Membranous septal aneurysm in a child with Noonan syndrome and hypertrophic cardiomyopathy

PRIYADARSHINI ARUNAKUMAR[1], Usha MK[1], RAMYA S[2], and Jayaranganath M[1]

[1]Sri Jayadeva Institute of Cardiovascular Sciences and Research
[2]Sri Jayadeva Institute of Cardiology

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Abstract

A four year old boy was diagnosed with hypertrophic cardiomyopathy with moderate degree of obstruction of the left ventricular outflow tract, secondary to asymmetric septal hypertrophy and no features of congestive heart failure, at the age of the 7 months. He was also found to have myxomatous mitral valve, systolic anterior motion of anterior mitral leaflet and moderate mitral regurgitation. There was no evidence of ventricular septal defect at this point in time.In view of phenotypic features suggestive of Noonan syndrome, he underwent genetic evaluation with target gene sequencing and was detected to have a novel heterozygous mutation in exon 13 of LZTR 1 (Leucine- zipper-like transcriptional regulator 1) gene. Echocardiogram on follow up showed increase in left ventricular outflow tract obstruction and appearance of an aneurysm in the membranous portion of interventricular septum. There were no features of right ventricular outflow obstruction, tricuspid regurgitation or aortic regurgitation. The appearance of the membranous septal aneurysm may be related to the direction of jet arising from the point of contact of the anterior mitral leaflet with the hypertrophied basal septum and hitting this part of the interventricular septum. It is an unusual mechanism for formation of membranous septal aneurysm, given its absence in the earlier echocardiograms and absence of any left to right shunt across the aneurysm. He has been initiated on beta blockers due to severe left ventricular outflow tract obstruction and is planned for septal myomectomy, resection of membranous septal aneurysm and mitral valve repair.

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MEMBRANOUS SEPTAL ANEURYSM IN A CHILD WITH NOONAN SYNDROME AND HYPERTROPHIC CARDIOMYOPATHY

AUTHORS

DR PRIYADARSHINI ARUNAKUMAR *****, DM

ASSISTANT PROFESSOR, DEPARTMENT OF PEDIATRIC CARDIOLOGY,

SRI JAYADEVA INSTITUTE OF CARDIOVASCULAR SCIENCES AND RESEARCH,

BANGALORE, INDIA (PIN -560069)

PHONE +91 8593841984, E MAIL priya_arun_2000@yahoo.com, FAX 080-22977261

DR USHA MK, MD

ASSOCIATE PROFESSOR, DEPARTMENT OF PEDIATRIC CARDIOLOGY,

SRI JAYADEVA INSTITUTE OF CARDIOVASCULAR SCIENCES AND RESEARCH,

BANGALORE, INDIA (PIN -560069)

DR RAMYA GS, DNB

SENIOR RESIDENT, DEPARTMENT OF PEDIATRIC CARDIOLOGY,

SRI JAYADEVA INSTITUTE OF CARDIOVASCULAR SCIENCES AND RESEARCH,

BANGALORE, INDIA (PIN -560069)

DR JAYRANGANATH M, DM

PROFESSOR, DEPARTMENT OF PEDIATRIC CARDIOLOGY,

SRI JAYADEVA INSTITUTE OF CARDIOVASCULAR SCIENCES AND RESEARCH,

BANGALORE, INDIA (PIN -560069)

RUNNING HEAD

MEMBRANOUS SEPTAL ANEURYSM IN HCM

*CORRESPONDING AUTHOR

ABSTRACT:

A four year old boy was diagnosed with hypertrophic cardiomyopathy with moderate degree of obstruction of the left ventricular outflow tract, secondary to asymmetric septal hypertrophy during infancy. Echocardiogram on follow up showed increase in left ventricular outflow tract obstruction and appearance of an aneurysm in the membranous portion of interventricular septum. The appearance of the membranous septal aneurysm may be related to the direction of jet arising from the point of contact of the anterior mitral leaflet with the hypertrophied basal septum and hitting this part of the interventricular septum.

KEYWORDS:

Hypertrophic cardiomyopathy, Noonan syndrome, Membranous septal aneurysm

MANUSCRIPT TEXT:

A four year old boy was diagnosed with hypertrophic cardiomyopathy with moderate degree of obstruction of the left ventricular outflow tract secondary to asymmetric septal hypertrophy and no features of congestive heart failure during infancy. He was also found to have myxomatous mitral valve, systolic anterior motion of anterior mitral leaflet and moderate mitral regurgitation (Fig 1, Video 1). In view of phenotypic features suggestive of Noonan syndrome (Fig 3), he underwent genetic evaluation with target gene sequencing and was detected to have a novel heterozygous mutation in exon 13 of LZTR 1 (Leucine- zipper-like transcriptional regulator 1) gene[1]. Echocardiogram on follow up showed increase in left ventricular outflow tract obstruction and appearance of an aneurysm in the membranous portion of interventricular septum (Fig 3 and 4, Video 2 and 3). There were no features of right ventricular inflow or outflow obstruction, tricuspid regurgitation or aortic regurgitation. Membranous septal aneurysms in children are usually seen in association with ventricular septal defects. The appearance of the membranous septal aneurysm in this boy may be attributed to the direction of jet arising from the point of contact of the anterior mitral leaflet with the hypertrophied basal septum and hitting this part of the interventricular septum. It is an unusual mechanism for formation of membranous septal aneurysm, given its absence in the earlier echocardiograms and absence of any left to right shunt across the aneurysm. For membranous septal aneurysms to become clinically important, they have to interfere with the closure mechanism of the cardiac valves, cause a mass effect, or cause flow obstruction[2]. Histopathology of resected aneurysms have shown almost total loss of elastic fibres and extensive accumulation of mucopolysaccharides[3]. This patient has been initiated on beta blockers due to severe left ventricular outflow tract obstruction and is planned for septal myomectomy, mitral valve repair and membranous septal aneurysm resection due to the fear of thrombus formation and rupture of the aneurysm.

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FIGURE LEGENDS:

Fig 1 – M mode of the mitral valve (MV) shows systolic anterior motion of the mitral valve (marked with blue arrow) and contact with the interventricular septum (IVS)

Fig 2 – Shows typical facial features of Noonan syndrome like broad forehead, hypertelorism,low set ears, bulbous tip of the nose and cupid bow upper lip

Fig 3 – Apical 4 chamber view (LA-left atrium, LV-left ventricle, RA-right atrium, RV-right ventricle) - Red arrow shows myxomatous mitral valve, blue arrows shows hypertrophied basal interventricular septum, green arrow and green star show membranous septal aneurysm (AN)

Fig 4 – Continuous wave doppler of the left ventricular outflow tract shows severe left ventricular outflow tract obstruction with a peak gradient of 100 mm Hg

Video 1 – Modified parasternal long axis shows myxomatous mitral valve, moderate mitral regurgitation into the left atrium (LA), systolic anterior motion of the mitral valve, hypertrophied basal interventricular septum (IVS) and resultant turbulence in the left ventricular outflow tract and Aorta (AO)

Video 2 – Apical 4 chamber view shows membranous septal aneurysm (AN) protruding into the right ventricular inflow region and absence of any ventricular septal defect (LA-left atrium, LV-left ventricle, RA-right atrium, RV-right ventricle)

Video 3 – Simultaneous 2D and colour imaging show contact of myxomatous mitral valve (AML) and hypertrophied basal interventricular septum (IVS) with resultant jet directed towards the membranous septal aneurysm (AN), absence of right ventricular inflow turbulence and absence of communication with the right ventricle