MORQUIO'S DISEASE

REPORT OF TWO CASES

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In 1929 Morquio¹ described an osseous condition characterized by a widespread, symmetrical deformity of the skeletal system. It was characterized by a shortening of height and an anterior posterior deepening of the thorax. In contrast, the extremities have normal dimensions, although there are deformities present such as flat feet and genu valgum. Since then, Ruggles², Meyer and Brennemann³, Barnett⁴, Davis and Currie⁵, and Coward and Nemir⁶, have published reports of similar cases. In all there have been eighteen cases reported. The familial occurrence of the disease is most marked, there being only three cases on record where only one member of a family was affected.

Case records.

Two brothers were admitted at different times to the hospital. The younger (S. S., aged 9 years) was admitted in October, 1934, while A. S., aged 11 years, was admitted in April, 1935. The parents who are not related are alive and well. There are also two sisters and two other brothers who are all normal. No information could be obtained of the presence of this deformity in the preceding generation. The children had always lived on a farm and had never received any cod-liver oil in infancy or early childhood. Both children were admitted with a complaint of deformity of the back from early childhood. In addition, during the past two years the gait of the younger brother had become waddling in character.

The physical appearance of both children was similar, though in the younger brother the changes in the osseous system were more marked (fig. 1 and 2).

HEADS: These appeared to be larger than normal and square in shape. The bridge of the nose was depressed. The facial expression was bright and alert. The teeth were malformed in the older patient.

TRUNK: The trunk appeared to be short due to changes in the vertebral bodies (see fig. 3). There was a marked kyphosis in the upper lumbar region. The sternum projected forward in the

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upper three-quarters and curved in below this. There was some flaring of the costal margins and the anterior posterior diameter of the chest was greater than the transverse diameter. The costal margin nearly rested on the crest of the ilium.

EXTREMITIES: There was a wide-base gait which in the younger brother resembled that of dislocation of the hip. In the standing position a marked genu valgum resulted in a separation of the

FIG. 1.—Photograph of S. S., aged 9 years.

FIG. 2.—Photograph of A. S., aged 11 years.

feet of four inches. The feet were very flat. The bones of the extremities were fairly straight. The finger tips nearly reached the level of the knee.

MUSCULAR DEVELOPMENT was fairly good but in the younger brother it was better on the right side than on the left. There was some muscular weakness but no limitation of movement.

The liver and spleen were not palpable. The eyes were normal.

FIG. 3.—X-ray of vertebrae (A. S.)—lateral view, showing deformity of vertebral bodies.

TABLE I.
PHYSICAL MEASUREMENTS.

	9 YEARS NORMAL (ENGLEBACK) 1932	9 YEARS S. S.	11 YEARS NORMAL (ENGLEBACK) 1932	11 YEARS A. S.	7 YEARS NORMAL (HOLT)	7 YEARS MEYER & BRENNEMANN
HEAD	20.9 inches	23 inches	21.1 inches	21½ inches	20½ inches	21 inches
CHEST	24.6 ,,	24 ,,	26.1 ,,	27 ,,	24 ,,	37½ ,,
ABDOMEN	22.3 ,,	22 ,,	...	...	...	...
TOTAL LENGTH	51 ,,	46 ,,	55 ,,	51 ,,	36 ,,	32 ,,
WEIGHT	57 lb.	50½ lb.	70 lb.	51 lb.	56 lb.	31 lb.
SITTING HEIGHT	...	...	26 inches	23 inches	23 inches	19½ inches

TABLE 2
CLINICAL AND LABORATORY INVESTIGATIONS

	S. S. 9 YEARS	A. S. 11 YEARS
URINE ANALYSIS	Normal	Normal
CONCENTRATION TEST	Normal limits	Normal limits
TUBERCULIN SKIN TEST 1/400 DILUTION	Negative	Negative
BLOOD WASSERMANN	Negative	Negative
INTELLIGENCE QUOTIENT	124	—
B. M. R.	99 per cent. (B. & T.)	100 per cent. (B. & T.)
BLOOD PRESSURE	108/82	120/90
CALCIUM MGM. PER 100 C.C. BLOOD ..	9.1—9.9	10.3—9.9
PHOSPHOROUS MGM. PER 100 C.C. BLOOD	4.9—5.0	4.4
PHOSPHATASE	12.6 units	19.2 units
N. P. N.	43 mgm.	36 mgm.
CREATININE	1.66 mgm.	1.25 mgm.
CALCIUM RETENTION FOR 4 DAY PERIOD RECEIVING AN INTAKE OF 1 GM. OF CALCIUM DAILY	+ 2.95 mgm.	+ 2.387 mgm.
CALCIUM RETENTION FOR 3 DAY PERIOD RECEIVING AN INTAKE OF 100 MGM. OF CALCIUM DAILY	- .378 mgm.	- .336 mgm.

In table 1 are seen the measurements of both boys and also the measurements of the case reported by Meyer and Brennemann³. It will be noted that the three children are three to four inches shorter than the normal total height. This shortening is chiefly due to changes in the vertebral column,

as the sitting height is less than normal for that age. The greater chest measurement of the case reported by Meyer and Brennemann is possibly due to the fact that the kyphosis was in the thoracic region, while in the two patients here described it occurred in the lumbar areas. All children were under weight.

FIG. 4.—X-ray showing changes in knees (S. S.).

In table 2 are seen the results of the clinical and laboratory investigations. The findings were all within normal limits. It is of interest to note that on an intake of 1 gm. of calcium per day there was a positive balance, while on an intake of 0.1 gm. of calcium per day there was a

FIG. 5.—X-ray of hands of A. S. and hands of normal child same age. This shows decreased number of centres of ossification and osteoporosis. There is malformation of proximal ends of metacarpal bones.

negative balance. A normal boy, ten and a half years of age, who was placed on similar diet containing 0.1 gm. of calcium per day at the same time also showed a negative calcium retention.

FIG. 6.—X-ray of pelvis and upper ends of femur (A. S.), showing changes in contour of pelvis and heads of femora.

X-ray findings.

Dr. A. Rolph, radiologist to the Hospital for Sick Children, Toronto, has made the following report of the skiagrams that were taken of the entire skeleton. The bony changes were similar in each case.

All the vertebrae show a considerable degree of osteoporosis combined with a curious change in shape. For the most part they are under-developed. In all cases the vertical diameter of the bodies is diminished and a considerable degree of lordosis has been produced in the lumbar region. The first and second lumbar vertebrae are short in the anterior posterior diameter (fig. 3).

Skiagrams of the long bones show a considerable degree of general osteoporosis and the epiphyses are very irregularly formed. At the elbows it is noted that the epiphyses on the medial and lateral aspects are

so developed as to make the lower end of the humerus almost symmetrical. The trochlea is distinctly under-developed. The medial epicondyle is abnormally developed, being an irregular mass of small centres of ossification which spread out in semilunar formation. At the wrists there is a distinctive under-development of centres of ossification. At the knees (fig. 4) the epiphyses of the femora and tibia are very irregular in outline, appearing almost angular at points along their articular surfaces.

Skiagrams of hands (fig. 5): The ends of the third and fourth metacarpals are deformed, some having a pointed base instead of the usual flat ends. These findings correspond to the skiagrams in the case reported by Meyer and Brennemann.

Skiagrams of the hips (fig. 6): The pelvis is distinctly deformed, having a pinched-in appearance from side to side. The acetabular cavities are unusually enlarged, and their articular surfaces are irregular. The heads of the femora show flattened epiphyses which are very poorly developed and mottled, and somewhat like those bones in Legg-Perthe's Disease. Both sides have the same changes.

Skull: This shows an unusually high cranium, with unusually thick bones forming the cranial vault. They have a slightly more granular appearance than normal. The sella turcica appears small.

Intravenous pyelograms of the urinary tract were normal in each boy.

Comment.

Clinically the outstanding findings are dwarfism and deformity of the body due to changes occurring chiefly in the vertebrae, pelvis and long bones. There is delayed epiphyseal development and retardation of the centres of ossification. At present this condition might be confused with a somewhat similar osseous condition described by Poynton⁷ and others in which dwarfism is associated with bony changes in the skull as well as the skeleton, splenomegalia, enlarged liver, and defective vision.

From the x-ray examination it would appear that some process was interfering with normal development of the epiphyses, the centres of ossification.

No explanation is offered for the etiology of this condition. It has already been suggested by others that the condition might be related either to a metabolic or endocrine disturbance. Ruggles has treated his cases with both pituitary and thyroid extract. The results were disappointing. Meyer and Brennemann gave their cases 90 drops of viosterol a day for five months, which caused a rise in phosphorus and a drop in the calcium values, but there was no improvement in the osseous system. The younger of the present patients was given three tablespoonsful of cod-liver oil daily and Mead's Mineral Mixture No. 5, receiving 1 tablet thrice a day. He did not take the cod-liver oil but did take the calcium tablets. The mother reports a gain in height but as yet he has not been admitted for a re-examination. The older brother was put on thyroid, $\frac{1}{2}$ grain daily.

Summary.

A report is presented of two brothers with a generalized, symmetrical osseous disturbance resulting in dwarfism and peculiarly shaped chest. Radiologically the chief changes appear in the vertebrae, pelvis, long bones, and delay in the development of the epiphyses especially of the head of the femur. The condition has a tendency to be familial and its etiology is unknown at present.

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