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THE MARFAN SYNDROME
A Report of an Atypical Case

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K. G., a 14 year old colored male, was first seen in the Henry Ford Hospital in September, 1956, by the Pediatric Department. He was brought in primarily for evaluation of his general physical condition, especially in regard to a spinal curvature.

Past History: The patient was normal full term child born following an uneventful pregnancy and a normal labor. His general health has been excellent.

Family History: There were three fairly tall maternal uncles but none with a body habitus similar to the patient's. No relatives have died suddenly or have other symptoms suggestive of a dissecting aneurysm. The father has bilateral cataracts which began at the age of 25. The patient has two normal siblings.

Pertinent Physical Findings: Height 176 cms. with an arm span of 196 cms. Weight 107 lbs. His head was somewhat tower-shaped (Fig. 1). There was a pronounced arachnodactyly and his extremeties were long and narrow, his fingertips reaching the knee level when he was standing erect (Fig. 2). There was a pronounced kyphoscoliosis with an associated thoracic cage asymmetry (Fig. 3). He showed none of the features usually seen in the typical Marfan syndrome, such as under-development of the muscles, sparsity of the subcutaneous fat, laxity of joint capsules and flat feet. Evaluation by the Cardiology Department showed no abnormalities. Mental development was normal.

Eye Examination: There was some slight facial asymmetry, the right eye being 3 mms. higher than the left and the right palpebral fissure being slanted slightly upward The corneas were clear. The pupils were round, regular, of equal size and reacted normally to light and accommodation. Visual acuity without correction: RE 20/30-2, LE 20/15.

Muscle balance without correction:

DV: 4 prism diopters of intermittent esotropia.

NV: 3 prism diopters of intermittent esotropia with 2 prism diopters of left hyperphoria.

Fixation with the left eye was preferred. Extraocular movements were normal. Near point of convergence was 3 cms.

Manifest refraction was unsatisfactory.

Cycloplegic refraction: RE: $-.25 + .75 \times 90 = 20/30$

LE: $+.25 + .25 \times 180 = 20/15$

Slit Lamp: Both corneas were clear. The anterior chambers were deep, well-formed and optically empty. The irides were normal. The lenses were of normal transparency and in normal position. There was a fine vitreous haze in the right eye. The vitreous in the left eye was normal.

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Fig. 1. Tower Shaped Head

Fig. 2. Long Narrow Extremities

Fig. 3 Kyphoscoliosis with associated Thoracic Cage Asymmetry

Ophthalmoscopic examination: No abnormalities were observed.
Visual Field examination: Full peripherally to the 3/330 white isopter. The red isopters, centrally, were likewise normal with the exception that in the right eye the 2 mm. red isopter did not appear to be quite as red at the point of fixation as it did about 2 degrees out. This is interpreted as being a partial scotoma to the 2 mm. red isopter from suppression amblyopia.
Tension: RE 17 mm. Hg.
LE 17 mm. Hg.

Classification of this case with respect to Marfan's syndrome involves some speculative aspects. A brief review of the Marfan syndrome therefore seems in order.

I. *Clinical Manifestations.*

The individual with Marfan's syndrome shows a typical body habitus consisting of long thin extremities. In general, the more distal bones of the extremities tend to demonstrate this excessive length more strikingly. Arachnodactyly is the result. The affected individual tends to be tall. Redundancy and weakness of joint capsules, ligaments, tendons and fascia are responsible for a large group of disorders which include pes planus, genu recurvatum, hyperextensibility of joints, habitual dislocation of the hips, patella, clavicles, mandible and other joints. Ganglia, hernias, synovial diverticula and kyphoscoliosis are also seen frequently. The skull and face bones are long and narrow with a resultant highly arched palate and prognathism. Muscular underdevelopment and hypotonia along with a pronounced sparsity of subcutaneous fat is a prominent feature.

The cardiovascular system is strikingly involved. An abiotrophy of the elastic tissue of the aorta and pulmonary artery may result in a diffuse dilatation of these vessels or in a dissecting aneurysm. This latter development has resulted in the early death of many individuals with this syndrome.

The eye — Ectopia lentis, almost always bilateral, is the hallmark of ocular involvement. Seventy percent of all cases of congenital ectopia lentis occur as components of the Marfan syndrome. The zonules, when visualized with the slit lamp, are redundant, attenuated and often broken. The lower ligaments are most often defective with a consequent upward displacement of the lens. The lenses are often small and may be spherical. Coloboma lentis has been reported. Secondary iritis and glaucoma often occur. Axial myopia is usually present and has been reported to be as high as twenty diopters in some cases. This tends to indicate involvement of the sclera, fundamentally a ligamentous structure, in a basic connective tissue defect. The scleral defect is occasionally expressed in the cornea as keratoconus, clouding or megalocornea. The sclera may be impressively blue. The pupil is often difficult or impossible to dilate due to hypoplasia of the dilator pupillae muscle.

Spontaneous retinal detachments occur in unusually high incidence and are a frequent complication of lens extractions. Severe limitation of visual acuity and even total blindness occurs all too frequently.

Some patients with advanced Marfan's syndrome, familial in that other members of the family were involved, and with characteristic body habitus, dissection of the aorta, and autopsy demonstration of the pathognomonic changes in the media, have

not shown ocular abnormalities even on the most careful examination. This part of the syndrome, it seems, may be repressed completely in a given family.

II. *Differential Diagnosis.*

In the Negro race particularly, the Marfan syndrome is often suggested by the skeletal proportions. A picture mimicking it in many respects can result from Rh incompatibility or from intra-uterine rubella infection. Ectopia lentis does not occur in these cases. Arachnodactyly *per se* is not to be taken to indicate Marfan's syndrome as it is a nonspecific manifestation with many possible causes. Atypical muscular dystrophys can be confused with Marfan's syndrome and may require histological differentiation. Given stigmata suggestive of the Marfan syndrome, one can be most confident of the diagnosis if ectopia lentis is present in the patient or if other members of the family are unequivocally affected.

III. *Incidence and Inheritance.*

The sexes are equally affected. There is no racial or subracial concentration of cases. It has been reported from all parts of the world. It is an uncommon but by no means rare disease. The connective tissue defect involving the aorta is the leading cause of dissecting aortic aneurysms in the younger decades. There is reason to believe that there is an appreciable number of very mild or *forme fruste* cases in which the connective tissue defect has little effect on the health and longevity.

Partial submersion of the manifestation of this syndrome can be observed in pyknic stock; the individuals thus affected may not have an impressively unusual habitus. However, no protection from ocular or aortic abnormality is afforded thereby.

The pattern of inheritance is that of a simple Mendelian autosomal dominant. Parenteral consanguinity has not been an impressive feature. Sporadic *de novo* mutations arise in perhaps 15% of all cases.

IV. *Pathology.*

The basic defect is an abiotrophy of the elastic tissue fibers. They are constitutionally inadequate and undergo degeneration, particularly at sites of stress, the ascending aorta, for instance. The smooth muscle elements, which normally originate and insert on the elastic lamellae, collapse together into disorganized whorls and undergo hyperplasia and hypertrophy. Reparative processes leave the tissues scarred and weakened. Exactly what the zonule of the lens has in common with the elastic tissue of the media of the aorta is unknown. If it were known, the basic defect might be better understood.

Comment.

The physical features in this case are compatible with a mild form of the Marfan syndrome, but the eyes showed none of the typical manifestations. However, as mentioned previously, the ocular abnormalities may be completely suppressed in certain cases. Our tentative diagnosis is: *Forme fruste* Marfan's syndrome, *de novo* mutation.

Summary.

A diagnostic problem concerning a questionable case of Marfan's syndrome is presented. The clinical manifestations, differential diagnosis, incidence, heredity and pathology of the Marfan syndrome are reviewed briefly.