THE SYNDROME OF MARCHESANI*

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MARCHESANI (1939) first described the syndrome which is named after him, and since the appearance of his report cases have been described in European and American literature. The characteristic features of this syndrome are:

1. Brachycephaly, small stature, short limbs, brachydactyly, short squat fingers and toes with broad hands and feet, limitation of movement at the joints, and well-developed musculature and subcutaneous fat;

2. Spherophakia with microphakia, iridotonesis, visibility of the zonule, lenticular myopia, and glaucoma.

These features are well developed in the two cases to be described, a brother and sister, the only children of parents who are first cousins. All the other members of the family are of short stature but do not exhibit brachycephaly.

Case Reports

Case 1, a girl aged 6 in 1953, was brought to the hospital with the complaint that she could not see the blackboard at school. The visual acuity in each eye was 6/24, with -7 D sph., -5 D cyl., axis 180°. She was seen at regular intervals after this and the myopia progressed. At the age of 12 years the visual acuity in each eye was 6/24, with -14 D sph., -5 D cyl., axis 180°. It was noted at this time that the equator of the lens could be seen in the pupils of both eyes. One year later the visual acuity of the right eye had deteriorated to hand movements, and that of the left was 6/24 with -20 D sph., -6 D cyl., axis 180°. The discs were noted to be cupped and the intra-ocular tension was raised.

She was admitted to hospital in March, 1959, and general examination revealed the skeletal and muscular features of Marchesani’s syndrome (Figs 1, 2, and 3, overleaf). There was evidence of congenital heart disease, and a haemorrhagic diathesis was present, caused by an abnormality of capillary contractibility.

The right eye (visual acuity hand movements with -20 D sph., -5 D cyl., axis 180°) was white and showed no inflammatory change. The cornea was clear and its horizontal diameter 13 mm. The anterior chamber contained vitreous. The axial region of the anterior chamber was shallow and the iris and lens tremulous. Persistent pupillary membrane remnants were present. The equator of the lens could be seen when the pupil was dilated, the lens being small and globular (Fig. 4, overleaf).

* Received for publication June 29, 1960.

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FIG. 1.—Case 1. The entire equator of the lens is visible under mydriasis.

FIG. 2.—Hands of Case 1.

FIG. 3.—Foot of Case 1.

FIG. 4.—Right eye of Case 1. The entire equator of the lens is visible under mydriasis.

The zonule was not visible and the vitreous could be seen passing anteriorly round the equator of the lens. The disc was cupped, but no degenerative changes of a myopic nature were seen in the fundus. A peripheral field could not be plotted, though a temporal island of vision was present.

The left eye could be corrected to 6/18 with −21 D sph., −6 D cyl., axis 180°. It was similar to the right, though the cupping of the disc was less and the peripheral field was full.

Gonioscopy revealed a false angle in the right anterior chamber; the angle of the left anterior chamber was narrow but with open clefts at 6 and 12 o’clock.
The intra-ocular pressure of the right and left eyes varied between 20 and 30 mm. Hg (Schiotz). That of the right eye settled at 22 to 26 mm. Hg on gutt. di-isopropyl fluorophosphate 0-01 per cent. twice daily and that of the left at 15 mm. Hg on gutt. pilocarpine 2 per cent. three times daily.

Tonography and aplanation tonometry were carried out. On gutt. pilocarpine 2 per cent. three times daily, aplanation tonometry of the right eye was 43 mm. Hg, and of the left 29 mm. Hg. Facility of outflow of the right eye was 0-10, and of the left 0-08.

With the mydriasis of gutt. homatropine and cocaine, aplanation tonometry of the right eye was 45 mm. Hg, and of the left 28 mm. Hg. Facility of outflow of the right eye was 0-03, and of the left 0-05.

On April 4, 1959, surface cyclodiathermy was performed on the right eye, twelve applications for 7 seconds at 70 milliamps, 7 mm. from the limbus. A temporary reduction in the tension followed this procedure, but 6 weeks later the intra-ocular pressure was 47 mm. Hg (Schiotz) in the right eye and 43 mm. Hg in the left. The aplanation tonometer readings were 38 mm. Hg in the right eye and 34 mm. Hg in the left.

On May 23, 1959, an inferior half penetrating cyclodiathermy was performed, each eye receiving 32 applications each of 5 seconds at 50 milliamps, at 6 mm. and at 8 mm. from the limbus. The tension then fell to 15 mm. Hg (Schiotz) in each eye and has since remained normal. During the month following the penetrating cyclodiathermy, the anterior chambers became deeper, and on October 28, 1959, the visual acuity in the left eye was 6/18, with -12 D sph., -4 D cyl., axis 180°.

Case 2, a brother of Case 1, was first seen in 1955 when he was 5½ years old. He was noted to have a similar physique to his sister (Fig. 5). With -6 D sph., -2 D cyl., axis 180°, the visual acuity was 6/36 in each eye. There was a gradual progression of the myopia and on May 19, 1959, when he was examined for investigation, the visual acuity was 6/24 in each eye with -21 D sph., -5 D cyl., axis 180°. The horizontal diameters of the clear corneae measured 14 mm. Vitreous was present in the right anterior chamber but not in the left. There was iridodonesis. The small globular lenses were dislocated forwards and medially so that the anterior chambers were very shallow axially. Persistent pupillary membrane remnants were present and the equator of the lenses could be seen on mydriasis. The discs were normal and the peripheral fields full. The intra-ocular tension was found to be 28 mm. Hg right and left. Gonioscopy was not possible.

This boy was phased on gutt. pilocarpine 2 per cent. 4-hrly and was found to be well controlled. Tonography has not been possible but a dark-room test while on pilocarpine 2 per cent. on May 14, 1959, produced a rise in intra-ocular tension of 16 mm. Hg (20–36) in one hour in the right eye and of 26 mm. Hg (24–50) in the left.

Discussion

These cases, like those previously reported, were consanguineous. They exhibit all the features detailed by Marchesani (1939) and are similar to cases reported by Meyer and Holstein (1941). Marchesani's syndrome
invites comparison with Marfan’s syndrome, in which the subjects are dolio-cephalic and tall, and have long limbs, a narrow thorax, deep-set eyes, and weak musculature. The lenses in arachnodactyly are usually dislocated upwards and medially but the zonule persists at least in part. The equator of the lens is not visible in its entirety and the lens is normal both in size and in the radii of curvature. The zonule in Marchesani’s syndrome, in contrast, appears to be almost entirely deficient and the lens is small and spherical.

Marchesani considered both brachydactyly and arachnodactyly to result from dysplasia of mesodermal structures and suggested the terms: “dystrophia mesodermalis hyperplastica” for the syndrome known after him and the term “dystrophia mesodermalis hypoplastica” for Marfan’s syndrome. The development of spherophakia is usually explained as a failure of development, or as an early degeneration of the zonule, the lens retaining its early form and never being subjected to the action of the ciliary body (Duke-Elder, 1938; Vogt, 1931).

While the relationship between skeleto-muscular changes and lens anomalies is debatable, Meyer and Holstein suggested a mechanism by which ciliary body hyperplasia could lead to spherophakia. In late childhood hyperplasia of mesodermal tissue is occurring throughout the body, and hyperplasia of the ciliary body results in a lack of tension in the zonular fibres, thus allowing the lens to assume its spherical form of maximal accommodation.

An interesting feature of both the cases here reported is that there has been a progression in the myopia, which has apparently occurred as the lenses have gradually assumed a more spherical shape. The first case, with the increase in depth of the anterior chamber and the consequent decrease in effective power of the lens after cyclodiathermy, showed some decrease in the myopia. That the myopia is lenticular is confirmed by the finding of a normal fundus and emmetropia in spherophakic eyes that have been subjected to lens extraction. The tonographic studies performed on the first case showed conclusively that the glaucoma was not of the inverse type (Urbanek, 1930) and that the outflow was improved by the administration of miotics. It is unlikely, therefore, that lens extraction would relieve the tension. In the first case reported by Meyer and Holstein (1941), pilocarpine produced a rise in tension and the lens was extracted. This lens measured 7 mm. in diameter and was 5.8 mm. thick.

In the first case, surface cyclodiathermy produced a temporary reduction in tension and supported the view that a penetrating cyclodiathermy would be a reasonable procedure to undertake. Penetrating cyclodiathermy also appears to control tension when this occurs in some cases of congenital ectopia lentis.
I am indebted to Mr. A. G. Cross for permission to study these cases and for his advice and encouragement in the preparation of this paper, and to the Medical Committee of Moorfields Eye Hospital for permission to publish the findings.

REFERENCES