Fracture of the Proximal Pole of the Bipartite Carpal Scaphoid: A Probable Holt–Oram-like Syndrome

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Abstract Bipartite scaphoid has been debated, and is of a traumatic origin for some authors and congenital for others. The aim of study is to distinguish clinical, radiographic and MR criteria for the diagnosis of congenital bipartite scaphoid. I describe a case of a patient with bilateral bipartite scaphoid admitted at my clinic following a traumatic accident which produced a fracture of the proximal pole of the right carpal scaphoid. The patient had a history of bipartite scaphoid. In conclusion, I believe that congenital origin is more probable and this rare pathology is a sporadic form of Holt–Oram-like syndrome.

Keywords The bipartite carpal scaphoid · The proximal pole · Fracture · Holt–Oram-like syndrome

Introduction

Bipartite scaphoid is a congenital anomaly of the carpal scaphoid which has already been described in literature. It is a rare deformity, with an aetiology that is still under discussion. There are two main hypotheses: congenital, due to an independent ossification of two separate nuclei [7, 11] and traumatic, following unidentified fracture of the scaphoid during childhood [9].

I report a case of a patient with bilateral bipartite scaphoid who was admitted to our Clinical Unit following a traumatic accident which caused a fracture of the proximal pole of the right carpal scaphoid.

Case Report

A 30-year-old right handed male motorcyclist and volleyball player was involved in an accident in which his motorcycle collided with the back of a stopping car while the motorcyclist was grasping the handlebars. The patient landed on his right hand with the wrist in extension and radial deviation.

He presented at our clinic 1 h after the injury with pain and swelling of the anatomical snuffbox in his right hand. The trauma, as described by the patient, did not seem particularly severe. Axial compression of the thumb, which compressed the scaphoid, elicited pain. The patient reported discomfort just with percussion of the tip of abducted thumb. Forced ulnar deviation of pronated wrist elicited pain. Hypoplasia of the thumb and ulnar deviation of the metacarpals was also clinically observed. A radiographic examination of the right wrist (Fig. 1), showed a fracture of the proximal pole of the scaphoid with a suspect bipartitism.

Radiographic characteristics of the fracture were irregular. Seeing the particular morphotypic features of the hands of the patient, we decided to look for the presence of a possible associated contralateral anomaly.

The contralateral radiographs of the left wrist (Fig. 2) confirmed the diagnostic hypothesis of a bilateral bipartite scaphoid.

The proximal pole fracture had a displacement of >1 mm and was considered unstable. Fracture had an interscapoid angle of >35°, a radiolunate angle of >15° or a scapholunate angle of >60°. I immobilized the proximal pole fracture in a long arm thumb-spica cast for 12 weeks.

The patient's medical history did not show any hereditary metabolic, endocrinologic, orthopaedic diseases or cardiac abnormalities. Following an accurate musculoske-

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Figure 1 Anteroposterior and lateral radiographs of the right wrist demonstrated a fracture of proximal pole of scaphoid with a suspect bipartitism.

letal examination, other anomalies were not present. Relevant data were: a fracture of the left wrist which had occurred at the age of 16, ineligibility for military service at the age of 18 due to the morphotype of his hands, the presence of a right inguinal hernia and a surgical intervention for phimosis at the age of 2. The patient was first seen at our hospital at the age of 3 years. At that time, he had sustained a superficial laceration of the base of the right

thumb. The diagnosis was made on the basis of clinical findings consisting with characteristics hand deformities. The right hand had a hypoplastic thumb and mild radial deformity. At same year of the age, the laceration healed uneventfully and patient was asymptomatic until he sustained a minor fall onto the right forearm and complained of mild right wrist and elbow pain. X-ray film of the right wrist demonstrated two separate centers of the scaphoid (Fig. 3).

At 16 years of age, the patient was seen for another control. He did not present any pain in his right hand. There was the absence of history of trauma. The total arc of motion for flexion and extension of wrist was 80°. The radiograph confirmed that the scaphoid was ossified in two portions (Fig. 4).

After 12 weeks, on the removal of the cast, clinical outcome was characterised by a slight pain at the anatomical snuffbox. The total arc of motion of his wrist was 75°.

I carried out a Magnetic Resonance Imaging examination at 12 weeks. This demonstrated the presence of two ossific centers in the distal part of scaphoid with signs of initial osteonecrosis of the proximal pole (Fig. 5). Treatment options were discussed with the patient, and his preference was to use a brace for support during heavy activities or sporting activities; he declined surgical intervention.

After 6 months, the patient was seen for another control. He did not show any pain on both hands and the total range of motion of wrists is optimal. MRI of right hand with use of gadolinium was interrupted because the patient was allergic to gadolinium. Subsequently, he declined MRI of both hands.

Figure 2 Radiographs of the left wrist demonstrated that the scaphoid was ossified in two portions.

Figure 3 Radiograph of bipartite scaphoid (right hand) obtained at age 3.

Discussion

It is evident that in medical literature bipartite scaphoid is extremely rare and is a controversial topic [7]. In reference to studies carried out by Pfitzner [11], bipartite scaphoid was of congenital origin, due to an independent ossification of two separated nuclei. Caffaratti [4] sustained that bipartite scaphoid was not caused by trauma. He demonstrated the bilaterality of this rare anomaly. According to Bunnell [3], the following factors were considered to be supportive of the diagnosis of congenital bipartite scaphoid: the absence of history of trauma, the presence of bilateral scaphoid bipartition, equal size and density of both ossicles, the absence of degenerative changes in the radial scaphoid carpal articulation and a clear space between the components with smooth edges at the joint surfaces. Louis et al. [9], however, in a study of 196 human fetuses were unable to find any example to support this theory of congenital bipartition. These same authors reviewed 5,365 radiograph studies of children between the ages of 4 1/2 and 12 1/2 years and found three examples between the ages of 7 and 8 years (all boys) of multiple ossification centers of the scaphoid. After reviewing an additional 11,280 X-ray

Figure 4 Radiograph of bipartite scaphoid (right wrist) obtained at age 16.

Figure 5 MRI of the right wrist after 12 weeks. The presence of two ossific nuclei for scaphoid (note the high intensity signal between the bipartite scaphoid indicative of bony tissue rather than fibrous tissue in distal part of scaphoid), with signs of initial osteonecrosis of the proximal pole (part), was confirmed.

studies of individuals more than 12 years of age and finding not a single case of bipartite scaphoid, they concluded that multiple ossification centers occasionally occur in the scaphoid, but these always coalesced to form the adult scaphoid. They did not, however, have the opportunity to follow up the cases that had multiple ossification centers and their conclusions were ones of statistical probability, not documentation. On the basis of this finding, they stated that with the exception of certain malformation syndromes, the so-called congenital bipartite scaphoid was, in actuality, a traumatically acquired condition.

There are numerous elements which suggest that it could be a congenital pathology, and among others, I cite the work of Doman and Marcus [7] who documented a single case of an apparent congenital bipartite scaphoid in a woman followed-up from childhood to young adulthood with serial radiographs and a MR. I also used serial radiographs and a MR to make the diagnosis of bipartite scaphoid. I sustain that the patient I examined, satisfies all the criteria suggested by Doman and Marcus and by Pfitzner, Caffaratti and Bunnell as well. Therefore, the patient can ultimately be diagnosed of congenital bilateral bipartite carpal scaphoid.

The extreme rarity of this pathology has led us to carry out a follow up of the patient in order to evaluate its progress and eventually understand if bilateral bipartite scaphoid is an isolated anatomical anomaly, rather than an element of a syndrome. There is a syndrome described in the literature which might be associated with a congenital bipartite scaphoid. Tate et al. [8] described a bipartite

scaphoid in a patient with Holt–Oram syndrome and demonstrated scaphoid ossification centers located in right wrist. Holt–Oram syndrome was first described by two British cardiologists in 1960 (Holt and Oram, 1960). Modes of inheritance is autosomal dominant with variable expressivity; penetrance of 100%; patients with skeletal defect alone can transmit full clinical manifestations of syndrome (cardiac and limb defects); sporadic in about 40% of affected individuals; transmission through an “unaffected parent” (Brault et al. 1991); locus 12q24; mutations in TBX5 gene (Huang, 2002) [8].

Clinical manifestations are: (a) congenital cardiovascular diseases: atrial septal defect (60%) (ostium secundum type the most common congenital heart defect); ventricular septal defect; hypoplastic left heart syndrome; hypoplastic peripheral vasculature; patent ductus arteriosus (PDA); (Sletten et al. 1996); cardiac conduction defects (Ogur et al. 1998); persistent left SVC (Cheng, 2000); (b) fingerlike or absent thumb, thenar abnormalities, other hypoplastic muscles (Spranger et al. 1997); (c) other reported abnormalities: no specific facial dysmorphism (Allanson et al. 2003); skin tag over the lower part of the sternum; deficiency of immunoglobulins and impaired cellular immunity; association with malignancy; aplastic anemia; severe myopia; spotty melanotic pigmentation of the skin; deletion of 14q; limitation of motion of the shoulder and elbow; narrow shoulder; postaxial and central polydactyly as variable manifestations in a four-generation family (Moens et al. 1993) [8].

Radiologic manifestations are: (a) hand anomalies (digitalized anomalies; digitalized; absent; hypoplastic; triphalangeal; bifid); first metacarpal anomalies (e.g., absent; hypoplastic, abnormal plane); finger anomalies (clinodactyly; syndactyly; absent; hypoplastic); second to fifth metacarpal anomalies; (b) carpal bone anomalies: delayed ossification/absence; hypoplastic; fused, irregular; extracarpal/os centrale; enlarged; bipartite (DeSmet 2002; Tate et al. 2000); (c) anomalies of forearm: hypoplasia or absence of the radius; anomalous of radial head, hypoplasia, absence of ulna, radio-ulnar synostosis, humero-ulnar synostosis; (d) anomalies of the humerus; hypoplasia or absence; (e) shoulder anomalies: hypoplasia of the clavicle, broadened clavicle; deformed humeral head; prominent coracoclavicular joint; rotated and small scapula; hypoplastic glenoid fossa; Sprengel deformity; accessory bones; (f) chest wall anomalies; deficiency of pectoral muscles; pectum excavatum; pectum carinatum; rib anomalies: scoliosis; fusion; hemivertebrae; (g) prenatal diagnosis: limb, heart (Tongsong et al. 2000) [8]. This syndrome is caused by mutations in TBX5 gene which can produce a wide range of disease phenotypes characteristic of the Holt–Oram syndrome. The TBX5 gene has an important role in both skeletal and cardiac development [1, 2].

Concerning my case, I noted the bilaterality of this congenital anomaly. There was no history of congenital anomalies on either side of the family. Atrial and ventricular septal defects, phocomelia and other carpal abnormalities were not present. The presence of scaphoid ossification centers located in right hand at the age of 3 years and the presence of a bilateral hypoplasia of the thumb satisfy some diagnostic criteria of this syndrome. Subsequently, I demonstrated that Holt–Oram syndrome is a genetic disorder inherited as an autosomal dominant trait highly penetrant and is caused by a mutation of 7-intron sequence in the transcription factor TBX5, a member of the T-box gene family, localized on the long arm of the chromosome 12. In conclusion, I supported the hypothesis of Debeer et al. [6]. I believe that bipartite scaphoid associated to Holt–Oram syndrome can be part of the spectrum of the radial longitudinal deficiency. This is the very premise and case description by Tate et al. in 2000 [12]. Also, Manske and others have written several extensive reviews of radial longitudinal deficiency and included in their analysis bipartite scaphoid and Holt–Oram syndrome [5, 10]. Phylogenetically, two carpal ossification centers, comparable in site to the carpal scaphoid can be found in certain primitive reptiles. In mammalian development, however, this appears as single centers. It is possible that the bipartition may arise from heterogenous ossification centers. The bipartite fragments are not equal in size and do not remain in the normal anatomical position and ulnar negative variant is transformed in ulnar positive variant during development. The case, in particular, lends support to possibility of heterogenous ossification centers which may be phylogenetic, neogenetic or anomalous in origin. In conclusion, I believe that bipartite scaphoid is not an isolated anatomical anomaly but is a sporadic form of Holt–Oram-like syndrome.

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