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Partial anomalous pulmonary venous drainage in two patients with Turner's syndrome

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SUMMARY We report two cases of partial anomalous pulmonary venous drainage in a series of 135 patients with Turner's syndrome.

The association of coarctation of the aorta, aortic valve disease, and pulmonary valve stenosis with Turner's syndrome is well established.[1 2] It is possible that many other congenital cardiac anomalies are more common in this disorder, but reported cases of these are so few that such associations may be no more than a coincidence. Uhrenholt *et al*[3] described one patient with Turner's syndrome and isolated partial anomalous pulmonary venous drainage. In this paper we describe two further instances of this disorder identified in a series of 135 patients with Turner's syndrome.

Case reports

CASE 1

This patient was born in Scotland in 1933 and although she never menstruated a diagnosis of gonadal dysgenesis was not made until the age of 35 when she was referred to an endocrinology clinic because of suspected hypothyroidism. At that time she complained of dry skin and loss of hair. Her height was 145 cm, weight 53·5 kg. There was no breast development, no axillary or pubic hair, and the external genitalia were underdeveloped. Buccal mucosal cells were chromatin negative and the karyotype of peripheral leucocytes and skin fibroblasts was 45,X. There was no webbing of the neck, no increase in carrying angle, and her chest was not 'shield-like'. She was of below average intelligence. It was noted that she had an early diastolic murmur (EDM) at the left sternal edge but she refused further cardiac investigation at that time. Oestrogen replacement therapy was not instituted.

At the age of 43 years a chest *x*-ray carried out after an episode of acute bronchitis showed cardiac enlargement with widening of the upper mediastinum and prominent vascular shadows particularly in the right upper and mid zones. Tomograms of the right hilum suggested that there was a large anomalous vessel in this region. When later seen at a cardiology clinic the patient was dyspnoeic and cyanosed, the jugular venous pressure was raised to 12 cm, there was slight ankle oedema, the pulse was 110/min, there was a right ventricular impulse, a fourth heart sound, and an ejection sound at the left sternal edge. The electrocardiogram showed sinus rhythm and complete right bundle branch block. The cardiac failure responded to treatment with diuretics and digoxin. At cardiac catheterisation there was a left to right shunt of 1·5:1 with anomalous drainage of the pulmonary veins from the upper lobes into the superior vena cava. No explanation was found for the EDM heard previously. She was later readmitted to hospital on a number of occasions for treatment of cardiac failure. It was not possible to identify any other factor contributing to the severity of the cardiac failure. She died later the same year. No necropsy was carried out.

CASE 2

This patient was born in Scotland in 1923 and never menstruated. A diagnosis of gonadal dysgenesis was not made until the age of 47 years when she was noted to have features of Turner's syndrome on admission to hospital for treatment of bronchitis. The only previous illness for which she had consulted a doctor was pneumonia, complicated by an empyema, at the age of 10 years. Her height was 147 cm, weight 36·9 kg. There was no breast development, axillary hair was absent, and pubic hair was sparse. She had slight webbing of the neck, bilateral cubitus valgus, and she was thought to be of below normal intelligence. Buccal mucosal cells were chromatin negative and the karyotype of peripheral leucocytes was 45,X. Two years later she was seen at a medical

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clinic because of retrosternal pain on exertion or excitement. Occasionally the pain was accompanied by dyspnoea. In addition to the findings already noted she had a loud first heart sound and an ejection sound at the left sternal edge. A chest *x*-ray showed pleural thickening on the left side, some dilatation of the main pulmonary vessels, and calcification of the coronary arteries. The electrocardiogram showed incomplete right bundle branch block and T wave inversion in lateral chest leads. A diagnosis of angina of effort as a result of ischaemic heart disease was made and she was treated with glyceryl tinitrate and oxprenolol. Her symptoms improved but six months later, after a further episode of pain, there was more marked inversion of T waves in anterior chest leads and she was considered to have suffered a myocardial infarct. Three years later the patient developed severe congestive cardiac failure and a loud apical systolic murmur which was initially attributed to a prolapsing cusp of the mitral valve. Cardiac catheterisation showed there was a left to right shunt of 2·5:1. Blood samples from the lower end of the superior vena cava were highly saturated with oxygen, indicating that the shunt was at this level. Right ventricular angiography showed large main and branch pulmonary arteries; the right lower lobe pulmonary veins drained into the superior vena cava. The drainage of the left lung was uncertain. On the strength of these findings a diagnosis of partial anomalous pulmonary venous drainage was made. Cardiac failure was controlled with digitalis and diuretic therapy and except for continuing angina her condition remains satisfactory.

Discussion

Noonan[4] and Tourniaire *et al*[5] have reported patients with Turner's syndrome who had partial anomalous pulmonary venous drainage, pulmonary stenosis, and an atrial septal defect, but the patient described by Uhrenholt *et al*[3] and the two reported here are the only instances known to us of isolated partial anomalous pulmonary drainage in Turner's syndrome. Our cases occurred in two patients from a series of 135 adults with Turner's syndrome included in the Edinburgh Medical Research Council register of patients with chromosome abnormalities. We can find no data on the prevalence of this abnormality in a population of otherwise normal women, but Hughes and Rumore[6] found two cases who had been symptomless during life in a series of 280 consecutive necropsy examinations. It is not possible to decide, therefore, if the frequency of 1·5% in our series of 135 patients is more than fortuitous.

Partial anomalous pulmonary venous drainage is usually symptomless and the development of severe congestive cardiac failure in both our patients is an unexpected feature. The inference is that this congenital anomaly has a more serious prognosis when it occurs in Turner's syndrome.

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