Bilateral Second Carpal Row Duplication Associated with Multiple Epiphyseal Dysplasia

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Abstract

We report a case of a 75-year-old woman presenting a hitherto undescribed condition of bilateral second carpal row duplication. She was diagnosed in childhood with both Marfan and Ehlers-Danlos syndromes, with no clear evidence and no further medical follow-up. She presented throughout her life with various articular symptoms, which appeared to be compatible with a diagnosis of multiple epiphyseal dysplasia, and underwent several surgical procedures on her knees and hips. Most recently, she was reporting pain at the base of the fifth metacarpal bone of the left hand. X-ray images and computed tomography (CT) were obtained for exploration and showed a total second row duplication in both carpi, with a total number of 18 carpal bones in each wrist.

Keywords

- carpal duplication
- epiphyseal dysplasia
- carpal malformation

The number of carpal bones may be increased in an individual as a result of anatomical variants or true congenital anomalies. Numerical increment can be due to additional bones or from split bones. Over 20 different accessory carpal ossicles have been described.¹ The presence of these additional bones usually results from a failure of fusion of their ossification centers. Splitting can also occur: the scaphoid, the lunate, the hamulus of the hamate, and less frequently the capitate and trapezium are known to be sometimes bipartite.¹ However, these anomalies have always been described either in isolation or as part of a wider syndromic condition. No case of bilateral total second carpal row duplication has ever been reported, whether associated with other general conditions or not.

Marfan syndrome and Ehlers-Danlos syndrome are two hereditary disorders involving collagen synthesis. These two conditions are known to provide ligamentous hyperlaxity and joint hypermobility, associated with wrist complaints in 70% of patients suffering from Ehlers-Danlos syndrome.² However, no osseous malformation has so far been reported in these patients.

Multiple epiphyseal dysplasia is a rare condition involving early-onset osteoarthritis (mainly involving hips and knees) and abnormally small and/or irregular epiphyses, causing a secondary metaphyseal flaring. This disease is clinically and genetically heterogeneous; six genes are known to be associated with this phenotype.^{3,4}

Case Report

A 75-year-old woman first presented with pain in the left wrist. Clinically, the pain was electively located at the base of the fifth metacarpal of the left hand, on its dorsal side. The pain was mechanical, slowly progressive over a few months, and of low intensity. No traumatism had been reported for this pain, and the wrist showed no sign of inflammation. The wrist mobility was subnormal, with 80° of extension and 90° of palmar flexion on both sides. No hyperlaxity was noted.

Wrist examination showed a Kapandji score of 10/10 on both sides, a K-pinch test of 4.5 kgf on the right side and 3.5 kgf on the left side, and Jamar test of 15 kgf on the right side and 14 kgf on the left side.

A standard X-ray exploration (➤Fig. 1) first revealed an abnormal number of carpal bones, which prompted a computed tomography (CT) scan. Global shape of the wrist anatomy was respected. The bilateral wrist CT scan (➤Fig. 2) showed a total second row duplication. In the left wrist, the trapezium and the trapezoid were each divided in four fragments of regular shape with symmetry in both frontal and sagittal planes. The capitate and the hamate were each divided in three regular fragments, with a sagittal alignment. No anatomical anomalies were noted regarding the first row of the carpus. The total count of carpal bones in

Fig. 1 (a–d) Standard X-ray images of both wrists.

the left wrist was 18. The anomalies were identical in the right wrist.

The CT scan also showed signs of bilateral chondrocalcinosis, with scapholunate dislocation on the left side, compatible with a scaphoid chondrocalcinosis advanced collapse (SCAC) wrist. The base of the left fifth metacarpal, which was the only initial complaint of our patient, was articulated with all three hamate fragments and presented degenerative arthritic changes.

No specific treatment has so far been undertaken, since our patient has low physical activity and is unwilling to undergo any kind of surgery. The pain can be controlled with on-demand oral painkillers.

In her medical history, the patient reported a diagnosis of both Marfan and Ehlers-Danlos syndromes in childhood with

no clear medical evidence, for which she was treated during a few years in Switzerland between the ages of 2 to 14 years, with no further medical investigation or follow-up ever since.

Further questioning of the patient revealed several ancient articular pains, responsible for bilateral total knee arthroplasty (TKA) about 30 years previously and left-side total hip arthroplasty at the same period. Old preoperative knee and hip X-ray images showed an early-onset severe osteoarthritis with irregular epiphysis (►Fig. 3).

The patient also complained of pain in both elbows. Standard radiographs showed irregular epiphysis with chronic radial head luxation on the left side. The mobility of both elbows was normal with minor pain. Examination showed a cubitus varus of 45° on the left side (►Fig. 4).

Fig. 2 (a) Axial view at the level of the hamate's hook. (b) Axial view at the level of the base of M1. M1, first metacarpal; TZ, trapezium; TO, trapezoid; S, scaphoid; C, capitate; H, hamate; T, triquetrum; M4, fourth metacarpal; M5, fifth metacarpal.

Finally, she complained of mechanical pain in both feet. Imaging showed tarsal duplication: each foot had six cuneiform bones, with an axial symmetry, as in the carpus (► Fig. 5).

These different clinical features, along with the standards radiographs, are compatible with the diagnosis of multiple epiphyseal dysplasia. We believe the patient was misdiagnosed in childhood and actually never had either Marfan or Ehlers-Danlos syndrome.

Discussion

In the embryo, bone, cartilage, and tendon of the upper limb come from a lateral plate mesoderm, forming the limb bud with the mesoderm (which will form muscles, nerves, and vessels) and the overlying ectoderm.⁵ The limb bud appears 26 days after fertilization, when the embryo is only 4 mm long. Cartilage precursor cells then accumulate in the center of the bud and differentiate into either cartilage-forming chondrocytes (for endochondral skeletal elements) or bone-forming osteoblasts (for membranous skeletal elements). Carpal bones are endochondral elements. Development of joints occurs by repression of chondrogenesis at the site of future joints. All chondrification centers and joint interzones are formed within the first 50 days after fertilization.

Fig. 3 (a,b) Standard X-ray images of knees (1980). The patient now has TKA.

The molecular mechanisms that orchestrate limb development are various, and the pathogenesis of limb anomalies is complex.^{6,7} Three signaling centers control development: the apical ectodermal ridge (AER) regulates proximodistal development, the zone of polarizing activity (ZPA) regulates radioulnar development, and an ectodermal signaling center regulates dorsoventral development.

The anomalies we observed in this patient present all the characteristics of bipartite bones, caused by a lack of fusion of the initial ossification centers, as it has been defined in other articles,¹ or in our case, quadripartite or tripartite bones. Especially, the wrist CT scan clearly shows a lack of fusion between the body of the hamate and its hook. Accessory bones present different features: smaller size than the normal carpal bones, no symmetry, and location between several normal bones. In our case, all fragments of each duplicated bone are about the same size, with a regular shape, a symmetrical arrangement, and fine cortical bone at the periphery.

No evidence can be found that the bilateral carpal duplication is related to either multiple epiphyseal dysplasia or

Fig. 4 Standard X-ray images of elbows. (a,b) Top. (c,d) Bottom.

Marfan or Ehlers-Danlos syndrome, since we found no published article suggesting a correlation could exist.

Some syndromes are known to produce carpal malformations,⁸ such as hand-foot-uterus syndrome, Holt-Oram syndrome, oto-palato-digital syndrome, Larsen syndrome, and Gorlin-Schlörff-Paparelle syndrome. These syndromes are associated with the presence of the *os centrale*—an extra ossicle located between the scaphoid, capitate, and trapezoid—but not with carpal duplication.

Accessory carpal bones of the distal row can be found in brachydactyly A-1, diastrophic dysplasia, Ellis-Van Creveld syndrome, Larsen syndrome, and oto-palato-digital syndrome. However these syndromes usually present many associated anomalies, such as expansion of the metacarpals and proximal phalanges and short distal phalanges in Larsen syndrome, or fusion and malpositioning of carpal bones, elongation of the ulnar side of the second metacarpal bone, and short distal phalanges in oto-palato-digital syndrome.

Fig. 5 X-ray and CT images of ankles and medial foot. (a,b) Top. (c,d) Bottom. C1, first cuneiform bone; C2, second cuneiform bone; C3, third cuneiform bone; M5, base of the fifth metatarsal bone; Cub, cuboid bone.

Conflict of Interest

None

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