CASE REPORT

GARGOYLISM

REPORT OF FOUR CASES

BY

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The first published description of the syndrome now known as gargoylism appears to be that given by Hunter (1917), although the late Dr. John Thomson, of Edinburgh, had in his clinical teaching recognized the condition as a disease entity as early as 1908 (Henderson, 1940). Some authors have adopted the eponym 'Hurler's syndrome,' though this is a misnomer, since Hurler's (1920) paper did not appear until two years after Hunter's. The literature relating to the condition has recently been extensively reviewed in this journal by Henderson (1940). The following reports add four new cases, and serve further to emphasize the familial nature of the syndrome.

Clinical records

Case 1. M.B. (fig. 1), female, aged four-and-a-half years, a sister of case 2, was admitted to hospital in October, 1933.

FAMILY HISTORY. The child is of French-Polish extraction. The other

FIG. 1.—Showing the two sisters D. B., aged three-and-a-half years, and M. B., four-and-a-half years.

siblings, with the exception of case 2, were normal, and the parents cannot remember any children like these amongst their immediate or distant relatives.

PRESENT ILLNESS. The child seemed normal until four or five months of age. At that time her mother noticed that her head seemed large and that she was continually perspiring. Her breathing became difficult and she developed a purulent nasal discharge. At times her respirations were difficult and were associated with a crowing sound. Her abdomen at this time was large and the navel protruded. A doctor was consulted, who stated that the infant was getting rickets and prescribed cod-liver oil, orange juice and thyroid tablets for a month. No improvement was noted during this time and the medicine was discontinued. The child's development was slow. She did not sit up until eleven months of age, did not stand until two years of age and was unable to speak at four-and-a-half years of age. Her first tooth appeared at twelve months.

PHYSICAL EXAMINATION. The general appearance is that of an under-developed, well-nourished, female child with coarse features, a large head, protuberant abdomen and obvious mental retardation. The head is symmetrically enlarged and the suture lines are palpable as grooves. There is a bulging area on the skull posterior to the ear on either side. The nose is broad, with a depressed bridge, and the lips are thick. The hair is thick and coarse and the eyebrows are thick and bushy. The eyes are widely spaced and protrude slightly; both corneae are uniformly cloudy. The pupils are equal, react to light, and there is no strabismus. The teeth are small and the incisors are separated. There is a profuse post-nasal discharge. The chest is symmetrical, with a rachitic rosary palpable. The lungs and heart are normal. The abdomen is large and the navel is protruding. The liver is palpable four finger-breadths below the costal margin in the right mammary line. The spleen is palpable two fingerbreadths below the costal margin. There is a moderate kyphosis over the lower dorsal and upper lumbar spine; no scoliosis is present. The scapulae are fixed. The forearm and fingers are thickened and there is some limitation of extension at the elbow, wrist and finger joints. The knees appear large and also show limitation of movement. The reflexes are normal.

MEASUREMENTS	PATIENT	NORMALS (ENGLEBACH, 1932)
	INCHES	INCHES
Length ..	37	39.5 to 42.3
Head ..	20½	19.9
Chest ..	20½	21.0
Abdomen ..	22	19.7

RADIOGRAPHIC EXAMINATION shows abnormal thickness of the long bones (fig. 2), with rachitic irregularities in the diaphyses. The second, third and fourth metacarpals show pointed proximal extremities, and the wrists show three carpal ossification centres at four-and-a-half years of age. The skull is irregular in thickness, heavy over the occiput and thin over the temporal and parietal areas. The general structure is turriccephalic in type and is associated with a bulging of the temporal and parietal bones. The sella turcica appears to be slightly enlarged.

LABORATORY EXAMINATION. Red blood-cells 4,500,000 per c.mm.; white blood-cells 9,000 per c.mm.; Polys., 68 per cent.; Lymphs., 32 per cent.; Haemoglobin, 85 per cent.

Blood cholesterol, 229 mgm. and 221 mgm.

FIG. 2.—M. B., showing rachitic changes in the long bones, the pointed metacarpals and three carpal centres of ossification.

Blood calcium, 9.4 mgm. ; Blood phosphorus, 4.0 mgm.; Blood Wassermann, negative.

Von Pirquet tuberculin test, negative.

- PSYCHOMETRIC EXAMINATION by Kuhlmann's test. Mental age one to two years, I.Q. 26.

Case 2. D. B. (see fig. 1), female, aged three-and-a-half years, a sister of case 1, was also admitted to hospital in October, 1933.

PRESENT ILLNESS. This child seemed normal until six months of age, at which time her mother noticed that she was not as active as other children of her age. At eight months of age she began to breathe through her mouth because of a purulent nasal infection, and at times her respirations were of a crowing type. She was seen at that time by a physician and was given cod-liver oil for one month with no appreciable improvement. The child's development was slow. She could not stand until two years of age and could not walk or talk until three-and-a-half years of age. She was seen by a physician three months before admission and given thyroid tablets, which produced no demonstrable change.

PHYSICAL EXAMINATION. On examination she presents findings which are similar to those in case 1. She is an under-developed, well-nourished, female child of rachitic appearance who is mentally deficient. The skin is dry and somewhat thickened. The head is large and symmetrical but shows an area of bulging above and posterior to each ear in the region of the parietal and temporal bones. Frontal bossing is also seen. The eyes appear widely separated, protrude slightly, and the corneae are uniformly cloudy. The

pupils react to light, and there is no strabismus. Vision appears good. The nose is wide and shows flattening of the bridge. The neck is short and thick. The lips are thick, the teeth small and the incisors separated. There is a profuse post-nasal discharge. The chest is symmetrical, but shows rachitic beading at the costochondral junctions. A faint systolic murmur is heard in the pulmonary area. The abdomen is large and there is an umbilical hernia. The liver is palpable four finger-breadths below the costal margin, and the tip of the spleen is also palpable. The scapulae are fixed and there is limitation of extension of the extremities. The bones of the extremities appear shortened and thickened on palpation. There is marked kyphosis of the lower dorsal and upper lumbar spine but no scoliosis. Reflexes are normal.

MEASUREMENTS	PATIENT	NORMALS (ENGLEBACH, 1932)
	INCHES	INCHES
Length ..	34½	36·9 to 39·5
Head ..	20	19·6
Chest ..	20½	20·4
Abdomen ..	21	19·3

RADIOGRAPHIC EXAMINATION shows irregular, almost ovoid, vertebral bodies, with a shelf-like projection on their anterior margins (fig. 3). The long bones are uniform in density and are thicker than normal. At the wrists, two carpal ossification centres are to be seen at three-and-a-half years of age. The skull is somewhat irregular in thickness, being unduly heavy in the occipital region (fig. 4). The bones of the temporal and parietal regions are thin and bulge prominently. The sella turcica appears to be normal in size.

FIG. 3.—D. B., showing the ovoid shape of some of the vertebra with the anterior projections of the bodies.

FIG. 4.—D. B., showing the marked bulging of the temporal and parietal areas of the skull.

LABORATORY EXAMINATION. Red blood-cells, 4,500,000 per c.mm.; White blood-cells, 1,500 per c.mm.; Polys., 74 per cent.; Lymphos., 26 per cent.; Haemoglobin, 90 per cent.

Blood cholesterol, 235 mgm. and 260 mgm.; Blood calcium, 9.2 mgm.; Blood phosphorus, 3.0 mgm.

Blood Wassermann, negative.

Von Pirquet tuberculin test, negative.

PSYCHOMETRIC EXAMINATION by Kuhlmann's test. Mental age 9 mos. I.Q. 22.

Attempts were made at different times to trace these children, but without success. Just recently, however, it was learned that both had died from pneumonia within a year after discharge from hospital.

Case 3. E. A., male, (fig. 5 and 6) aged nine years, a brother of case 4, was admitted to hospital in November, 1939.

FAMILY HISTORY. These two children are of English extraction. Their parents and the other siblings are normal. There is no history of similar children in the immediate or remote relatives.

PRESENT ILLNESS. This child appeared to be normal until two years of age, at which time the family noticed that his development was slow when compared

FIG. 5.—Showing E. A., aged nine years.

FIG. 6.—Showing a close-up of E. A., aged nine years.

with that of the average child. He was examined by the family physician, and thyroid tablets were prescribed and were administered at sporadic intervals for four years. There was, however, no improvement with this medication. In 1935 the boy had a severe attack of measles and later in the year a hernial repair. In 1939 the right patella began to slip laterally, and this orthopaedic deformity increased in severity until he was unable to walk. The early developmental history of this boy seemed normal. He had two teeth at seven months of age. He walked at one-and-a-half years, he said a few words and fed himself at two years. Some deterioration, however, gradually appeared and at three years of age he became unable to talk or feed himself. At the present time he is almost bed-ridden, incontinent and speechless.

PHYSICAL EXAMINATION. On examination he shows some degree of dwarfism, which particularly involves the extremities. He has a large head, coarse, ugly features, dry, coarse hair and coarse, shaggy eyebrows. The skin is dry and thickened: there are no pigmented areas. The secondary sexual characters are not present. The eyes are rather widely separated, and the nose is broad,

with a depressed bridge. The lips are thick and the incisor teeth are separated ; the other teeth are carious. On ophthalmoscopic examination the optic discs are pale, showing nerve fibres, and there are no corneal opacities. The neck is short and thick, the thyroid is not palpable. The chest is broad and symmetrical and the lungs and heart are normal. The abdomen is protuberant, and scars of bilateral herniotomies are present in the inguinal region. The umbilicus is herniated. The lower border of the liver is four finger-breadths below the costal margin in the mammary line. The spleen is not palpable. The extremities are short in proportion to the trunk and the hands are small and spade-like. There is limitation of extension of the elbows and knees and limitation of movement at the shoulders. On flexing the right knee the patella is seen to be displaced laterally and overlies the lower part of the condyle of the femur. On extending the limb it is returned to the mid-position. The reflexes are present and there are no cranial nerve lesions.

MEASUREMENTS	PATIENT	NORMALS (ENGLEBACH, 1932)
Length of body	47 $\frac{1}{4}$ inches	49.5 to 52.9 inches
Circumference of head ..	22 "	20.9 inches
Circumference of chest ..	25 "	24.6 "
Circumference of abdomen	22 "	22.3 "
Span	44 $\frac{1}{2}$ "	49.1 to 52.9 inches
Upper longitudinal measure	24 $\frac{1}{2}$ "	24.7 to 27.1 "
Lower longitudinal measure	22 $\frac{3}{4}$ "	23.8 to 26.3 "
Weight	54 lb.	51.6 to 64.4 lb.

RADIOGRAPHIC EXAMINATION of the vertebrae (fig. 7) shows ovoid bodies, with a shelf-like projection of the anterior margins. The long bones (fig. 8)

FIG. 7.—E. A., showing the ovoid shape of some of the vertebra with the anterior projections of the bodies.

FIG. 8.—E. A., showing the rachitic changes in the long bones and seven carpal centres.

FIG. 9.—E. A., showing the dilatation of the lateral ventricles as demonstrated by an encephalogram.

show normal structure, and the wrists have seven carpal centres at nine years of age. The skull shows a slightly enlarged sella turcica with well-defined clinoid processes. An encephalogram (fig. 9) shows bilateral dilatation of both lateral ventricles with some slight cortical atrophy.

LABORATORY EXAMINATIONS. Red blood-cells, 5,300,000 per c.mm.; Haemoglobin, 85 per cent.; White blood-cells, 6,200 per c.mm.; Polys., 80 per cent.; Lymphos., 20 per cent.

Blood calcium, 9.7 mgm. per cent.; Blood phosphorus, 4.1 mgm. per cent.; Cholesterol, 150 mgm.; Phosphatase, 22 units.

PSYCHOMETRIC EXAMINATION by Kuhlmann's Pre-school Test indicated a mental development of $6\frac{1}{2}$ mos. and I.Q. below 10.

Case 4. F. A., male (fig. 10 and 11), aged nine years, a brother of case 3, was admitted to hospital in January, 1935.

PRESENT ILLNESS. During the first two weeks of life this child had many slight convulsions. At two weeks of age the thymus was irradiated and the convulsions then ceased. The early development of this child appeared to be

FIG. 10.—Showing F. A., nine years.

FIG. 11.—Showing a close-up of F. A., aged nine years.

normal. The first tooth appeared between seven and eight months of age. He sat alone at twelve to fourteen months, said single words at fourteen months, but could not say short sentences until he was over two-and-a-half years of age. He was seen by a physician at that time and a diagnosis of cretinism made. Thyroid was given irregularly for over two years and was accompanied by a short course of pituitary injections. There was, however, no improvement on this treatment. At four years of age the child began to lose the power of speech and the parents thought that he also became hard of hearing. At five years of age he had an attack of bronchopneumonia, and following this his teeth became extremely carious. At the present time the child is drowsy and sleeps a great deal of the time. When roused he is uneasy, restless and difficult to manage. Control of the bladder and bowel are now lacking and he is frequently incontinent. His appetite is variable, at times poor and at others ravenous. His gait is unsteady and he is unable to walk without support.

PHYSICAL EXAMINATION. This boy appeared short for his age and of low mentality. He did not respond to sounds, muttered unintelligibly and violently

resented examination. He had coarse, dry, red hair and the eyebrows were thick. The skin was thick; moderate hirsutism was present over the body and there was slight growth of pubic hair. His head was large and asymmetrical, with some flattening of the right forehead and a prominent bulging over the left ear involving the temporal and parietal bones. A similar prominence occurred on the right side, but to a lesser degree. The orbital ridges were prominent. The eyes did not react to light and accommodation. No corneal opacities were present. The nose was flat and wide, with prominent external nares. The lips were thick and everted. The teeth were markedly carious and the incisors were separated. The lower jaw was more prominent than normal. Respiratory and cardiovascular systems showed no abnormalities. The abdomen was not enlarged and no herniae were present. The liver margin was three finger-breadths below the costal margin in the mammary line but the spleen was not palpable. There was some limitation of movement of the scapulae and also of the knee and elbow joints. The reflexes were normal and there were no lesions of the cranial nerves.

MEASUREMENTS	PATIENT	NORMALS (ENGLEBACH, 1932)
	INCHES	INCHES
Length of body	47	49.5 to 52.9
Circumference of head ..	22	20.9
Circumference of chest ..	25	24.6
Circumference of abdomen	21½	22.3

RADIOGRAPHIC EXAMINATION showed thickened long bones of uniform density. The wrists showed seven carpal centres at nine years of age. The skull showed thinning of the parietal and temporal areas, with slight external bulging. The sella turcica was slightly enlarged. Encephalogram (fig. 12) showed dilatation of both lateral ventricles and poor cortical markings.

FIG. 12.—F. A., showing the dilatation of the lateral ventricles as demonstrated by an encephalogram.

LABORATORY EXAMINATIONS. Red blood-cells, 5,300,000 per c.mm.; White blood-cells, 9,100 per c.mm.; Polys., 74 per cent.; Lymphos., 26 per cent.; Haemoglobin, 88 per cent.

Blood Wassermann, negative; spinal fluid Wassermann, negative.

Von Pirquet tuberculin test, negative.

A formal psychometric test was not done, but the child was grossly retarded and would be classed as an idiot.

This boy was discharged from hospital without further treatment but died at home within four months.

Comment

One feature of these cases which has not been described as yet is the prominence of the skull above and posterior to the ears. On radiographic examination the parietal and temporal bones in this area appear to be quite thin, and this, with the associated prominence, results in a characteristic radiological appearance. This abnormality was particularly noted in cases 1 and 2, and to a lesser degree in case 4.

Conclusion

Four cases of gargoylism have been reported in this article. The first two cases (sisters) conform in all details to the classical description and are considered as complete forms of the syndrome. The latter two (brothers) did not show corneal opacities but conformed in all other respects and are considered as incomplete forms of the syndrome. No direct evidence was obtained to support the belief that gargoylism is due to a disturbance of lipoid metabolism, since none of these cases came to autopsy.

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