MARFAN'S SYNDROME WITH RETINAL DETACHMENT*

BY

JAMES R. HUDSON

London

SINCE the account of arachnodactyly by Marfan (1896), and the observation of its association with ectopia lentis by Salle (1912), a large number of cases has been described. The subject has been fully reviewed by Rados (1942), whose paper includes a statistical survey of all cases (numbering 204) described in the literature up until 1940, and an extensive bibliography. The prominent features of the syndrome are well known—elongation of the long bones, with long slender fingers and toes, spinal deformity, congenital heart disease, and ectopia lentis accompanied by miosis (Ormond, 1930). Opinion as to the hereditary nature of the condition varies. Strebel and Steiger (1915) reported a family in which ectopia lentis was associated with high myopia and congenital heart disease, Mann (1937) states that ectopia lentis linked with arachnodactyly is seldom an hereditary condition, and several other authors (Weve, 1931; Duke-Elder, 1937; Doggart, 1949) comment on the strongly hereditary character of the disease, which affects both sexes and is transmitted as a dominant characteristic. A perusal of the literature has revealed a description of only two cases of retinal detachment in association with the disease (Fischbach, 1937; Cassidy and McFarland, 1947). These were in a male child aged one-and-a-half years, and in a male adult aged 52 years, respectively. It is, therefore, hoped that the present case report may be of interest. The early recurrence of the detachment with spontaneous resolution following a short period of rest was a striking feature.

CASE REPORT

Miss D. B., aged 24 years, a typist, attended Moorfields Hospital on September 2, 1949, complaining of blurring of vision in the upper temporal field of the left eye during the previous three months, and of the appearance of a shadow over this area of the field, which had extended to obscure most of the field of the affected eye during the three weeks prior to her examination at the hospital. The patient exhibited the general physical features of Marfan's syndrome, including arachnodactyly, poor muscular development, and congenital heart disease.

Ocular Examination.—Movements full. Right eye 6/12 with correction (+4.00 D. sph.); eye white, anterior chamber deep, pupil active, iris tremulous, lens

* Received for publication February, 15, 1951.

transparent and dislocated downwards, vitreous opacities, fundus normal, tension normal.

Left eye 6/36 with correction (+8.00 D. sph.); eye white, anterior chamber deep, pupil active, iris tremulous, lens transparent and dislocated downwards, vitreous opacities, protrusion of the vitreous face through the pupillary aperture, almost total retinal detachment deep nasally and shallow temporally.

Further examination of the fundi under a mydriatic revealed nothing abnormal in the right eye. The left eye showed an almost total detachment of the retina, deep nasally and shallow temporally, with the appearance of a dialysis extending from 7 to 10 o'clock.

Operative treatment was delayed for two reasons, first to allow time for any possible improvement in the detachment, and secondly because the patient was anxious to have a general anaesthetic. The opinion of a physician was sought and Dr. J. S. Stead reported the presence of aortic regurgitation. Although the congenital heart disease was not of a cyanotic type, he considered the risk of a general anaesthetic unjustifiable, and the patient later decided to submit to operation under local anaesthesia.

Operation.—This was carried out on September 19, 1949, under topical and retro-ocular anaesthesia. The internal rectus was exposed and divided, and a barrage of surface diathermy was laid down, extending back from the ora at 7 o'clock, to 15 mm. from the limbus, and forward to the ora at 10 o'clock. Ophthalmoscopic examination then revealed what appeared to be a further small dialysis at the lower edge of the diathermy reaction, and this was included in a further small barrage extending to the insertion of the inferior rectus. Diathermy puncture was performed within the area of the original barrage. The internal rectus was re-attached and the conjunctiva sutured.

The patient was discharged from hospital on October 7, 1949, four weeks after admission. The right eye was unchanged. The left eye was whitening with a clear cornea, pupil half-dilated and inactive, slight vitreous haze, retina in place in all areas with pigmented diathermy reaction peripherally in the nasal half, tension low to normal, and visual acuity 6/18 with +7.00 D. sph. +1.00 D. cyl. at 10°.

Recurrence and Spontaneous Resolution.—Two weeks later the patient was seen again in the out-patient department with a recurrence of her symptoms. On examination the left eye showed a large balloon detachment, the corrected visual acuity being reduced to 4/60. On October 28, this had increased to a total detachment. On re-admission to hospital after 9 days, during which the patient had rested in bed at home, the retina was observed to be in place, and it remained so after the patient was allowed up. When she was seen again on January 17, 1950, the visual acuity of the right eye was 6/18 with glasses, and of the left eye 6/12 with glasses. The retina was in place in all areas. The tension of the eyes was normal.

The patient unfortunately developed heart failure and died on February 23, 1950.

I should like to thank Mr. R. C. Davenport, under whose care this case was admitted, and members of the Medical Committee at Moorfields Hospital for their permission to make use of the hospital records of the case.

REFERENCES

- CASSIDY, J. V., and MCFARLAND C., B. (1947). *Amer. J. Ophthalm.*, **30**, 469.
 DOGGART, J. H. (1949). "Ophthalmic Medicine". Churchill, London.
 DUKE-ELDER, W. S. (1937). "Textbook of Ophthalmology", vol. 2, p. 1351-2. Kimpton, London.
 FISCHBACH, H. (1937). *Z. Kinderheilk.*, **58**, 630.
 MANN, I. (1937). "Developmental Abnormalities of the Eye". University Press, Cambridge.
 MARFAN, A. B. (1896). *Bull. Soc. méd. Hôp. Paris*, **13**, 220.
 ORMOND, A. W. (1930). *Guy's Hosp. Rep.*, **80**, 68.
 RADOS, A. (1942). *Arch. Ophthalm.*, Chicago, **27**, 477.
 SALLE, V. (1912). *J. Kinderheilk.*, **75**, 540.
 STREBEL, J., and STEIGER, O. (1915). *Arch. Augenheilk.*, **78**, 208.
 WEVE, H. (1931). *Ibid.*, **104**, 1.