HEREDITARY HYPERTELORISM WITHOUT MENTAL DEFICIENCY

BY

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Some years ago Greig¹ described 'a hitherto undifferentiated congenital cranio-facial deformity.' To the condition he gave the name 'Ocular hyper-telorism.' The salient peculiarity of the condition is the great breadth separating the inner canthi. The abnormality results from the mal-development, commencing in early embryonic life, of the portion of the sphenoid which is laid down in cartilage. The great wings are undersized, and the lesser wings are hypertrophied and pass outwards to produce the characteristic changes in the face and skull. Some consequent degree of mental deficiency would be expected to coexist. Mental defect is a frequent concomitant of imperfect sphenoidal development. Strabismus is commonly associated with the disorder.

In the case which I have the opportunity of describing, the cranio-facial distinctions are marked but mental deficiency is absent.

Marion B., (Fig. 1), aged two years and one month, was admitted to my wards at Booth Hall Infirmary for Children, Manchester, on the fourth of June of this year suffering from acute bronchitis, a condition to which she is especially disposed.

FIG. 1.

Family History. The parents were not blood relations. The mother was hypertelorism. Although active and sensible she was very emotional. She was thirty-nine years of age when the patient, her first child, was born. Unfortunately I was unable to see her for more than a few moments as she was literally hurrying to hospital to be confined. And, again it is regretted, the mother's death took place a few days later from a puerperal complication. There is no photograph of this parent extant, but, in the words of the father, 'When you look at Marion's face you look at her mother's also.'

The second child, a male, presents no anatomical peculiarities. There is no history of miscarriage. The father was forty-nine years old at the time of the birth of the patient. He is alert and of fair intelligence. There is some evidence of old rickets. Except for the emotionalism of the mother there is nothing indicative of nervous stigmata or instability.

The pregnancy was normal and the child was born without undue difficulty. The weight at birth was two kilogrammes.

FIG. 2.

The child is 81 cm. in height and weighs 11.8 kgm. She is able to walk well but stands with the weight of her body on the left foot and the right leg slightly abducted (Fig. 3). There is some right genu valgum. The epiphyses are enlarged and the radiologist reports that the X-ray of the wrist shows early evidence of rickets. The bones of the skull are thin. The skin is sallow but smooth. There is no acrocyanosis. While the hands and feet are plump they show no disproportions. Subcutaneous tissue is fairly well marked (Fig. 3). A small umbilical hernia is present. The face appears symmetrical. It is interesting to mention that Lightwood and Sheldon² describe a case of right-sided (unilateral) hypertelorism, and Fridolin³ published a case of plagiocephaly which Greig considers to be partly hypertelorism.

Although at times the facial expression is bright and happy it is generally one of apathy. The mouth is open, but this is probably related to the presence of hypertrophied tonsillar and adenoid tissue. The forehead is broad and the frontal eminences are unusually prominent. There is some flattening of the occipital region but the vertex is only slightly raised (Fig. 2). Her eyes are placed widely apart. There is no ptosis nor definite strabismus. A longitudinal mesial depression, 30 mm. in length, passes to the tip of the broad nose. The nasal bridge is flattened and the tip is retroussé. On examining the mouth it is found that the teeth are good but the incisors have the appearance of molars, the palate is not unduly arched, and the tongue is active and not enlarged. The anterior fontanelle can be felt, but is closed. Sight, hearing

FIG. 3.

and articulation are good and the vocabulary is no more limited than would be anticipated in a child of two years. The second mitral sound is accentuated but no murmur can be detected.

Measurements of head: The maximum horizontal circumference of the skull, measured round the most prominent parts of the glabella and occiput, is 480 mm. The greatest length, estimated from the most prominent part of the glabella to the most prominent part of the occiput, is 150 mm. The greatest parietal breadth, taken behind and above the temporal line, is 120 mm. The skull is thus brachycephalic, the cephalic index being 80. The facial length, from pogonion to nasion is 70 mm. The maximum bizygomatic width is 115 mm. The total facial index is thus 61. The highest point of the vertex is 70 mm. above the external auditory meatus. The distance between the inner

canthi is 40 mm. The two sets of orbital measurements are identical. The supraorbital notch is 35 mm. from the sagittal line. The greatest width of the orbit is 40 mm. and the greatest height 30 mm.

Mental condition: The child is extremely emotional. The appearance of a stranger at once excites and frightens her. She weeps on any provocation. She prefers to sleep upon the floor, and by her father alone can she be prevailed upon to go to bed.

With the exception of these features no evidence of impaired mentality can be detected. There is no history of convulsions.

DISCUSSION.

The important features of this case are the indications of hereditary origin of the anatomical peculiarities and the absence of mental deficiency. The ages of the parents at the time of the patient's birth are noteworthy.

Ogilvie and Posel⁴ cite a case of oxycephaly whose mother and mother's cousin, also a female, showed similar characteristics. The more remote relative is reported to be mentally deficient.

A hypertelorid child without mental defect is described by Abernethy⁵. The mother, the mother's brother, and possibly the maternal grandfather show evidence of ocular hypertelorsim.

Mental deficiency and hypertelorism were present in a case quoted by Reuben and Fox⁶. Two cousins had hypertelorid features and the writers emphasize a familial tendency to myopia.

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