Case Report Section

Aneurysmal Venous Dilatation in Marfan’s Syndrome*

NAIP TUNA, M.D.**
Minneapolis, Minnesota

Since the original description of Marfan’s syndrome in 1896, over 350 cases have been reported in the literature.1 It has been accepted as an hereditary disorder of the connective tissue with widespread malformations of the musculoskeletal, the cardiovascular, and the ocular systems.2 The exact etiology and pathogenesis is not known. Of special interest is the experimental production, in rats fed Lathyrus odoratus seeds, of musculoskeletal and cardiovascular changes similar to those seen in patients with Marfan’s syndrome.3-5 A variety of manifestations associated with this syndrome have been reported. The most common ones are the following:

I. Musculoskeletal system: Dolichostenomelia, dolichocephaly, high arched palate, cleft palate, pectus excavatum, pectus carinatum, kyphoscoliosis, spina bifida, hemivertebra, winged scapula, hyperextension and subluxation of joints, pes planus, hernia, muscular hypotonia.

II. Cardiovascular system: Diffuse aneurysm of aorta, dissecting aneurysm of aorta, coarctation of aorta, aneurysm of innominate and common carotid arteries, patent ductus arteriosus, dissecting aneurysm of pulmonary artery, congenital and idiopathic dilatation of pulmonary artery, involvement of the semi-lunar valves, aneurysmal dilatation of aortic sinuses of Valsalva, involvement of atrio-ventricular valves and chordae tendineae, patent foramen ovale, interatrial septal defect, tetralogy of Fallot, and varicose veins.

III. Ocular system: Ectopia lentis, coloboma of lens, retinal detachment, abnormalities of lens and cornea (microphakia, spherophakia, megacornea, microcornea, keratoconus).

IV. Pulmonary system: Susceptibility to respiratory infections, developmental malformations of the lung, cystic disease of lung, spontaneous pneumothorax.

V. Renal system: Ectopic kidney with hydronephrosis, polycystic kidneys, atresia of the ureter.

This paper reports a case of Marfan’s syndrome with aneurysmal dilatation of a neck vein, venous varicosities, intractable stasis ulcer of the leg, testicular atrophy, and other more commonly seen abnormalities.

Case Report

V. R. No. 891028. A 42 year old farmer was hospitalized from November 23 to December 23, 1955, because of severe congestive heart failure, a mass in the right supraventricular fossa, and intractable leg ulcer. He first knew he had some difficulty with his heart when in 1940 he was rejected from military service because of a heart murmur. Prior to 1949, he was engaged in heavy farm work with no symptom. In 1949 his heart was apparently fibrillating and he was treated with digitalis and bed rest. He improved in about two weeks and has been on digoxin (0.15 mg. daily)

*From the Department of Medicine, University of Minnesota Hospitals.
**Instructor in medicine, University of Minnesota Medical School.
since that time. He had not worked much for the past four years because of inter-
mittent bouts of congestive heart failure. About four weeks prior to admission his
symptoms gradually increased. He developed paroxysmal nocturnal and marked exer-
tional dyspnea, retrosternal pain coming upon exertion, and increasing peripheral
edema. Three months prior to admission, during an episode of cardiac decompensation,
he noticed a swelling in the right supraclavicular fossa, about the size of a tennis
ball. This mass would become more marked on exertion, talking, lying down flat,
coughing or sneezing. There was no pain associated with the mass, but there were
pressure symptoms manifested primarily as a choking sensation in "getting in breath"
("seems to shut off my breath in one side").

His past history was significant in that at age six he had acute tonsillitis and ton-
sillectomy followed by Sydenham's chorea and an ill-defined illness of one year. There
was no history of joint and cardiac involvement. About four months prior to admission
he developed an ulcer on his right leg which seemed to heal for a few days to break
open again later. He was never married and apparently had never had sexual inter-
course or masturbated. Family history revealed that the immediate members of his
family, including the parents and six brothers and five sisters, were of average body
build. There was no history of cardiovascular disease in the family except for the
mother who had high blood pressure. Two sisters and three children (out of five
examined) of these two sisters had skeletal abnormalities such as pigeon breast, high
arched palate, kyphoscoliosis, arachnodactyly and pes planus. Cardiac examination
including electrocardiograms and cardiac fluoroscopy was normal.

Physical examination on admission revealed a tall slender man who appeared
chronically ill. There was mild cyanosis of lips. He was 74½ inches tall and weighed
180½ pounds. Fingertip-to-fingertip span was 78 inches and pubic symphysis-to-heel
dimension was 41½ inches (over half his total height).

The extremities including the phalanges were long and thin. There was marked
arachnodactyly of fingers and toes and bilateral pes planus. The palate was high-
arched and the auditory canals were wide and very short. Both ears were long and
pointed. The nasal passages were narrow. There was moderate kyphoscoliosis of the
thoracic spine. The left hemithorax was slightly larger than the right one. There
was a nontransluminate cystic mass, about the size of a tennis ball in the right
supraclavicular fossa (Fig. 1). This mass could be reduced by gentle pressure while
the patient was in a sitting position. There was a fine intermittent thrill and a to-
and-fro bruit of the venous hum type at the lower and medial extremity of the mass,
but there were no intrinsic pulsations. There were venous varicosities in the right
lower leg and a 1.5 cm. round, punched-out right pretibial ulcer that extended to the
periosteum. This was surrounded by a pigmented indurated area. There was a two
plus bilateral leg edema. The blood pressure was 138/72, the pulse irregular, and the
temperature 96.60°F. The heart was enlarged both to the left and to the right. The
point of maximum cardiac impulse was at the left anterior axillary line in the fifth

FIGURE 1: Venous aneurysm in the right supraclavicular fossa
FIGURE 2: Photomicrograph, testicular biopsy (X 175).

The left border of the cardiac dullness extended 3 cm. to the left of the anterior axillary line, and the right border was 4 cm. to the right of the sternum in the fourth intercostal space. The rhythm was irregular. There was a systolic thrill and a grade III systolic murmur at the apex which was transmitted to the left axilla. The first mitral sound was not accentuated. In addition, there was a grade IV harsh systolic murmur over the third intercostal space, at the left sternal border. P' was louder than A'. There were rales in both lung fields posteriorly. The liver was pulsatile and was four finger breadths below the right costal margin. Examination of the abdomen was otherwise negative. The penis was small and the testes were mushy and atrophic. The hair distribution appeared normal. Verbal scale I.Q. was found to be 77. Laboratory studies including red-cell and white-cell counts, hemoglobin, erythrocyte sedimentation rate (Westergren), blood urea nitrogen, glucose, electrolytes, basal metabolic rate, protein bound iodine, urinary excretion of follicle-stimulating hormone, and blood Kline test were within normal limits.

Urinalysis revealed one plus albumin and occasional red cells. The 24 hour urinary 17-Ketosteroid excretion was 4.9 mg. (N: 10-24 mg.). The venous pressure was 23.5 cm. citrate and the arm-to-tongue circulation time was 65 seconds. Needle aspira-
tion of the mass in the neck yielded dark red blood and there was no external distortion on gentle rotation of the needle through a wide arc. The oxygen saturation of this blood was 57.6 per cent, that of the right arm vein 55.5 per cent and the femoral artery blood 96.6 per cent. Biopsy specimens (Fig. 2) obtained from both testes revealed the seminiferous tubules to be completely lacking in spermatogenic cells. Intertubular cells were present and appeared normal. In some of these cells ceroid pigment was found. Electrocardiograms showed atrial fibrillation numerous ventricular extrasystoles from multiple foci, right axis deviation and right ventricular hypertrophy. Cardiac catheterization done on December 6, 1955, showed a pulmonary artery pressure of 87/42 mm. Hg, a right ventricular pressure 86/0 mm. Hg, and a right atrial mean pressure of 10 mm. Hg and no evidence of intracardiac shunt. Angiocardiography done at the same time did not reveal shunts at either the atrial or the ventricular level. Cardiac fluoroscopy showed generalized cardiomegaly with right and left ventricular enlargement and slight left atrial enlargement. No calcifications were seen. In addition, there was basilar emphysema and poor motion of the diaphragma. Postero-anterior and lateral chest X-rays revealed marked cardiomegaly with apparent generalized enlargement (Fig. 3). Both hilar shadows were prominent. There was rather marked pulmonary congestion and edema, granular appearance throughout both lungs, and considerable basilar emphysema. There was a large soft tissue mass in the lower neck on the right side. In addition there was marked degree of kyphosis of the dorsal spine. X-rays of the esophagus, stomach, duodenum, and intravenous pyelograms were negative. Barium enema showed several diverticula of the sigmoid colon.

He was treated with low sodium diet, oral potassium chloride (2 grams three times daily), oral proestinyl (250 mg. four times daily) and parenteral mercurial diuretics. Digoxin was discontinued. On this regimen, the patient showed considerable improvement. The ectopic beats disappeared and he lost 20 pounds in the first week. After his general condition improved the mass in the right neck disappeared completely, and the chest X-rays revealed marked reduction in heart size and considerable clearing of the congestion and of the granular appearance of the lungs. He was discharged from the hospital on December 23, 1955 on a maintenance dosage of digitals. The stasis ulcer showed some improvement on conservative treatment, but did not heal completely. During 1956, he was admitted to the hospital three more times, each time in congestive heart failure. During each admission the large mass in the right fossa supraventricularis would become prominent upon correction of the congestive heart failure. The leg ulcer persisted as an intractable stasis ulcer although there was marked reduction in its size.

DISCUSSION

This man has many of the characteristic clinical features of Marfan's syndrome which permit diagnosis at a glance. Ocular abnormalities which are seen in up to 75 per cent of cases having this syndrome are absent in this case. The cardiovascular system involvement which is seen in 30 to 60 per cent of cases appears to be limited to heart and veins. Aorta, pulmonary arteries, and the other major arteries are grossly normal. The main lesion in the heart appears to consist of mitral valvular disease with mitral incompetence. The pulmonary hypertension and the left and right heart failure are most likely secondary to mitral incompetence. There were no intracardiac shunts demonstrable by cardiac catheterization or angiocardiography.

It is not possible to tell whether the mitral lesion was due to rheumatic valvulitis or to noninflammatory process as often seen in Marfan's syndrome. It has been shown that the main valvular changes in Marfan's syndrome consist of thickening and shortening of valves and the chordae tendinae and microscopically the lesions have been described as being of a fibromyxomatous character. There is a high incidence of aortic involvement in this syndrome. The nature of aortic lesions has been well outlined. Cases of Marfan's syndrome have been reported with death due to dissecting aneurysm of the aorta developing after trauma. The unusual hazards of trauma in this condition have recently been emphasized. Rupture of aortic valves and death due to dissecting aneurysm of inominate and carotid arteries have also been reported.

Several authors have mentioned the association of varicose veins and varicose ulcers with Marfan's syndrome; however it is not possible from the reported cases, to get an exact idea about the incidence of venous anomalies in this condition. It has been assumed that the loss of the subcutaneous support to the veins encourages the production of varicosities and intractable varicose ulcers. The large aneurysmal venous dilatation in the right supraclavicular fossa of this patient is an interesting finding. To our knowledge, this type of venous anomaly has not been reported in Marfan's syndrome. From the behavior of this mass, one is forced to conclude that it is probably due to weakness and not obstruction and the connective tissue and the vascular wall. It is interesting to note that this mass becomes largest at the height of congestive heart failure when the venous pressure is elevated and disappears completely after the patient improves.

From the reported cases one gets the impression that patients with Marfan's syndrome do not have abnormalities referable to their sexual organs. The case reported here has severe testicular atrophy and sexual impotence.
REFERENCES


