MARCHESANI’S SYNDROME*

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Since this condition was first described by Marchesani (1939), several cases have been reported in the continental and American literature, but none hitherto in Great Britain.

The syndrome is characterized by certain skeletal and ocular abnormalities, the former being in striking contrast to those present in Marfan’s syndrome. The stature is short, usually 5 feet or a little less, the figure stocky with well-developed musculature, and the skin and subcutaneous tissue thick. The hands are spade-shaped with broad palms and short stubby fingers, and in childhood, radiography shows delayed carpal ossification. The shape of the feet follows the same pattern. The head tends to be broad and square and most of the cases which have been illustrated show a strong resemblance in facies as well as build.

The important ocular features are spherophakia and ectopia lentis, giving rise to lenticular myopia and iridodonesis, and glaucoma which is probably also due to the spherophakia. The visual outcome is usually poor because of the resistance of the glaucoma to treatment, including surgery. The condition is generally familial and in a high proportion of cases reported there is consanguinity of the parents, suggesting a rather rare autosomal recessive gene. Two families described (Rousseau and Hermann, 1949; McGavic, 1959) show dominant inheritance. Short stature is usually found in one or both parents and not uncommonly some members of the affected family show the brachymorphy without the eye signs. Brachydactyly itself is generally dominant (McNutt, 1946). Cases of spherophakia complicated by glaucoma, sometimes familial, were reviewed by Shapira (1934), but not in association with brachymorphy.

Treatment of the glaucoma is difficult. The condition is usually paradoxical in that the use of miotics may lead to a rise in tension which can be reversed by mydriatics. This was not a feature in our case although miotics failed to lower the ocular tension. Extraction of the lens, generally by means of a vectis, has frequently been advocated but has not always led to relief.

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of the glaucoma. Needling of a spherophakic lens does not lead to absorption. Eyes which have had a filtering operation generally do badly.

Iridectomy ab externo performed at an early stage, before secondary changes have occurred in the filtration angle, would appear to be a simple and rational procedure. There is support for this view in the literature (Rosenthal and Kloepfer, 1956) and in our case.

Case Report

A boy aged 16 first attended the Eye Out-Patient Department in September, 1958. He had previously attended a School Eye Clinic where spectacles to correct his myopia had been ordered. He had no symptoms apart from defective vision.

Examination.—The pupils were dilated with Cyclogyl and the refraction and visual acuity were as follows: right eye 6/12, with −15 D sph., −1·5 D cyl., axis 180°; left eye 6/24, with −15 D sph., −2 D cyl., axis 180°. The left eye showed stretching and thinning of the sclera above. It was possible to see through the dilated pupils that both lenses were dislocated downwards.

The right optic disc was of good colour and showed slight cupping, possibly physiological. The left disc showed glaucomatous cupping and pallor. The fundi were otherwise normal in appearance and in particular there was no sign of myopic change.

The intra-ocular pressure was 34 mm. Hg in the right eye and 46 mm. Hg in the left (Schiotz). The right visual field was normal; the left showed arcuate scotomata with a nasal step.

Treatment.—Pilocarpine and Diamox produced no significant change in the intra-ocular pressure. A cyclocialysis was performed on the left eye without benefit, and later an iridectomy ab externo was carried out, but with difficulty because of the staphylomatous condition of the sclera. About this time the patient was recognized as being a case of Marchesani’s syndrome.

Progress

Left Eye: The tension has remained generally a little raised and there has been further loss of the visual field. The visual acuity is now 6/36+, with −13 D sph., −3·5 D cyl., axis 125°.

Right Eye: Because of a rise in tension an iridectomy ab externo was carried out on the right eye. The tension has now remained normal for 10 months with no loss of visual field. The visual acuity is now 6/9 ptly, with −10 D sph., −4 D cyl., axis 175°.

Slit-lamp examination revealed nothing abnormal in the right eye apart from the marked convexity of the lens with bowing of the overlying iris and shallowness of the temporal side of the anterior chamber. Gonioscopy of the right eye before operation confirmed the bowing of the iris but the angle of the anterior chamber was normal. Gonioscopy of the left eye after operation showed peripheral anterior synechiae all round the angle. Tonography of the right eye indicated a normal facility of aqueous outflow.

The patient’s height is 4 ft 10½ in. and he weighs 126 lb. His hands and feet show the characteristic appearance and he has well-developed muscles and subcutaneous tissues. His head is broad and rather square (Fig. 1, opposite). X-ray examinations have revealed no abnormality apart from an unusual shortness of the metacarpals and metatarsals. He is of normal intelligence and placid temperament.
The only ocular abnormalities found among other members of his family are hypermetropia and squint. His sister, aged 12, is 2½ inches taller than he is and has long narrow hands and fingers which are almost arachnodactylic. One maternal uncle (Fig. 2, II, 3) is in a mental hospital with reputed postencephalitic behaviour disorders dating from age 8, but there is no history of an overt attack of encephalitis. In build and facies this man closely resembles his affected nephew but his eyes are normal. The patient’s father was an adopted child and nothing is known about his relatives.

**Summary**

A case of Marchesani’s syndrome is described in which the glaucoma appears to have been brought under control in one eye after an iridectomy *ab externo*.

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**Fig. 1.**—Patient’s general appearance, showing height.

**Fig. 2.**—Family tree showing reported case (III, 2), and his mother (II, 2), maternal uncle (II, 7), and maternal grandfather (I, 1).
REFERENCES


ADDITIONAL BIBLIOGRAPHY