

Congenital Pseudoaneurysm of the Mitral Aortic Intervalvular Fibrosa in neonatal Marfan syndrome – Transthoracic Echocardiogram and Computed Tomography images

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Abstract

Congenital Pseudoaneurysm of the mitral aortic intervalvular fibrosa (P-MAIVF) is extremely rare condition. We report a case of neonatal Marfan syndrome with unique and rare diagnosis of case of congenital pseudoaneurysm of the mitral aortic intervalvular fibrosa, where the transthoracic echocardiogram and computed tomography images described a congenital pseudoaneurysm of the mitral aortic intervalvular fibrosa.

Keywords: Pseudoaneurysm, Mitral Aortic Intervalvular Fibrosa, Neonatal Marfan syndrome

A female infant with birth weight of 2500 grams was delivered at 33 weeks of gestation due to incessant fetal supraventricular tachycardia and hydrops fetalis. The neonate required significant cardiorespiratory support including mechanical ventilation, multiple antiarrhythmic and inotropic

support. She displayed unique dysmorphic features of low-set, dysplastic ears, long palms, arachnodactyly in upper and lower extremities, and long fingers with small nails. Her cardiac auscultatory findings of significant mitral regurgitation murmur and to-fro murmur from mitral aortic intervalvular fibrosa aneurysm.

Transthoracic echocardiography revealed large aneurysmal structure noted posterior to the aorta, extending into the right and left atria. This chamber communicates to the left ventricular outflow tract in the region of the mitral-aortic fibrosa with to-and-fro flow. The location is suspicious for a pseudoaneurysm of the mitral-aortic intervalvular fibrosa. Redundant mitral valve leaflets with mild-moderate insufficiency and no stenosis. (Fig 1A, B, C) The left ventricle was severely dilated with severely depressed systolic function.

Further cross sectional imaging with cardiac computed tomography (CT) showed multilobulated structure at the inferior aspect of the heart between the right and left atria measuring 6 x 5 mm diameter apparent connection to the left ventricular outflow tract, filling with contrast of the same density as that of the left ventricle. This is consistent with pseudoaneurysm of the mitral aortic intervalvular fibrosa. (Fig 2A, 2B)

Her rapid exome genomic sequencing revealed a pathogenic de novo variant within FBN1 gene (c.3257G>C within exon 27), consistent with a diagnosis of early onset Marfan syndrome. She was deemed to be extremely high-risk surgical candidate for repair of pseudoaneurysm. Parents opted for compassionate care and declined autopsy.

Neonatal Marfan syndrome patients with significant cardiac lesions generally carry poor prognosis. The pseudoaneurysm of mitral-aortic intervalvular fibrosa (P-MAIVF), mostly seen following infective endocarditis in adults and older children. Diagnosis of congenital P-MAIVF

is rare and there has been no cases reported with this combination of neonatal Marfan syndrome and congenital pseudoaneurysm of mitral-aortic intervalvular fibrosa[1]. Diagnosis can be made with 2D and 3D echocardiograms and often needs additional cross sectional imaging in the form of cardiac CT or MRI needed prior to attempted intervention[2].

Figure Legends:

Transthoracic echocardiogram (TTE)

1A. Parasternal long -axis view with color comparison demonstrating the typical location of pseudoaneurysm (P-MAIVF) in the left ventricular outflow between the aortic and mitral valves. Ao= aorta; AoV= aortic valve; LV=left ventricle; RV=right ventricle.

1B. Apical 4 chamber view demonstrating pseudoaneurysm (P-MAIVF) between the LV outflow tract and mitral valve. The pseudoaneurysm is seen as large globular structure within the left atrium. On color Doppler profile to-fro flow in the pseudoaneurysm and posteriorly directed mitral regurgitation.

1C. Apical 4 chamber image with color mapping delineating the opening of P-MAIVF. Arrow pointing towards the opening of pseudoaneurysm.

Computed tomography (CT):

2A: Computed tomography scan axial stack image showing large pseudoaneurysm seen as large globular structure within the left atrium with contrast level similar to left ventricle. LV= left ventricle; LA= left atrium; RV= right ventricle; Arrow pointing towards the neck of pseudoaneurysm.

2B: Computed tomography coronal image showing large pseudoaneurysm seen as large globular structure within the left atrium. LV= left ventricle; Ao=Aorta; Arrow pointing towards the neck of pseudoaneurysm.

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