CHONDRODYSPLASIA IN TWINS

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Our colleague, L. G. Parsons¹, has briefly outlined the historical facts regarding a disorder of the skeleton bearing the various names—hereditary deforming chondrodystrophy, multiple cartilaginous exostosis, diaphysial aclasis (Keith) or Ollier's disease; and Brailsford² has recorded the radiological details of a similar condition under the term chondro-osteodystrophy. The disease produces a striking clinical picture and is of special interest in relationship to its congenital origin, familial tendency, and its more frequent occurrence in males. The essential disorder lies apparently in the abnormal process of bone growth, affecting the skeletal parts derived from a cartilaginous or membranous basis. Its cause is unknown. Although certain theoretical endocrine and peripheral vascular derangements have been suspected, no true evidence of such is established in cases carefully studied. From the pathological aspect 'the growth of cartilage appears to be excessive and the calcification irregular.' Radiographic studies of the long bones show the metaphysis and the end of the diaphysis to be occupied by cartilage, and the bone cortex in the same areas to be thin and irregular. The cartilaginous masses may thus, in some areas, give a pseudo-cystic appearance. The epiphysis is notably misshapen and reveals an unusual direction, usually oblique. Parson's description of the clinical picture epitomizes the significant features and he states that the clinical picture depends to a large extent on the presence or absence of secondary skeletal deformities; indeed, the presence of multiple exostoses may pass completely unnoticed by the patient, or attention may only be drawn to them by limitation of movement at a joint, or by pain from a pointed exostosis. All sorts of variations exist; thus in some cases there may be exostosis in normal bones, in others exostosis in bones with slight deformities, or in others again deformed bones without exostosis. If deformed bones are present the deformities become marked as the child grows, and there is always some stunting of growth, this being due to shortness of the legs. The arms are also shortened. The unequal growth in length of the paired bones of the legs and forearms produces characteristic deformities and radiographic changes. 'The radius becomes a bent bow; the ulna serves as its tight string. In about one-third of the cases the bow becomes unbent by a spontaneous dislocation of the proximal end of the radius' (Keith). Less

frequently the distal end of the radius becomes dislocated, throwing the hand over to the ulnar side (an inch or more above the wrist). This produces a characteristic deformity, the wrist appearing to be unduly long and mobile so that the hand can be bent far back on the forearm. This deformity is so characteristic as to suggest at once the correct diagnosis. Sometimes as a result of this dislocation the hand may be deviated so that it forms almost a right angle with the forearm. As might be expected, pronation and supination are often limited. As a result of relative shortening of the fibula, pes cavus and genu valgum may occur. Also scoliosis, deformities of the pelvis, scapulae, clavicles, fingers and ribs, even a well-marked rosary, may be present. The phalanges of the feet and hands show enchondromata more frequently than exostoses, and 'radiographs show some or all of the various forms of the disease: exostoses, curves, irregularities, epiphyseal obliquities, bending and unequal growth of the paired long bones masses of cartilage in the metaphysis, dense stippling in some of the tarsal, carpal or phalangeal bones, and frequently an assymetrical distribution of the changes is revealed.' In some cases the vertebrae, jaws and skull also reveal evidences of the diseases.

Brailsford's case (which we have abstracted from his paper) was that of a boy aged 3 years 9 months.

The patient was a full term child and appeared to be normal until he began to walk at 13 months of age. The mother then noticed that the child could not stand erect, but stood and walked by the body supported by the hands on the knees. On this account he was taken to a hospital, but no definite abnormality was detected. Some months later he was examined at another institution and was provided with a posterior spinal support. Similar appliances have been worn continuously since. The boy is a bright and intelligent little fellow and looks relatively well nourished. He is 2 ft. 9 in. in height and weighs 26 lb. 11 oz. He can stand almost erect without support, and walks with a normal gait, but readily gets tired, and then lies down and assumes the hand-on-knee position during sleep. His joints are all on the large side; there is no evidence of wasting. The neck is short and thick and he has a double inguinal hernia. All his teeth show marked caries. He had pneumonia when 20 months old, but since has had no other illness. He has a good appetite, and for nearly three years has been given daily doses of cod-liver oil. The parents, who are normal in stature and appearance, had one other child (a girl) previously, which appeared to be quite normal, but died of pneumonia at the age of 21 months. No history of any deformity in any member of the parents' families could be obtained from them. On radiographic examination the most noticeable features are the large joint spaces; the irregularity and fragmentation of the epiphyses particularly of the metacarpals and metatarsals; the irregular shape and size of the vertebral bodies and the dislocation, of the lumbar dorsal vertebrae; the short, thick, long bones and the coarse, irregular reticulation of the cancellous tissue and the absence of the regular lines of the lamellae.

Brailsford in his paper shows excellent radiographs of his case, and states that there can be no question that in this disorder there is some systematic affection which produces a disturbance of the normal growth of cartilage and bone. His case illustrates well the thoracic and vertebral dislocation which is usually present.

Authors' cases.

The two cases which are recorded in the present paper are specially interesting because the disease is shown in twin boys, each being an exact

duplicate of the other in every physical and mental particular. We have been unable to find any previous record of such bone disorder in twins.

Gerald and Charles R., aged 6 years. Although the parents appear perfectly normal their offspring have been singularly unfortunate. The first baby died of

Charles and Gerald R.

Charles and Gerald R., and a normal sized boy of the same age.

Radiograph showing the characteristic epiphysial changes in the forearm bones and in the hands.

Radiograph of spinal column showing the peculiar shape of the vertebral bodies and the displacement of the lower dorsal vertebrae.

CHONDRODYSPLASIA IN TWINS

55

Radiograph of the knee showing the epiphyseal irregularity.

Radiograph of the femoral epiphyses showing the irregularity and flattening.

spina bifida. The present twins were the next born, and the last baby aged 5 years has bilateral large inguinal herniae. It has not been possible to make any efficient enquiry into the family tree regarding the bone disease.

Gerald and Charles were brought to the hospital at the age of 6 years for diagnosis. They had been born in a normal confinement, and the parents confess that they did not notice anything very unusual until the age of $2\frac{1}{2}$ years, when the prominence of the boys' chests attracted attention. Each was a somewhat delicate child at that time, and both suffered from sickness ('vomiting like a fountain') but this disability has now completely disappeared, and the boys are now well nourished and of distinctly ruddy appearance. Feeding in infancy was satisfactory and no evidence of any ordinary rickets was noted.

The photographs illustrate well the physical posture and outline in this disease, the small stature, prominent joints, chests and spinal deformities (especially the prominence of the lower dorsal region), and the curving of the forearms. In remarkable contrast the head is of reasonably good shape, and the boys possess a pleasant countenance and an almost perpetual smile. Their mentality is good. The teeth (in contrast to Brailsford's case) are in magnificent condition, and are all present, and quite regular in arrangement.

The measurements of the twins, with those of a control of the same age, are as follows:—

	Age.	Height.	Weight.	Head circumference.
Charles	6 yr.	2 ft. 8 in.	2 st. 3 lb.	21 inches
Gerald	6 yr.	2 ft. 8 in.	2 st. $4\frac{1}{2}$ lb.	21 „
Normal	6 yr.	3 ft. 7 in.	3 st. $2\frac{1}{4}$ lb.	$19\frac{1}{2}$ „

Each has bright auburn hair and the skin is normal. No abnormal visceral signs are present. Urine normal. Blood counts in each were quite normal and no unusual features in the blood biochemistry was found. Their gait is slow, but walking is painless, and they are quite able to sit up comfortably and to carry out ordinary physical movements. Respiration is normal and easy; the blood vessels are normal.

Radiographs reveal the characteristic and typical features of this remarkable disease. The epiphysial deformity is well illustrated in the forearm bones. The spine shows a peculiar tongue-like shape of the vertebral bodies, and a forward dislocation of the dorsal vertebrae is apparent. The spinal articular processes are also somewhat irregular and the spinous processes stunted.

We are indebted to Dr. Harold Black for the radiographs of these boys.

REFERENCES.

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