CONGENITAL ABSENCE OF THE HUMERAL HEAD

Report of Two Cases

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The purpose of this article is to present clinical data, review the literature, and speculate as to the origin of a rare condition. The first case here recorded had been passed over with the remark that "the condition is probably congenital." Attention was aroused when a second case was seen a few months later.

CLINICAL MATERIAL

Case 1. B. S., aged 26 years—Of normal appearance, with powerfully developed muscles. Abduction and medial rotation movements of the right shoulder joint were almost completely lacking. The right humerus was ten inches long and the left eleven and a half inches long. There was no history of injury and no other explanation of the condition which had been present as long as the patient could remember.

Radiographs showed that in place of the usual head of the humerus there was a flat surface with a small central boss projecting about 4 mm. (Fig. 1). The articular surface was twice the length of that of the glenoid. The greater and lesser tuberosities were well developed and the inter-tubercular sulcus was seen clearly on the outer surface of the bone. The shaft had a distinct outward bow with its summit at the deltoide insertion. The cortex was abnormally thick and dense on the outer side, but it was thin on the inner concave side. The glenoid was flattened, with a slight concavity opposite the central boss of the humerus. A tubercle was present above the glenoid. The coracoid process was developed normally. The left shoulder joint was normal.
Case 2. Z. M. K., a healthy Sepoy, aged 28 years—This patient stated at once that disability of the shoulder joints had been present since childhood but not to such a degree as to affect his capacity as a fighting man. He was somewhat indignant that his fitness for service should be questioned. On examination he was found to be normal except for the upper arms and shoulder joints. He was well developed. Both upper arms were short but the forearms were of normal length (Fig. 2). The right humerus measured nine inches and the left ten and a quarter inches. Judging by the position of the lateral condyles there was lateral rotation deformity of the right humerus. Abduction was limited by bone block (Fig. 3). Radiographs showed gross deformity of both humeral heads and glenoids. As in the first case, the humeri were shortened and bowed outwards and the cortex was thicker on the convexity and thinner on the concavity. In order to gain information as to the shape of the joint surfaces radiographs were taken in the position of adduction, abduction and lateral rotation, and also from the axillary view. The findings were:

Fig. 2

Fig. 3

Fig. 4

Fig. 5

Case 2. Bilateral congenital absence of the heads of the humerus. Note the shortening of the upper arms (Fig. 2). Abduction movement of the shoulder is limited by bone block (Fig. 3). In both arms the shaft of the humerus is bowed, there is alteration of cortical density, and the glenoid boss is a convex protrusion fitting into a concave humeral head (Figs. 4 and 5).
CONGENITAL ABSENCE OF THE HUMERAL HEAD

Right shoulder—The glenoid was represented by a boss of bone at the upper end of the axillary border of the scapula. Above and below this rudiment, separated from it by well-defined grooves, could be seen the supraglenoid and infraglenoid tubercles; these structures and the coracoid process appeared to be of normal size. The upper end of the humerus was represented by four bone elevations surrounding a concavity, the elevations representing the greater tuberosity, the lesser tuberosity, and the rather exaggerated facets of the infraspinatus and teres minor muscles. The shaft of the humerus showed bowing and alteration of cortical density (Fig. 4). The axillary view showed that the glenoid boss was a convex projection fitting into a concave humeral head. Left shoulder—The glenoid boss was larger than on the right side, and the humeral joint surface was more shallow and more vertical. Its lower lip was composed of dense bone and was pressed down. The supraglenoid tubercle was not differentiated. The lesser tuberosity was more pronounced than in the right humerus but the greater tuberosity, and the bowing, thickening, and trabeculation of the shaft, were similar (Fig. 5). An axillary view showed similar reversal of the ball and socket arrangement of the joint.

REVIEW OF LITERATURE

Extensive search of the literature has disclosed only six cases with deformity similar to that now described. Lewin (1931) reported two cases. The first had bilateral but asymmetrical deformity. On one side there was a convex glenoid, and a concave humeral head, suggesting absence of only part of the capital epiphysis (Fig. 7). On the other side the joint was formed of a humeral surface which was slightly S-shaped, filling a corresponding glenoid surface (Fig. 6); there was no vestige of the head and the glenoid was more developed than its fellow. Lewin's second case showed unilateral deformity; very similar to that of his first case, but it was complicated by ankylosis of the elbow and Madelung's deformity (Fig. 8). Walter Müller (1939) described three cases with deformities of varying degree (Figs. 9, 10, 11). All three patients had other congenital bone and joint deformities. Brailsford (1944) recorded another case. No details were given but it will be seen from the radiographic tracing that the appearances are similar to those of Lewin's first case (Fig. 12).

DISCUSSION

This group of cases represents varying degrees of a typical but hitherto unrecognized deformity of the shoulder joint. The essential features are complete or incomplete absence of the humeral head with a rudimentary glenoid. The deformity may be unilateral or bilateral; it may be isolated or it may be one of a number of deformities in the same patient. It is associated with relatively slight disability. Both patients whose cases are now recorded fought as combatants through the 1945 Burma campaign. The ball and cup arrangement of the shoulder joint is reversed or it is replaced by a saddle-shaped articulation. There seems to be correspondence between the degree of capital deficiency and the state of development of the glenoid (Müller's three cases—Figs. 9, 10, 11). It appears that lack of the capital epiphysis, or of its cartilaginous and precartilaginous stages, causes corresponding lack of the upper epiphysis of the glenoid-complex while the main body of the glenoid continues to develop.

These observations suggest that the defect occurs at a very early stage of embryonic life and that it is due to failure of development of the tissue in which the capital epiphysis should form. The observation that centres developing late and outside the joint capsule are present and well formed, while those developing early and inside the capsule are not present, or are present only in rudimentary form, also indicates that the defect occurs in the early period of development of the primitive joint.

Further support for this hypothesis is gained from consideration of the experimental work of Murray, Fell, and Robison. Murray (1926) showed that joints develop characteristic shape even when grafted elsewhere so that they are remote from the normal influence of contiguous structures. Clearly the general form of a joint is determined by intrinsic influences, although structural details may be the result of "mechanical" forces. Fell and Robison (1929) came to similar conclusions. The problem as to how two opposing rudiments, developed
from a common mesodermal mass, are able to separate and become movable and yet be interdependent and complementary, was considered by Fell and Canti (1934) using Canti's technique of cinematography in observing and recording in vitro the development of tissues (1928). Fell and Canti showed that joint formation is not caused by the presence of a layer of non-chondrogenic tissue across the site of the future articulation as suggested by the studies of Murray (1926) and Warren (1934). These studies demonstrated that the avian limb bud is a mosaic system and suggested that separation of the articular surfaces might be an expression of this already existing mosaic. Fell and Canti, in the same report, produced evidence for important conclusions. 1) The earlier stages of joint formation can proceed in the complete absence of muscular action and of nerve and blood supply. 2) Joint formation can take place in the absence of the presumptive joint region. 3) Joint formation in the absence of the presumptive joint tissue is controlled by two factors—(a) the amount of tissue removed, (b) the degree of development of the blastema up to the time of operation. 4) The articular ends of the long bones cannot develop their normal shape after removal of the presumptive articular tissue.

From these considerations it may be supposed that the deformity under consideration arose at the presumptive stage, since only those parts entering into the formation of the primitive joint appear to be affected. Extra-capsular structures developed normally except
for minor variations which may be ascribed to altered mechanics. At a very early stage, when the primitive joint had just begun to form, an accident occurred which altered the process of separation in the mesenchyme to form the articular ends. The brunt of force of the accident seems to have fallen upon the humeral head, because in all these cases the glenoid is at least present in rudimentary form, whereas the complementary head of the humerus is seriously deficient in every case. When the cases are set out in order it is seen that the degree of development of the glenoid bears inverse relationship to the extent of humeral defect. If the humeral defect is minimal, the glenoid is well formed; if the humeral element is missing completely, the glenoid is a mere rudiment.

The presumptive joint stage appears to have been well advanced when this unknown factor exerted its influence, causing virtual removal of the caput area and leaving the glenoid without possibility of developing its normal shape. If this is a true reading of the picture, these cases present clinical evidence in support of the conclusions reached experimentally by Fell and Canti.

That the damage is inflicted on the shoulder alone, or only a few joints, might suggest that the accidental factor was present for a limited time which was coincident with formation of the presumptive shoulder joint. But it would appear also that some parts of the body may be more sensitive than others even when exposed simultaneously, and this would account for unilateral cases.

It is possible that the cases collected here on the basis of similar radiographic appearances in the shoulder may not form a true group, because they may not have a common cause. The clinical evidence of similar appearances is, however, supported strongly by experimental data, and it seems reasonable to accept these cases as a group which show a congenital deformity of the shoulder joint, hitherto unrecognised.

**SUMMARY**

1. Two cases of an unrecognised congenital defect of the humeral head are described and the cause is discussed.
2. Only six cases with similar radiographic appearances could be collected from the literature. In most of these cases other skeletal deformities were present, whereas in those now reported only the shoulder was affected.
3. Examination of radiographs suggests that the main deformity consists of lack of development of the capital epiphysis of the humerus.
4. Consideration of the cases, together with experimental data from the studies of Fell and Canti, suggests that the time in development at which the fault occurred was the presumptive joint stage, just when the articular rudiments had separated.
5. A "nociferous agent," acting only for a limited period, and only on certain tissues, is postulated.
6. It is suggested that the defects recorded should be recognised as a group of congenital deformities of the shoulder joint.

**REFERENCES**

Canti, R. G. (1928): Archiv für experimentelle Zelforschung, 6, 86.