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ORIGINAL ARTICLE

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Aortic dilatation in Turner syndrome: the role of MRI in early recognition

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Abstract ***Background***: Aortic dilatation and dissection are rare but important complications of Turner syndrome that increase the risk of sudden death in young patients. ***Objective***: To assess the value of aortic MRI in patients with Turner syndrome; in particular to demonstrate early aortic dilatation. ***Materials and methods***: A total of 21 patients with Turner syndrome underwent MRI of the thoracic aorta with measurement of vessel diameter at four levels. ***Results***: Measurements were normal for age in 15 cases, two patients presented with values at the upper limit of normal and four had obvious dilatation of the ascending aorta. All were symptom free. ***Conclusions***: MRI allows the non-invasive demonstration of early aortic dilatation, which may lead to earlier surgery in asymptomatic individuals.

Keywords Aorta · Dilatation · Turner syndrome · MRI · Children

Introduction

Turner syndrome may be complicated by aortic dilatation, dissection and rupture [1], but studies attempting to establish the prevalence of these complications are uncommon. One of the earliest publications reporting the frequency of aortic dilatation in Turner syndrome recorded aortic ectasia in 8.8% of a population of 57 patients [2]. In 1998, the same group of workers reported ectasia of the ascending aorta in 6.8% of patients with Turner syndrome, two-thirds being complicated by dissection [3].

Although the prevalence of these aortic anomalies in populations of Turner syndrome patients has been studied only recently and is poorly documented, the importance of specific risk factors is well understood [4]. These predisposing factors are aortic dilatation, severe aortic valve stenosis related to bicuspid aortic valve, aortic valve dysplasia and systemic hypertension. In most cases, cardiovascular anomalies have been detected before the discovery of a clinical complication. Thus, girls with Turner syndrome may present before 30 years of age with a classic pathological sequence of fusiform dilatation, dissection and rupture.

The aim of this study was to determine the benefit of aortic MRI in young patients regularly attending a department of endocrinology and without specific cardiac follow-up. We wanted to use a non-invasive method to demonstrate the incidence of aortic anomalies, accessible to preventive therapy, in a population without any significant symptoms.

Material and methods

Our study group comprised 21 patients with Turner syndrome, initially identified because of short stature and/or delayed puberty. The diagnosis was established by karyotype analysis. All the patients were followed up in an endocrinology department, but not in a cardiology unit. The age range at diagnosis was 2–14 years 9 months (mean, 8 years and 3 months). Patients underwent MRI between 5 years 6 months and 19 years (mean, 14 years and 9 months). All patients underwent complete evaluation, including clinical examination, karyotype, ECHO and MRI of the thoracic aorta. Informed consent was obtained from the parents or directly from the patient if she had reached the age of majority.

For the MRI study we employed a 1.5-T system (General Electric Medical Systems, Milwaukee, WI, USA) and executed 'black blood' turbo-spin-echo (TSE) T1-weighted (T1-W) sequences (TR/TE, 700 ms/60 ms; field of view, 46×34.5 cm; matrix, 256×192; slice thickness, 8 mm; number of excitations, 4). The duration of the examination was approximately 15 min. Contrast medium was not administered and no patient required sedation. Images were obtained in the axial and parasagittal planes. Scan interpretation and measurements were made by two radiologists who were blinded to the other results of cardiovascular assessment. Cardiac structures, aortic wall and aortic diameter were assessed.

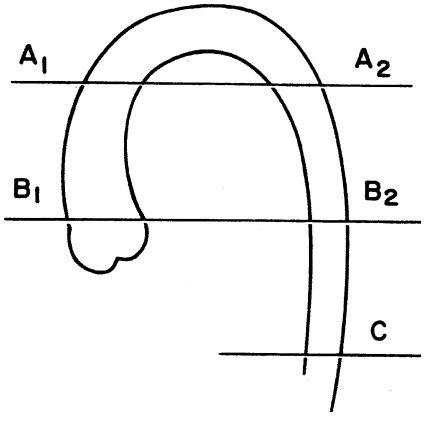

Fig. 1 Reference planes

The ascending and descending aorta diameter was measured at four levels, as indicated in Fig. 1 [5]. Data were compared with reference values obtained from CT of a normal paediatric population ranging from 2 to 19 years of age [5]. Measurements over the 95th percentile were considered pathological, while diameters equal to the 95th percentile were called 'limit values'. MRI was always undertaken soon after the ECHO, without significant delay.

Results

All 21 patients had proven Turner syndrome; the karyotype distribution was XO monosomy ($n=11$; 58.4%), XO mosaicism ($n=9$, 42.9%) and constitutional chromosome anomaly (isochromosome; $n=1$, 4.8%).

Normal aortic dimensions were measured in 15 (71%) children (Fig. 2). Aortic diameters were at the upper limit of normal in two (10%) patients. True dilatation of the aorta was identified in four (19%) patients (Figs. 3, 4). In the latter group, two patients (a 19-year-old girl with a bicuspid aortic valve and a 7.5-year-old girl with no other cardiac abnormality) showed obvious fusiform dilatation of the ascending aorta. In the other two patients (a 5.5-year-old girl and a

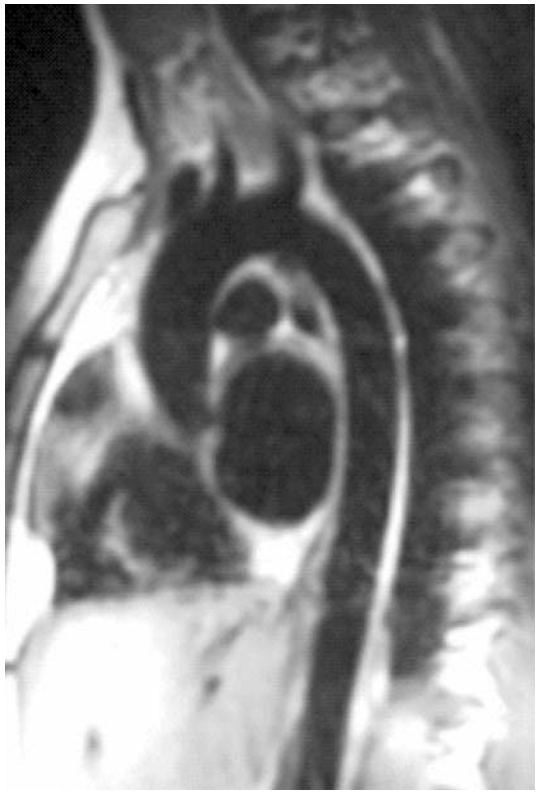

Fig. 2 Sagittal T1-weighted MRI of the normal thoracic aorta

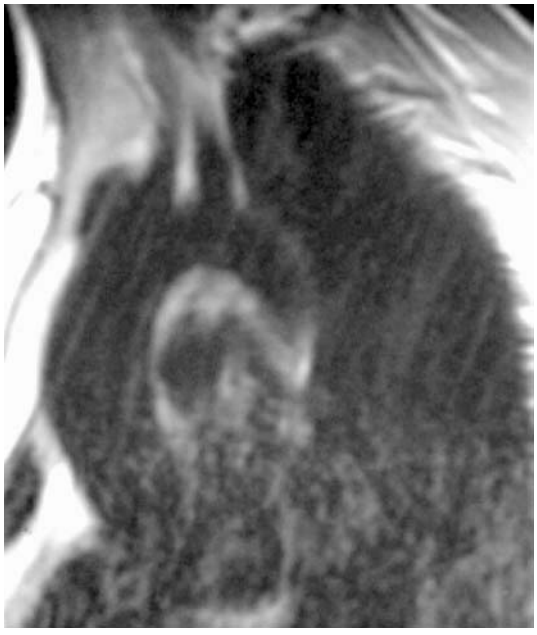

Fig. 3 Parasagittal T1-weighted MRI showing proximal ascending aortic ectasia

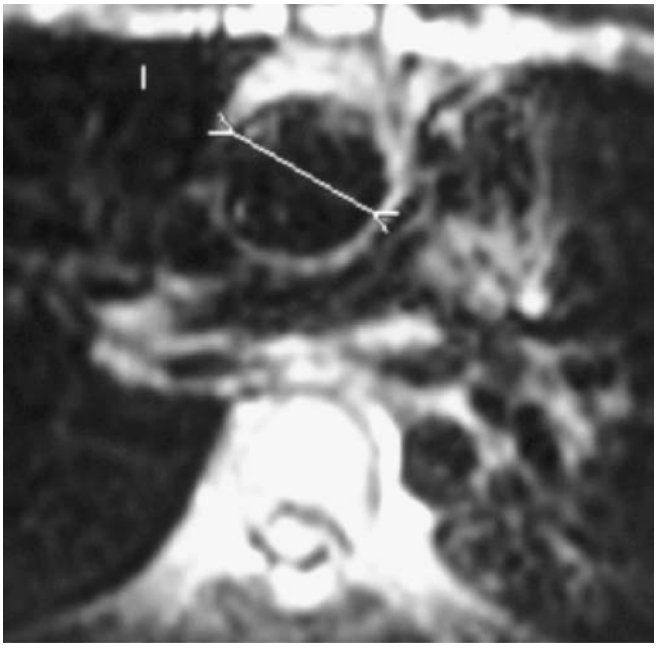

Fig. 4 Axial T1-weighted MRI showing proximal ascending aortic ectasia

15-year-old girl), aortic dilatation was mild. Neither aortic coarctation nor parietal cyst suggestive of cystic medial necrosis was observed. Only one of the four dilatations seen on MRI was detected by ECHO.

Three of the four patients with aortic ectasia were shown to have XO mosaicism. There was no ectasia in the patient with an isochromosome.

The US examinations revealed some pathological findings. One patient had both an asymptomatic bicuspid aortic valve and ascending aortic ectasia. A mildly stenotic subaortic membrane was identified in one patient. Two patients had mitral valve dysplasia, one having additional aortic valve dysplasia. Overall, US abnormalities were found in 23.8% of the patients.

In this study, no patient had aortic coarctation.

From a clinical perspective, there was no functional disorder among this group. Moreover, every girl had normal systemic arterial blood pressure. Cardiac auscultation found two B2 clicks related to mild aortic regurgitation and an ejection midsystolic murmur.

Discussion

Turner syndrome was first described in 1938 [6]. Its main features are delayed puberty or infertility, short stature, osseous disorders, facial dysmorphism, urinary tract malformations and cardiovascular diseases. In the last group of anomalies, we mainly find aortic coarctation or pseudocoarctation (kinking) and bicuspid aortic valve (about 20–50% [7]) as well as mitral and aortic valvular dysplasia, atrial or ventricular septal defects, partial anomalous pulmonary venous return, dextrocardia, aberrant subclavian artery and single coronary artery. These pathological elements are found exclusively in the left-sided circulation, whereas the anomalies encountered in Noonan's syndrome affect the right-sided circulation [8].

In our series of 21 patients, we did not encounter any individual with aortic coarctation, either with ECHO or with MRI, despite its high incidence in the population with Turner syndrome. This may be due to the fact that our patients are followed up for endocrine disorders and never had any cardiac or vascular symptoms. This result can be compared to the study of Campbell and Polani [9], which reported only 8% of patients with aortic coarctation in a non-selected group. Our other results are much more in accordance with the literature. Indeed, we found cardiovascular anomalies in 23.8% of patients using ECHO (4.8% with aortic bicuspid valve and 19% with other valvular pathologies).

Transthoracic ECHO seems to be more sensitive than MRI for the detection of valvular pathologies, but may be less efficient for aortic screening [10, 11]—one proximal aortic dilatation detected by ECHO versus four dilatations observed by MRI. This incidence is particularly high if we consider that no patient had elevated blood pressure or coarctation, classic risk factors for aortic ectasia in this context. We could have used

transoesophageal ECHO, but it is too invasive for a screening examination [12].

Even if we exclude risk factors such as coarctation, bicuspid aortic valve, severe aortic valve stenosis and elevated systemic blood pressure [13], these young patients seem to present with a specific mesenchymal pathology—cystic medial necrosis [14]. These patients may have a dramatic lethal event before the third decade, even younger than those observed in Ehlers-Danlos and Marfan syndromes (populations also with a predisposition for aortic ectasia). Aortic ectasia may occur in an asymptomatic girl [15] or young woman without any risk factor [16, 17].

With regard to karyotype, we note that in our series aortic anomalies are more frequently discovered in patients with XO monosomy than in those with mosaicism [18]. This is not surprising, knowing that dysmorphic and endocrine disorders are more severe in XO monosomy than in mosaicism and constitutional chromosomal anomalies.

We report two aortic diameters at the upper limit of normal. It should be noted that the reference values for aortic diameter in the paediatric population are correlated to age and not to body surface area or weight. Thus we can speculate that our two limit values are in fact above the upper limit of normal because of the reduced height of our patients.

In our opinion, monitoring should be undertaken using MRI, the most appropriate modality allowing complete evaluation of the thoracic aorta [19]. Moreover, in about 15 min we obtained accurate measurements with this non-invasive technique requiring neither sedation nor contrast medium injection. Aortic screening should be undertaken regularly, even if the first examination is normal. In fact, physiopathological mechanisms of the disease may be slowly progressive. We suggest performing MRI every 2 years, in order to establish the diagnosis as soon as possible and eventually to propose surgical repair before rupture. However, this interval can be modified depending on additional risk factors. In conclusion, because of the high risk of thoracic aortic dilatation in patients with Turner syndrome, we believe that a global screening of the thoracic aorta is of utmost importance. This screening can been undertaken using MRI, which allows complete evaluation of the thoracic aorta. Further studies need to be undertaken in order to determine precisely the incidence of thoracic aortic ectasia in a large population of patients with Turner syndrome and to determine the periodicity of MRI examination.

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