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SHORT REPORT

Sudden Infant Death Syndrome and Left Ventricular Hypertrabeculation-Hidden Arrhythmogenic Entity?

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Abstract: Left ventricular noncompaction/hypertrabeculation is a condition which is characterized by a highly trabeculated, "spongy" myocardium.
It can present at any age with heart failure, arrhythmia and/or thromboembolic events.
A wide variety of mutations have been found to be a cause of hypertrabeculation and it is possible that there is a continuum of hypertrophic cardiomyopathy, dilated cardiomyopathy and hypertrabeculation/noncompaction.
We present a case of left ventricular hypertrabeculation which presented as sudden infant death syndrome and we propose that this entity may be a hidden cause of arrhythmic death in some infants presenting as sudden infant death syndrome.

Keywords: sudden infant death syndrome, hypertrabeculation, noncompaction

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Introduction

The cardiac syndrome of isolated, left ventricular noncompaction is characterized by a persistence of the embryonic pattern of a highly trabeculated myocardium in the left ventricle.[1] This condition can be familial or sporadic and can be due to a variety of mutations in any one of the following proteins: mitochondrial, cytoskeletal, Z-line or sarcomeric.[1]

Chin et al[2] were the first to describe the entity and the complications of heart failure, ventricular arrhythmias and thromboembolic events. An association with Wolff-Parkinson-White syndrome was also noted.[2]

This condition can present during early life as fetal hydrops[1] or as sudden infant death syndrome due to neonatal heart failure and/or ventricular fibrillation.[1,3–6]

Case Report and Discussion

We present a case report of a three month old male infant who presented with sudden infant death syndrome. This three month old non-caucasian died suddenly and unexpectedly at his day care centre. He did not have any known medical problems and no surgical procedures were ever performed. No known allergies were present and no known family history of sudden, unexpected death were present. Unfortunately the family members were lost to follow up, before electrocardiographic and echocardiographic screening could be performed to assess the risk for sudden unexpected death.

Postmortem examination of the heart revealed numerous apical trabeculations of the left ventricle (see Fig. 1). The right ventricle appeared perfectly normal. The left ventricular wall thickness measured 1 cm and the histological assessment was perfectly normal. No abnormalities were detected in any other organs during the postmortem examination and no thrombi were detected in the arterial system or the left ventricle.

Current consensus on the mechanism of left ventricular noncompaction is that an arrest of myocardial maturation occurs during embryogenesis.[7] Before the eighth week of fetal life the myocardium consists of a network of fibres, washed by deep recesses, communicating with the left ventricular cavity.[1] The reason for this is that there is no coronary vasculature yet, therefore the trabeculations of myocardial tissue increases the myocardial surface area in contact with the ventricular lumen, as the myocardium is nourished directly from the endoventricular column of blood at this stage.[1]

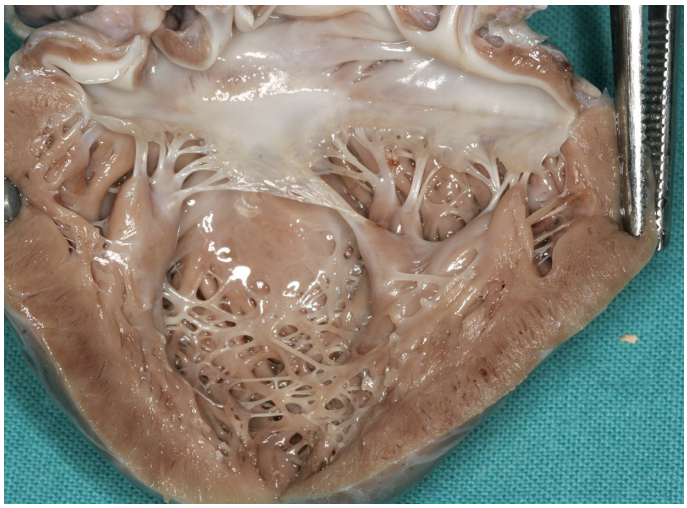

Figure 1. Left ventricular hypertrabeculation in a three month old infant, presenting as sudden infant death syndrome. It is postulated that the apical area of hypertrabeculation acted as the source of a fatal arrhythmia.

The coronary vasculature develops during the fifth to eighth week of fetal life and after this the meshwork of myocardial fibres will become compacted.[1] This "compaction process" advances from the base of the heart to the apex and from the epicardium to the endocardium as this is the direction of coronary arterial development.[1]

Various echocardiographic criteria have been proposed for the diagnosis of left ventricular noncompaction.[2,8] In reaction to these proposed echocardiographic criteria Stöllberger et al[1,9,10] named this peculiar left ventricular phenotype "left ventricular hypertrabeculation" and defined the condition as the presence of more than three trabeculations in the left ventricle in a location distal (apical) to the papillary muscles. A postmortem study by Boyd et al[1,11] in 474 normal human hearts have shown that while 68% of these hearts displayed prominent trabeculations, only 4% of these hearts had more than three and none had more than five trabeculations.

Left ventricular noncompaction/hypertrabeculation can occur in a sporadic or familial form and both types can be due to a variety of mutations in various sarcomere, mitochondrial, Z-line or cytoskeletal proteins.[1,12–15]

Figure 1 clearly demonstrates more than three trabeculations in the left ventricle in a location apical to the papillary muscles. According to

Boyd et al[11] this case which presented as a sudden infant death syndrome thus fulfills the criterion for left ventricular hypertrabeculation. The association of hypertrabeculation/noncompaction with various sarcomere mutations supports the concept that this entity is a cardiomyopathy and furthermore, that there is a spectrum from hypertrophic cardiomyopathy, especially apical hypertrophic cardiomyopathy, dilated cardiomyopathy and hypertrabeculation/noncompaction.[14]

The natural history of left ventricular hypertrabeculation/noncompaction varies widely. Presentation with heart failure, arrhythmias and/or thromboembolism is described in patients of all ages.[15] Mild cases may remain asymptomatic.

We propose that this case of sudden infant death syndrome is due to an episode of fatal arrhythmia due to underlying left ventricular hypertrabeculation and that the presence of more than three trabeculations apical to the papillary muscles should be specifically excluded during postmortem examination in cases of sudden infant death syndrome.

We acknowledge the possibility that the left ventricular hypertrabeculation may be an incidental finding as this entity may be found in asymptomatic, "healthy" individuals, but due to the observation that arrhythmias may occur at any age[15] we propose that this may be an underrecognized cause of sudden, unexpected death, mimicking sudden infant death syndrome.

Disclosure

This manuscript has been read and approved by all authors. This paper is unique and is not under consideration by any other publication and has not been published elsewhere. The authors and peer reviewers of this paper report no conflicts of interest. The authors confirm that they have permission to reproduce any copyrighted material. Written consent was obtained from the patients parents for publication of this study.

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