A Case of Medionecrosis Aortae Cystica Idiopathica (Erdheim) with Congenital Hypoplasia of Myocardium

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MEDIONECROSIS aortae cystica idiopathica (Erdheim, 1929)1) has been rarely reported in the literature. As this lesion of the aorta is also often noted in Marfan syndrome, the concept that the idiopathic dilatation of the aorta is a forme fruste2) seems a possibility3) and is a matter of recent interest from the point of view of the chemical alterations of the aortic wall.

The hypoplasia of the myocardium in this case is also extremely rare.4) Such a case with these two lesions is reported in this paper and the etiologies are discussed.

CASE REPORT

The patient was a 43-years-old, male, school teacher, who was transferred from Sakamachi hospital on Nov. 15, 1963, because of dyspnea, palpitation and edema for 2 months. He had been healthy and exercised well while he was young. He became to develop the shortness of the breath one year ago when he exercised, though he was able to go to school without much difficulties for 10,000 M. on a path crossing the mountain in the snow-storm in the last winter. In August in 1963, he had to take a rest for about 2 hours, when he arrived at the school, because of the above mentioned complaints. At that time he was pointed to have the cardiomegaly and came to our clinic on Sept. 16, 1963. At the clinic the physical examination revealed the cardiomegaly with systolic murmurs over the left precordial area. The chest X-ray film (Fig. 1) showed diffusely enlarged cardiac silhouette and the B-line of Kerley was noted at the right lower lung area. The electrocardiogram (Fig. 2) showed left axis deviation, rsR’ complex in the lead V1, SV1+SV6>65 mm., RV6>50 mm. and the depression of the ST segment in the lead II, aV6, V6 and V6. T waves were decreased in height or inverted in all leads. Proteinuria was noticed.

On Sept. 21, dyspnea and arrhythmia appeared at home, and he was admitted to Sakamachi hospital. The electrocardiogram revealed atrial fibrillation. Digitalis, diuretica and other cardiotonic substances were ineffective, and the patient was transferred to our hospital on Nov. 15.

Past history: Sinusotomy, appendectomy and acute nephritis at the age of 16. V. D. was denied.

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Family history: No special ocular disease, including myopia and blindness, and the height and the body build were within normal limits in the 3 generations. Grand-father and grand-mother died at the age of 70. Father died at 72 years old because of acute cerebrovascular accident. Mother died at 50 years old because of acute abdomen. The siblings were 5 and one of them died at 10 days after birth because of malnutrition, though the rest of them were healthy.

Physical examination on admission revealed poorly nourished (weight, 49 Kg.) and moderately developed (height, 159 cm.) male with well balanced constitution.
in acute distress. Head: normal. Eye: myopia. Mouth, throat and neck: normal. Lips were cyanotic. The thoracic wall was normal. Cardiovascular: cardiomegaly (Fig. 1) and cardiac murmur (Fig. 3), which will be described below, were noticed. B.P.: 120/50 mm. Hg. Chest: dull by the percussion at the bases of both lung fields and there the crepitant rales were audible. Abdomen: palpable liver of 4 cm. at the right mid-clavicular line. Otherwise, normal. Edema at the lower part of the anterior tibia was noticed. Neurological: normal. Lymph nodes: not palpable. Musculoskeletal: normal. No arachinodactyilia.

Laboratory examinations: On admission, RBC 6.15 × 10⁶, Hb. 100% (by Sahli method), Ht. 49%, WBC 8,300 with differential, N. 78, Band 3, M.2, L. 17; Urinalysis, proteinuria (from 0.1 to 1.0 Gm. per day) and normal sediment; Coprology, negative occult blood reaction and no parasite ova; ESR, 2 mm. in one hour; Liver function tests, normal; Electrophoresis, T.P. 5.1 Gm./100 ml., alb. 68.4%, alpha-globulin (G.) 6.8%, beta-G. 9.0%, gamma-G. 15.8%; Cholesterol, 158 mg./100 ml.; Electrolytes, normal; Wassermann’s test, negative; CRP, negative; ASLO, less than 100 u.; Latex test, negative. The chest X-ray (Fig. 1) revealed the more diffusely enlarged cardiac silhouette. The phonocardiogram (Fig. 3) showed at the apex the decreased I sound in the intensity and the diastolic murmur of decrescendo after the III sound was seen more clearly in the low frequency record. At the left 3rd intercostal space the systolic and the diastolic sounds were noticed. The maximal point of these murmurs was at the aortic area and there the 2nd cardiac sound increased in the intensity. The electrocardiogram (Fig. 2) showed left axis deviation, atrial fibrillation, same QRS complex and more depressed ST and T segments in all leads.

Fig. 3. Phonocardiogram. HF: High frequency, LF: Low frequency.
Hospital course: The diagnosis was made from the above mentioned data as aortic valvular insufficiency with mitral valvular insufficiency of the obscure origin, though Austin-Flint murmur could not be ruled out by the phonocardiogram. Digitalis, diuretica and coronary vascular dilators were used. Though he was improved temporarily, the congestive heart failure was resistant to these therapeutic maneuvers. On the 24th hospital day he became unconscious suddenly. The external cardiac massage was given immediately. The electrocardiogram revealed ventricular fibrillation. The external defibrillation was applied and the cardiac asystole was induced. The idioventricular rhythm appeared and the blood pressure of 120/80 mm.Hg was maintained by norepinephrine dripping intravenously, but the pressure decreased gradually and these maneuvers became ineffective after 3 hours.

Autopsy Findings:

Gross findings: The important findings related to the aorta and the heart were as follows. A saccular aneurysm of 6.5 cm. in width and 4 cm. in length was found just above the aortic valves. The aortic valves were depressed below and the edges of them were thickened as the club and resulted in the aortic valvular insufficiency. Other valves appeared macroscopically to be intact. The heart weighed 585 Gm. and was enlarged as a whole. The left ventricle of the heart (Fig. 4) was dilated remarkably. The hypertrophy of the distal parts of the outflow tracts of the both ventricles was slightly seen. The myocardium of the ventricles became aneurysm-like dilatation (Fig. 4). A thrombus of the tip of the thumb-size was seen in the left auricle. The coronary arteries were slightly arteriosclerotic and the anomalies of them were not seen. The haemopericardium of
Fig. 5. The left figure is the microscopic film of the aorta of hematoxylin-eosin stain showing the aortic idiopathic medial necrosis. The top is the intima and the bottom is the adventitia. The upper two-thirds of the figure is the affected media in which the tone becomes lighter because the collagenous fibers increase instead of the elastic fibers. At the middle of the figure, the spaces are formed by the deficit of the elastic fibers and are filled with the mucoid substances. The lower one third is normal.

The section of the thinned myocardium near the apex of hematoxylin-eosin stain is shown at the right. The left side of the figure is the endocardium and the right is the epicardium. No fibrosis is shown, though the infiltration of the small round cells are seen. The myocardium itself is thin and the muscle fibers are scarce in numbers.

150 ml. of the effusion was noted with the perforation of a pea-size which might be caused by the cardiac massage, and the fracture of the sternum and ribs were also seen. The pleural cavities contained 1,050 ml. of the effusion in the left and 300 ml. in the right. Congestion was seen in the liver, spleen and kidneys.

Microscopic findings: Aorta: The inner two-thirds of the media of the aortic aneurysm contained much mucoid substances. There were shown the fragmentation and the sparsity of the elastic and muscle fibers and the increase of the collagenous fibers without the inflammatory cells. The outer one-third of the media was normal, and in the border of these two parts the cystic degenerations were seen remarkably (Fig. 5). The mucoid substances in these lesions showed toluidine blue metachromasia. These changes were diagnosed as a "medionecrosis aortae cystica idiopathica (Erdheim)." These changes, though slight in degree, were seen throughout the aorta down to the common iliac arteries. The pulmonary arteries were also affected.

Heart: The muscle fibers were hypertrophic but they were particularly small in numbers in the apical part of the ventricular myocardium as the Fig. 5. The small ischemic scars and the infiltrations of small round cells were found in the myocardium in the slight degree, though it was thought that the scars were not enough to produce the thinning of the ventricular wall. The mucoid substances were found rich in the ventricular endocardium and in all valves especially at the edges of the aortic valves.

The bony tissue with marrows was noted in the bronchial cartilage.
DISCUSSION

Clinically this case was thought to have aortic insufficiency with possible mitral stenosis of the unknown etiology. The aortic aneurysm was only suggestive from the aortic failure. The usual causes of the aortic lesion including syphilis, rheumatic disease and so forth, were denied from the clinical and laboratory findings. His family history was negative for Marfan syndrome and the muscular diseases. Even in retrospect, the Erdheim’s necrosis\(^2\) was a possible diagnosis in the differential diagnosis of this case.

The etiology of the Erdheim’s necrosis\(^1\) is going to be elucidated. The hypoxia\(^5\) especially by the hypotension was suggested. Also the medial hypertrophy and the intimal hyaline thickening of the vasa vasorum by arteriosclerosis were thought to be the exact findings of this disorder,\(^5\)\(^-\)\(^7\) though other papers\(^8\),\(^9\) contradict to it presenting the cases without noticeable changes of the nutrient vessels in the aortic wall and also the dissection of the aorta in the medial degeneration in the younger group than in the arteriosclerotic disorder.\(^10\) The developmental anomalies of the nutrient vessels of the aorta\(^11\) are doubtful.\(^3\) Syphilis has no relation to this disorder.\(^5\) The recent development in the investigation in the chemical and metabolic changes of the connective tissue of the aortic media became to reveal the more exact mechanism involved in this disorder comparing with the aortic lesion in Marfan syndrome.\(^5\),\(^12\) The predominant vascular lesion in the lathyrism which may be induced experimentally\(^13\)-\(^16\) is the destruction of the elastic fibers, accompanied by the edema and the proliferation of the fibroblasts which may result in the dissecting aneurysm. This disruption of the elastic fibers is the result of the disturbance in the formation of the new fibers rather than the elastolysis,\(^17\) and the increase of mucopolysaccharide in the aortic media is noted.\(^18\) The microscopic findings of the aorta in this case was typical to the Erdheim’s necrosis. Involvements of the arteries were also seen throughout the aorta to the common iliac arteries and in the pulmonary arteries, though the media that was cardiopetal from the innominate artery was usually involved.\(^3\) It should be noted that these medial changes may occur in the normal persons, especially in those of old age group.\(^19\) But in this case the arteriosclerotic changes of the nutrient vessels were only slight and the mucopolysaccharide in the aneurysm was found by toluidine blue (metachromasia) stain to be increased remarkably.

The ideas that the idiopathic medial necrosis of the aorta is a forme fruste of Marfan syndrome or not\(^2\) deserve further investigation.\(^9\) The Marfan syndrome is thought to be a heritable disorder of the connective tissue, characterized by the widespread malformations affecting the musculoskeletal,
cardiovascular and ocular systems. A gene may have or reduced penetrance and the latter will produce the so-called forme fruste of a given syndrome, though this is only rarely reported. If the sporadic form with only cardiovascular involvement similar to the Marfan syndrome occurs, is it right to give the diagnosis to this case as a forme fruste of the Marfan syndrome? The idiopathic medial necrosis may be a disorder of this rare type because it is a non-heritable disease with the aortic aneurysm due to the cystic medial degeneration. This point is difficult to prove or disprove as the pathological studies of the media of these 2 diseases are the same results. The exact relation of these 2 disorders is still unsettled, but it would be thought that these may be the same lesions caused by the metabolic disorder of the aortic media.

The cardiac lesions in this case were the myocardial hypoplasia, the cardiomegaly due to the aortic insufficiency and the small myocardial fibrosis due probably to the coronary ischemia. The congenital myocardial hypoplasia is a rare disorder. The disorders seen only in the left ventricle or the right ventricle have been reported in 7 cases. Also the diverticule or the aneurysm of the heart was reported in 21 cases (Kucsko, 1964) in the left atrium, in the left ventricle or the right ventricle. Only the description of the parchment heart (Osler) is similar to this case about the thinning of the myocardium, though its description is incomplete. In this case, the myocardium was thought slightly hypoplastic and the muscle fibers were small in number especially in the apex. The heart, however, weighed 585 Gm. and was hypertrophic as a whole. Neither the aortic insufficiency nor the small myocardial fibrosis was enough to explain the excessive thinning of the myocardium at the apex. So the congenital factor would be the true nature as a cause of the hypoplasia rather than the same moment to the aortic media, though the mucoid substances were found in the endocardium. The combination of these 2 lesions was difficult to explain from the same etiology because even in the Marfan syndrome no such a combination has not yet been reported. But it will be permitted to say that the muscular metabolism in the cardiovascular system in the fetal period played an important role in these disorders.

**Summary**

A case of medionecrosis aortae cystica idiopathica (Erdheim) with congenital hypoplasia of the myocardium is presented with the pathological studies. A review of the literature disclosed no similar case.
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REFERENCES