

## Gargoylism in a Chinese Boy

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Since Hunter (1917) and Hurler (1919) first described gargoylism there have been a number of cases of this syndrome reported throughout the world. No Chinese case, however, has ever been reported. This is the first report of gargoylism affecting a Formosan Chinese. The case history and results of examinations are presented here.

### Case History

A boy of 5 years and 10 months was admitted to the Department of Neurology and Psychiatry, National Taiwan University Hospital, Taipei, on 11 August 1969. Indications for admission were physical and mental retardation. Slow development, both physical and mental, was observed in infancy, but he has shown obvious deterioration since the age of 3.

The patient is the first child of a Formosan Chinese coal-miner from a small village in north-eastern Formosa. He was the product of a full-term pregnancy. Delivery was normal and spontaneous. The neck was encircled by the umbilical cord, but no cyanosis or delayed crying was reported. Body weight was said to be average, and no significant disproportion of the body was noticed by the parents. He was breast-fed by his mother and the sucking response was reported to be strong.

In early infancy and thereafter frequent diarrhoea and fever were noted. The mother was told by a paediatrician that the fever was due to 'upper respiratory tract infection'. An inguinal hernia, resulting in an egg-sized mass in the right scrotum, was noted at 3 months. The mass was especially noticeable when he was crying loudly. However, the mass disappeared without surgery after a belt was applied to the right scrotum at the age of 2 years.

At 10 months, the patient could support his head. Dentition started at 11 months. An umbilical hernia developed at about this time and is still present now. Before 1 year, the patient responded to loud noises, but this became dulled after the first year, particularly when the sound originated on the right side.

He could sit alone at 1 year. Weaning from the breast began at 2 years, and then the diarrhoea became more frequent and abdominal distension was noticed. He

was able to speak four words clearly at about the age of 2 years, but after the age of 3 the four words were forgotten, and up to the present time, no more intelligible words have been spoken, though articulation of sounds remains good.

At 3 years the patient learned to walk unaided, and could run six months later. Since then he has been described as restless and hyperactive. He showed a strong desire to go outdoors, but he was rejected in his attempts to play with neighbourhood children. If not supervised, he repeatedly fell into ditches and holes. These falls resulted in wounds on his knees and forehead. At about this time he learned to spoon-feed himself, though he did not do so skilfully, and he tended to spread the food on the table or ground. When he had finished eating, he threw the bowl on the floor. He was indiscriminate about objects he placed in his mouth, and attempted to eat non-edible items. Increase in body height slowed after the age of 3. He is reported to be destructive and has not achieved bowel and urine control. There is no history of dyspnoea, cyanosis, corneal opacity, or seizure.

Owing to a sustained high fever at the age of 3 he was taken to a paediatrician, who drew the attention of the parents to the overgrown head and enlarged ears. Retrospectively, the parents felt that the large head and ears had developed gradually from the age of 2. On the advice of the paediatrician, he was brought to our clinic, where an examination showed physical and mental retardation, hepatosplenomegaly, and inguinal hernia. Hospitalization was advised for further examination, but the father was under treatment for hepatitis at that time, and consequently the patient was taken home.

After returning home, the patient's condition remained unaltered until eight months before admission, at which time the diarrhoea became continuous. The stool was green and foul. His mother became concerned when the condition showed no improvement after many months, and brought him to the National Taiwan University Hospital clinic once more in early August 1969.

**Family history.** Family history, encompassing the last three generations, disclosed no one having the same peculiar body configuration as the patient. No mental retardation or death in infancy was revealed. There was no consanguinity between the parents. Both parents were healthy and of normal mentality. Two younger

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sisters of the patient showed no abnormalities. The mother had never aborted and was now in her fourth pregnancy.

**Condition on admission.** The patient was underdeveloped, though adequately nourished. Body measurements were: body weight, 17.5 kg.; height, 95 cm.; sitting height, 54 cm.; head circumference, 54 cm.; chest girth, 55 cm.; abdominal girth, 56 cm. (An average Chinese boy of this age has these body measurements: body weight, 17.09 kg.  $\pm$  1.67; height, 107.48 cm.  $\pm$  4.31; head circumference, 50.67  $\pm$  1.59; chest girth, 53.44  $\pm$  2.13.) Fig. 1 shows his characteristic physical features. The head was excessively large and the antero-posterior diameter particularly so. The eyebrows were heavy and the eyes wide set. Mild exophthalmos was found, but no corneal opacity was noticed. Fundus examination revealed clear disc margins. The nasal bridge was depressed. Lids, lips, and tongue were thick. The ear lobes were prominent. Response to noise was dull, particularly when the sound originated on the right side, but a more detailed hearing test could not be performed. No other significant cranial nerve involvement was found. The neck was short but supple, but the shoulders were usually hunched, giving the impression that the head was sitting directly on the chest. Despite the barrel-shape of the chest, there was no audible heart murmur and no râles. The abdomen was distended and the liver and spleen were enlarged (liver 4 finger breadths below costal margin, spleen 2 finger breadths). A thumb-tip sized umbilical hernia was noted. Genital organs were underdeveloped. The skin was scarred owing to frequent falls and was coarse and dry, and body hair was excessive. Extremities were short and there was limitation of extension of the distal phalangeal and the elbow joints. The hands were short and wide. No cyanosis was observed on the nail beds. Deep tendon reflexes were normal and no pathological reflexes were found.

The patient was restless and hyperactive. His mother finds it necessary to constrain him with a harness most of

the day to prevent him from harming himself or bothering others. He needs to be fed and otherwise cared for. He has achieved neither bowel nor urine control. There is no spontaneous verbalization except for some meaningless noises. He is unable to follow either verbal or gestured orders. Feelings of satisfaction or frustration can be seen on his face. When he wants something that is denied him, he goes to his mother, but does not scream or shout and does not show any other signs of excitement. He sleeps poorly at night. He shows much interest in looking at the cars passing in the street outside the ward.



FIG. 2. Skull x-ray, antero-posterior view. Increase in A-P diameter of the calvarium with marked widening of the diploë especially in the bilateral parietal regions. The pituitary fossa is elongated.

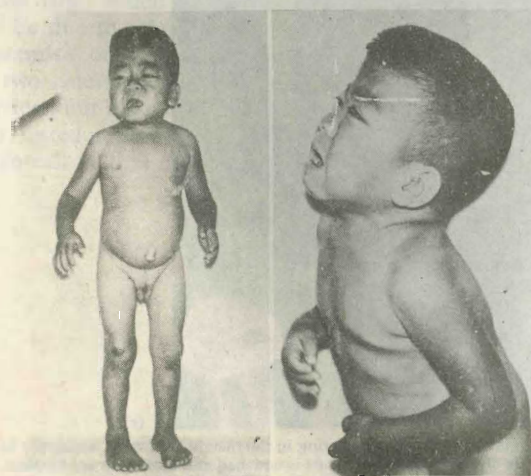


FIG. 1. Characteristic physical features.

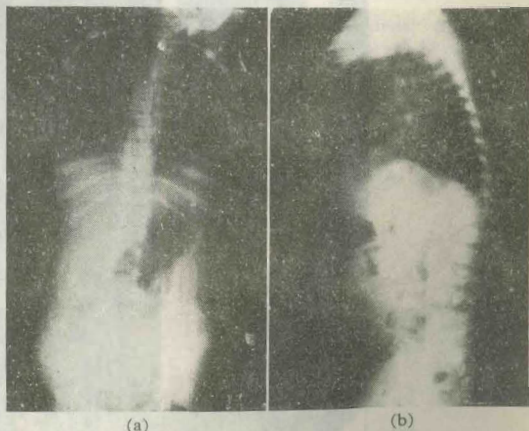


FIG. 3a and b. Chest and spine x-rays, antero-posterior and lateral views. Spine shows kyphosis at L1-L2 with inferior tonguing of the vertebral bodies of L1-L2. Ribs are widened, and the heart is enlarged.

**Laboratory Findings**

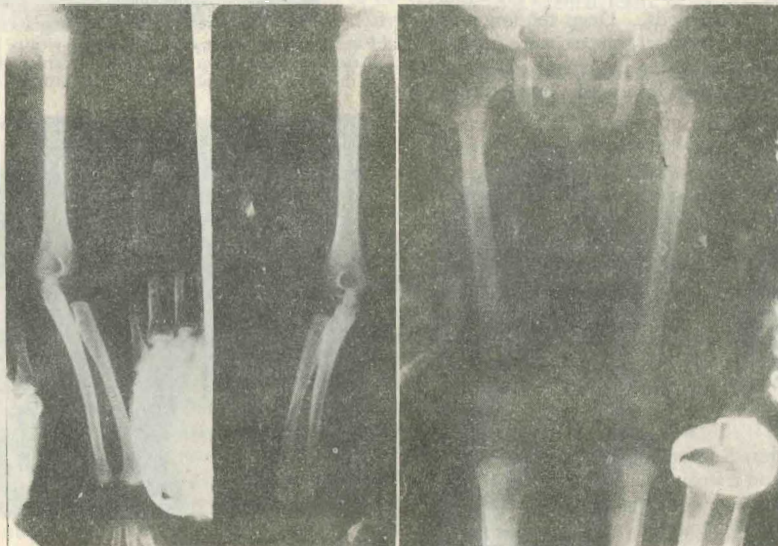
Mental age was 11½ months when measured by the Bayley scale of mental development. During the examination he was very hyperactive and showed an extremely poor capacity for learning. His language development was most seriously retarded.

Routine examination of blood, urine, and stool showed no significant abnormality except for the presence of ascaris eggs in the stool. Blood serological test for syphilis was negative. Blood chemistry, including liver function test and protein-bound iodine (5.1 µg./100 ml.), proved to be within

normal limits. ECG and sleep record of EEG were also within normal limits. Chromosome analysis of peripheral blood leucocytes revealed no abnormality. Fig. 2-4 illustrate and describe characteristic bone deformities. Results of toluidine spot urine test (Berry and Spinanger, 1960) for the patient and his relatives are shown in Fig. 5. The test was performed 'blind'—the origin of each sample was not made available to the examiner. The patient's test showed a strong spot (positive); his sisters' tests a purplish ring ('weakly positive'); and parents and a control negative.



(a)



(b)

(c)

Figs. 4a, b, and c. X-rays of long bones and hands. Long bones are slightly shortened with flaring in the metaphysal ends, especially in the knee and wrist areas. Acetabulum is shallow on each side. Diaphysis of small bones of hands is widened and both ends are blunted. There is also obvious retardation of bone growth under 3 years of bone age. (a) One carpal bone at the age of 3; (b) two carpal bones at the age of 5 years and 10 months.

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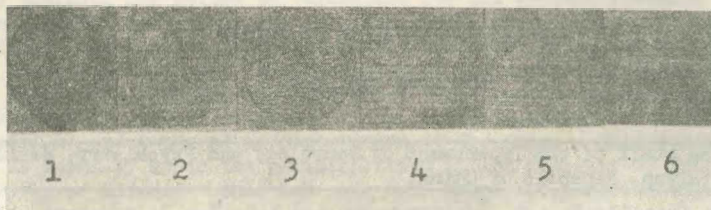


FIG. 5. Results of toluidine paper spot urine test. (1) Patient, showing a strong purple spot; (2) and (3) sisters, a purplish ring; (4) (5) and (6) father, mother, and a control, negative.

### Comments

From the case history and clinical features, as well as the laboratory analysis, one must conclude that this patient is a gargoyle. Reports of gargoylism have mainly been divided into two major genetically distinct groups. In type I (Hurler's disease), transmission is by an autosomal recessive gene; males and females are affected equally; consanguinity is common, and corneal clouding and dwarfism are present. In Type II (Hunter's disease), transmission is by a sex-linked recessive gene, and though dwarfism may be present, the preceding characteristics are not usually observed, but the individual may be deaf, and hyperactivity is common, as well as longer life expectancy. In this case the patient is male, with the female sibs normal; there is no consanguinity between the parents; there is no corneal clouding; dwarfism is minimal; hearing is impaired; hyperactivity is noted; and the patient survives. On this basis, then, this case may be regarded as Hunter's disease with a sex-linked recessive inheritance. The toluidine paper spot test showed a strong purple spot (metachromasia) for the patient. This was due to the presence of chondroitin sulphuric acid (Brante, 1952). It is quite strange that the tests of both sisters showed a purplish ring, which might be weakly positive or might be due to other non-specific elements. On the premise of sex-linked recessive inheritance, these two sisters with weakly positive findings may be carriers, but the mother, who is supposed to be a carrier, tested negative. Tests to detect the carriers through cell culture of skin fibroblasts (Danes and

Bearn, 1967) were not performed, since facilities for culturing skin fibroblasts have not yet been set up in this hospital.

### Summary

This paper represents the first report of a Formosan Chinese boy with gargoylism. The patient shows characteristics common to Hunter's disease probably inherited through a sex-linked recessive gene. The patient's urine when tested with toluidine blue showed metachromasia, and the same test on his two sisters showed 'weakly positive', indicating the possibility that they are carriers. A fibroblast culture to detect carriers was not available at the time this work was done.

We would like to extend special thanks to Dr. Jean C. Y. Hsu of the Radiological Department, National Taiwan University Hospital, for her help in re-examining the x-ray film of this case, and to Dr. Shing Ming Sung of this department for his assistance in the toluidine spot urine test.

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