

Pseudoxanthome élastique : une cause rare de valvulopathie mitrale

Pseudoxanthoma elasticum: A rare cause of mitral valvular disease

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ABSTRACT

Le pseudoxanthome élastique (PXE) est une maladie autosomique récessive très rare du tissu conjonctif qui touche la peau, la rétine et le système cardiovasculaire.

L'atteinte cardiovasculaire comprend la vasculopathie périphérique, l'hypertension artérielle, la coronaropathie, la cardiomyopathie restrictive mais aussi les valvulopathies. Nous décrivons dans ce rapport de cas un patient atteint de PXE et d'une valvulite mitrale sévère, avec une sténose mitrale prédominante.

SUMMARY

Pseudoxanthoma elasticum (PXE) is a very rare autosomal recessive connective tissue disorder with involvement of the skin, the retina, and the cardiovascular system.

Cardiovascular involvement includes peripheral vascular disease, hypertension, coronary artery disease, restrictive cardiomyopathy but also valvular disease. We describe in this case report a patient with PXE and severe mitral valvulitis, predominantly mitral stenosis.

MOTS-CLÉS

Pseudoxanthome élastique ; Maladie systémique ; Sténose mitrale

KEYWORDS

Pseudoxanthoma elasticum; Systemic disease; Mitral stenosis

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INTRODUCTION

Pseudoxanthoma elasticum (PXE) is an autosomal recessive connective tissue disorder with involvement of the skin, the retina, and the cardiovascular system. The estimated prevalence of PXE is 1 in 50 000.

Cardiovascular involvement includes peripheral vascular disease, hypertension, coronary artery disease, restrictive cardiomyopathy and is mainly characterized by mineralization and fragmentation of elastic fibers of blood vessels and premature atherosclerosis.

However, clinical cardiovascular manifestation may also include valvular involvement which is seldom mentioned in literature.

We describe here a patient with PXE and severe mitral valvulitis.

CASE PRESENTATION

This 22 year old white woman with history of first degree consanguinity was referred to our department at the neonatal age for cardiac murmur.

Several transthoracic echocardiographic (TTE) exams were performed and the conclusion was always nearly the same: congenital mitral valvular disease with calcifications over the posterior leaflet and severe left ventricular hypertrophy (LVH). Diagnosis of genetic Hypertrophic cardiomyopathy (HCM) with early positive phenotype was done.

In 2007, she had cardiac MRI scan which showed severe infiltration of the basal segment of posterolateral wall extending to the mitral valve and subvalvular apparatus.

In 2008, she had developed yellow cobblestone lesions in flexural areas and she was referred to the dermatology department, they performed skin biopsy and the patient was diagnosed as having pseudoxanthoma elasticum. (Photo 1)



Photo 1. Pathognomonic skin lesion of PXE in our patient: Bilateral yellow streaks and plaques, particularly in the neck.

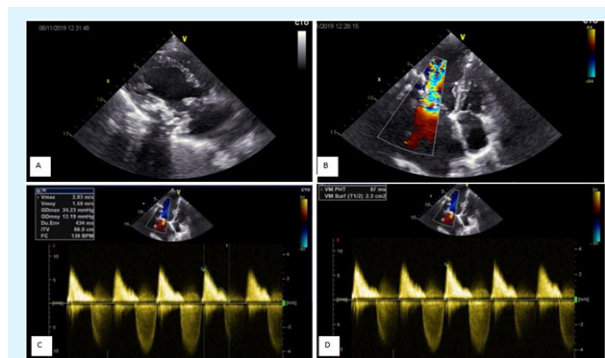


Figure 1. Echocardiographic findings: A: Parasternal long axis view showing severe calcification of the basal inferolateral segment extending to the posterior leaflet of mitral valve with a shadow cone. B: 4 apical chamber view (Left ventricle in the left) showing aliasing flow due to mitral stenosis diastolic gradient. C: Continuous wave doppler applied on the mitral valve inflow showing an elevated gradient: mean gradient = 12 mmHg. D: Continuous wave doppler across the mitral valve inflow, PHT=97 ms.

During yearly follow up, her visual acuity had impaired due to retinal hemorrhage.

Her electrocardiogram revealed a normal sinus rhythm with ST depression and negative T waves in inferior leads.

24- hour Holter monitoring objected two short bursts of supraventricular tachycardia.

She underwent coronary angiography which showed total chronic occlusion of the left circumflex artery.

DISCUSSION

Previous studies have revealed that cardiac involvement of pseudoxanthoma elasticum includes coronary artery disease, peripheral vascular disease, hypertension, and restrictive cardiomyopathy due to subendocardial fibrosis[1].

Valvular involvement was also reported by many authors. Mitral and aortic valves are typically invaded, and mitral regurgitation due to mitral prolapse can be observed. However, whether it directly involves the mitral valve has not been well clarified as most of data were based on case reports. Autopsies of PXE cases revealed direct involvement of the myocardium with endocardial lesions, characterized by degenerated elastic fibers with calcification in the subendocardium. In 1884 Balzer reported on the autopsy finding of 49 year old male having PXE the presence of yellowish thickening of the right endocardial surface of the heart which histologically

showed degeneration of elastic fibers[2]. McKusick also described a 63 year old black male with PXE who had a yellowish thickening of the endocardium of the right atrium, the posterior leaflet of the tricuspid valve and the anterior leaflet of the mitral valve at the edge with many calcifications at its attachment [3].

The above pathological findings suggested that valvular disease can occur directly related to PXE. The association of pseudoxanthoma elasticum and mitral stenosis particularly was reported by Coffman and Sommers in 1959. They showed the pathologic changes of the mitral valve in a necropsy case, and suggested that the degeneration of elastic fibers, disorganization with fragmentation and clumping, and calcification of elastic fibers might cause mitral stenosis[4]. Another interesting point in our patient's history is LVH without arterial hypertension which was especially marked in infancy, it was found in 4.5% of a cohort of 67 PXE patients without known cause.[5]

CONCLUSION

Valvular involvement is one of the cardiovascular manifestations of PXE, explained mainly by endocardial and subendocardial extension and elastic fibers degeneration.

PXE should be considered one of the uncommon causes of mitral stenosis and should be differentiated from rheumatoid mitral stenosis.

PXE may also cause LVH and in context of mitral valvulitis it should be differentiated from HCM.

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Figure 4. Coupe axiale d'une angio-TDM thoracique montrant une dilatation post-sténotique de l'Aorte thoracique