Brachydactyly type C associated with shortening of the hallux

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Abstract

A four generation autosomal dominant pedigree of brachydactyly type C is presented with its radiological features. The hands and feet were similarly affected. All the subjects showing these changes had shortening of the big toes and, in addition, had cupped ears.

Brachydactyly was classified into five main types by Bell¹ in 1951. Type C involves shortening of the index, middle, and little fingers with deformity of the proximal and middle phalanges. Characteristically hypersegmentation produces an extra, wedge shaped bone at

the base of the proximal phalanx in the index and middle fingers. This typically produces ulnar deviation of the index finger.

Associated abnormalities of the feet have only rarely been described before and no previous cases also having bilateral cup shaped ears have been reported.

Case reports

The male index patient (case B, fig 1) presented at 50 years of age. He was of normal intelligence and height. The index and middle fingers of both hands were shortened with ulnar deviation of the index fingers (fig 2A,B). No functional deficit was apparent. Both right and left halluces were shortened, the former more than the latter (fig 2C,D). He had small, cup shaped ears resulting primarily from a deficiency in the length of the helical rim and scapha in the upper part of the auricle (fig 3). There was also some unfurling of the antihelical fold. He remembered his mother having similar deformities.

He stated that his sister's younger son's hands (case E) were similarly affected (fig 4E)

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Figure 1 Pedigree of family with brachydactyly type C. Subject B is the index case.

Figure 2 Hands and feet of index patient case B.

Figure 3 Small, cup shaped ear of the index case. Unfurling and shortening, in particular of the antihelical fold, is shown.

Figure 4 Radiographs of hands and feet of cases C, D, and E. Case D has had attempted corrective surgery to his right index finger.

while his sister (fig 4A) and older nephew (fig 4C) had shortening of both index fingers only. The younger nephew's son, 7 months old at the time of investigation, also had brachydactyly of the right and left index and middle fingers.

The big toes were deformed in cases C, D, E, and F as well (fig 4B,D,F). These family members all had similar shaped ears but refused facial photographs.

Discussion

Bell's grouping of 1951 is considered the standard reference for shortening of the digits.¹ Wood² in his case report described brachydactyly type C under the title hyperphalangism, drawing attention to the occurrence of extra phalanges. He also commented that a range of features is usually found in this form of brachydactyly as is the case in our reported pedigree. The spectrum of changes illustrates clearly the variable expression of this autosomal dominant condition.

The extra bone is seen to be wedge shaped, occurring at the base of the proximal phalanx. Its C shaped epiphysis is consistent with the pathology of 'longitudinally bracketed diaphysis' as described by Theander and Carstam.³ These supernumerary ossicles are usually described as fusing with the proximal phalanges but, most interestingly, in case E (fig 4E) it is the middle two phalanges of the middle fingers that appear joined.

The appearance of the thumbs is also worthy of note. While brachyphalangism is usually seen as affecting the middle phalanx of the index, middle, and little fingers, cases C, D, and E show marked shortening and irregular outline of the proximal phalanges of the thumb (fig 4A,C,E). Involvement of this digit is rare and more commonly presents with hypoplasia of the first metacarpal. The epiphysis of this bone may be absent causing shortening, and it may develop a triangular shape.

The anatomy of the big toes in all the subjects is also abnormal. Shortening of the hallux is contributed to by reduction and irregularity of both the metatarsal bone and proximal phalanx. Most interestingly in case C an extra wedge shaped phalanx has appeared in the feet as well as in the hands (figs 4A,B).

Foot deformities such as talipes valgus have been described in association with brachydactyly type C in the hand before,⁴ though Bell¹ noted that in most published cases no foot anomalies were reported. Our examples all clearly show a similar spectrum of foot involvement. Of note, malformation of the big toe associated with shortening of the fingers has also been described in fibrodysplasia ossificans progressiva.⁵

The transmission of ear changes in our subjects is very unusual. Dysmorphic facial features occurring with such hand abnormalities is extremely rare. Manzke⁶ reported the case of an 8 year old boy with the Robin sequence and type C changes but this was an isolated case. The association with ear abnormalities suggests that the bone shortening in type C brachydactyly may result primarily from a defect in chondrification, as has been shown in diastrophic dysplasia.⁷ Cartilaginous abnormality of the pinna, leading to calcification, has been seen with brachydactyly in the Keutel syndrome.⁸

In conclusion our pedigree presents a clear example of the digital manifestations found in type C brachydactyly. The similar changes in the big toes of all the subjects are of particular note and show an affinity with the changes in the fingers. The presence of cupped ears may provide evidence for the condition resulting from abnormal chondrification rather than bone matrix changes.

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