thyroid; 41.1% in the lower left; 9.1% in the upper right, and 7.1% in the upper left. In the present case, the adenoma was in the upper left parathyroid, consequently in the rather rare group.

According to Norris, the average duration of the disease before the diagnosis is between 5 and 7 years; here, the duration was of 16 years.

Finally, we would like to emphasize the following point: it is always possible, when one is searching for a parathyroid adenoma, to take an aberrant thyroid adenoma for a parathyroid one, as in the present case. Probably there have been cases in which another operation was necessary on account of that error. That is why, in hospitals where it is possible, the surgeon, before closing the wound, must ask the pathologist for an immediate examination in order to know if the tissues removed are of parathyroid origin; if not, the operation must be continued.

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A CASE OF GARGOYLISM*

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UP TILL NOW, only about 150 cases of gargoylism have been described in literature and the pathological findings were recorded only in a small number of cases. Because of the rarity of this condition, it was thought worth while to record yet another case of this disease, which was characterized by dwarfism, enlargement of the head, skeletal deformities, hepatosplenomegaly, sexual infantilism and mental retardation. Cirrhosis of the liver and portal congestion resulted in profuse hæmatemesis and melæna and death of the patient. Such an unusual termination of gargoylism has not been reported so far in literature. Striking cerebral changes demonstrated on post-mortem examination are also described.

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General considerations.—Skeletal abnormalities, enlargement of liver and spleen, corneal clouding and mental changes were first described by Hunter[1] in 1917 and by Hurler[2] in 1919. The grotesque appearance of these patients led Ellis Sheldon and Capon[3] to name this disease gargoylism after the quaint statues on the water spouts of mediæval Gothic cathedrals. Although in its classical form gargoylism is generally easy to diagnose, a number of cases have been described which do not conform to the typical picture of the disease. Jervis[4] who has made a study of these intermediate or formes frustes of gargoylism describes cases with gargoyle-like facies and mental deficiency, but lacking other features of the disease. However, in some of these cases, there was ballooning and vacuolation of the nerve cells of the brain, identical with the changes found in classical forms of gargoylism. Also atypical cases have been described with no corneal changes and absence of hepatosplenomegaly, and some gargoyles were found to possess a normal mentality.

Kressler and Aegerter,[5] and Ashby *et al.*[6] found vacuolated cells in organs of the body in this disease, resembling in many respects those found in certain lipoid storage diseases. Washington[7] assumed that skeletal deformities of gargoylism were due to the abnormal lipoid deposition hence naming it a lipochondrodystrophy. The same view was shared by Schmidt,[8] who concluded that lipoid storage interfered with the formation of calcified matrix at the epiphyses and also led to impaired osteoblastic activity. Lindsay *et al.*[9] suggest that swelling of the cells and their vacuole-like appearance is due to an abnormal glyco-protein deposition.

However, the assumption that gargoylism is a lipid storage disease, in common with Nieman-Pick's disease (sphingomyelin), Tay-Sach's and amaurotic familial idiocy (ganglioside) and Gaucher's disease (kerasin) cannot be entertained as long as the stored material is unknown. Extensive extractions of the dried tissue by Tanhausser[10] failed to demonstrate any changes in neutral fat, cholesterols, phospholipids, lecithin, cephalin and sphingomyelin in any organs involved in the disease. Strauss *et al.*[11] have confirmed these negative findings in their analyses. Thus, it would be premature to speak of gargoylism as a storage disease until the abnormal substance has been found.

In discussing the etiology of gargoylism,

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Jervis[4] believes it to be due to a single autosomal recessive gene, which is responsible for a defect in a single biochemical reaction. A considerable amount of evidence has now accumulated to show that single genes which are associated with specific hereditary traits control single biochemical reactions. From Jervis' study it appears that a change of an existing gene responsible for certain growth requirements might precipitate the syndrome of gargoylism.

First admission.—Miss G.L., aged 37, French-Canadian was admitted to the Hospital on December 27, 1949 because of a bout of vomiting. The vomitus had contained dark red blood clots and undigested food. She was never sick prior to her admission to the hospital. Her appetite was good, bowels and micturition regular, and she had not lost any weight. However, she has never had any menstrual periods, never experienced a sexual desire and spent most of her time at home reading comics and knitting. She has never been out to work and did only a minimum amount of housework. Born one month prematurely and weighing 8 lb. She appeared normal in every respect. When six months old, for no apparent reason, she started losing weight. At 9 months her big head was noted. When she was one year old, her mother noticed that her left hand was smaller and shorter than the right one, and since about that time her body growth was slow and the physical development retarded. She started walking at 2 years, speaking at 3, and feeding herself when 3¼ years old. She learned to read at 9, three years after going to school, which she attended from 6 years of age until she was 15. At school she was never good at anything. She never participated in any of the school games or in physical exercises. At 19 progressive shortness of vision compelled her to use spectacles and at about the same time she lost all her teeth through decay.

There was no consanguinity in patient's family. Her mother suffered from severe hypertension and some œdema few months before the patient was born. Two other sibs were premature, both born at 8 months of gestation, another sib was born at 6½ months of gestation and lived only for 2 days. This was followed by a miscarriage at 5½ months of gestation. Subsequent examination of the mother did not reveal any abnormality, except for a hypertension of 170/90; Rh testing of the mother was negative. Patient's father died in a railway accident; apparently he was very short sighted since childhood. There was no dwarfism even in the remote branches of the family traced four generations back, and none of the present members of the family appear to be in any way affected.

Examination of the patient revealed a very short, poorly developed female, height 4 feet 5 inches, span 45 inches, weight 75 lb., of a very dark complexion and deep pigmentation under both eyes. The head was large, scapho-cephalic in shape and appeared to be out of proportion to the dwarfish body, to which it was joined by a very short neck.

This unusual appearance was accentuated by a wide forehead, prominent supraorbital ridges and high cheek bones, a protuberant lower jaw, depressed nasal bridge and a complete absence of teeth. All the extremities appeared rather disproportionately short, there was some bowing of the legs, and the left forearm was found to be one inch shorter than the right, as measured from the olecranon. Gastric analysis: normal. Lumbar puncture: Queckenstedt test: normal. C.S.F.: normal.

Radiological findings.—Skull: enlarged, scaphocephalic in shape. There is a very marked thickening of the outer table. The frontal sinuses appear markedly developed, particularly to the right. The pituitary fossa is roofed in and appears to be small in proportion to the large skull. Chest: no abnormality, except for broadening and osteoporotic changes in the ribs. Arms: osteoporosis and cyst at the head of left humerus. Wrists: right wrist—the epiphyseal line is still present at the lower end of the radius and ulna. The left wrist presents an abnormality consisting in wide separation of the radius and ulna, and the ulna being somewhat longer than the radius. The same osteoporosis is noted. Flat plate of the abdomen: generalized osteoporosis of the ribs, lumbar spine and pelvis; enlargement of the hepatic and splenic shadows. Femora and knees: no abnormality, except for the osteoporotic changes. G.I. series: no significant changes.

Eventually the patient left hospital at her own request and before all examinations were completed on January 10, 1950, and on that day she was apyrexial and was feeling well.

Second admission.—She was readmitted on February 2, 1950, because of swelling of abdomen, legs and feet of two weeks' duration. Physical examination revealed free fluid in the abdominal cavity and gross œdema of the extremities. About 4 litres of fluid were removed by abdominal paracentesis. The ascitic fluid, however, reformed slowly and a fortnight after admission she had severe hæmatemesis and melæna. Despite transfusions the R.B.C. dropped from 4,300,000 on admission to 1,600,000 on February 17, with a corresponding fall in hæmoglobin. Jaundice appeared, the patient became comatose and died on February 20, 1950. A clinical diagnosis was made of gargoylism, sexual infantilism and cirrhosis of the liver.

Summary of autopsy findings.—The thoracic cavity, lung and mediastinal structures revealed no abnormality. The heart weighed 180 gm. and was small and flabby but was otherwise normal. The coronary arteries showed very slight thickening by a few scattered, raised, yellowish atheromatous plaques, but there were no areas of stenosis. The aorta was normally elastic. Its intimal surface was raised by a few longitudinally arranged yellowish fatty streaks, particularly in the abdominal aorta. The thyroid was small and weighed only 15 gm. after fixation; cut surfaces showed colloid to be present in scant amounts. One section showed a slight increase in the amount of intra-acinar fibrous tissue. The acini were small, lined by cuboidal epithelial cells, and for the most part empty; only some contained colloid. A few nests of lymphocytes were present between the acini. Two parathyroid glands were located, they were small, measuring less than 0.5 cm. in diameter, but of a normal histological appearance. The abdominal cavity contained about 3,400 c.c. of an amber coloured fluid. Aside from an enlarged liver and spleen the contained viscera appeared to be of usual size, shape and colour and were free from any gross pathological change.

The liver weighed 900 gm. and measured 21 x 18 x 6 cm. The capsule was rough and bore a rather fine nodularity throughout and was of a peculiar light orange-brown colour. Histologically the liver cells were divided into small irregular lobular masses by interlacing fine strands of fibrous tissue. The liver architecture was largely lost and liver lobules were replaced by regenerating masses of liver cords. A fine fatty droplet accumulation was seen in a few irregular zones. The gallbladder, bile ducts, and pancreas were entirely normal. The spleen weighed 350 gm. and measured 14 x 8 x 5 cm., the capsule was smooth, but near one pole there was a bluish-red nodular elevation, about 2.5 cm. in diameter, which on section was found to be a recent infarct. Histologically there was a diffuse fine increase in the amount of fibrous tissue in the pulp. The adrenals, kidneys and bladder were normal. The uterus was small measuring 3 x 2.5 x 1.5 cm. but had become converted to a thin-walled saccular structure containing greenish-yellow, purulent material. One section of the uterus showed marked atrophy, the myometrium being reduced to a thin atrophic muscle and fibrous tissue. The endometrium was represented by a single layer of epithelial cells. No gland crypts were present. A few macrophages and polymorphonuclear leukocytes clung to the lining of the uterus. The Fallopian tubes

were small and were lightly adherent to surrounding structures. Ovarian tissue was very scant in amount and difficult to locate on gross examination. Histologically no ovarian tissue could be found. Two sections from the ovarian region showed only fatty tissue containing a few para-ovarian cystic structures. The mesenteric and retroperitoneal lymph-nodes were normal. The inferior vena cava, common, external and internal iliac and vertebral veins were normal and free from thrombi. Milking of the femoral vein failed to dislodge any thrombus material.

The brain weighed 1,380 gm. Over the lateral surface of the left cerebral hemisphere, centred about the mid point of the motor gyrus, was a zone 8 x 6 cm. in which there was a dark red discoloration of the pia-arachnoid. In this zone thrombosed twig-like vessels were seen. Otherwise cerebral hemispheres, cerebellum, midbrain, pons and medulla were essentially unremarkable on external examination and on slicing, save for slight œdema of the white matter. Cortex and subcortical white matter—left precentral gyrus. The veins of the subarachnoid space were filled with fairly recent thrombus. The subarachnoid space itself was filled with a loose fibrin mesh, in which were masses of inflammatory cells. The majority were polymorphonuclear leukocytes, but mononuclears were also seen. The brain substance showed œdema and a few small blood vessels were filled with recent thrombus. Thalamus and internal capsule—left. A tiny blood vessel contained recent thrombus. Several neurons contained a yellowish-brown pigment in their cytoplasm, but showed no distension. The pituitary was normal.

Specimens of bone removed from the left humerus in the region of the anatomical neck showed some irregular thinning of cortex suggestive of small cystic formations. Bone marrow in this region was dark reddish in colour. one section of the head of the humerus showed marked rarefaction of cortex and cancellous bone. The marrow was fatty. The calvarium was thick and contained red marrow. One section of the calvarium showed condensation of cortical bone. The trabeculæ were thickened and the marrow was actively hæmopoietic.

Differential diagnosis of gargoylism. — The characteristic picture of dwarfism, skeletal deformities, corneal opacities, hepatosplenomegaly and mental retardation not found in any other disease, differentiate gargoylism from such conditions as pituitary dwarfism, hydrocephaly, cretinism, congenital syphilis, achondroplasia, cœliac disease, renal dwarfism, juvenile hepatic cirrhosis, Rh positive children born of Rh negative mothers, Albright's syndrome, and Hand-Schuller-Christian, and Gauchner's diseases. Also in the differential diagnosis of gargoylism one has to consider a very similar disease described by Morqio.[12] Morqio's disease closely resembles gargoylism in its almost identical external appearance, such as dwarfism, monstrous facies, various bony deformities, etc. However, mental symptoms, clouding of the cornea, and hepatosplenomegaly are absent in the latter. Also, the joints in Morqio's disease are supposed to be abnormally movable and there is no limitation of extension or flexion deformities as described in gargoylism. Some authorities believe the two diseases to be identical.

Of the main group of clinical and pathological changes present in our case, one may say that dwarfism, enlargement of the head, skeletal abnormalities, hepatosplenomegaly, mental retardation and sexual infantilism are compatible with a diagnosis of gargoylism or formes frustes of the disease.

The great enlargement of the frontal sinus, marked shortening of one limb, persistence of epiphysis, generalized osteoporosis and cystic changes in the bone form unusual features in this syndrome. Also extensive cirrhotic changes in the liver leading to portal hypertension, hæmatemesis and death of the patient and the striking cerebral changes have not been reported in a case of gargoylism before.

SUMMARY

A case of gargoylism is reported in which cirrhosis of the liver resulted in hæmatemesis and death of the patient. Some unusual clinical features, not described in gargoylism before, are also mentioned.

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CHYLOTHORAX*

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CHYLOTHORAX or chylous ascites, is not common. Goldman[1] gave a comprehensive review, and Everhart and Jacobs[2] gave a review of the literature up until 1939. Of 69 cases reviewed, trauma was responsible in 25, and in the next largest group of 22, secondary neoplasm and tuberculosis. Whiteside, Stewart and Cuthbertson,[3] Beatty,[4] Davis,[5] Shackelford and Fisher,[6] and Cellan-Jones and Murphy[7] all discussed traumatic chylothorax. Nowak and Barton[8] reported

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