CASE REPORT

A Young Girl with Brachymesophalangia V

Waseem Mehmood Nizamani[1], Farheen Raza[2]

ABSTRACT

Brachymesophalangia is the most common hereditary anomaly of the middle phalanges. Brachymesophalangia–V (BMP-V) is the terminology used for a short and broad middle phalanx of the fifth digit which can present both alone and in a large number of complex brachydactylies with congenital disorders. Its frequency varied in different populations between 3.4% and 21%. To our knowledge no case of BMP-V has been reported from our country. We incidentally found a case of BMP-V who presented to us with the history of trauma which is presented in this case report. Literature search showed that this condition is caused by heterozygous mutations in the Indian hedgehog (IHH) gene.

KEY WORDS: *Brachydactyly; BMP-V; Phenotype; Skeletal Dysplasia; Hand.*

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[1] Waseem Mehmood Nizamani

Senior Registrar, Department of Radiology, Ziauddin University and Hospitals, Karachi.

[2] Farheen Raza

Resident, Department of Radiology, Ziauddin University and Hospitals, Karachi.

Corresponding Author

Farheen Raza

INTRODUCTION

Brachymesophalangia is the most common hereditary anomaly of the middle phalanges. Brachymesophalangia–V (BMP-V) is the terminology used for a short and broad middle phalanx of the fifth digit which can present both alone and in a large number of complex brachydactylies with congenital disorders. Despite this fact, it is not common in all populations. When this feature appears alone, it is clinically known as brachydactyly type A3 (BDA3).[1]

It is more common in the small finger seen in up to 20% of the Japanese population and about 1% of the European population. About 2/3 of patients with Down syndrome have brachymesophalangia. Brachydactylies constitute a heterogeneous group of inherited digital abnormalities classified into the five clinical types A, B, C, D and E .[2]

To our knowledge no case of BMP-V has been reported from all over Pakistan hence so far this is probably the first case report from our region.

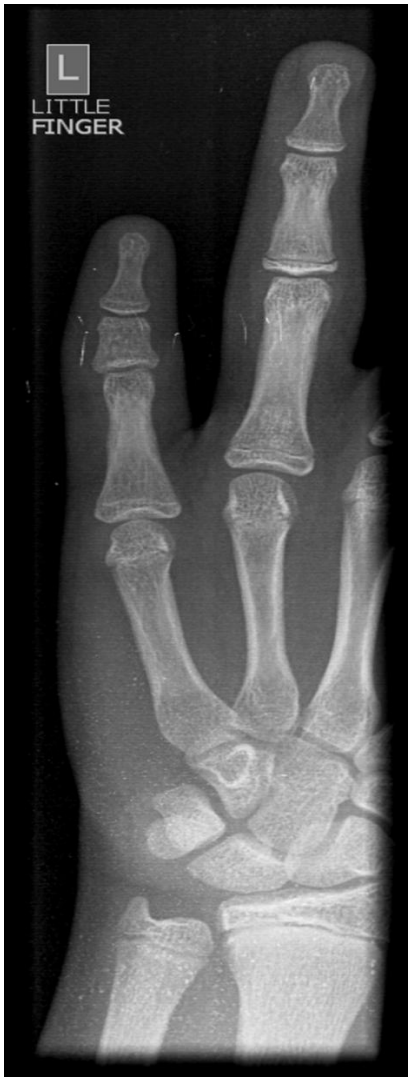
CASE

An 18 year young girl presented to radiology department with history of fall and X-rays of hand, shoulder, knee and lumbar spine were obtained for any fracture or dislocation. On examination she was quite and calm, vitals were normal. Her BMI was 20.5. No lacerations or bruises were seen on skin. She was advised skeletal X-rays by ER physician for any fracture or dislocation. The X-ray of hand showed a marked shortening of the middle phalanx and a normal distal, proximal phalanx with normal appearing metacarpals (Figure 1), no other abnormal finding was noted in other radiographs.

DISCUSSION

The review of the literature reveals plenty of research and case reports internationally but lack of research in Pakistan. Brachydactyly was the first human anomaly to be recognized as an example of Mendelian inheritance. Brachydactyly type A (BDA) is divided into the subtypes A1, A2 and A3 of which type A1 is characterized shortening confined mainly to the middle phalanges, which may be fused to the distal ones, in type A-2 only the middle phalanx of the index finger is abnormal and in type A-3 only the middle phalanx of digit 5 is abnormal.[3] Hertzog defined Brachydactyly A3 as middle phalanx V less than half the length of middle phalanx IV.[4,5]BMP-V has three variants: BMP-V alone, BMP-V with clinodactyly, and BMP-V with a cone-shaped epiphysis.[6]

Figure 1: AP radiograph of left hand 4th and 5th finger show shortening of middle phalanx of 5th digit and a normal appearing proximal distal phalanx and metacarpals.



The condition is caused by heterozygous mutations in the Indian hedgehog (IHH) gene or a yet unidentified gene on chromosome 5p13.[7] Mutations in the IHH gene have been implicated

in two human genetic diseases, brachydactyly type A1 and acrocapitofemoral dysplasia.[8] The hedgehog family of proteins is conserved in vertebrates and they have exclusive properties in the regulation of different developmental processes. The proteins act as key regulators for growth, patterning and morphogenesis of limbs during embryogenesis.[9]

The exact frequency of Brachydactyly is not known in our population because of lack of research its frequency could be between 3.4% and 21% which is a range estimated between different population of the world. With the help of research and case report we can estimate the exact frequency in our country.

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