic examinations were normal.

On family history her grandfather suffered from same 4th toe brachydactyly bilaterally. Details or photographs and radiograms were not available. Also there was family history of her son suffering from 4th toe brachydactyly of left foot only with right foot normal and

Fig 1b: Show bilateral shortening of fourth metatarsal

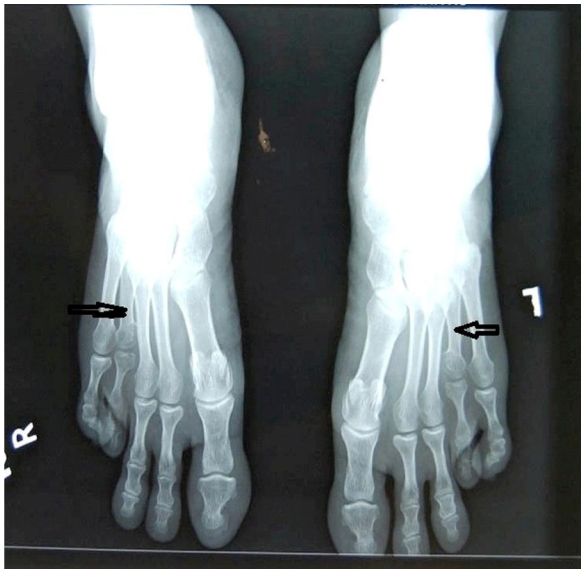
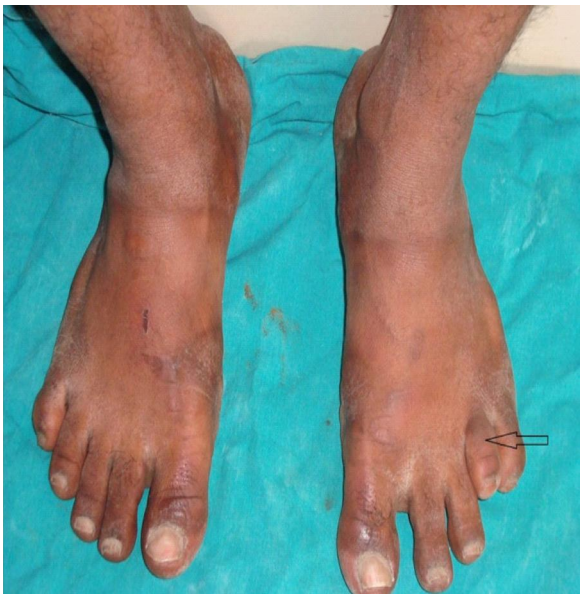


Fig 2a: Show left foot with 4th toe brachydactyly right foot normal

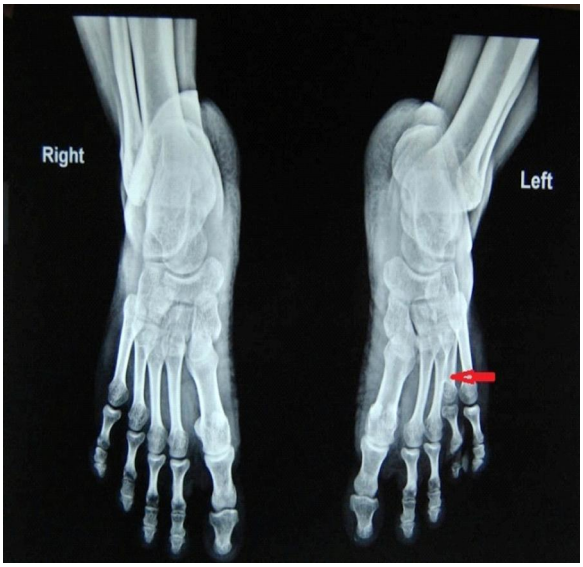


that appeared to be very rare occurrence. On general and systemic examination no other abnormality found (remove examination no other abnormality found).

From the fig 1a and fig 1b it is evident that the 4th toe appears shortened bilaterally because of Shortened 4th metatarsal. Rest all other bones in both feet were normal.

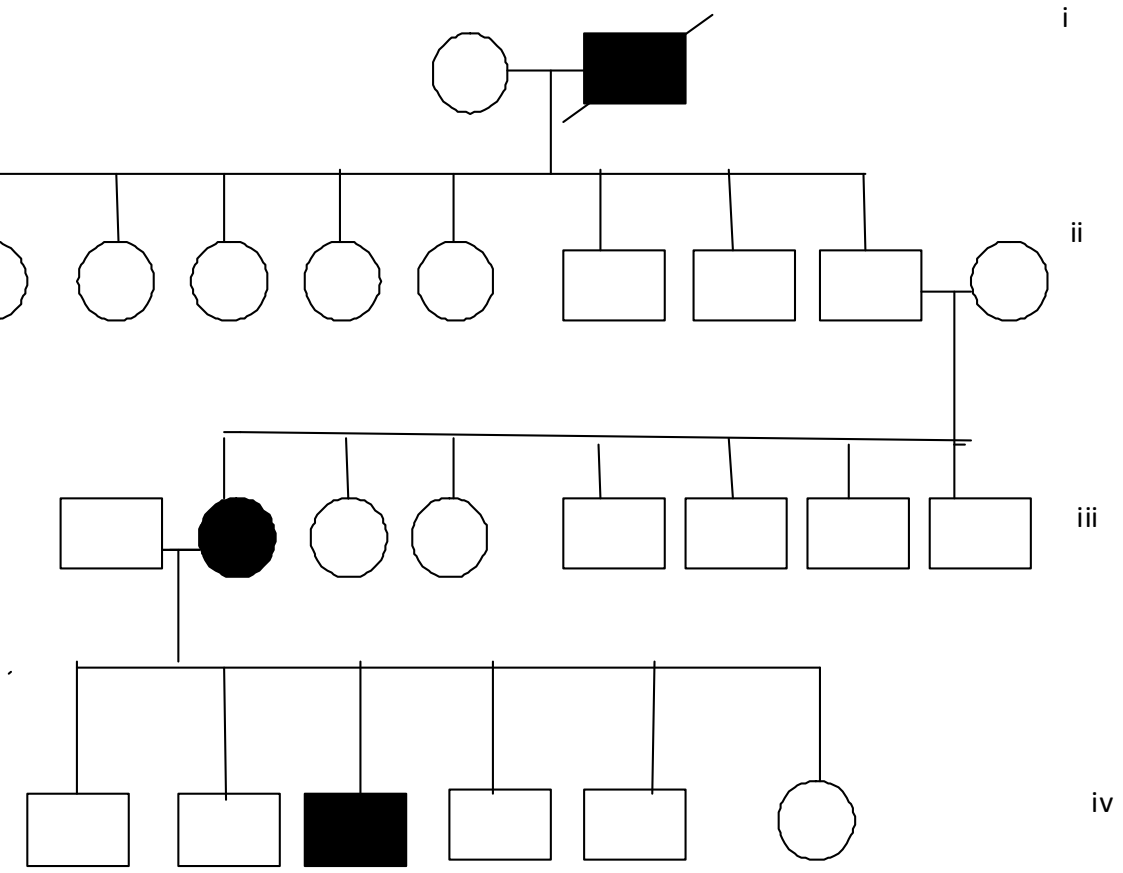
From fig 2a and fig 2b it is evident that the 4th toe appears shortened on left side because

Fig 2b: Show shortened 4th metatarsal. On right side no variation evident



of shortened 4th metatarsal on left side. Rest all other bones of left foot were normal. Also right foot appears absolutely normal in photograph as well as in radiogram.

The pedigree plotted as follows:



From above pedigree it is clear that in the first generation the Type–E brachydactylous trait in the grandfather of the patient. In the IInd generation no one suffered from the concerned trait indicating non-penetrance of the concerned trait. In the IIIrd generation 1st female child was suffered from the concerned trait bilaterally. In the IVth generation the IIIrd male child affected from the Type–E brachydactyly unilaterally indicating variable expression of the brachydactylous trait. Such a pedigree is more common with an autosomal dominant trait.

Discussion

Brachydactyly can involve any of the phalanges, metacarpals, metatarsals in many different combinations. The shortening of these bones may range from mild to severe. Sometimes certain bones are completely absent. Shortening of bones may occur in one or many or all the bones. For a particular digit or finger the entire digit may be short or the particular phalynx may be short. When brachyactyly involves the distal phalynx the fingernails or toe nails may be small or absent. Type-E brachydactyly is rare as an isolated anomaly. As an isolated anomaly it inherits as autosomal dominant trait with variable expressivity.[1] Trauma is the most common cause of this deformity.[4] It can also be seen in cases of Pseudopseudohypoparathyroidism (PPHP), neurofibromatosis and congenital adrenal hyperplasia due to 11-â hydroxylase deficiency. Pseudopseudohypoparathyroidism was excluded as the patients are not short statured, no evidence of cataracts. Moreover the phalanges of forth toes were not shorter as was the case with PPHP.[5,6,7] No e/o

neurofibromatosis was there.

In the present case report it is evident that this is a case of Type-E brachydactyly according to Bells classification.[8] It is clear from the above case report that there is positive family history of the same deformity inherited as autosomal dominant inheritance with non penetrance and variable expression of the trait. McKusick[1] reported a family with 17 affected members in three generations confirming autosomal dominant inheritance and variable expressivity. Non penetrance and variable expressivity are the major pitfalls in genetic counselling and so it is very necessary for the clinicians and counsellors to know the usual degree of penetrance and variable expressivity of the concerned autosomal dominant condition. Also non-penetrance and variable expressions are more conspicuous in humans than in animals and plants so the genetics must understand its importance when studying animal models of human diseases[9]. There are number of surgical techniques for correction of the brachydactyly including lengthening of the shortened metacarpal or the metatarsal[10,11], but in our case as the brachydactyly did not affect the function of those limbs the surgical role was limited for only cosmetic purpose.

Acknowledgment

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