MARCHESANI'S SYNDROME*

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THIS syndrome was first described by Marchesani (1939), and no case has hitherto been reported from Pakistan. The following patient came under observation recently.

Case Report

A Bengali Muslim boy aged 14 years was brought by his father with the complaint of defective vision, especially at night, since childhood. The parents first noticed the disability when the boy was 3 years old. He was born at full-term, but started walking very late at the age of 5 years. Both the parents were of short stature (father—5' 2"; mother—5'), but the patient's other brothers and sisters were of average height. None showed any ocular anomalies of congenital origin. No history of night blindness could be elicited in the family. There was no consanguinity.

Examination.—The boy was short (4' 4") and well-developed (Fig. 1, opposite), and weighed 68 lb. The head was broad and square, cephalic index 84, and the fingers and toes were short and stubby. The heart and lungs showed no abnormality. He was of average intelligence but slow in movement.

The visual acuity in each eye was only counting fingers at 2 ft. Retinoscopy under homatropine mydriasis showed myopia (right eye - 3.5 D sph.; left eye - 2.5 D sph.) not improved with glasses. A mild degree of iridodonesis was present in the upper segment in both eyes. The left pupil was eccentric, with a shallow notching of the pupillary margin at the 7 o'clock position on the left side. The pupillary reactions were normal. When the pupils were dilated both lenses were found to be partially dislocated downwards. The fundi showed atrophic discs, with slight cupping of the left disc, and mild attenuation of the arteries in both eyes. Gross chorio-retinal degenerative changes were seen in both eyes. Bone-corpuscule-like pigmentation was more marked over the mid-periphery of the fundi.

The ocular tension was 22 mm. Hg (Schiötz) in the right eye and 29 mm. Hg in the left. Slit-lamp examination revealed a slight degree of hyperconvexity of the lens. Confrontation tests showed the visual fields to be contracted.

Routine laboratory investigations of the blood, urine, and stools showed no abnormality. The Wassermann reaction and Kahn test were negative.

Treatment.—The patient was put on 1 per cent. pilocarpine drops twice daily, intramuscular injections of vitamin B₁ daily, and vitamin A and D capsules orally.

Result.—The ocular tension is now 18 mm. Hg (Schiötz) in the right eye and 24 mm. Hg in the left. There has been no improvement in the visual acuity.

Discussion

Marchesani's syndrome as described in the literature presents certain characteristic features; skeletal abnormalities include broad and square head

* Received for publication May 4, 1962.

FIG. 1.—General appearance of the case of Marchesani's syndrome.
FIG. 2.—A typical case of Marfan's syndrome.

(brachycephaly), short stature with well-developed musculature, thick skin and subcutaneous tissue, short limbs, short stubby fingers and toes (brachydactyly), and broad hands and feet.

The appearances are in sharp contrast to those of Marfan's syndrome (Fig. 2).

The characteristic ocular features are spherophakia, iridodonesis, ectopia lentis, lenticular myopia, and glaucoma.

In the case reported here, we observed besides the usual characteristic features eccentricity of one pupil with shallow notching of the margin, pigmentary degenerative fundus changes, and optic atrophy. Unlike other cases there was no consanguinity. With the use of miotics the ocular tension was lowered and no paradoxical phenomenon was observed.

Summary

A case of Marchesani's syndrome is described, which showed optic atrophy and pigmentary degeneration of both fundi.

We wish to thank Dr. S. M. Rob, Associate Professor of Medicine, Dacca Medical College, for the photograph of the case of Marfan's syndrome.

REFERENCE

MARCHESANI, O. (1939). *Klin. Mbl. Augenheilk.*, 103, 392.