CASE REPORTS

PARAPLEGIA AND MONGOLISM IN TWINS

BY

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Clinical record

M. G., aged four years and six months, was an undoubted mongol, showing the typical signs of round head, slanting eyes, broad face and incurved little finger. She had an internal strabismus and chronically inflamed conjunctivae. She suffered from a paraplegia, a condition rarely seen amongst mongols. On examination of her nervous system the following data were obtained: There was marked spasticity of the legs, especially of the ankles. She walked unsteadily in the equino-varus position, the heels only occasionally reaching the ground. On attempting to rise off the floor she showed a marked 'cuisse-tronc' sign and then could only raise herself by getting on to all fours. Her arms were normal as to muscular tone, reflexes and voluntary movements, but showed a slight degree of choreic movement. Her legs were hypertonic, paretic, showed exaggerated tendon reflexes, the knee-jerks being obtained from well down the shins (i.e. Strumpell's sign was positive). The plantar reflexes were extensor, but the Oppenheim reflexes were negative. No clonus was obtained. There was no evidence of cerebellar dysfunction.

Physical examination of the twin sister

On enquiry it was found that she had a twin sister (G. G.) who was declared by the mother to be normal. This child was brought by her mother for examination, and although she might not be remarked as a mongol when seen casually, several experienced observers agreed that she should be diagnosed as such, although the characteristics were certainly less pronounced than in the case of her sister. A careful examination of her nervous system showed only a slight degree of choreic movement in her arms, a slight degree of the 'cuisse-tronc' phenomenon on rising to a sitting from a recumbent position and a doubtful extensor plantar response on the right side. Otherwise there was no appreciable disability in her various systems. The slight positive 'cuisse-tronc' sign and doubtful plantar reflex on one side are insufficient to warrant a diagnosis of a clinical syndrome of pyramidal defect or damage in a child of this age, though such disturbance cannot be wholly excluded.

The nature of the twinning

The question as to whether the twins were identical or not was obviously of the first importance. The hair and eye colour were strikingly similar, but in other respects there was no difficulty in distinguishing between the two. However, as the chief differences could be attributed to the presence of paraplegia and a greater degree of mongolism in M. G. and as they did not seem to exceed those sometimes found in accepted identical twins, the diagnosis could only be made by means of further tests :

1. The blood groups were kindly determined by Dr. H. C. Heathcote, who found that both twins were A. and both were M.

2. Next a series of skiagraphs were taken by Dr. G. D. Steven of the limbs. The lower limbs showed no appreciable difference in the stage of ossification, but in the upper limbs there were distinct differences. In M. G. the centres for the heads of the radii had appeared, but not in G. G. ; further, while M. G. had well developed centres for the medial epicondyles, G. G. had an extremely small one on one side only. It is interesting to note that so far as there were differences the more obvious mongol was more advanced in this respect than her twin sister. This finding made it unlikely that the twins were identical, for it is known that differences in rate of ossification of any appreciable degree are not found in identical twins (Bushke, 1935).

3. Finger-print investigation. The finger-prints of both children were submitted to Dr. Percy Stocks, whose extensive work (1930 and 1933) on this question has shown that the determination of the type of twinning can be made with considerable confidence. Dr. Stocks reported fully, and we are greatly indebted to him for his assistance. One thumb print was not clear enough for examination, but a comparison of the other nine prints showed only one pair similar (defining similar as in the paper by Dr. Stocks). Dr. Stocks states that with one exception only, he has never seen twins with less than four similar pairs of prints, whose physical characteristics suggested identity. These exceptional twins had only one similar pair, but he has suggested that the case was one of those instances in which close physical resemblance occurs in fraternal twins, as indeed it may do occasionally in brothers or sisters born at different times.

The evidence from finger-prints alone, therefore, makes it almost certain that M. G. and G. G. are fraternal and not identical twins, and this confirms the evidence afforded by the different stages of ossification at the elbow and the general lack of close physical similarity.

Mental examination of the twins

M. G. (paraplegic) : Chronological age four years six months ; Mental age two years eight months ; I.Q. 59.

This result was made up as follows : She passed all the two-year tests standardized by Gesell. She passed three only of the Binet three-year tests. She could name objects shown to her, name objects in pictures and give her last name, but failed the other three-year tests. She could count four pennies, but failed all the other four-year tests. She passed nothing above the fourth year.

G. G. : Chronological age four years six months ; mental age four years ; I.Q. 89.

These tests were made on the ordinary Binet scale.

WHOLE FAMILY. Mother. D.G. M.G. M. G. G. G.

The remainder of the family.

FATHER, aged 38, was healthy. His occupation was a builder's labourer. His family history was reported negative.

MOTHER, aged 31, was healthy. Her family history was reported negative.

HALF-BROTHER, D. G., son of the mother, aged eight years nine months, was healthy. His mental age was nine years one month ; I.Q. 104.

BROTHER, M. G., aged five years nine months, was healthy. His mental age was six years six months ; I.Q. 113.

Many normal persons show traits which might be considered at least mongoloid, but it was notable that in general appearance the parents and the two brothers were not in any way of this type, which increased our confidence in diagnosing G. G. as a mongol.

Discussion

Paraplegia in mongols.—It would appear from the literature that paralysis is very rare amongst mongols.

Brousseau (1928) states that she has seen two cases of mongols with paralysis, one being a girl of four years seven months with congenital spastic paralysis ; no details are given with regard to the other. Stevens (1915) in an article on the relationship of syphilis and mongolism describes two cases of spastic paralysis out of a series of twenty, one in a girl of two years six months and the other in a boy of seven years. Tredgold (1929) in his description of mongolism makes no mention of the occurrence of paralysis.

There is, therefore, no reason to suppose that the paraplegia in this case has any relationship with the mongolism as such.

No certain knowledge exists as to the precise cause of either infantile paraplegia or of mongolism. Paraplegia is generally supposed to be of

(1) Spinal origin due to birth injuries of the spinal cord, but in these cases there is no reason to expect mental deficiency unless it is coincidental, and almost all such cases occur in breach presentations. In this case there was no history of abnormal labour.

(2) Cortical origin due to injury to both leg areas presumably at least in some cases consequent upon lesions of the longitudinal sinus at or about the vertex.

(3) It is generally agreed that at least a proportion of cases of diplegia (Little's disease) is congenital in origin, i.e. due to failure of development, and so it is possible that some cases of paraplegia with no appreciable involvement of the arms may be due to the same cause.

So far as mongolism is concerned many theories have been advanced, most of which have proved untenable on more extensive examination. Penrose (1932) concludes that the condition has a hereditary basis, but that whether a potential mongol becomes a mongol in fact, depends on non-genetic post-fertilization factors the nature of which is unknown, except that advanced maternal age is sometimes a factor.

Mongolism in twins.—The familial incidence of mongolism is small. Penrose

(1933) states that about one mongol in fifty has a similarly affected brother or sister. It would be expected, therefore, that in the case of fraternal twins one of which was a mongol, the other would nearly always be normal. In the case of identical twins, however, both would be affected.

Penrose (1932) states ' . . . the curious fact that although cases of like-sexed twins, both affected, have been observed, there is no instance on record of twins both affected and of different sexes. The explanation seems to be that while in the case of identical twins neither is more likely to miscarry than the other, in the case of twins which are both affected but binovular, one twin nearly always miscarries.'

It should be explained that Penrose brings forward evidence to show that the mothers of mongols show a relatively high rate of miscarriage and that deaths of their children in infancy, before a diagnosis of mongolism could be established, are also disproportionately numerous. The deduction is that the number of mongols eliminated by pre-natal and early post-natal deaths may be greater than the number of mongols observed, perhaps considerably greater. As regards twins, Penrose quotes two of his own cases in each of which a mongol was born following the previous miscarriage of a twin foetus. While, therefore, fraternal twins both of whom are mongols would in any case be rare owing to the low familial incidence of the condition, the additional complication of early death so reduces the proportion that no proved case appears to be on record. The present case, therefore, is of interest as being an example of fraternal twins both of whom are mongols and both of whom have survived the period of infancy.

Variation of degree of mongolism.—No authority has yet succeeded in laying down absolute criteria for the diagnosis of mongolism.

Tredgold (1929) remarks ' . . . these anomalies are not distinctive in themselves. As a matter of fact, many ordinary ailments, and even normal individuals, possess one or more of the peculiarities which go to the make up of mongolism. It is the combination which is so characteristic of this condition. But where this combination occurs it produces a picture which is so distinctive as to constitute a definite and readily recognizable clinical variety.'

It must therefore be a matter to some extent of personal judgement as to whether any particular individual should be diagnosed as a mongol or not, but for reasons given above we are satisfied that the second twin G. G. should be so regarded. If this diagnosis is accepted not only is the occurrence of mongolism in a pair of fraternal twins of interest, but also the relatively high mental age of G. G.

This is in accordance with the observation of Tredgold (1929) who states that ' the degree of mental defect varies very considerably and on the whole I am inclined to think that there is a direct relation between this and the intensity of the physical signs.'

Most studies of mongols in the literature have been undertaken on mental defectives and nearly always those in institutions. Even so Tredgold notes

that some mongols may be merely feeble-minded. Even allowing for more than usual retardation in mental growth an I.Q. of 89 at four years six months makes it improbable that G. G. will fall into the category even of the feeble-minded, as this raises the question as to how far some degree of mongolism may not occur in persons usually regarded as normal. It is striking that if an observer sets out to look for the presence of mongolian characteristics in the members of communities other than those in institutions for mental defectives, it is possible to select a certain number of individuals who might well be included within the diagnostic bounds of mongolism. We are not aware that any observations on these lines have been systematically made ; nor is this surprising since all observers probably tacitly assume that mental deficiency is an essential criterion of mongolism. Further, if the contention of Tredgold is accepted that the physical features become less marked as intelligence rises, mongols of normal mentality, if they exist, would not be likely to possess such marked features as to excite the attention of the ordinary clinical observer unless he were specially on the look-out for such cases. None the less, observations of such cases, if they can be found, might provide information of great importance for the elucidation of the causation and nature of this very obscure condition.

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