A Pedigree with Unusual Anomalies of the Elbows, Wrists and Hands in Five Generations

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SUMMARY

Five members of a White family with previously undescribed congenital anomalies of the elbows, wrists and hands, are presented.

The condition is transmitted as an autosomal dominant trait and is limited to the upper extremities.

The clinical and radiological features are described and the possible development is discussed.


Although congenital deformities of the upper extremity occur commonly, their classification is difficult because they display numerous individual variations and combinations.

It is generally thought that these anomalies are inherited, and we have been able to record a series of 5 members of a White family with unusual symmetrical bilateral congenital anomalies of the elbows, wrists and fingers.

The family pedigree (Fig. 1) clearly shows this to be a case of autosomal dominant inheritance with complete penetrance. Ten members of the family were known to be affected, 4 males and 6 females.

We have examined 3 males aged respectively 31 years, 24 years and 7 years, and 2 females aged 6 years and 2 years.

There was a close family resemblance, with a relatively large cranial vault, but no abnormalities of the skull could be demonstrated radiologically.

All were of normal intelligence, and except for the upper limbs, no other congenital anomalies could be found.

The elbow joints: There was prominence of the radial head and an unusual shape of the olecranon process, both combining to give the appearance of dislocation of the elbow (Fig. 2).

Movements at the elbow joint were not markedly limited. Flexion and extension through a range of 120 degrees from the habitual position of 90 degrees of flexion, was possible. The forearm was, however, fixed in a position of 30 degrees of supination in both adults.

Roentgenograms of the elbows (Fig. 3) showed dysplasia of all the bony components of the joint. The humeral condyles were underdeveloped and the radial head displayed a bulbous appearance. The olecranon and coronoid processes were underdeveloped to the extent that the proximal extremity of the ulna presented no more than a flat surface for articulation with the humerus.

The radiological appearances are also well demonstrated in the case of the 7-year-old boy (Fig. 4), who already displayed the full-blown features of the syndrome.

The wrist joints: Slight radial deviation was noted in all cases, together with a flexion deformity of 20 degrees (Fig. 5). No ulnar deviation was possible, and extension took place from a position of 20 degrees of volar flexion to the...
neutral position only. Active flexion was possible from 20 degrees to 70 degrees.

The thenar and hypothenar muscles were underdeveloped, but in each case the patient enjoyed a normal range of movements of the digits, including the thumb. The pisiform presented as a bony prominence in the adult male cases.

The digits: Marked brachydactyly was found in all the cases (Fig. 5). The distal phalanges were club-shaped—with the nails small and grooved. The little fingers of the boy showed pronounced strebloomicrodactyly.

X-rays of the wrist (Fig. 6) showed the presence of radial deviation with abnormally shaped carpal bones, especially the navicular and triquetrum. There was fusion of the triquetrum and the pisiform.

The features of brachydactyly and strebloomicrodactyly were most clearly demonstrated in the 7-year-old boy (Fig. 7). In the elder children, the epiphyses of the phalanges and metacarpals were not fused, nor was symphalangism present.

The radiological appearance of the wrist of this boy deviated from the normal in many respects. The most striking feature was the large triquetrum (Fig. 7). Other
differences were the round instead of triangular hamate, the smaller capitate and the small or late development of the trapezium, trapezoid and navicular.

Very little is known about triquetropisiform fusion. The ossific centre of the triquetrum appears at about 3 years, and when the ossific centre of the pisiform appears at 10-12 years, the triquetrum is already well developed.

Although the pisiform normally articulates with the triquetrum, and triquetropisiform fusion may be due to failure of the formation of articular interzones, there may be other unknown factors.

The ossific centres of the bones appearing between 5-6 years (trapezium, trapezoid and navicular), are late in appearance and development in our cases, and the much larger triquetrum and hamate may be responsible for the early and persistent radial deviation of the wrist.

As all the carpal bones in both adults and children appear to be abnormal, the basic defect may be present in the cartilaginous anlage or ossification of each individual carpal bone.

Dislocation of the radial head in association with carpal anomalies is not unusual. In our series, the picture is not of simple dislocation of the radial head, but rather of abnormal development of the bony components of the elbow joint.

Clinically and radiologically it does not resemble the elbow deformity of hereditary onycho-osteo-dysplasia. The latter is characterized by hypoplasia of the capitellum and commonly by secondary dysplasia and dislocation of the radial head.

Nievergelt (1944) cited by Pearlman et al. described a syndrome of familial malformation, consisting of bilateral elbow dysplasia with radial head subluxation, massive carpal coalition with brachydactyly and clinopodactyly and tarsal synostosis, together with club feet.

In our series, the elbow deformities differed markedly from Nievergelt’s description, and both the massive carpal and tarsal coalitions with their associated deformities were also absent.

Brachydactyly is less common than polydactyly and syndactyly. It is more frequently encountered in conjunction with other digital anomalies. The 7-year-old (Fig. 7) also displayed streblomicrodactyly of the little fingers, that is, claw deformity of the little finger. This must be differentiated from clinodactyly, which may be described as an incurving of the distal interphalangeal joint of the finger.

Differences of opinion exist in regard to the aetiology of brachydactyly. Shafar considered the cause to be premature ossification of the epiphyseal cartilages of the metacarpals and phalanges. He believed a partial arrest of development occurred at an early embryonic stage when the digits were still united by a web, and the distal phalanges had not become differentiated.

Burrows and Bell both suggested premature epiphyseal fusion rather than absent epiphyses, as a cause of the condition. Barsky’s observations, which applied almost entirely to children, failed to confirm this suggestion.

Radiographs of the children in our series did not display premature epiphyseal fusion or absent epiphyses.

We hope to report on the progress of the individuals in our series after a number of years. The pattern of carpal

DISCUSSION

Carpal fusions appear to be more common in Negro races. Cockshott found lunatotriquetral fusion to be 10 times more common in a Nigerian tribe than in any group of the White races.

Fusion of the triquetrum and the pisiform is rarely seen. Bogart described one case in a series of 1452 roentgenograms of the wrist. He stated that it very rarely occurred as an isolated feature and that, in general, pathological fusion it was the last to occur. We have been unable to find reference to a similar case in the literature.

It would seem that variations in carpal anomalies are genetically determined. The nature of the fusion between the lunate and the triquetrum in particular, has been extensively studied. In the opinion of O’Rahilly, the term ‘fusion’ is probably incorrect. It would be more accurate to refer to it as a failure of separation.

Normally the cartilaginous anlage of the bones form at about the 5th intra-uterine week. Subsequently a jelly-like substance appears at the site of future joint cavities and the cartilage becomes cleft into separate portions, which will later form the individual bones. Failure of the formation of these articular interzones will result in the continuity of cartilage, which will then later become bone.

The age at which the ossific centres join shows a wide variation. In lunatotriquetral fusion it varies between 6-15 years, but in capitatemamate fusions, union of the bony centres occurs early. This is to be expected as the ossific centres of the two individual bones are the first to appear.

Fig. 7. Roentgenograms of wrist and hand of the 7-year-old boy, demonstrating large triquetrum, abnormal hamate and ‘late appearance and development of trapezium, trapezoid and navicular. Metacarpal and phalangeal epiphyses appear to be normal and streblomicrodactyly of little fingers is apparent.
development, fusions and deformities, and possibly of the development of brachydactyly, may be shown after a prolonged period of observation.

REFERENCES
2. Bogart, F. B. (1932); Ibid., 28, 638.

Transvaal Society of Pathologists:
Abstracts of Papers

The following are the abstracts of papers presented at a meeting of the Transvaal Society of Pathologists held in the New Lecture Theatre, SAIMR, Johannesburg, on 14 October 1972:

VIRAL HEPATITIS WITH PARTICULAR REFERENCE TO AUSTRALIA ANTIGEN

W. B. BECKER, Department of Medical Virology, University of Stellenbosch, CP

Several viruses may produce hepatitis as a dominant feature of infection but the main problem is virus A (infectious hepatitis) and virus B (serum hepatitis) infection. Regarding the current status of specific tests for virus A infection, Deinhardt’s group maintains that certain marmoset species provide a suitable and reliable laboratory host for the isolation of virus A, despite criticism, particularly from Melnick’s group. A collaborative study between the two laboratories to resolve the issue is in progress. The report from Ferris and co-workers on a specific faecal antigen in virus A hepatitis needs further investigation. Specific tests for virus B depend on the specific association of Australia antigen (HAA) with virus B infection. The evidence is weighted in favour of certain of the particles which are seen in HAA preparations being virus B. The pathogenesis of virus B infection can presently be best understood on the basis of regarding HAA as infectious, as well as antigenically related to Blumberg’s Ag system of serum protein polymorphism. The availability of sensitive specific tests for HAA and HAA-antibody is helpful in instituting measures to prevent the spread of virus B infection. Preliminary information indicates that immunoglobulin prepared from convalescent cases of virus B infection may prove useful prophylactically. The principle of a vaccine prepared from HAA warrants further investigation. The urgent need is for in vitro methods of culturing virus A and virus B.

YERSINIA ENTEROCOLITICA SEPTICAEMIA

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Infection by Yersinia enterocolitica produces a wide range of clinical forms. The septicaemia form of this condition, however, is rare, and only 20 cases have been reported in the literature.

In our series of cases about 20% (7 out of 34 cases) presented with septicaemia. Most of these patients were Bantu with underlying cirrhosis and siderosis, and the effect of iron on this organism was assessed. In vivo experiments suggested that iron may inhibit the bactericidal effect of specific antiserum. Mice were injected with increasing quantities of ferric ammonium citrate followed by Yersinia enterocolitica and a significantly greater mortality was observed in those animals with iron overload. It is postulated, therefore, that iron overload as found in adult Bantu males could increase the virulence of, or diminish the host’s resistance to, Yersinia enterocolitica, allowing the organism to invade more readily.

THE ENTEROHEPATIC CIRCULATION OF VITAMIN B12 IN THE NON-HUMAN PRIMATE

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The biliary excretion of radioactive vitamin B12 (\(^{57}CoB_{12}\)) was studied in the Chacma baboon (Papio ursinus). \(^{57}CoB_{12}\)-labelled B12 baboon serum was injected directly into the portal vein at laparotomy and bile was collected through a tube placed in the common bile duct. There was a rapid passage of the vitamin through the hepatic parenchymal cell, as shown by the appearance of \(^{57}CoB_{12}\) in the bile 30 minutes after injection. In several experiments between 2% and 5% of the injected dose was excreted in the bile over 6 hours. In further experiments it was shown that a totally gastrectomized baboon failed to absorb this biliary B12 unless it was given together with normal baboon gastric juice. Baboon gastric juice was shown to contain intrinsic factor, which cross-reacted with blocking antibodies to intrinsic factor obtained from a patient with Addisonian pernicious anaemia. The absorption of biliary vitamin B12 is, therefore, intrinsic factor-dependent, and this offers proof for the hypothesis that endogenous B12 loss, superimposed upon a