MRI OF A CHILD WITH MARFAN SYNDROME

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ABSTRACT

We report an 8-year-old girl with Marfan syndrome who had dilatation of the aortic root and ascending thoracic aorta, seen on MR imaging. Besides these anatomical abnormalities, aortic regurgitation was also seen on MR imaging, which is a good noninvasive method to diagnose aortic abnormalities in children with Marfan syndrome.

INTRODUCTION

Marfan syndrome is a generalized connective tissue disease primarily involving elastic tissue which results in ocular, skeletal, and cardiovascular anomalies. This syndrome is diagnosed more often in adults than children. One reason is that most manifestations of Marfan syndrome become more evident with time.[1] Echocardiography, computed tomography (CT) and magnetic resonance (MR) imaging are noninvasive methods to diagnose thoracic aorta abnormalities in these patients. We present a child with Marfan syndrome whose MR imaging showed not only dilatation of the aortic root and ascending thoracic aortic but also aortic regurgitation.

CASE REPORT

An 8-year-old girl was first evaluated for impaired vision 3 years ago. There was no history of Marfan syndrome in her family. Pertinent results of physical examination included dislocation of the lens of both eyes and signs of moderate to severe aortic regurgitation. Her weight was proper for her height (weight to height ratio = 91%). She had no apparent tall features (height to age ratio = 96%) but had arachnodactyly and a decreased upper to lower trunk ratio. On palpation cardiac examination revealed the apex was downward. Aortic regurgitation murmur was heard at the upper left sternal border. Chest radiographs showed cardiomegaly with an enlarged left ventricle and dilatation of the ascending aorta (Figs. 1 and 2). Echocardiography showed a huge dilated aortic root and moderately severe aortic regurgitation. Cardiac MRI with T1-weighted spin echo technique showed dilatation of the aortic root and ascending aorta and dilatation of the left ventricle (Figs. 3 and 4). A cine phase contrast study showed aortic regurgitation jet extending all the way to the cardiac apex (Fig. 5). She underwent aortic root graft and aortic valve replacement with uneventful recovery.

DISCUSSION

Marfan syndrome is a hereditary disorder of connective tissue, inherited as an autosomal dominant. In approximately 15 per cent of cases no other family member has signs of the syndrome.[2] The clinical expression of this syndrome varies. In its classic form, there are

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abnormalities of the eye (ectopia lentis), aorta (aneurysm of the ascending aorta and aortic regurgitation) and skeleton (limbs disproportionately long compared with the trunk, arachnodactyly, pectus deformity and mild joint laxity). Pyeritz and McKusick[2] have based the diagnosis of this syndrome on the four criteria of characteristic family history and ocular, cardiovascular, and skeletal features. They thought that it was prudent to require at least two of these criteria to make the diagnosis.

In our patient, the diagnosis of Marfan syndrome was made based on 3 criteria: ocular (ectopia lentis), cardiovascular (aortic root dilatation and aortic regurgitation), and mild skeletal signs (arachnodactyly and increased length of the limbs compared with the trunk (decreased upper to lower trunk ratio).

In the past the term "Forme fruste" was used when a person had this syndrome with all the manifestations except tall features.

Phornphutkul et al[3] examined 36 patients younger than 16 years of age and found that mitral regurgitation was the most frequent cardiac lesion (47%). Dilatation of the aortic root was also present in a significant number of children (28%) with Marfan syndrome with no apparent aortic regurgitation. This regurgitation will usually develop later on in life.

Chest radiographs are usually not helpful in detecting enlargement of the aortic root. Since the aortic root is within the cardiac silhouette, dilatation or an aneurysm of it is usually not recognized in chest radiographs unless they are very large.

Both echocardiography and CT have been used to make initial measurements and monitor the diameter of the thoracic aorta. Echocardiography is noninvasive and relatively inexpensive, but without good resolution it may not be able to demonstrate the entire thoracic aorta.[4] CT allows one to diagnose an aortic aneurysm and aortic dissection, which are important causes of death in these patients. Relative disadvantages of CT are exposure of patients to ionizing radiation and the need for contrast media.

MR imaging has been shown to be useful in diagnosing aortic abnormalities in these patients.[4,5] It is a noninvasive method showing anatomical abnormalities, as in our patient. It also shows pathophysiology, such as aortic regurgitation.

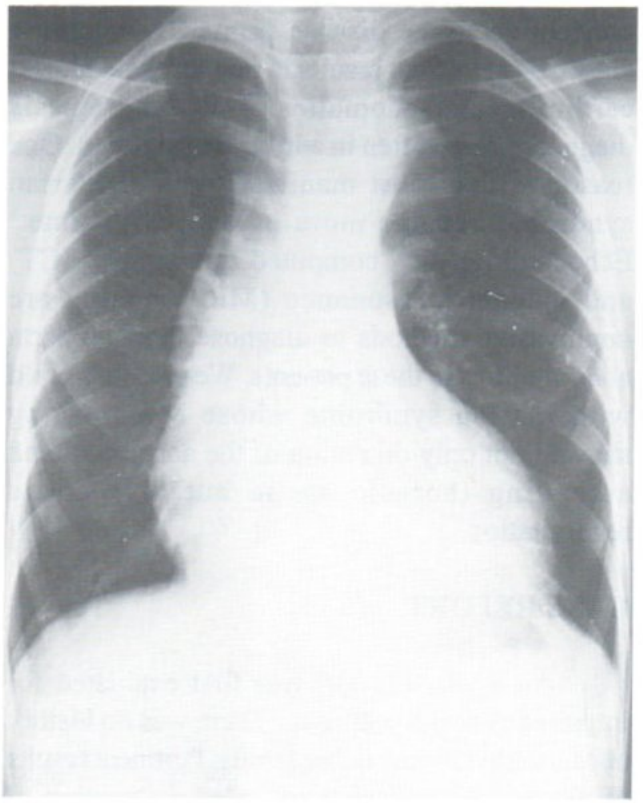

Fig.1 Frontal chest radiograph shows cardiomegaly and dilatation of the ascending thoracic aorta.

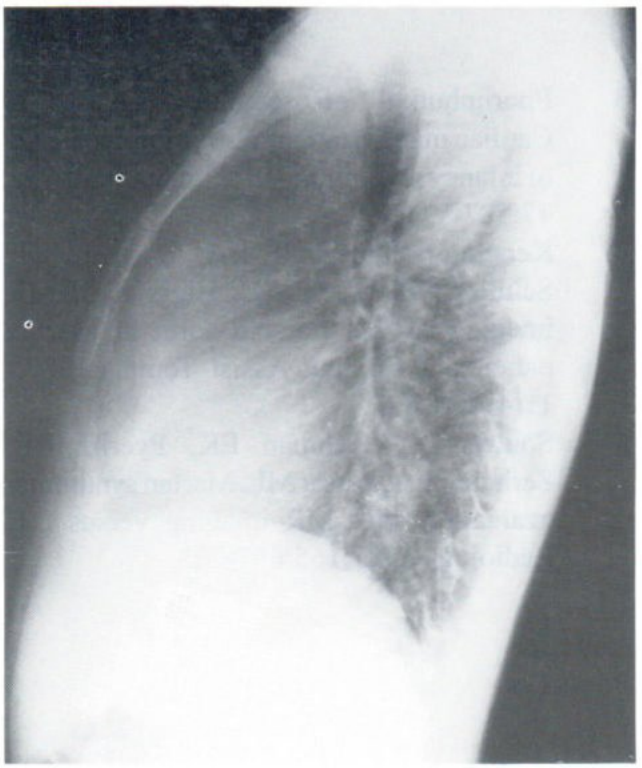

Fig.2 Lateral chest radiograph shows obliteration of the retrosternal clear space by the dilated ascending thoracic aorta.

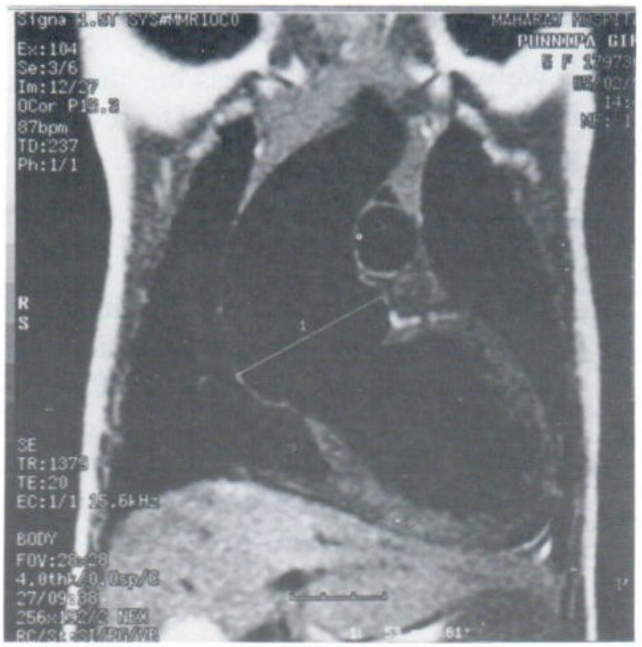

Fig.3 Coronal MR image shows a huge dilatation of the aortic root and the ascending thoracic aorta. Note the dilated left ventricle.

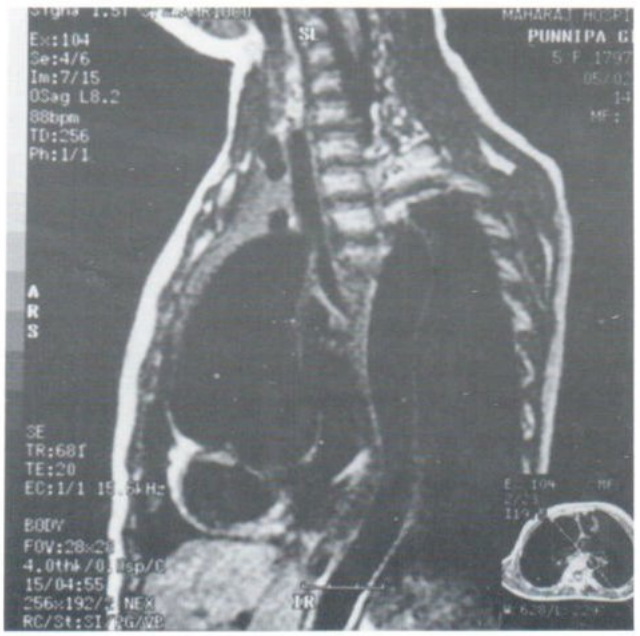

Fig.4 Oblique sagittal MR image shows a huge dilatation of the aortic root and the ascending thoracic aorta.

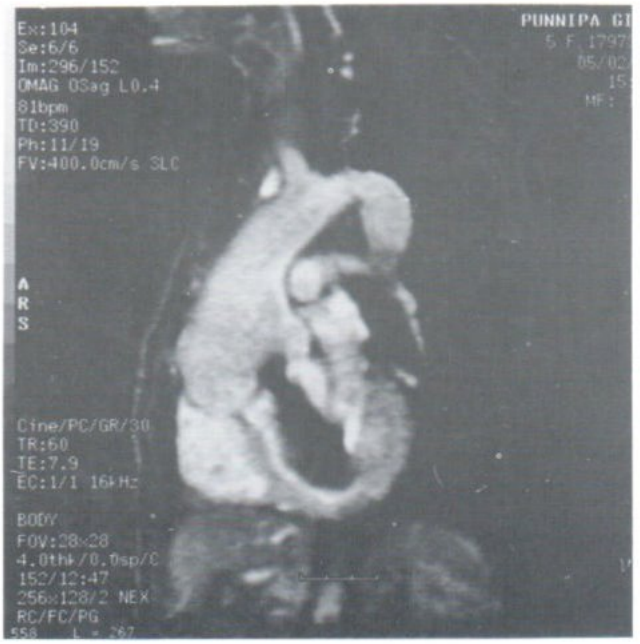

Fig. 5 Phase contrast oblique sagittal MR image shows regurgitation of blood from the aortic root into the left ventricle (signal void in left ventricle).

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