HYPERTELORISM IN SEVERAL GENERATIONS.

BY

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Hypertelorism as defined by Greig in 1924 is a cranio-facial deformity of the sphenoid and nasal bones, often associated with mental deficiency and abnormalities of other parts of the skeletal and muscular systems.

The case which I am about to describe is in certain aspects peculiar, for in it there is no evidence of mental defect while there is good evidence of the inheritance of the cranial deformity through several generations. For the use of this case I am indebted to Mr. Dodds Parker, to whom the boy was brought on account of an inguinal hernia.

Fig. 1. Boy, aged 10, showing hypertelorism facies.

Fig. 2. Patient's mother and baby sister.

Fig. 3. Patient's maternal uncle.

Fig. 4. Patient's maternal uncle as a baby.

Case Record.

T. M. (Fig. 1), a boy aged 10 years was admitted to the Radcliffe Infirmary for the cure of a right inguinal hernia. On examination he was found to show the typical signs of hypertelorism.

He is the eldest of six children; the other five are all girls, the eldest of whom is 8 years of age. Of these one only, the youngest, shows a similar abnormality.

The *family history* is of distinct interest as the maternal grandfather, the mother and the mother's brother were all recognized by their friends as having their eyes abnormally widely spaced. This is quite noticeable in the photographs of the mother (Fig. 2), who is shown with the youngest baby, and in that of her brother (Figs. 3 and 4), but the photograph of the grandfather showed but little abnormality. There is no history of any cranial abnormality in the father's family.

The pregnancy ran a normal course: the child was breast-fed for five months. He had measles at 18 months and mumps at 4 years of age, but no other illnesses.

The peculiar breadth between the eyes was noticed in early infancy and was attributed to inheritance from the mother's family. The parents are not blood-relations.

General condition is excellent. The skin is soft and healthy: the subcutaneous fat is not abnormal in amount or distribution. His muscular development is good: he is a "Wolf-cub" and plays the ordinary games of boys of his age.

Mental Condition. The child seems to be remarkably intelligent: he attends school and is in Standard IV in which the average age of the boys is about 10 years. He finds his school work neither particularly easy nor too hard. He is interested in locomotives and knows the names and the functions of their various parts. He draws very well, much better than most boys of his age.

Fig. 5. Skiagram of patient's skull

Physical Examination. Age 10 years, height 133.1 cm. The skull is somewhat square, and broader behind than in front, the greatest breadth being the bi-parietal, 147 mm. The forehead is broad and approximates to the vertical. The upper facial index is 46, the lower 77. The occiput is broad and steep and the lambda rather high. The glabello-occipital length is 179 mm. and the minimum frontal diameter 105 mm.

The fundi oculorum are normal. There is slight ptosis of the upper lids of both eyes, slight nystagmus and external strabismus. Binocular vision is present. The external ocular breadth is 87 mm. and the internal 47 mm., the intra-orbital width is 19 mm., the external orbital breadth is 99 mm.

The ears are normal in shape and lie close to the head. The bridge of the nose is broad and flat but the organ is well shaped below, the nostrils being small and regular in outline. The nasal height is 40 mm., nasal length 35 mm., and breadth 27 mm. There is no nasal obstruction. The inter-zygomatic breadth is 124 mm. and the inter-malar 96 mm. The upper face length is 57 mm., and the total face length 95 mm. The nasal index is 67 and the cephalic index 82.

The tongue is normal in size, shape and activity. Of the teeth, four sound permanent incisors are present above and below, the permanent canines are erupted in the lower jaw. The first pre-molar is present on both sides in the upper jaw. All the remaining milk teeth are carious in some degree. The palate is narrow and highly arched: the tonsils show no abnormality. There are no enlarged lymphatic glands nor other abnormality in the neck. The heart and lungs are normal. In the abdomen the only abnormality is the presence of a right inguinal hernia, congenital and reducible, with a small hydrocele of the tunica vaginalis of that side. There is no hernia nor protuberance of the umbilicus which lies mid-way between the pubic symphysis and the xiphisternum. There is no phimosis, and both testes are in the scrotum. The arms and legs are well formed, symmetrical and muscular. The fingers are normal in shape, neither stumpy nor thickened, though the hands are somewhat broad. Both feet show a minor degree of pes cavus, and both great toes are rather unusually far distant from the second digits. There is a very slight webbing of all the toes but no sign of this in the hands. X-ray photographs of the hands and feet show no such abnormalities as were found in Greig's and Cockayne's cases, the broadness of the hands being accounted for by the soft parts. The tendon reflexes in the arms are brisk, those in the legs normal, except for the knee-jerks which are sluggish. The plantar responses are flexor. Deep muscular pressure pain and tendon pressure pain are practically absent in the calves. The Sigma reaction is negative. The X-ray photograph (Fig. 5) of the skull corresponds with what would be expected from the cranial measurements, but the bony deformities described by Greig and recognised as characteristic of the condition are present, if at all, in a minor degree. The short broad nasal bones, however, the massive sella turcica and the unusual size and density of the lesser wings of the sphenoid are sufficiently definite in this case to put the diagnosis beyond doubt.

DISCUSSION.

The main point of interest in this case lies in the apparently indisputable hereditary transmission of the deformity through three generations of both sexes, unaccompanied by stigmata of mental deficiency.

Further, this child and his hypertelorid maternal uncle share a fondness for and a marked proficiency in draughtmanship, which is not found in any of the other ateloric members of the family.

While it is possible, as Greig suggests, that the advanced cases of the disease are the sporadic results of degeneracy in the stock producing them, there are no obvious grounds for applying that hypothesis to these milder cases which present no mental or physical infirmity.

Braithwaite suggests that the sphenoidal deformity may in mongolism be either the cause or the result of dyspituitarism, and further that a lesion of the pituitary gland may be a factor common to hypertelorism and mongolism.

In the case under review there is no evidence of any functional pituitary lesion, although marked sphenoidal deformity is present.

The present case, though it throws no light upon the causes of mongolism, clearly shows that dyspituitarism is no necessary consequence of sphenoidal deformity: it suggests rather that hypertelorism and mongolism are probably not inter-related, the former being perhaps primarily of endocrine origin, the latter a somatic deformity.

The justifiable inference seems to be that in these cases we are dealing with a Mendelian "sport" which, as it is not sufficiently removed from the normal to impair the efficiency of the organism has, like the majority of such sports become fixed, and so a normally hereditary Mendelian asset. For obvious reasons the grosser examples of the malformation fail to be propagated: these may be the results of racial degeneracy which is, perhaps, the most obvious factor in the production of mentally and physically abnormal individuals.

At the same time abnormal members do occur in the soundest stocks, and it is possible that the shape of the sphenoid, like the number of digits, may be developmentally one of the "weak spots" which, although normally fixed, are more liable to extra-normal variations, whether advantageous or disadvantageous to their possessors, than the other members of the body.

I wish to thank Mr. Dodds Parker for permission to publish this case, and Sir Archibald Garrod for his kindness in putting at my service the literature of the previously recorded cases. I am indebted to Mr. Dudley Buxton for his anthropometric study of the boy's head and other measurements, and to Dr. Sankey for the taking and reproduction of the X-ray photographs.

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