

Case Report

Presence of Pseudoepiphyses in All the Metacarpal and Metatarsal Bones of a 9 Year-Old Girl: An Extremely Rare Case

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Abstract: Presentation of a 9 year-old girl case. An X-ray of her left hand, requested by her paediatrician to evaluate her bone age, revealed pseudoepiphyses in the non-epiphyseal ends of the five metacarpal bones. For this reason, X-rays were taken of the opposing hand and of the feet, which revealed that they had the same pseudo-epiphyseal pattern. X-rays of the rest of her skeleton did not reveal any other pathology. A clinical evaluation performed by a Geneticist did not show any syndromic association. The conclusion is that this is an extremely rare case of an isolated bone variant.

Keywords: Pseudoepiphyses, Physeal Notches, Supernumerary Epiphyses

1. Introduction

The long bones of the extremities, with the exception of the bones of hands and feet, have two epiphyseal ends. The diaphysis or shaft of a long bone begins ossification in utero from a primary ossification center, which is demonstrable in the skeleton of the newborn. During infancy, the physis or growth cartilage and the epiphysis or secondary ossification center in each end of the diaphysis can be seen Figure 1. The same does not occur in the long bones of the hands and feet: the phalanges and the metacarpal and metatarsal bones have only one epiphyseal end, which in the phalanges and in the first metacarpus and first metatarsus is found in the proximal end and in the other metacarpal and metatarsal bones in the distal end Figure 2. Bones grow lengthwise at the epiphyseal end. In the non-epiphyseal end, the bone of the diaphysis invades the hyaline cartilage until ossification is complete, a fact that was demonstrated with X-rays and MRIs by Tal Laor [1].

Occasionally, an aberrant physeal cartilage and epiphyseal ossification centers can be seen or only remnants of the aberrant physeal cartilage in the non-epiphyseal end of some of the long bones of feet and hands. This happens more frequently in the distal end of the first metacarpus and of the first metatarsus, and with less frequency in the proximal end of the second and fifth metacarpal bones. These aberrant physeal cartilages and ossification centers lead to the formation of complete and incomplete pseudoepiphyses and physeal notches.

In “complete pseudoepiphyses” [2, 3, 4] the aberrant physeal cartilage occupies the entire diameter of the bone crosswise, and separates the metaphyseal bone tissue completely from the epiphyseal ossification center, which is very rare Figure 3. In “incomplete pseudoepiphyses,” the aberrant physeal cartilage is crossed by one or more bony bridges from the metaphysis to the epiphyseal center [5], leaving two bilateral grooves as cartilage. Incomplete pseudoepiphyses are more frequent than complete epiphyses

Figure 4. In the physal notches, which are the most common, a remnant of the aberrant physal cartilage interrupts one of the sides of the bone metaphysis [7] Figure 5. These notches can be the final stage of evolution of the pseudoepiphyses, or they can appear already as a notch at a certain age and then fuse and disappear [2].



Figure 1. The long bones of the skeleton are bi-epiphyseal, they have a diaphysis (arrow head) also called shaft, originating in the primary ossification center, two physes, or physal cartilages or growth cartilages (straight arrows), and two epiphyses or secondary ossification centers (curved arrows).



Figure 3. X-ray of left hand of 7 year-old girl (case in files) shows, in the proximal end of the second metacarpal bone, a complete pseudoepiphyses (arrow). The physal cartilage crosscuts the entire diameter of the bone.

Some authors argue that the difference between the so-called complete pseudoepiphyses and incomplete pseudoepiphyses is not important, as these are only different stages of the same process [8]. The longitudinal study of X-rays of hands by Lee and Garn [2] show the evolution in the development of these epiphyses (Figure 7, male case 223). An X-ray taken at the age of 1 year shows a supernumerary epiphysis. In X-rays taken at the ages of 2, 3, and 5 years, they appear as pseudoepiphyses, and whereas at 7 and 10 years of age as notches. Lee and Garn call the complete epiphyses supernumerary epiphyses [2].



Figure 2. The long bones of hands and feet have an epiphyseal end and a non-epiphyseal end. The epiphyseal end of the phalanges and of the first metacarpal bone is proximal (arrow heads) and in the four last metacarpal bones it is distal (straight arrows).



Figure 4. X-ray of hand of a 10 year-old girl (case in files). In the proximal end of the second metacarpal bone an incomplete pseudoepiphyses can be seen (Straight arrows); the physal cartilage is crossed by a bony bridge.



Figure 5. X-ray of hand of a 4 year-old boy (case in files). In the ulnar side of the proximal end of the second metacarpal bone a physal notch can be seen.

One of the characteristics of pseudoepiphyses is that they have no influence over the longitudinal growth of the bone. Ogden [6] shows that in pseudoepiphyses, there are remnants of a physal structure, incapable of producing longitudinal growth of the bone, due to the penetration of bone tissue through the physal cartilage up to the epiphyses. In essence, this is comparable to the bony bridges created between the metaphyses and the epiphyses of children after trauma or infection, which limit the growth of the bone; however, Tjeerd [3] presents the case of two siblings where the pseudoepiphyses in the bones of hands and feet increased bone growth.

Numerous authors associate the presence of pseudoepiphyses with pathological conditions, such as Down syndrome and hypothyroidism [9], achondroplasia and non-endocrine dwarfism [10], cleidocranial dysostosis and Laurence-Moon-Biedl-Bardet syndrome [11], Wolf-Hirschhorn syndrome, [12], flat feet [13], Larsen syndrome, and Otopalatodigital syndrome [14].

The presence of pseudoepiphyses in hands and feet has been extensively reported in the medical literature in healthy children. In a longitudinal study of X-rays of the left hand of 234 healthy children, Lee and Garn [2] find incomplete pseudoepiphyses in the second metacarpus in 26% of males and in 29% of females; in the fifth metacarpus in 42% of males and in 51% of females, and complete pseudoepiphyses in the second metacarpus in 2% of children. Posoner [15] found pseudoepiphyses in 93 out of 100 healthy school children studied. In a review of 610 children, Limband-Lougenbury [4] found complete pseudoepiphyses in the first metacarpus in 1.97% of children and in the second metacarpus in 1.31%, incomplete pseudoepiphyses in 15.25% of the second metacarpus, and in 7.21% of the fifth metacarpus and 0.49% in the third metacarpus. Pizonas [13] described the presence of pseudoepiphyses in the proximal end of the second metatarsus in 6.3% of 271 children studied. In the same publication, he presented the X-ray of the feet of a

child aged 4 years and 7 months with pseudoepiphyses in the five metatarsal bones. Tjeerd [3] reports the presence of complete pseudoepiphyses in the first metacarpus, in the proximal and medial phalanges of the hands, and in the proximal phalanges of the feet, with increased growth in hands and feet in a couple of siblings. Nakashina [16] reported the casual finding of pseudoepiphyses in all the metacarpal and metatarsal bones of a healthy 7 year-old girl.

2. Presentation of the Case



Figure 6. Presentation of the case: X-ray of hands of the girl showing incomplete pseudoepiphyses in the five metacarpal bones.



Figure 7. Presentation of the case: X-rays of the feet of the girl showing incomplete pseudoepiphyses in the four last metatarsal bones and circular physal notches in the first metatarsal bones.

Case of a 9 year-old girl, product of the first pregnancy of non-consanguineous parents, with no relevant personal or family history. A paediatrician requested for X-ray of left hand to evaluate bone age, X-ray showed that the bone age was consistent with the chronological age according to Greulich

and Pyle's Atlas [17] but it also reveals pseudoepiphyses in the distal end of the first metacarpus and in the proximal end of the other metacarpal bones. Because of this finding, X-rays are made of the opposing hand and of the feet, which reveal the same pseudo-epiphyseal pattern Figures 6 and 7. A radiological study of the entire skeleton was conducted and no other abnormalities are found.

2.1. Prenatal Background

First pregnancy, normal monthly sonogram controls, amniocentesis in second quarter due to alteration in biochemical screening, with normal karyotype results: 46, XX in twenty analyzed cells. Cesarean delivery without complications, APGAR 8-9, anthropometry evaluation consistent with gestational age.

2.2. Psychomotor Development

Development milestones were achieved according to age. At present in elementary school with good academic performance.

2.3. Physical Examination

Height 117 cm (10th percentile); weight 21kg (15th percentile); arm span 122 cm; arm span/height ratio 1.04; upper segment 59 cm; lower segment 58 cm; upper segment/lower segment ratio 1; cephalic perimeter 50 cm (25th percentile).

Normocephalic cranium, non-dysmorphic facial cranial region with no evident alterations, average forehead, horizontal palpebral fissures, average nasal bridge and root, mouth with no evident alterations, normal palate, pinna in normal position, normal neck, thorax and abdomen with no evident alterations, thoracic, dorsal and lumbar regions with no evident alterations. Symmetrical upper extremities, with no shortening, normotrophic, adequately formed hands and phalanges, no alterations in flexion creases, ratio of length of middle finger to palm bilaterally preserved, no hyper-mobility of joints reported. Symmetrical lower extremities with no shortening, normotrophic, feet and phalanges with no evident alterations. Female genitals consistent with age. Normoreflexia.

Beighton's criteria for hyper-mobility of joints 2/8.

3. Discussion

The 9 years old girl presents pseudoepiphyses in the non-epiphyseal ends of all the metacarpal and metatarsal bones. This bone alteration could correspond to a syndromic entity or a developmental variant. The Genetic evaluation ruled out cleidocranial dysplasia, Larsen syndrome, Otopalatodigital syndrome, Dyggve-Melchior-Clausen Dysplasia and pseudo-hypo-parathyroidism. It is believed that the case corresponds to an isolated bone variant.

Although it is true that the isolated finding of pseudoepiphyses in metacarpal and metatarsal bones is frequent in healthy children [2, 4, 13, 16], finding

pseudoepiphyses in all the metacarpal and metatarsal bones is so infrequent that only one case has been found reported in medical literature. Nakashima [15] reported the case of a 7 year-old girl who presented pain in one of her feet. The X-ray revealed complete pseudoepiphyses in all the metatarsal bones, the study was completed with X-rays of the opposing foot and hands, finding pseudoepiphyses in all the metacarpal and metatarsal bones. This girl, like our patient, had no other developmental anomalies, and no syndromic entity.

Numerous works link the presence of pseudoepiphyses with pathological conditions [9, 10, 11, 12, 13, 14], but finding pseudoepiphyses in the bones of hands and feet of healthy children is frequent [2, 4, 15, 13, 16]. The case reported here falls in this group of healthy children.

Nakashima describes complete pseudoepiphyses in the metacarpal and metatarsal bones. The case submitted by us has incomplete pseudoepiphyses in the four last metacarpal and metatarsal bones, and physeal notches in the first metacarpal and first metatarsal bones. The girl presented is one year older than the girl in Nakashima's case, and it was not possible to ascertain whether she had a pattern similar to Nakashima's one year earlier. According to Lechman, these variations are different stages of the same ossification process [8].

4. Conclusions

The presence of pseudoepiphyses in all the metacarpal and metatarsal bones is extremely rare. There is only one other reported case in medical literature. Both cases are anomalies in healthy girls.

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