

## Case Report

# A rare case of bilateral idiopathic brachymetacarpia and brachymetatarsia

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### ABSTRACT

We present a rare case of bilateral idiopathic 4<sup>th</sup> metacarpal and metatarsal shortening. Patient was thoroughly investigated to rule out syndromic association. No intervention was done as the patient was asymptomatic.

**Keywords:** Brachydactyly, Brachymetatarsia, Brachymetacarpia, Metacarpal shortening, Metatarsa shortening

### INTRODUCTION

Brachydactyly is a disorder of the hand characterized by apparent shortening of digits.<sup>1</sup> It occurs in diverse congenital or acquired disorders, either as an isolated malformation or with other skeletal manifestations.<sup>2</sup> Brachymetatarsia and brachymetacarpia are respectively abnormal shortening of metatarsal and metacarpal bones.<sup>3,4</sup> The cause of which may be due to a syndrome, may be acquired from diseases in childhood, or may be idiopathic.<sup>4,5</sup> The deformity reportedly is caused by premature closure of the epiphyseal plate, but the etiology of which is not known.<sup>6,7</sup>

### CASE REPORT

16 years old female presented with complaint of deformity in both the hands and feet. Deformity was not present at birth; however her parents observed it during early childhood which is increasing since then. No complaint of pain or discomfort. No history of similar complaints in parents or other two siblings. Patient was examined wherein found to have bilateral shortening of 4<sup>th</sup> finger in hands (Figure 1) and 4<sup>th</sup> toe in feet (Figure 2). On clenching fist, deformity in hand becomes obvious with dimpling of dorsum of hand instead of knuckles

suggesting metacarpal shortening where right side is more affected than left (Figure 3). X ray of both the hands and feet showed shortened 4<sup>th</sup> metacarpals (Figure 4) and shortened 4<sup>th</sup> metatarsals (Figure 5) respectively. Laboratory investigations: blood count, thyroid and parathyroid profile including serum calcium and phosphorous were normal. As the patient was not symptomatic in terms of pain in hand or foot, and also was not keen about the cosmetic appearance, no intervention was done.



**Figure 1: Shortened 4<sup>th</sup> finger in both the hands.**



Figure 2: Shortened 4<sup>th</sup> toe in both the feet.



Figure 3: On clenching fist deformity becomes obvious with dimpling of dorsum of hand instead of knuckles.



Figure 4: Radiograph showing bilateral 4<sup>th</sup> metacarpal shortening (right side more than left).



Figure 5: Radiograph showing bilateral 4<sup>th</sup> metatarsal shortening.

## DISCUSSION

Brachydactyly is a hereditary disorder of the hand characterized by apparent shortening of digits.<sup>1</sup> The word brachydactyly is derived from the Greek, meaning “short fingers” which occurs in diverse congenital disorders, either as an isolated malformation or with other skeletal manifestations.<sup>2</sup> There are three types: idiopathic congenital, associated congenital, and acquired;<sup>4,5</sup> the idiopathic congenital etiology seems to be the most frequent.<sup>5</sup> Some causes of associated congenital brachymetatarsia are Down’s, Turner’s, Larsen’s, and Albright’s syndromes and diastrophic dwarfism. The acquired etiology refers to traumas, neurotrophic disorders, poliomyelitis, and surgery during growing age.<sup>8</sup>

Our patient did not have any finding other than hand and foot deformity to suggest any syndromic association. The deformity reportedly is caused by premature closure of the epiphyseal plate, but the etiology of which is not known.<sup>6,7</sup>

Brachymetatarsia is abnormal shortness of a metatarsal bone.<sup>3</sup> It is a rare deformity, with incidence ranging from 0.02% to 0.05%, with a clear female preponderance of 25:1.<sup>9</sup> Brachymetacarpia is abnormal shortness of the metacarpals.<sup>4</sup> The incidence being less than 1 in 1000 and the occurrence rate in females is 5 times greater than in males.<sup>10</sup> Occurrence of bilateral brachymetacarpia and brachymetatarsia in a single patient is extremely rare.

This deformity can be unilateral or bilateral, and any of the five metatarsals/metacarpals can be affected.<sup>5,11</sup> The deformity is seen particularly in females and usually affects the 4<sup>th</sup> metacarpal or metatarsal after the age of 4 years.<sup>11</sup> The deformity is a useful clinical marker, but is not a specific sign for particular disease.<sup>12</sup> Brachymetacarpia/brachymetatarsia usually does not impair function as in our case, and surgical treatment is usually for cosmetic reasons.<sup>4,10</sup>

## CONCLUSION

Idiopathic shortening of 4<sup>th</sup> metacarpals and metatarsals bilaterally is a rare condition. Syndromic association is possible. The condition is usually asymptomatic and does not impair function.

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