CONGENITAL HYPOPLASIA OF THE CARPAL SCAPHOID BONE

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Three cases of absence of the carpal scaphoid bone have previously been reported in this country (Hodgson 1943, Hanley and Conlon 1957). All three had similar deformities, namely absence or hypoplasia of the carpal scaphoid bone, associated with retardation in the development of the first metacarpal bone, the styloid process of the radius and thenar muscles.

While agreeing with these authors that their cases are examples of a rare congenital lesion, I think that they must be considered as only one form of a group of lesions which are not so rare as has been previously supposed. These lesions consist in a congenital hypoplasia of the lower end of the radius and a hypoplasia of the muscles of the radial aspect of the forearm, and in a congenital hypoplasia of the carpal scaphoid bone, associated with abnormalities of the thumb and of the thenar muscles. The form of presentation extends from absence of the thumb to a hypoplasia of the first metacarpal bone, styloid process of the radius, the thenar muscles and the muscles of the radial aspect of the forearm. The recognition of these lesions is of importance from the point of view of reconstructive surgery.

In America in 1958 Barsky described the gradations in which ectrodactyly of the thumb may present, and said that they ranged from complete absence of the thumb ray to absence of only the distal phalanx. He did not mention in any of his patients the absence or hypoplasia of the carpal scaphoid bone, though several of his radiographs did in fact demonstrate this absence.

CLASSIFICATION

The gradations by which hypoplasia of the carpal scaphoid bone and its associated abnormalities may present are broadly as follows.

Type 1—A short thumb of atrophic appearance associated with hypoplasia of the thenar muscles. The adductor of the thumb and the long flexor and long extensor are present. The absence or hypoplasia of the carpal scaphoid bone causes a radial displacement of the carpus and accentuates the hypoplasia of the musculature of the radial aspect of the forearm.

The bony anomalies consist of the absence or hypoplasia of the carpal scaphoid bone and a poorly developed first metacarpal bone lacking the tubercle for the insertion of the abductor pollicis brevis and having a rounded base instead of the usual saddle-shaped articular surface. There is absence of the radial styloid process, and hypoplasia of the lower end of the radius. The first metacarpo-phalangeal joint has only a single sesamoid bone.

Type 2—The thumb is small and its position more proximal than normal; it has two phalanges and a metacarpal. The scaphoid bone is absent or hypoplastic and there may be absence of the trapezium. The lower end of the radius is hypoplastic. The muscular attachments of the thumb are almost completely deficient and it cannot be placed in the position of opposition.

Type 3—The "thumb" is, in effect, a finger; its metacarpal resembles that of a finger and there are three phalanges. The long extensor and long flexor are present; there is no thenar eminence, and the thumb cannot be opposed. The scaphoid bone is absent or hypoplastic, and there may be absence of the trapezium. There is hypoplasia of the lower end of the radius.

Type 4—The “floating thumb”: the thumb is small and is situated more distally than normal. The long extensor and long flexor of the thumb and the thenar muscles are absent. The scaphoid bone is absent or hypoplastic, and there may be absence of the trapezium. There is hypoplasia of the lower end of the radius.

Type 5—The thumb is represented by a rudimentary nodule on the radial side of the index finger. There is no thenar eminence, and the thumb has no muscular attachment. The skeletal elements of the first ray are absent and the scaphoid bone is absent or hypoplastic. There is marked hypoplasia of the lower end of the radius.

Type 6—The four-fingered hand: there are four normal fingers but no thumb or thenar eminence. The skeletal elements of the first ray are absent and the scaphoid bone is absent or hypoplastic. There is marked hypoplasia of the lower end of the radius.

CASE REPORTS

Thirteen patients demonstrate the six modes of presentation described.

Type 1—There were six examples of this type.

Case 1—A man aged forty-three attended the casualty department with minor bruises and abrasions following a fall from his motor cycle. On examination it was observed that there was radial deviation of the hands and atrophy of the muscles on the radial aspect of the forearms and of the thenar eminences. Both thumbs were smaller than normal and were tapered. The deformity was worse on

FIG. 1

FIG. 2

Case 1—Type 1 deformity.

the right (Fig. 1). Both the long flexor, the long extensor and the adductor of the thumb were present and of normal strength on both sides; the abductor pollicis brevis was absent and the opponens and flexor pollicis brevis were weak and gave a poor response to faradism. The radial pulse was absent. In spite of these deformities and muscular deficiencies, the patient managed to do heavy work.

Radiological examination showed absence of the scaphoid bone in the right wrist and a hypoplastic scaphoid bone in the left (Fig. 2). Both hands showed a poorly developed first metacarpal with a rounded base and with absence of the tubercle for the insertion of the abductor pollicis brevis. Both had only a single sesamoid bone at the first metacarpophalangeal joint, and on both sides the radial styloid process was absent. The terminal phalanx of the right index showed an old ununited fracture.

Other congenital abnormalities present in this patient were bilateral cervical ribs, a smaller clavicle on the right than on the left, and fusion of the bodies of the second and third cervical vertebrae.

FIG. 3

FIG. 4

Case 2—Type 1 deformity.

Case 2—A boy aged thirteen years was first seen at the age of six with radial deviation of the left hand and a rudimentary left thumb, which was atrophic in appearance and smaller than that on the right. There was atrophy of the muscles of the thenar eminence and slight atrophy of the musculature of the radial aspect of the forearm. The long flexor and long extensor of the thumb were present, though the power of flexion was weak. The child could not oppose the thumb nor abduct it, but adduction was normal. Flexion of the metacarpo-phalangeal joint by the flexor pollicis brevis was absent, and the metacarpo-phalangeal joint showed marked laxity.

When re-examined at the age of thirteen years it was found that he had a similar condition in the right hand. Figure 3 shows the two hands.

In the right hand there was wasting of the thenar muscles and the thumb was a little tapered. The long flexor and long extensor of the thumb were present, and the muscles of the thenar eminence were all present in spite of the wasting of the eminence. There was also slight wasting of the muscles of the radial aspect of the forearm, but there was no obvious radial deviation of the carpus.

Radiological examination of the left hand (Fig. 4) showed absence of the scaphoid bone. The first metacarpal bone and phalanges of the thumb were small and hypoplastic. There was no epiphysis at the proximal end of the first metacarpal. The changes in the metacarpo-phalangeal joint were caused by a previous operation. The lower end of the radius was hypoplastic.

The right hand showed a scaphoid bone which was hypoplastic in that it was slimmer and smaller than normal. The first metacarpal bone and phalanges of the thumb were normal in appearance, as was the lower end of the radius (Fig. 4).

FIG. 5

Case 3—Type 1 deformity.

Case 3—A girl aged nine years had, on the left side, a small and hypoplastic thumb with hypoplasia of the thenar eminence and absence of the thenar muscles, with the exception of adductor pollicis. The long extensor and the long flexor of the thumb were present but they flexed and extended the metacarpo-phalangeal joint and not the interphalangeal joint. There was radial deviation of the wrist and hand.

Radiological examination of the left hand showed the absence of the scaphoid bone; the first metacarpal bone was poorly developed and had an epiphysis at its distal end but no epiphysis at its proximal end. There was hypoplasia of the radial aspect of the lower end of the radius and absence of the trapezium (Fig. 5).

Case 4—A boy aged eight years was seen with congenital deformities of both hands and a congenital scoliosis with

wedging of the first thoracic vertebra and a second thoracic hemivertebra. The first and second ribs were fused on both sides.

Examination of the right hand showed slight hypoplasia of the radial aspect of the forearm with no obvious radial deviation of the hand. There was a hypoplastic tapering thumb with hypoplasia of the musculature of the thenar eminence. The long extensor and long flexor of the thumb were both present; there was absence of the abductor pollicis brevis and opponens pollicis.

Radiological examination of the right hand showed absence of the ossific centres for the scaphoid bone and trapezium; the first metacarpal bone was shorter than normal and had no epiphysis at the proximal end. The phalanges were hypoplastic, and there was hypoplasia of the lower end of the radius (Fig. 6).

FIG. 6

Case 4—Type 1 deformity.

Case 5—A girl aged five years, first seen two years earlier, had hypoplastic thumbs and hypoplasia of the thenar eminence on both sides. The right thumb had a lax metacarpophalangeal joint. The flexor pollicis longus and extensor pollicis longus were both present as was adductor pollicis. The opponens pollicis was absent. There was radial deviation of the carpus and hand and slight hypoplasia of the muscles of the radial aspect of the forearm.

The left thumb had a lax metacarpophalangeal joint. The extensor pollicis longus and flexor pollicis longus were both present. The adductor pollicis was present but the opponens pollicis was absent. There was hypoplasia of the muscles of the radial aspect of the forearm and radial deviation of the wrist and hand (Fig. 7).

Radiological examination showed absence of the ossific centres for the scaphoid bones, trapezoid bones and trapezia (Fig. 8). The first metacarpal bones were both hypoplastic and were without an epiphysis at the proximal end. They appeared to have had an epiphysis at the distal end. There was hypoplasia of the lower end of the radius on both sides. The right hand showed an operation scar.

FIG. 7

FIG. 8

Case 5—Type 1 deformity.

Case 6—A girl, aged twelve years, was first seen at the age of ten with a history of scoliosis since the age of eighteen months. Examination showed a congenital flexion contracture of the right elbow in addition to the scoliosis. The left hand showed webbing between the little and ring fingers and hypoplasia of the thumb. Examination of the left hand and forearm at the age of twelve showed

FIG. 9

FIG. 10

Case 6—Type 1 deformity.

that all the muscles of the forearm were less well developed than on the right and there was hypoplasia of the thumb and of the thenar muscles. There was slight radial deviation of the wrist and hand. The long extensor and the long flexor of the thumb were both present (Fig. 9).

Radiological examination showed hypoplasia of the lower end of the radius, hypoplasia of the scaphoid bone, and hypoplasia of the skeletal elements of the thumb. There was only a single sesamoid bone at the first metacarpo-phalangeal joint. In addition, there was fusion of the fourth and fifth metacarpal bones and fusion of the lunate and triquetral bones. There was radial deviation of the wrist and hand (Fig. 10).

Type 2—There were two examples.

Case 7—A boy aged five years was first seen as an infant of one month with a rudimentary right thumb and radial deviation of the hand. The thumb was small and atrophic, but had two phalanges and a hypoplastic metacarpal bone. There were no thenar muscles, nor was there a long extensor or a long flexor to the thumb.

The thumb was not used by the child, and as he grew it became obvious that the thumb was useless; he used the index finger instead.

Radiological examination showed absence of the ossific centres for the scaphoid bone and trapezium, and hypoplasia of the first metacarpal bone and phalanges of the thumb. The first metacarpal bone had an epiphysis at its distal end and no epiphysis at its proximal end. There was hypoplasia of the lower end of the radius (Fig. 11). The child also had bilateral talipes calcaneus.

FIG. 11

Case 7—Type 2 deformity.

Case 8—A girl aged six years was first seen because of deformities of both hands. She also had a spastic gait and walked on a wide base. On examination of the right hand it was seen that there were four normal fingers. The thumb was small and hypoplastic; there was hypoplasia of the thenar eminence and hypoplasia of the musculature of the radial aspect of the forearm with radial deviation of the carpus and hand. There was a skin shortage holding the interphalangeal joint of the thumb in a 30-degree contracture with some radial swing. There was no long extensor or long flexor of the thumb (Fig. 12).

FIG. 12

FIG. 13

Case 8—Type 2 deformity.

Radiological examination of the right hand showed absence of the ossific centres for the scaphoid and trapezium bones and the trapezium. The first metacarpal bone was small and hypoplastic and its phalanges were smaller than normal. There was slight hypoplasia of the radial aspect of the lower radial epiphysis (Fig. 13).

Type 3—There were two examples.

Case 9—A girl aged fourteen years was first seen as an infant with congenital deformities of the hands and double big toes. In the right hand the thumb was very small and atrophic (Fig. 14); it had three phalanges, and there was a flexion contracture at the proximal interphalangeal joint. The long flexor and the long extensor of the thumb were both present but had no action on the thumb distal to the proximal interphalangeal joint, but flexed and extended the thumb at the metacarpophalangeal joint. The thenar eminence was absent, and movements of adduction, abduction and opposition were absent. There was wasting of the muscles of the radial aspect of the forearm and the hand was deviated radially.

In the left hand the thumb was longer than normal (Fig. 14). It was tapered, and it had three phalanges and a metacarpal. The long flexor and long extensor to the thumb were present. There was marked wasting of the thenar eminence. Adduction was present but opposition was effected to a small extent only by a trick movement of flexion. There was wasting of the musculature of the radial aspect of the forearm and radial deviation of the hand.

Radiological examination of the right hand showed absence of the scaphoid bone and trapezium. The first metacarpal bone was long, slim and hypoplastic, as were its three phalanges. The first metacarpal had an epiphysis at both its proximal and distal ends. There was hypoplasia of the lower end of the radius (Fig. 15).

In the left hand the carpal scaphoid bone was absent. The other bones of the first ray did not show the hypoplasia that was shown on the right side. The first metacarpal bone had an epiphysis at both ends. The lower end of the radius was hypoplastic (Fig. 15).

FIG. 14
Case 9—Type 3 deformity.

FIG. 15
Case 9—Type 3 deformity.

FIG. 16

FIG. 17

Case 10—Type 3 deformity.

Case 10—A woman aged thirty-two years was the mother of the previous patient (Case 9) and she said that her father's hands were the same as those of her daughter. At the age of eleven she had had an additional thumb removed from the right hand. This thumb was situated at the level of the head of the metacarpal bone of the true thumb.

Examination of the left hand (Fig. 16) showed that the thumb was long and tapered and had three phalanges. The long extensor and long flexor of the thumb were present. There was absence of the muscles of the thenar eminence with absence of adduction, abduction and opposition. There was slight atrophy of the muscles of the radial aspect of the forearm but little radial deviation of the hand.

The appearances of the right hand were the same as those of the left hand except that there was a healed scar at the site of amputation of the accessory thumb. Movements were exactly the same.

Radiological examination of the left hand showed hypoplasia of the scaphoid bone; absence of the radial styloid process and hypoplasia of the lower end of the radius. The thumb had three phalanges (Fig. 17). Radiological examination of the right hand showed appearances similar to those on the left.

Type 4—There were three examples.

Case 3—A girl aged nine years had a Type 1 deformity of the left hand which has already been described. The right hand had a rudimentary thumb, with no muscular attachments and no active movement.

FIG. 18
Case 3—Type 4 deformity.

FIG. 19
Case 11—Type 4 deformity.

There was radial deviation of the wrist and hand. Radiological examination showed a floating thumb with two hypoplastic phalanges and a rudimentary distal portion of the first metacarpal bone. There was no scaphoid bone or trapezium. There was hypoplasia of the lower end of the radius (Fig. 18).

Case 11—A boy aged nine years was first seen as an infant with congenital deformities of both thumbs. The right thumb then consisted of a nodule of soft tissue on the radial aspect of the base of the index finger and had no bony skeleton, while the left thumb had a rudimentary structure with two phalanges and was also situated on the radial aspect of the base of the index finger. These two digits were removed at the age of two years.

The radiographs taken before amputation of the thumbs have unfortunately been destroyed. They are reported to have shown absence of the metacarpal bones on both sides although there were two phalanges present in the rudimentary thumb on the left. This appearance is similar to that shown in Figure 18. Later radiographs showed a similar appearance on both sides, namely, absence of the scaphoid bone and of the first ray distal to this, though the trapezium was present on the left. There was hypoplasia of the lower end of the radius on both sides (Fig. 19). This patient had been deaf in the right ear since birth and had congenital absence of the left testis.

FIG. 20

FIG. 21

Case 12—Type 4 deformity.

Case 12—A girl aged seven years had normal muscles of the left wrist and fingers, and there was no radial deviation of the wrist. There was hypoplasia of the muscles of the thenar eminence, and the long extensor and long flexor of the thumb were absent, as were extensor pollicis brevis and abductor pollicis longus (Fig. 20).

Radiological examination of the left hand showed evidence of a previous operation, and that the scaphoid bone and trapezium were absent. The thumb consisted of two phalanges and a rudimentary distal portion of the first metacarpal bone. There was no obvious hypoplasia of the lower end of the radius (Fig. 21)

Type 5—There was only one example.

Case 13—A boy aged five years was seen shortly after birth with a left talipes equinovarus, congenital absence of the right radius and thumb and congenital hypoplasia of the left thumb and thenar eminence.

On examination the left hand showed hypoplasia of the musculature of the radial aspect of the forearm with radial deviation of the wrist and hand. The thenar muscles were absent. The thumb was represented by a small rudimentary nodule situated at the level of the metacarpal heads of the other fingers. There was no musculature to the thumb rudiment which had no movement (Fig. 22). The right forearm and hand showed congenital absence of the radius with hypoplasia of the musculature of the radial aspect of the forearm and radial deviation of the carpus and hand. There was no thumb (Fig. 23).

Radiological examination of the left hand showed absence of the ossific centre for the scaphoid bone and of the bones of the first ray distal to it. There was marked hypoplasia of the lower end of the radius (Fig. 24). Radiological examination of the right hand showed the appearances of a typical congenital absence of the radius with absence of the ossific centre for the scaphoid bone and the bones of the first ray distal to it (Fig. 25).

FIG. 22

FIG. 23

FIG. 24

FIG. 25

Case 13—Type 5 deformity.

Type 6—There were two examples.

Case 4—A boy aged eight years had a Type 1 deformity of the right hand which has already been described. The left hand had four fingers with no thumb. There was hypoplasia of the muscles on the radial aspect of the forearm and slight radial deviation of the wrist and hand. There was absence of the thenar eminence. Radiographs showed the absence of the thumb elements and absence of the ossific centre for the scaphoid bone. There was hypoplasia of the lower end of the radius (Fig. 26).

FIG. 26

Case 4—Type 6 deformity.

FIG. 27

Case 8—Type 6 deformity.

FIG. 28

Case 8—A girl aged six years had a Type 2 deformity of the right hand which has already been described. The left hand had four normal fingers and no thumb or thenar eminence, and hypoplasia of the musculature of the radial aspect of the forearm with a little radial deviation of the wrist and hand (Fig. 27).

Radiological examination showed the absence of the skeletal elements of the thumb, a very small ossific centre for the trapezoid bone, but no ossific centre for the scaphoid bone. There was marked hypoplasia of the lower end of the radius, and the radius was shorter and more curved than normal (Fig. 28).

DISCUSSION

The condition of congenital hypoplasia of the carpal scaphoid and its associated abnormalities appears to be an entity, being mainly confined to the bones and musculature of the radial aspect of the hand, but absence of the carpal scaphoid bone and of the thumb often occurs with absence of the radius in the condition of congenital absence of the radius. According to O'Rahilly (1953) the incidence of absence of the carpal scaphoid and thumb in this condition is 80 per cent.

In congenital hypoplasia of the carpal scaphoid and its associated abnormalities the hypoplasia of the lower end of the radius and the hypoplasia of the musculature of the radial aspect of the forearm are indicative of the relationship between these two conditions. Riordan (1955) stated that congenital absence of the radius can present either as complete absence or as a partial absence, the latter usually being an absence of the distal end. He had not seen a case of absence of the proximal end. A suggested classification of the congenital deformities of the radial aspect of the forearm, carpal scaphoid and thumb is as follows: 1) Absence of the radius with absence or hypoplasia of the carpal scaphoid bone and thumb. 2) Absence of the lower end of the radius with absence or hypoplasia of the carpal scaphoid bone and thumb. 3) Hypoplasia of the lower end of the radius with absence or hypoplasia of the carpal scaphoid bone and its associated abnormalities of the thumb and of the thenar muscles.

This suggested classification is in accordance with our knowledge of phylogeny and embryology. The classic work by Gegenbaur (1898) explains the phylogeny of the limb. Figure 29 is a redrawing of Gegenbaur's original diagram showing "the scheme of a pentadactyl limb skeleton," and clearly shows the bones which are concerned in the formation of the rays of the limb. The first ray consists of radius, radiale, first carpal bone, first metacarpal bone and two phalanges. Wood Jones (1941), discussing Gegenbaur's work, considered that the scaphoid bone is a compound bone derived from both the radiale and the centrale—the radiale forming its proximal part, and the pre-axially displaced os centrale forming its distal part. He also considered that from a functional point of view this fusion was an expression of the specialisation of the functions of the human index finger. The trapezium is derived from the first carpal bone.

Embryologically, organisation of the limb is effected in a proximo-distal sequence, and absence of a proximal portion would lead to absence of or hypoplasia of the portions distal to it; for example, absence of the radius leads to absence—or hypoplasia—of the scaphoid bone, and therefore of the thumb.

FIG. 29
 The scheme of a pentadactyl limb skeleton (after Gegenbaur).

Wood Jones considered that the scaphoid was phylogenetically a recent acquirement and was a structural expression of the need for stability at the base of the important index finger. All the cases in this series showing absence or hypoplasia of the carpal scaphoid have a normal index finger; it is the thumb alone which is hypoplastic. Thus the development of the highly specialised human index finger is not dependent on the presence of a normal scaphoid.

Two of the cases in this series, mother and daughter, show what is apparently a thumb with three phalanges. Gates (1946) in his review of the literature of the three-jointed thumb says that this is one of the rarest malformations of the hand and is due to the failure of the two terminal phalanges to fuse. It is a hereditary condition. Wood Jones states that a first digit possessing the normal two phalanges is an ancient and stereotyped feature of the digitate vertebrate. If this is true, the thumb with three phalanges is not a thumb at all, but a finger. Examination of the radiographs of Case 9 tends to confirm this (Figs. 14 and 15). The normal

index metacarpal has an epiphysis at its distal end only, the normal thumb metacarpal has an epiphysis at its proximal end only. The metacarpal bone of the "thumb" in this patient has an epiphysis at both proximal and distal ends—it is therefore neither a true index metacarpal bone nor a true thumb metacarpal bone. The appearance and function of this pre-axial three-phalanged digit are more suggestive of its being a finger than a thumb, and it is possible that the digit is the index finger, and that there is a post-axial addition of a small finger to compensate for the loss of a pre-axial digit or thumb.

Several of the radiographs of hypoplastic two-phalanged thumbs in this series show that the thumb metacarpal has an epiphysis at its distal end only. This again is not typical of a normal first metacarpal bone and cannot be readily explained.

The bony and soft-tissue defects involving the first ray can be considered to be due to a suppression, either complete or partial, of the ray. The fact that in many cases there are other congenital abnormalities present suggests that there is a common denominator in the development of all the deformities.

SUMMARY

Thirteen patients with congenital hypoplasia of the carpal scaphoid and associated abnormalities have been described and the significance of the condition discussed.

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