ADDENDUM

REPORT OF AN ADDITIONAL CASE OF GARGOYLISM

BY

RICHARD W. B. ELLIS, M.D., F.R.C.P.

The following case of gargoylism is reported as an addendum to Dr. Henderson's paper, as it illustrates a number of the clinical and radiological features of the condition referred to in the text.

L.C., a girl, aged two years at the time of examination (June, 1940), was born at term by an easy low-forceps delivery. Her parents and grandparents were normal, and no similar condition was known in either family. There was no consanguinity of parents. There had been a previous miscarriage at ten weeks; no other children.

The patient weighed seven and a half pounds at birth; the umbilical cord was said to have been exceptionally large. She was breast fed for three months; breast feeding was discontinued owing to failure of the baby to gain weight after two months old and because of the mother's 'nervous state.' The baby was subsequently reared on dried milk, followed by mixed feeding. She sucked well in spite of severe snuffles and purulent nasal discharge dating from birth. She cut her first teeth at thirteen months of age, and walked alone at twenty-three months. She had been well except for colds and cervical adenitis during the first year. At the time of examination, she had been clean both by day and night for some months. She was feeding herself, holding cup and spoon, and was beginning to talk. She was said to be 'very observant' and would always go to fetch her father if her mother appeared in a new dress! She was obviously interested in her own clothes, and though frightened of strangers, played normally with her mother and understood simple requests. She recognized friends and animals with which she was familiar.

On examination (June, 1940). The child appears of normal, or almost normal, intelligence for age, though she is intensely resentful of examination by strangers. She is of average height ($32\frac{1}{2}$ in.) and weight (27 lb.) for age (fig. 1).

THE HEAD appears slightly larger than normal, but is not obviously hydrocephalic. Circumference $19\frac{1}{8}$ in. (average normal $18\frac{1}{2}$ in.). Intermeatal measure (over vertex) $12\frac{1}{2}$ in. The anterior fontanelle is patent. The hair is light brown in colour, and rather thick and coarse. The frontal veins lie in deep grooves in the frontal bones. There are fourteen teeth present.

THE FACIES is typical of the condition, the features having a peculiarly gross and heavy appearance. The lips are large and thick; there are deep nasolabial grooves, heavy cheeks, and depression of the nasal bridge, with profuse purulent nasal discharge. The eyebrows are sparse, coarse and black.

THE EARS are somewhat flattened and set unusually low.

THE EYES show bilateral clouding of the corneae, which though less marked than in many previously recorded cases, prevents satisfactory examination of the fundi. Visual acuity appears moderately good, though accurate testing is impossible.

THE ABDOMEN is distended and there is a small umbilical hernia. The liver is enlarged two finger-breadths below the costal margin, and the spleen is just palpable. Genitalia normal.

SKELETAL SYSTEM. There is an angular kyphosis in the region of the first lumbar vertebra and a deep dimple in the mid-line of the back immediately below it (fig. 2).

FIG. 1.—L. C., showing facies and characteristic flexion deformity of left hand.

FIG. 2.—Lumbar kyphosis.

The thorax is normal except for slight flaring of the lower ribs. The scapulae are normally placed.

There is some limitation of extension of the elbows, knees and hips, and more marked flexion deformity of the interphalangeal joints of the hands. There is a deep dimple over the head of the radius on either side.

Radiological examination

SKULL. The anterior fontanelle is patent, and the coronal sutures abnormally wide. The sella turcica appears shallow and elongated; the clinoid processes are not clearly defined, but there is no evidence of erosion.

SPINE. The body of the first lumbar vertebra is diminished in size and its upper and anterior portion is defective, giving rise to an angular kyphosis in this region (fig. 3).

RIBS. The lower ribs are expanded anteriorly.

HIPS. Both acetabula are poorly formed (fig. 4), the roof being slanting. The femoral necks are valgoid and there is partial subluxation.

RIGHT SHOULDER. The glenoid fossa is poorly formed, and the head of the humerus flattened.

FIG. 3.—Spine showing deformity of body of first lumbar vertebra and kyphosis.
Anterior expansion of ribs.

FIG. 4.—Hips.

FIG. 5. Hands. Widening and irregularity of metacarpals and phalanges.

HANDS. The metacarpals and phalanges show the widening and irregularity of outline characteristic of the condition. The interosseous spaces are increased in size (fig. 5) and the epiphyses small.

Comment

Although the changes are not so marked as in many of the published cases, this patient shows the following characteristic features of gargoylism: the peculiar facies, which in itself is diagnostic; hepatosplenomegaly; chondro-osteodystrophy, giving rise to flexion deformities, particularly of the hands; defect of the body of one of the upper lumbar vertebrae, resulting in kyphosis; clouding of the corneae; depression of the nasal bridge, with purulent nasal discharge; an elongated sella turcica; low-set ears; and hernia. Minor stigmata which I have also seen in other cases are the deep grooves lodging the frontal veins, and the sparse eyebrows composed of coarse dark hairs. A point in which this child differs from most of the classical cases is in her mentality. This appears almost normal for her age, and though it is too soon to predict that mental deterioration will not occur, the prognosis seems relatively more hopeful. Since the familial incidence of the condition is established, it was necessary to warn the parents that, even in the absence of consanguinity, there was a possibility of a subsequent child being affected.