THE RELATION OF HYPERTELORISM TO MONGOLISM;

With Description of a Case.

BY

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Since D. M. Greig⁽¹⁾ first described the cranial deformity called by him ocular hypertelorism seven other cases have been recorded. The two most recent are those of F. M. B. Allen⁽²⁾ and W. B. Drummond⁽³⁾. The most salient and constant feature recognisable clinically is the great breadth between the eyes. Other characteristic signs are mental deficiency (present in eight of nine cases), brachycephaly, the presence of a median groove in the frontal bone, and depression of the bridge of the nose.

The following case, although not very marked, shows some interesting points:—

G. L., girl, aged 15 months, was brought up to the out-patient department for backwardness. She could not stand, talk, nor feed herself, and she made no attempt to play with toys. The parents were healthy, and had had one other normal child.

The striking points about the appearance of the child were the widely separated eyes, the flat face, and the receding chin. The distance between the inner canthi was 1·2 ins., and that between the pupils 2 ins. The orbital height was 1·25 ins., the orbital width 1·5 ins.; the orbital index was therefore 83·3 or microseme. The eyelids were thick, and the lower lids everted.

The skull was flattened behind and looked brachycephalic. However, the length was 6.4 ins., the breadth 5 ins., and the cephalic index 76.9, or mesatocephalic. The circumference of the head was 18.75 ins., the frontal eminences were prominent, and there was a vertical groove running down the middle of the frontal bone. The hairy scalp showed a V-shaped peak in the middle line. The anterior fontanelle was widely open, the length being 2.5 ins., and the breadth 2.6 ins. The ears were large and stood out from the side of the head. The bridge of the nose was depressed, and the nose itself small. The length of the face was 3.3 ins., and the breadth 4.5 ins. The tongue was apparently normal. The palate was highly arched, and the child was a mouth breather. Eight teeth were present, the central and lateral incisors. They had appeared at the eighth month.

The fingers were thin and tapering, and the little finger was normal. The toes were similarly shaped to the fingers, but the gap between the hallux and the second toe was considerably greater than that between the other toes. There was slight cyanosis of the feet. The abdomen was prominent, and there was a large umbilical hernia. Muscle tone was markedly deficient; the child could put her head between her heels like a case of amyotonia congenita.

The costochondral junctions were enlarged, and there was a rickety kyphosis of the dorsal and lumbar spine. A skiagraph of the wrists showed marked rickety changes.

Hearing and sight were apparently normal. The fundi oculorum showed no abnormality.

A skiagram of the skull showed that the clinoid processes were rather close together, and that the sella turcica was a little on the shallow side. "It is probably not justifiable to assume that this is beyond the limits of the normal." (Dr. A. E. Payne.) The blood sugar three hours after a meal was 0.07%.

In this case there are certain peculiarities which are usually associated with mongolian deficiency. Perhaps the most striking is the abnormal gap between the hallux and the second toes. In addition there is mental dulness, amyotonia, flatness of the face, depression of the bridge of the nose, smallness of the nose, flattening of the occiput, projecting ears, widely open fontanelle, and a highly arched palate.

Study of the previously published cases shows that many of them had characteristics usually associated with mongolism. Thus, Greig's first case had a flat nose and a fissured tongue. Also she was strikingly fond of music, which is very characteristic of mongolism. She had tubercular adenitis of the neck, and died of pulmonary tuberculosis.

His second case had a depressed nasal bridge, an oblique right palpebral fissure, a flattened occiput, a highly arched palate, and the little fingers were disproportionately small. She also had tubercular adenitis of the neck, and died of spinal caries.

Muir's⁽⁴⁾ case showed a highly arched palate, flattening of the occiput, and large ears. This case also showed flattening of the pituitary fossa by radiography.

Of Cockayne's⁽⁵⁾ two cases, the first showed flattening of the occiput, while the second, a boy, had only one small testicle in the scrotum. In addition his fingers were short and square, and there was dwarfing of the big toe.

Drummond's case showed flattening of the bridge of the nose, a high and narrow palate, a fissured tongue, while her hands "reminded one of those of a mongol."

Allen's patient, also a boy, was a cryptorchid. His palate was high and arched, and the bridge of the nose was depressed.

No doubt most of the bony peculiarities are due to the malformation of the sphenoid which, according to Greig, is the primary lesion in these cases. But even so, the presence of so many common characters is suggestive that the pathology of the two conditions overlap, and that an explanation of one would help considerably in the understanding of the other.

In this connection it is interesting to note that both the male patients suffering from hypertelorism were cryptorchids. Jodiche⁽⁶⁾ studied the serum of mongols by Abderhalden's sero-diagnostic method, and found evidence of degeneration of a number of organs, especially the reproductive glands. That the reproductive glands are not always affected in hypertelorism is, however, suggested by the normal sexual development and menstrual function of Greig's first case.

If there is a common lesion in mongolism and hypertelorism, it seems most likely to be in the pituitary gland. A deformed sphenoid might conceivably be either the cause or the result of dyspituitarism, and this would explain the disturbance of growth and infantilism.

The idea that the pituitary is to blame in mongolian deficiency is no new one. Schüller⁽⁷⁾ in 1907 pointed out that mongolism was in many respects the direct opposite of acromegaly, and suggested that the condition might be called "acromiery." He also called attention to the frequency of cryptorchidism in this affection. Timme⁽⁸⁾ found an excavation under the anterior clinoid processes communicating with the sella turcica in 23 out of 24 cases of mongolism, which was not present in normal children. Clift⁽⁹⁾ working with a larger number of cases, was unable to confirm this. He found that the condition was just as common in normal children, but his work by no means put the pituitary out of court, as he observed that normally the anterior clinoid processes are widely separated, and can only be involved in such extraordinary enlargement of the pituitary body as is found in tumours.

Of the other theories of mongolism, that of Stæltzner⁽¹⁰⁾ is interesting. He regards it as a result of thyroid exhaustion of the mother during pregnancy. He reports three cases of myxædematous women who became pregnant. In each case the child was born with markedly mongoloid characteristics.

While it is not suggested that either hypertelorism or mongolian deficiency is due to a primary lesion of the pituitary body, it does not seem unlikely that some secondary affection of this gland is responsible for those phenomena common to both conditions.

SUMMARY.

1. A mild case of hypertelorism is reported.
2. The presence of characteristics usually associated with mongolism in this and other cases is discussed.
3. It is suggested that dyspituitarism is a lesion common to the two conditions.

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