Congenital total lipodystrophy and peripheral pulmonary artery stenosis

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Abstract

Multiple peripheral pulmonary artery stenoses were detected in three patients with congenital generalised lipodystrophy. This association, which has not been described before, may be clinically important in patients with lipodystrophy who present with impaired exercise tolerance or heart murmurs.

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Congenital total lipodystrophy is a rare inherited disease with multiple abnormalities.¹ Cardiovascular involvement is rare but includes hypertrophic cardiomyopathy^{2,3} and idiopathic increase in pulmonary artery pressure.⁴ We report three patients with this syndrome who have peripheral pulmonary artery stenoses.

Case reports

CASE 1

A 2 year old boy, known to have congenital lipodystrophy, presented with an asymptomatic heart murmur. He had signs of right ventricular hypertrophy and a pulmonary systolic murmur with a loud pulmonary second heart sound. The electrocardiogram (ECG) showed right ventricular hypertrophy. His echocardiogram showed a hypertrophied right ventricle supporting a diagnosis of pulmonary hypertension. At cardiac catheterisation the right ventricular systolic pressure was suprasystemic (85 mm Hg compared with a systolic blood pressure of 75 mm Hg). Pulmonary angiography (fig 1) showed multiple severe peripheral pulmonary arterial stenoses.

CASE 2

The 6 year old sister of case 1 was well but, on examination, had congenital total lipodystrophy. She had a systolic murmur radiating to both lung fields. Her ECG showed mild right ventricular hypertrophy. Cardiac ultrasound showed increased flow velocities in the right and left pulmonary arteries suggestive of pulmonary artery stenosis. The right ventricular pressure estimated from the tricuspid regurgitation was 50 mm Hg.

CASE 3

A 14 year old girl with insulin resistant diabetes mellitus, total lipodystrophy, renal failure, and systemic hypertension was referred because of respiratory distress associated with chest pain. Her ventilation/perfusion scan had shown mismatch suggesting a left sided pulmonary embolus. Examination was unremarkable except for a systolic murmur widely heard over both lungs. Her ECG was normal. Cardiac ultrasound revealed mildly increased left and right pulmonary artery velocities of 2.8m/s and 2m/s respectively. Her chest pain resolved spontaneously but she has mildly impaired exercise capacity.

Discussion

Total lipodystrophy is characterised by muscular hypertrophy, complete absence of subcutaneous fat, insulin resistant diabetes mellitus, hepatomegaly, hypertriglyceridaemia, acanthosis nigricans, nephropathy, hypertension, and hirsutism.¹ Although the pathogenesis is unknown, excess of circulating hypothalamic releasing factors has been suggested. Cardiac involvement has been reported with asymptomatic murmurs, cardiomegaly on chest radiography and hypertrophic cardiomyopathy.^{3,4}

Peripheral pulmonary artery stenosis may occur as an isolated (sometimes familial) anomaly or in association with syndromes⁵ such as rubella, Williams, Ehlers-Danlos, cutis laxa, Noonan's, and Alagille's but its association with congenital total lipodystrophy, has not been recognised previously. One study suggested that idiopathic pulmonary hypertension may occur in some cases.⁴ Our findings suggest that when pulmonary hypertension is detected in a child with congenital total lipodystrophy, it may be due to peripheral pulmonary artery stenoses rather than being idiopathic. The condition may mimic the ventilation/perfusion scan mismatch seen with pulmonary embolism or thrombosis⁶ and may affect prognosis.

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Figure 1 Left (A) and right (B) pulmonary angiograms from case 1. Multiple stenoses are seen in the distal branches of both pulmonary arteries (arrowed).

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