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A Case of Morquio-Brailford Syndrome

By FREDERICK EDWARD KRATTER

The principal clinical features of this rare, familial disease are the triad of skeletal deformities, structural anomalies of the cardiovascular system, and mental subnormality.

The syndrome is attributed to a recessive gene, and Morquio (1929) held the view that the underlying pathology consisted of mucoid changes occurring in the cartilages in place of their normal calcification.

Boecker (1942) and his associate (De Rudder, 1943) expressed the opinion that the disorder is related to gargoylism. There appears, however, to be no clinical or laboratory evidence that these two conditions are in fact identical (Washington, 1940), interchangeable or connected with each other. Gargoylism is a progressive metabolic disorder (Farrell, 1942) of the mucopolysaccharides, associated with mental subnormality of varying degree and a shortened life expectancy. The Morquio-Brailford syndrome, on the other hand, is less rapidly progressive, and shows milder degrees of mental subnormality and a life span reaching into the fourth or fifth decade (Catel, 1944; Zellweger, 1948; Fanconi, 1947).

Coffey held the view that this syndrome is a variant of chondrodystrophic dwarfing; Kuzma and King (1948) described a patient who showed a combination of diffuse chondrodystrophy with angiomatosis, a symptom-complex now known as Malfucci syndrome. Another patient, a girl, was described by Brailsford (1943); she showed an unusual bony dystrophy, was dwarfed in stature, and had severe malformations of the chest and limbs. Jensen (1934) investigated a similar case displaying a serious pathological condition of the metaphyses.

The abnormalities detectable in the bones, cartilages and joints of patients with Morquio syndrome are generally considered to be pathognomonic. These changes are osteochondrodystrophic in nature and are not infrequently mistaken for rickets. Typical of these changes is the short neck which appears sunken into the chest; the thorax is reduced in height owing to the flattened, wedge-shaped vertebral bodies. The antero-posterior diameter of the chest is increased, and its base is broadened; severe kypholordotic and/or scoliotic deformities are likewise prominent.

The stature is stunted and the face and cranium appear rather large in comparison with the short body. The joint surfaces of the small wrist and ankle bones are frequently irregular in shape and enlarged in size; occasionally some of these bones may be missing altogether. The feet are flat and are held wide apart on standing or walking, which accounts for the patients' ungainly waddling gait.

History of Patient

Joseph A., born 15 April, 1930, a white male, was admitted on 18 July, 1937, to Pineland Hospital and Training Center, Pownal, Maine.

His family history revealed deaf-mutism in a paternal uncle and mental deficiency in a maternal cousin and aunt. The latter died when aged seven months, and she also showed severe deformities similar to those exhibited by the patient.

The personal history disclosed that the patient had been the sixth child in order of birth, had weighed six pounds and eight ounces and was born in the eighth month of pregnancy. As a baby he was said to have had a large frame but a rather poorly nourished body. It was further recorded that the mother had been very irritable, nervous and emotionally unstable during the patient's pregnancy and that she had cried readily at the slightest provocation. It was also stated that her membranes had ruptured spontaneously ten days

before her delivery, that the birth was non-instrumental and that the doctors did not expect the baby to survive.

During his early infancy and childhood he apparently did not develop as he should; he was a frail and seedy child, and he seemed unable to digest his food properly. His first teeth appeared at nine months and he began to walk with assistance in his third year; his speech developed with great difficulty in his fourth year. Childhood diseases included mumps, measles, whooping cough and rickets; the records, however, did not reveal any serious accidents, major illnesses or operations.

He was never capable of attending school regularly, and could not dress himself without assistance, but acquired fair toilet habits in his fourth and fifth years. He was reportedly an obedient co-operative and agreeable boy. The hospital history further disclosed that he had made a rapid and successful adjustment to his new environment and that he had been clean in his personal habits and of cheerful disposition. He conversed readily with everybody, and it was noted that there had been some improvement in the articulation of his speech since his admission. His powers of retention were considered to be fair for a boy of his age (7 years); he was, however, extremely sensitive to correction and often would get quite upset when reprimanded about his occasionally noisy, boisterous and sulky behaviour.

He attended special school at the basic level provided by the hospital's teaching centre for a period exceeding ten years. He was described as a good-natured and well-behaved pupil who made slow but steady advancement. He demonstrated particular interest in music and singing classes, and was able to retain most of the tunes and lyrics. On reaching his 29th year of life, he was transferred to another unit of the hospital, where he was very well liked for his cheerfulness, willingness to do simple chores and sociable conduct. He still spends most of his spare time playing the banjo and in singing.

Physical Examination

State of nutrition good; general health fair; cranial circumference 24½ inches (macrocephaly); some asymmetry of face and head; extremities chondrodystrophic, fingers and toes stumpy; hair black and coarse; gothic-shaped palate; thyroid gland palpable; skin of coarse texture, thickened but of light colour; scalp redundant and loose over calvarium, thorax pigeon-chested and dorsal spine kyphoscoliotic. Height 3 feet 1 inch and weight 4 stone 1 pound (57 pounds).

Central nervous system: eyes: strabismus convergence, left side, associated with some clouding of left cornea; bilaterally sluggish pupillary reactions to light; bilateral, horizontal nystagmus; deep tendon reflexes slightly increased especially knee jerks.

Cardiac system: blood pressure 110/90; 1956, 120/95; 1958, 115/95; pulse rate 68, regular, fair volume; soft, blowing systolic murmur over pulmonary area; cardiac outlines enlarged.

Respiratory system: breath sounds prolonged in expiration over both sides of the chest in more or less equal measure associated with bilateral hyper-resonance to percussion. Wheezing sounds present over upper frontal lung fields and lower portions of back of chest.

Genito-urinary system: right testicle undescended.

Psychological Findings

Wechsler Adult Intelligence Scale: VIQ 59, PIQ 52, FIQ 53.

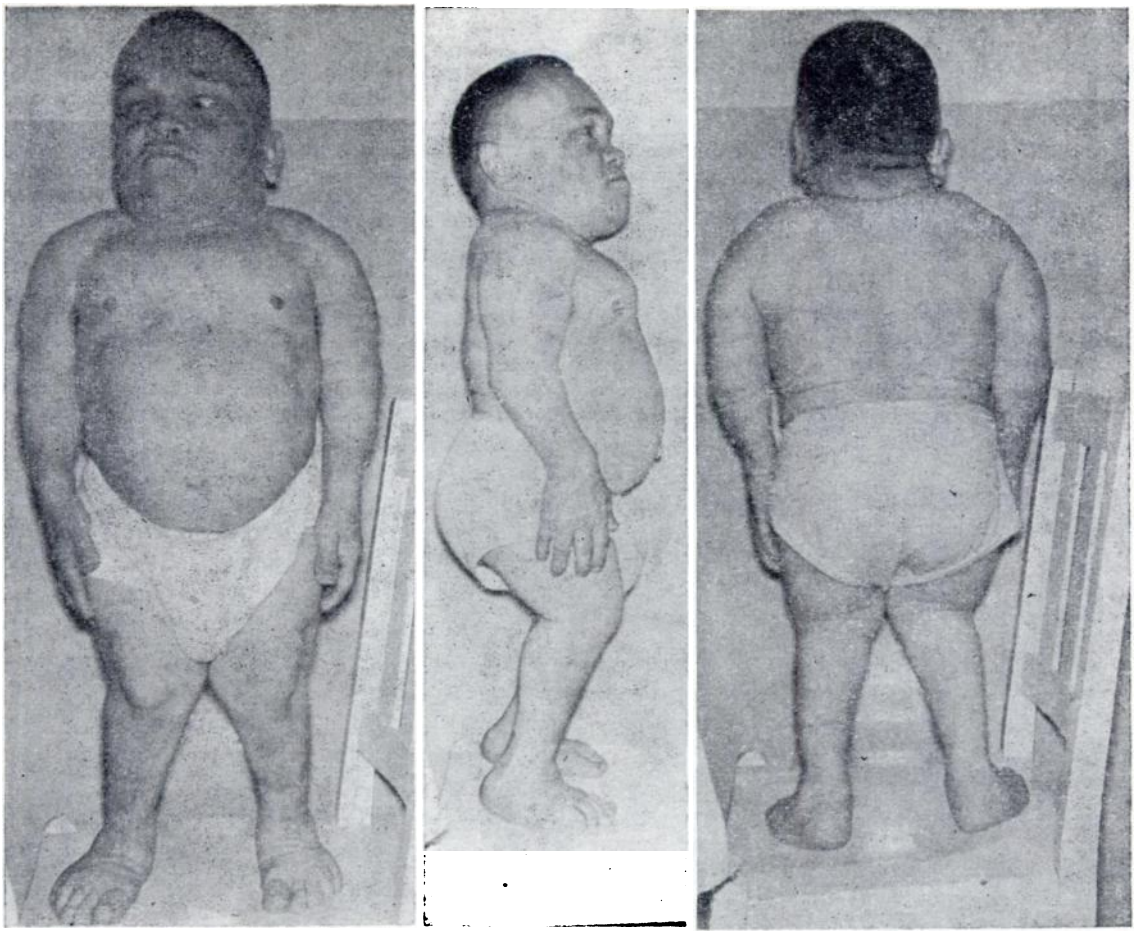
Raven Progressive Matrice Test: 50 percentile, MA 7–9; Goodenough Test: MA 5–9, I.Q. 44.

The patient's test results revealed that he might have the potential to perform somewhat higher than his I.Q. would indicate. However, his general activity was limited because of his dwarfed stature. All drawing tests were somehow distorted, which may in part be due to a sensory, spatial motor defect or just to poor vision.

The Bender drawings were quite small, with several distortions. Maturationally, they were between the sixth and seventh level. Small details such as fingers were enlarged in his drawings.

Electrocardiogram

Sinus arrhythmia, occasional premature beat in lead III.



Roentgenological Findings

X-ray examination revealed some fragmentation and distortion of the epiphyses of long bones as well as considerable shortening of shafts in both upper and lower extremities. Intervertebral discs were flattened and irregular in appearance. causing considerable shortening of the trunk. The third, fourth and fifth lumbar vertebral bodies displayed a pronounced narrowing in their outlines, and wedge-shaped deformities; some fish-hook-like exostotic excrescences were also observed. The thoracic spine showed a kypho-lordotic configuration.

The pelvic bones were narrower than is usually the case, particularly in the sagittal diameter, and the ribs showed small roots with flared and oar-shaped enlargements in the mid-portions. The epiphyseal margins of the shoulder blades presented some stippling, and the tarsal and carpal bones were irregular and distorted in contour.

Summary

This clinical study endeavours to present a description of the physical, psychological, psychiatric, electrocardiographic and roentgenological aspects of the Morquio-Brailsford syndrome in a male patient of Caucasian race and of subnormal intelligence (I.Q. 53).

A short review of the literature is given with special reference to the pathology of this symptom-complex.

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Frederick Edward Kratter, M.B., Ch.B., B.Soc.Sc., C.M.D., D.P.M., *Clinical Director, Western State Hospital, Hopkinsville, Kentucky, U.S.A.; now Consultant Psychiatrist and Medical Director, Brockhall Hospital, Langho, near Blackburn, Lancashire*

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