ADDENDUM

On October 1, 1959, Dr. W. G. Bigelow closed this defect using the heart-lung bypass, and a heat exchanger to lower body temperature. It was described as "something under a centimetre in diameter lying anteriorly and fairly low".

The patient's postoperative course was uneventful, but it is too soon to know whether or not his angina is improved.

REFERENCES


BILATERAL ECTOPIA LENTIS IN MARFAN'S SYNDROME: REVIEW OF FEATURES WITH REPORT OF TWO CASES

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Marfan's syndrome is an uncommon condition. More than 350 cases have been reported since Marfan,20 in 1896, described a five-year-old girl with spider fingers (pattes d'araignée) and poorly developed musculature. Subsequent reports have indicated that it may be associated with other anomalies. The syndrome is now well recognized in its classical form, but frequently goes unnoticed in its incomplete or atypical form ("forme fruste"). The purpose of this paper is to present the clinical features of Marfan's syndrome in the light of a brief survey of the reported anomalies which may be found and in the reports of two cases with skeletal and ocular manifestations. These patients belonged to two unrelated families. Some of the other members of these families had manifestations of this syndrome.

BRIEF SURVEY OF CLINICAL FEATURES

Although knowledge of the manifestations of Marfan's syndrome is widely disseminated, often only the classical form of the syndrome is diagnosed.

Members of the proband's family may present only some of the main features of the "forme fruste". Generally, the patient is underweight, lean, thin, lanky and perpetually tired and often has a melancholic look. Poor development of the musculature and subcutaneous tissue exaggerates the apparent emaciation.

Skeletal Manifestations

Usually the skull is dolichocephalic, with a long narrow face, high forehead, and underdeveloped and underslung lower jaw. The palate is usually narrow and often high-arched,4 and the external ears are sometimes deformed. The extremities are unusually long, thin and slender. Both clinically and radiologically, the phalanges—particularly the terminal phalanges—and metacarpal and metatarsal bones are long and thin. The feet are long like "fox-feet". Anomalies of the thorax occur in about one in four cases. Pectus excavatum or funnel breast, and scoliosis, kyphosis or lordosis have frequently been recorded. Pectus carinatum or pigeon breast, winging of the scapulae, hemivertebrae and "fanning" of the ribs have also sometimes been observed.

The joints are often lax and hyperextensible. Subluxation of the knees and dislocation of the hips may occur. Méry and Babonneix24 drew attention to "high patella" in a further report of Marfan's original case. Booth et al.,5 similarly, recorded increase in length of the patellar ligament. Occasionally the syndrome is combined with arthrogryposis (such a case was observed in the Ottawa General Hospital).1

The muscles are usually poorly developed and flaccid. Amyotonia congenita was noted by Young28 in a patient with arachnodactyly. Dystrophia myotonica has been recently described by Booth et al.2 Pterygium colli (webbing of neck, an anomaly not generally associated with this syndrome) was present in two children with Marfan's syndrome, recently described by Golden and Lakin.16

Cardiovascular Anomalies

Piper and Irvin-Jones32 stressed the frequency of congenital cardiac anomalies. Baer et al.2 associated arachnodactyly with developmental abnormalities of the media of the aorta. Coyette and Palmer41 reviewed the cardiovascular anomalies in 33 cases of Marfan's syndrome from the literature and one of their own. McKusick25 summarized such anomalies as follows: (1) Aortic defects: (a) dilatation of aortic ring; (b) dilatation of ascending aorta; (c) dissecting aneurysm; (d) combination of a, b, and c; (e) coarctation; (f) patent ductus arteriosus. (2) Anomalies of pulmonary arteries: (a) dilatation; (b) dissecting aneurysm; (c) microscopic alterations of media. (3) Septal defects: (a) atrial; (b) tetralogy of Fallot. (4) Valvular abnormalities: (a) stretching and sacculcation of the aortic cusps; (b) other gross and microscopic
changes; (c) subacute bacterial endocarditis. (5) Dysrhythmias and conduction defects.

The association of aneurysm of the aorta and aortic insufficiency has been frequently reported in Marfan’s syndrome. The aneurysm may produce indentation of the oesophagus demonstrable by fluoroscopy after ingestion of barium. Such a case was reported by Nath and Mittal,26 Dimond et al.,11 published the report of an attempt at surgical repair with a Hufnagel plastic valve in a patient with this syndrome who had developed aortic insufficiency secondary to trauma. Miller and Pearson25 recently reported mitral insufficiency simulating aortic stenosis, an unusual manifestation in Marfan’s syndrome. They found a “jet lesion” consisting of thickened and elevated endocardium on the atrial wall opposite the mitral orifice. In another recent report, Burry1 described supra-aortic stenosis with this syndrome. Hardin18 reported rupture of an abdominal aneurysm and fistula formation between the aorta and vena cava: repair with nylon prosthesis was attempted.

Ocular Anomalies

Ocular manifestations of Marfan’s syndrome are of interest and often present as problems of visual disability in young children. Most important of these is luxation of the lens. Börger reported in detail a patient who had dislocation of lens and iridodonesis with arachnodactyly. Rados34 found typical ocular manifestations, including ectopia lentis in 127 and ocular symptoms of questionable significance in 40 among 204 cases of Marfan’s syndrome collected from the literature. Other associated anomalies reported are: cataract of lens, coloboma of lens, coloboma of iris, congenital mydriasis or miosis, poor response of pupil to the action of atropine, myopia, megalocornea, strabismus, and nystagmus. Arcus juvenilis was associated in a case of this syndrome described by Golden and Lakin.16 Black and Landy3 observed blue sclera in their cases. Gibson13 described extreme myopia with loss of convergence in one, and increased epicanthic folds in another case of arachnodactyly. King21 reported ptosis with ectopia lentis and arachnodactyly. Baer et al.7 observed unexplained exophthalmos. Colour blindness was associated in the cases reported by Neresheimer27 and Booth et al. Ougard and Bédard29 reported two cases of Marfan’s syndrome with bilateral ectopia lentis: they found old detachment of retina in one eye.

Urogenital Anomalies

Urogenital defects have rarely been described in cases of Marfan’s syndrome. Genital hypoplasia in males was described by Börger,4 and undescended testes, by Haridas19 and Stern.26 Renal ectopia has been found by Olcott28 and Fischl and Ruthberg.33 Booth et al.5 reported one case with polycystic kidneys, and another with congenital ureteric stricture.

Respiratory Anomalies

Anomalies of the lungs are rare and have been described by Börger.4 Imperfect lobation of the lungs was recorded by Rados.34

Mental Defects

The majority of patients appear mentally normal. However, Dax10 observed mental defect in a number of instances. Gibson15 described four cases of arachnodactyly in an institutional population, whose associated mental defect varied in grade from moron to idiot.

Case Reports

Case 1.—D.S., a six-year-old French-Canadian boy, was admitted to the Ottawa General Hospital on August 29, 1957, because of diminution of vision, lacrimation, photophobia and pain in the left eye for one week. The onset had been fairly sudden. He was unable to see properly in bright light but could watch television. He had a normal full-term birth, weighing 7 lb. Developmental history was apparently normal. Intelligence and behaviour were unremarkable. He appeared thin since birth. His long and slender limbs, fingers and toes had always caused difficulty in obtaining shoes of proper size. The right ear was congenitally deformed. The mother also had a deformed and “shrunken” right ear. One of the patient’s sisters had long thin fingers, toes and limbs; hypotonic musculature; high-arched palate, and myopia, but without any other apparent ocular abnormality. There was no heart murmur. The rest of his 10 brothers and sisters were said to be normal; no known history of abnormalities on the father’s side or in the preceding generation.

Physical examination revealed a lean and thin boy with a boat-shaped head and an elongated face. The left external ear was normal but the right one appeared prominent and deformed, was flat and flap-like in its upper part, and felt soft as if there was no cartilage. The palate was high-arched.

There was photophobia. Right eye: tremulous iris with dilated pupil; the lens, which appeared small, was found dislocated towards the temporal side (Fig. 1) and floating with a pendulum-like movement in the vitreous. Left eye: the lens was dislocated into the anterior chamber and was “trapped” by a well-constricted pupil (Fig. 1). It appeared like a floating drop of oil, changing its position with the position of the head. Considerable ciliary injection was present. No increase in intraocular pressure was apparent.
The limbs were long, slender and hypotonic; fingers and toes were unusually long and thin. A mild degree of scoliosis was present on the left side. The examination of other systems revealed no abnormality.

The lens was removed from the anterior chamber of the left eye. Eserine had been instilled before operation to keep the pupil constricted and the lens in the anterior chamber. Recovery was uneventful. In July 1959, he was wearing glasses which gave him useful vision in spite of some degree of amblyopia.

Case 2.—F.H., a six-year-old Irish-Canadian boy, was seen on May 14, 1959, in the outpatient clinic of the Ottawa General Hospital because of poor vision. He had a normal full-term birth and development. His intelligence and behaviour were normal. He had been of thin build since birth. One of his sisters, aged 14, was said to have long and thin limbs, fingers, feet and toes. His brothers and other sisters, seven in all, seemed normal.

Physical examination showed a slender boy with long and slender limbs, hands, fingers, feet and toes (Fig. 2). All limbs were hypotonic. Each patellar ligament measured approximately an inch (2.5 cm.) in length. The face was elongated and the palate was high-arched. There was no photophobia. When the pupils were dilated, the lens on each side was found to be dislocated posteriorly, upwards and to the temporal side (Fig. 3). Intraocular tension was apparently not increased.

Comments

In both of these cases the presenting symptoms were related to the dislocation of the lens. In Case 1, the anterior dislocation of the lens in the left eye was presumably spontaneous and gave rise to considerable ciliary reaction and constriction of the pupil which responded poorly to atropine. In cases of Marfan’s syndrome, it is difficult to detect any luxation of the lens in the presence of poor pupillary dilatation, sometimes even after instillation of atropine. Often there is amblyopia even with a lens of normal transparency and it is apparently caused by optical changes due to the fact that the equator of the lens occupies the optical centre of the eye. There is also superadded astigmatism of the lens.

Extraction of the lens is indicated in adults when cataract or loss of visual acuity interferes with occupation. In children, the problem is more difficult. Glasses should be prescribed, taking advantage of the aphakic portion of the eye and extraction of the lens delayed until the vision becomes inadequate. Choyce reported a case of Marfan’s syndrome, with spontaneous dislocation of the lens in the anterior chamber of the eye, where an attempt to replace the lens in the posterior chamber by the technique described by Das Gupta and Basu led to the onset of acute congestive glaucoma. In cases of anterior dislocation, he suggests removing the lens without delay. In the first case presented here removal of the lens from the anterior chamber greatly relieved the considerable ciliary reaction.

The etiology of Marfan’s syndrome is obscure. The disorders have been explained on the basis of congenital mesodermal dystrophy, determined early in embryonic life. Marfan’s syndrome is also ascribed to hormonal disturbance. Stern suggests a genotype change affecting both the mesoderm and endocrines. McKusick presumed it to be an abiotrophy involving the elastic tissue which wears out prematurely under the usual stress and strain. It has been found that about 40% of those with arachnodactyly, like the cases presented here, also had bilateral congenital ectopia lentis. Pino et al. explain it as a dystrophic change in the tunica vascularis lentis, a mesodermal derivative which invests the embryonic lens, or as due to a defective suspensory ligament.

A reasonable interpretation would be that two dominant genes are linked in this syndrome, one affecting the lens and the other the skeleton. Ectopia lentis, which is a strict dominant by itself, is frequently missing in histories of arachnodactyly, but this absence might be explained by an intergenic effect in which the ectopia is suppressed. If this explanation is correct, as Gates suggested, one would expect to find families with arachnodactyly but without ectopia lentis.
Scoliosis, dissecting aneurysms of the aorta, and other cardiovascular lesions, similar to those seen in Marfan's syndrome, have been experimentally produced in rats fed *Lathyrus oratus* (sweet pea) seeds, as reported by Posneti and Baird, and Walker and Wirtschafter. McKusick pointed out that although the basic defects were not identical in the natural and the experimental syndromes, such studies may provide a lead to the inborn errors of metabolism which might have formed the basis of the abnormality of the aorta in Marfan's syndrome.

Passow endeavoured to link up this syndrome with that of status dystrophicus, a mild form of syringomyelia, or faulty closure of neural tube. He noted its frequent association with Horner's syndrome and heterochromia iridica, which are both dependent on disturbance of the cervical sympathetic. Bremer had found long span, funnel chest, kyphoscoliosis, winged scapula and unequal mammary associates associated with syringomyelia in families. Ellis pointed out that in several other syndromes, skeletal and ocular abnormalities are both present, namely, gargoyleism or chondro-osteo-dystrophy and corneal opacity, apical dystrophy and coloboma, punctate epiphyseal dysplasia and cataract, and polydactyly with retinal dystrophy. Gates suggested the addition of brittle bones and blue sclerae. The similarity of some of these features of Marfan's syndrome with those of Ehlers-Danlos syndrome is interesting, and includes also hyperextensibility of the joints. Sometimes congenital lenticular anomalies are present in the Ehlers-Danlos syndrome (recently pointed out in a previous paper), for example, lenticular opacities, observed by Johnson and Falls.

**Summary**

A brief survey of the abnormalities which may be found in patients with Marfan's syndrome is given. The two patients described, from two different families, had bilateral ectopia lentis. The lens of the left eye of the first patient was dislocated into the anterior chamber.

I am indebted to Dr. J. D. Allen, associate professor of ophthalmology, for his help and permission to include his cases, to Dr. D. J. Conway, professor of pediatrics, University of Ottawa, for his suggestions, and to Miss M. Gauthier, for her secretarial assistance.

**Addendum**

Since this paper was submitted for publication, a case of Marfan's syndrome has been reported (Tuna, N.: *Dis. Chest*, 36: 204, 1959), in which the patient had a large aneurysmal venous dilatation in the right supraclavicular fossa about the size of a tennis ball, which used to enlarge at the height of congestive cardiac failure and disappear completely after improvement. Apparently, this type of venous anomaly has not been reported with this syndrome. The patient also had severe testicular atrophy and sexual impotence.

**References**

7. Cited by Gates.

**SHORT COMMUNICATION**

THE USE OF A RECTAL SUPPOSITORY OF BISACODYL (DULCOCAP) IN GERIATRIC PATIENTS* 

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In a large geriatric unit the maintenance of proper bowel control becomes an important aspect of patient care. Inadequate evacuation can frequently result in serious consequences, notably diarrhea, impaction, obstruction and, as well, urological problems secondary to the pressure of a loaded colon upon the bladder and sometimes upon the ureters. In order to prevent these problems, a careful review of bowel function is necessary in any unit which has a considerable number of bedridden patients. The use of repetitive enemas has been a standard method of ensuring adequate evacuation. These, however, are time-consuming.

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**Bisacodyl** is marketed under the trade-name of DulcoLax by Geigy Pharmaceuticals.