MONGOLISM IN ONE OF TWINS.

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It has been suggested that mongolism results from damage to the germ plasm from gametic mutation or the fertilization of an abnormal ovum, the theory being mainly based on the study of twins, one or both of whom has been found to be mongoloid (Benda, 1947).

Although some authors claim that mongolism in twins is more frequent than might be expected, Graham (1950), describing an instance of mongolism in one of twins, emphasized the comparative rarity of such reports in British literature and was able to find only two references to the condition in the preceding twenty-two years (Armstrong, 1928; Russell, 1933). He has since been cited by others, but the contribution of Penrose (1934) was overlooked. Subsequently Crozier and Campbell (1950) reported a case from Ireland, while in Australia two cases were recorded by Cook (1950) and one by Robertson (1952). Engler (1952) has recently reported three more English cases, but does not state the sexes.

The condition is thus uncommon and of considerable interest. In the United States, Warner (1949) collected from the world literature 101 sets of twins, and added a further example of his own of a normal boy and a mongol girl; in twenty cases both twins were mongoloid. To these should be added the cases of Graham (1950), Crozier and Campbell (1950), Cook (1950), Robertson (1952) and Engler (1952), bringing the total to 90 sets where one twin was affected.

The first report of a normal female with a mongol male twin is that of Neumann (1899), and since then 20 cases have been described. Another example will now be given.

Case 1.

The father was born in 1903, the mother in 1906, both being highly intelligent. The family history is clear of psychosis and mental deficiency. There is a history of twinning on the maternal side but none on the paternal. The parents married in 1927 and there are two normal children born in 1928 and 1932. There were no miscarriages, and there was no gynaecological pathology. In the fifth month of the last and significant pregnancy the mother suffered from anaemia, breathlessness and oedema of the feet associated with considerable anxiety and depression, in contrast with the other pregnancies, which were normal. The twins were born in 1944, first the girl weighing 7½lb. as a vertex presentation, the boy 6 lb. 7 oz. 25 minutes later as a breech. There were two placentae. The boy is a typical

mongol of imbecile grade. The skull is brachycephalic with flattening of the occiput. The eyes show epicanthus, internal strabismus and nystagmus. His milestones were delayed, he sat up at 18 months, walked at 24 months, developed sphincter control at 3 years and formed speech at 4 years.

Both twins were examined at the age of 6$\frac{8}{12}$ years in 1951. The mongol had an I.Q. of 34 on the Terman Merrill Form L Scale and a mental age of 2$\frac{4}{12}$ years. His vocabulary was limited to a few simple monosyllables and he could not attempt the Mannikin Test or the Seguin Form Board. Socially he was more advanced by reason of an exceptional home and his Vineland Social Maturity rating was at the 4-year level. His sister was of normal intelligence, with a mental age of 7 years and an I.Q. of 102. Her Seguin Form Board score was 7 years.

Mongolism in one of twin girls was first described independently by Halbertsma (1923) in the United States and by Moro (1923) and Siegert (1923) in Germany. Fifteen further examples have subsequently been reported, to which the following should be added.

Case 2.

The father, born in 1879, is normal and still in fair health; his brother and sister are the parents of normal twins. The mother likewise was born in 1879 and died in 1931 of hemiplegia. It is of interest that her brothers were too dull for enlistment in the 1914–1918 War. The parents were first cousins and married in 1900, having nine children up to 1920, all of whom were of normal intelligence; there were no miscarriages. The tenth pregnancy, which was normal and happy resulted in the birth of female twins in 1922. Labour was precipitate; the mongol was born first and her sister four hours later. There were two placentae. The mongol was deeply cyanosed at birth and her milestones were much retarded; she was admitted to an institution in 1933. The patient is now 4 ft. 2 in. in height and weighs 4 st. 8 lb. The facies is characteristic, there is epicanthus, fissured tongue, low set ears and brachycephalic skull; her eyes are blue and her hair is fair. The little fingers are short and incurved and her feet show a large cleft between the first and second toes. There is a harsh systolic murmur over the praecordium, most pronounced at the mitral and pulmonary areas. Mentally she is an imbecile, with a Terman Merill Form L mental age of 3$\frac{1}{2}$ years, and an I.Q. below 30. She cannot give her age or birthday and is unable to name common objects at the 3-year level. The twin sister is of normal appearance, and unlike the mongol has brown eyes and dark hair. Her intelligence is average and she has a normal child.

Mongolism accounts for 5 per cent. of oligophrenia of all ages, and no aspect of the subject has attracted more attention than causation Twin studies are important and it is to be hoped that more will be reported, for although over 80 cases had been recorded up to 1947, Benda concluded that only 63 of these were reliable and could be used in discussion. The occurrence of mongolism in both of dizygotic twins (Russell, 1933; MacKaye, 1936) has made the theory of germinal mutation improbable, as this could hardly be expected to occur simultaneously in two different ova; it does, however, tend to support the theory of environmental factors operating at or soon after fertilization (Benda, 1947).

The above cases well illustrate certain of the factors which have been alleged to be of aetiological significance (Benda, 1947; Tredgold, 1952). Case 1 shows two of these; firstly the importance of a period of sterility between the birth of the last child and the mongol, which in this patient was twelve years, and secondly the appearance of anxiety, depression and illness during the pathological pregnancy itself.

In case 2 there was consanguinity, a family history of mental retardation,

followed by the birth of discordant dizygotic twins in an elderly woman after a long series of pregnancies. The child in Case 2 evidenced congenital heart disease, the increased incidence of which in mongolism is well recognized (Benda, 1947; Wylie, 1949; Evans, 1950).

The first description of mongolism in one of twins was given by Fraser and Mitchell (1876) in the columns of this Journal. In addition to the cases mentioned by Graham (1950), subsequent British reports were made by Shuttleworth (1909), Coupland (1922), Jewesbury (1925), and Myers (1925). These, including the two patients mentioned above, amount to 14 sets of twins compared to a total of 78 sets reported elsewhere. As the British Isles have a population of only about 50 millions, it is suggested that the emphasis on the rarity of this occurrence in Britain is incorrect, for the incidence is not unduly low taking into consideration the relative sizes of the populations involved.

Conclusions.

(1) It is submitted that the occurrence of mongolism in one of twins is not so rare in the British literature as has been suggested.

(2) The relevant literature is briefly reviewed.

(3) Two further cases are reported.

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