HEREDITARY MYOPIA IN IDENTICAL TWINS

BY

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THE S—sisters are twins, aged 51 years. They suffer from high myopia inherited from their father. Both are unmarried.

Twin Diagnosis

They have been classed as identical twins because of their close resemblance both physically and mentally. Rose and May are an intelligent and cheerful pair of elderly women, indistinguishable to a stranger. They have often had the same illnesses at the same time, they object to being parted, and they tend to answer questions in unison.

They went to school at about the age of five, always had to sit close to the blackboard, and were not allowed to do needlework. At about that time they were first examined and found to be myopic. They had measles together at the age of seven. Shortly after that, both started to wear glasses, and still do so.

Certain characteristics were examined in detail, with the following results:

Hair. Exactly the same as regards quantity, quality and colour. The crown whorls were mirror images of each other.

Eyebrows. Thickness, hair whorl and distribution were identical, distance between the inner ends 2 cm. in each case.

Teeth. Rose has six false teeth — three on each side above. May has seven — four lower central incisors and three upper teeth. The remaining corresponding teeth resemble each other closely.

Noses. Identical in shape.

Eyes — Iris. The colour and structure of the irides are identical, including a small radial yellow streak at five o'clock in the right eye, which could only be seen with the lens and loupe.

Lens. No opacities in either.

Vitreous. Rose (R) Large snaky opacity. (L) Small snaky opacity. May (R) Vitreous clear. (L) Numerous snaky vitreous opacities.

Fundi. The fundi, although not identical as regards vascular pattern and the characters of the disc, resemble each other very closely. The most noticeable difference was the presence of spontaneous venous pulsation in both eyes in the case of Rose, and its absence in the case of May.

The common fundus picture, equally developed in all four eyes, was as follows:

1. High myopia at the posterior pole — moderate only at the periphery.
2. Extensive choroidal atrophy of the posterior pole — much less marked at the periphery. Large myopic crescents surrounding the disc but falling short of the macula.
3. Marked pigmentary mottling at the macula.
4. Vessels narrow and irregular. No other sign of arterio-sclerosis apart from moderate crushing at venous crossings.
5. No haemorrhages or exudates.

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Ocular.
(a) Refraction under homatropine and cocaine.

Rose: (R) +1.5/95° (L) −15.0
+2.0/80°
May: (R) +22.0 (L) −24.0
+6.0/100°

(b) Visual acuity: was only tested for near vision.

May: (R) without glasses. Reads J1. (L) without glasses. Reads J1.

Interpupillary distance ... ... Rose 55 mm. May 55 mm.
Horizontal measurement of palpebral fissure — inner to outer canthus ... (R) 25 mm. (R) 25 mm.
(L) 25 mm. (L) 25 mm.
Vertical measurement of palpebral fissure in normal distance gaze ... (R) 11 mm. (R) 11 mm.
(L) 11 mm. (L) 11 mm.
Intra-ocular pressure (Schiotz) ... (R) 16 mm. Hg. (R) 15 mm. Hg.
(L) 17 mm. Hg. (L) 17 mm. Hg.

Other.
Circumference of head ... ... 51 cms. 50.5 cms.
Glabella to external occipital protuberance ... ... 32 cms. 32 cms.
Height ... ... ... ... 145 cms. 144.5 cms.
Tip of olecranon to ulnar styloid ... (R) 23 cms. (R) 23 cms.
(L) 23 cms. (L) 23 cms.
Blood pressure ... ... ... 160/70 158/80

The only significant difference is in the refraction. The right eyes correspond closely, but between the left eyes there is a difference of 9 D. spherical, 4 D. cylindrical, and 20° in the axes of the cylinders. If the eyes are considered as mirror images, then the greatest difference between corresponding eyes is 7 D. spherical — i.e., between May’s right eye and Rose’s left. In either case, Rose shows 5 D. more anisometropia than May, and this probably accounts for the relatively poor visual acuity in her right eye.

Pedigree

The parents were not related. Both the father and paternal grandfather are reputed to have had severe short-sight. One paternal uncle and one paternal aunt are also reported to have been affected. None of the mother’s relatives was known to be affected.

The patients’ sibship consisted of two pairs of twins, of which the elder pair was dizygotic. The sister died in infancy from an
unknown cause. The brother is alive and is myopic; he has six children and one grandchild, all apparently normal. Apart from the twins Rose and May, the only other members of the family examined have been the brother George S. and two of his children.

George S. Aged 48 years. Metal machinist.

Both eyes are myopic, but in his case the right eye is markedly, and the left noticeably, less severely affected than in his sisters. With his right eye corrected, he is able to carry on his job, which demands considerable precision. The left is amblyopic.
Refraction under homatropine.

- (R) \(-5.0\) \(+2.0 \times 65^\circ\)
- (L) \(-15.5\) \(+1.5 \times 65^\circ\)

Fundi. (R) There is a small myopic crescent limited to the temporal and inferior aspects of the disc, no myopic degeneration, a normal macula, and normal vessels.

(L) There is high myopia with considerable choroidal atrophy limited to the posterior pole. The periphery is much less myopic and shows no degeneration. There is a large myopic crescent enveloping the disc and falling just short of the macula, which itself shows pigmented mottling similar to that of the twins. The vessels are normal.

The left fundus is in fact a milder example of the same condition as that of the twins.

The two children examined, Robin and Jean, aged 10 and 12 years respectively, have normal vision. The fundi are normal. Robin has slight hypermetropic astigmatism, and Jean's refraction under homatropine is (R) \(-1.0\) D, (L) \(-0.5\) D. The remaining children could not be examined. One is training at a Police College, and the other three were said to have normal sight. Apparently none of George S.'s descendants has high myopia.

Discussion

The occurrence of high myopia in identical twins with a pedigree showing direct transmission over three generations is evidence of its genetic origin.

The lesion is equally developed in all four fundi, which correspond closely. The refractions, however, do not. Such differences of refraction in monozygotic twins with high myopia are well recognised (Bücklers, 1939; Marchesani, 1935). They suggest that, while the general picture of high myopia in these cases is genetically determined, the exact type and degree of refractive error may perhaps be influenced by environmental factors.

In view of the clearly dominant type of transmission, one would have expected the condition to reappear in the six children of George S. That it has not done so emphasises the variability of transmission in an undoubtedly genetic condition.

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References

Marchesani (1935).—Ibid., 94, 97.
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