Case Report

Indian Journal of Forensic Medicine and Pathology
Volume 8 Number 3, July - September 2015

Sudden Death during Recreational Sports Activity

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Abstract

Sudden death of an elite young during recreational sports activity is a devastating event for both the family and community. Sudden cardiac death (SCD) is one of the leading causes of death in such cases. Hypertrophic cardiomyopathy (HCM) is a primary disease of the myocardium in which the portion of the myocardium is hypertrophied without any obvious cause. It is the leading cause of sudden cardiac death in young athletes. The obstructive variant of HCM is most commonly involved in sports related sudden death. A 13 year old healthy adolescent without any family history brought dead to the emergency. Autopsy revealed an enlarged heart with asymmetrical left ventricular hypertrophy. Microscopy of left ventricular free wall showed hyperplastic muscle bundle and wavy muscle fibers. Sections of the interventricular septum showed extensive myocytic hypertrophy, myocytic fiber disarray and enlarged nucleus. No evidence of acute ischemic changes. The present report highlights a rare condition which is rarely reported by a forensic pathologist, which confirms that HCM is one of the types of cardiomyopathy causing sudden death during recreational physical activity in an elite young individual. Recommend standard autopsy guidelines and screening for first degree family members with necessary treatment / lifestyle modifications and further study on screening and genetics related to HCM.

Keywords: Sudden Death; Sports Activity; Cardiomyopathy; HCM; Autosomal Dominant; Genetic Screening.

Introduction

A growing number of people are involved in recreational physical activity [1]. Sudden death of an elite young during recreational sports activity is a devastating event for both the family and community. It is not uncommon for a medical examiner / forensic pathologist to encounter such sports-related sudden deaths and to be faced with the legal implications. Sudden cardiac death (SCD) is one of the leading causes of death in such cases[2-4]. In persons over the age of 35 years, the predominant cause of SCD is coronary artery disease (CAD), while in younger age groups cardiomyopathy (CM) and congenital heart disease comprise the majority of cases[5]. Cardiomyopathies include a variety of diseases such as hypertrophic cardiomyopathy, dilated cardiomyopathy, restrictive cardiomyopathy, arrhythmogenic right ventricular cardiomyopathy and the channalopathies. Hypertrophic cardiomyopathy (HCM) is a primary disease of the myocardium in which the portion of the myocardium is hypertrophied without any obvious cause [6-9]. It is the leading cause of sudden cardiac death in young athletes[10-15]. If such hypertrophy is observed in interventricular septum, the hypertrophied septum will obstruct the left ventricular outflow and is called as obstructive type of hypertrophic cardiomyopathy, death due to such condition are rarely reported. This communication reports the sudden death of a young boy due to hypertrophic obstructive cardiomyopathy (HOCM) and enlighten about the cardiac pathology, pathophysiologic mechanisms, optimal screening strategies, and prevention.

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Case Report

A 13 year old boy was brought dead to the emergency. He had a history of breathless and sudden collapse to the ground during the play in the school playground. Incidence took place within few minutes to an hour. He had no significant family and past history and was not on any medication.

At autopsy moderately built and nourished male child with cyanosed extremities, measuring 140 cm in length and 28 kg in weight. External examination was unremarkable except an abrasion on left knee. Stomach filled with 100 ml partially digested food. Viscera for chemical analysis came negative for any poisons. The heart weighed 280 gm. The normal heart weight reference range for that age is 150-200 gm, for body weight is 122 gm and for height is 144 c0m. The coronary arteries were patent.

The right and left atrium (RA & LA) were normal in size and wall thickness. The right ventricular (RV) wall showed myocardial hypertrophy varied from 6 to 9 mm. The left ventricle (LV) showed asymmetrical hypertrophy and varied from 18 to 20 mm along the anterolateral wall to 26 mm within the interventricular septum. The septum showed maximum hypertrophy of 26 mm at the outlet (just below the origin of aorta) leading to outflow obstruction to the LV. The papillary muscles were thickened in LV. No evidence of any recent or old infarct was noted (Fig 1).

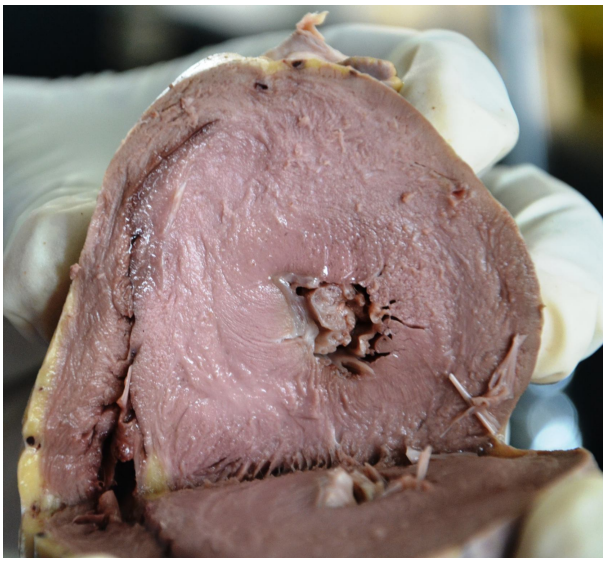

Fig. 1: Gross section showing asymmetrical hypertrophy of left ventricle with septum and thickened papillary muscle

Microscopy of RA, LA and RV showed hyperplastic muscle bundles. Microscopic sections of the LV free wall showed hyperplastic muscle bundle and wavy muscle fibers (Fig 2 & 3). Sections of the interventricular septum showed extensive myocytic hypertrophy, myocytic fiber disarray & enlarged nucleus (Fig 4 & 5). Fibrin thrombi in muscle fibers were noted in few sections (Fig 6). No evidence of acute ischemic changes. Microscopy of other organs and toxicological analysis was unremarkable.

Cause of death opined as death is due to acute cardiac failure as a result of hypertrophic cardiomyopathy (HCM).

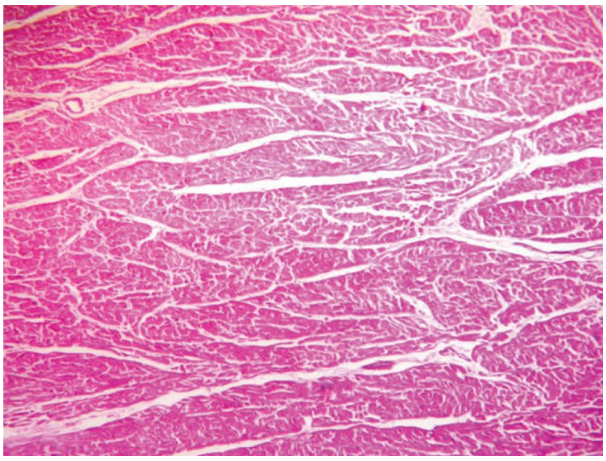

Fig. 2: Hyperplastic muscle bundles (haematoxylin & eosin, 10X)

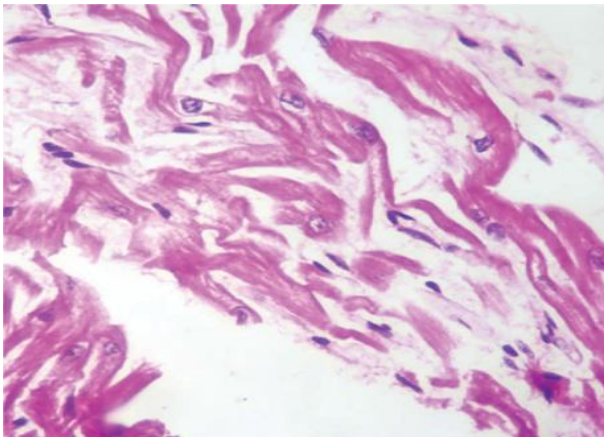

Fig. 3: Wavy muscle fibers (haematoxylin & eosin, 40X)

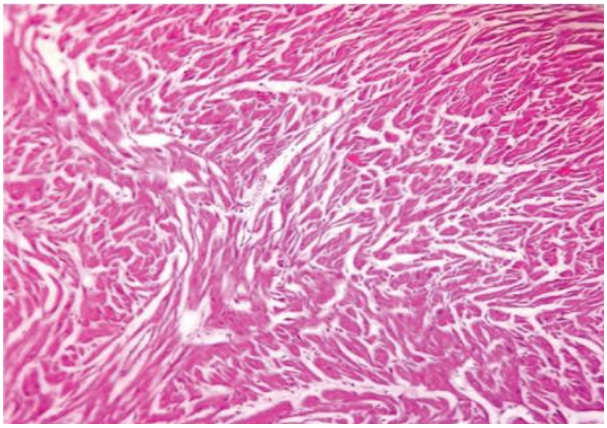

Fig. 4: Myocytic hypertrophy & fiber disarray (haematoxylin & eosin, 10X)

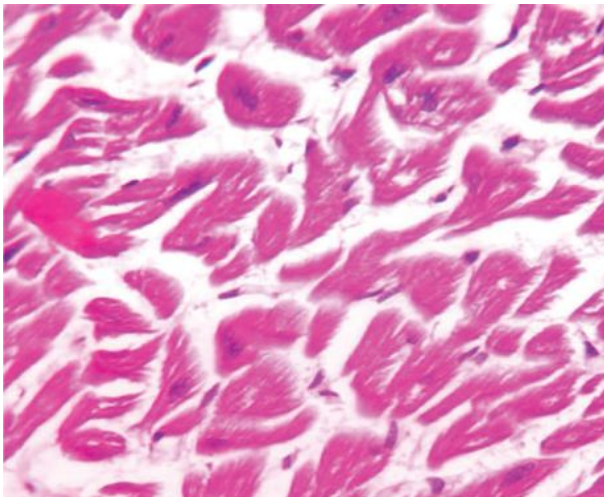

Fig. 5: Myocytic fiber disarray & enlarged nucleus (haematoxylin & eosin, 40X)

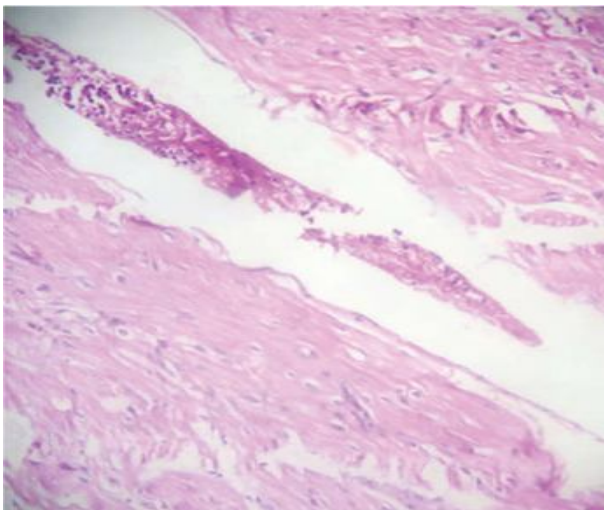

Fig. 6: Fibrin thrombi in muscle fibers (haematoxylin & eosin, 10X)

Discussion

Hypertrophic cardiomyopathy (HCM) is most often associated with sudden death in young athletes[10-15]. It has a prevalence of approximately 1 in 500 adults[16,17]. It is a primary disease of myocardium characterized by a hypertrophied but nondilated left ventricle in the absence of another cardiac or systemic disease that may produce left ventricular hypertrophy. Hypertrophic cardiomyopathy affects young individuals[18]. The mortality in hypertrophic cardiomyopathy is more in children and adolescents than in adults[11]. Approximately one half of young athletes who die suddenly have hypertrophic cardiomyopathy[19]. Unfortunately, they usually have no history of preceding symptoms[10-15].

Depending on whether the distortion of normal heart anatomy causes an obstruction to the outflow of blood from the left ventricle of the heart as seen in present case, HCM can be classified as obstructive and non-obstructive. The obstructive variant of HCM, hypertrophic obstructive cardiomyopathy (HOCM) was historically known as idiopathic hypertrophic subaortic stenosis (IHSS) and asymmetric septal hypertrophy (ASH) [20]. The non-obstructive variant of HCM is apical hypertrophic cardiomyopathy, also called Yamaguchi Syndrome or Yamaguchi Hypertrophy [21]. The majority of HCM cases are familial. Familial hypertrophic cardiomyopathy is inherited as an autosomal dominant trait and is attributed to mutations in one of a number of genes that encode for one of the sarcomere proteins[20]. About 50-60% of patients with a high index of clinical suspicion for HCM will have a mutation identified in at least 1 of 9 sarcomeric genes. Approximately 45% of these mutations occur in the β myosin heavy chain gene on chromosome 14 q11.2-3, while approximately 35% involve the cardiac myosin binding protein C gene. Since HCM is typically an autosomal dominant trait, children of a single HCM parent have 50% chance of inheriting the disease-causing mutation. Whenever a mutation is identified through genetic testing, family-specific genetic testing can be used to identify relatives at risk for the disease[22]. In individuals without a family history of HCM, the most common cause of the disease is a de novo mutation of the gene that produces the β -myosin heavy chain.

An insertion / deletion polymorphism in the gene encoding for angiotensin converting enzyme (ACE) alters the clinical phenotype of the disease. The D / D (deletion / deletion) genotype of ACE is associated with more marked hypertrophy of the left ventricle and may be associated with higher risk of adverse outcomes[23,24]. Some mutations could have more malignant potential compared to others (β myosin heavy chain). For example, troponin T mutations were originally associated with 50% mortality before the age of 40. However, a more recent and larger study found a similar risk to other sarcomeric protein mutations[25]. The pattern of left ventricular wall thickening is characteristically asymmetric, with portions of ventricular septum disproportionately thicker than most of the left ventricular free wall. The increase in left ventricular mass usually results in impaired ventricular filling and compliance; some patients may also have obstruction to left ventricular outflow produced by systolic anterior motion of the mitral valve. The non-obstructive form is more prevalent than the obstructive form. Histologic examination shows bizarre cellular architecture with a markedly disordered arrangement of cardiac muscle

cells (the so-called myocyte disarray) and more abnormal intramural coronary arteries with thickened walls and narrowed lumen often associated with fibrosis[13,26].

In the present case recorded mass of the heart indicates that the heart is enlarged for age, height and weight of the individual (cardiomegaly). This finding would correlate with the significant LV hypertrophy (increased wall thickening) along with extensive asymmetric hypertrophy of interventricular septum leading to obstruction to the left ventricular outflow.

Conclusion

The sudden deaths due to HCM are not less common, but they are poorly reported because of improper autopsy guidelines followed[26]. Thus authors recommend a standard autopsy guideline by The Royal College of Pathologists to be followed in case of sudden death with likely cardiac pathology [27]. As this cardiomyopathy is inherited as an autosomal dominant trait, diagnosis at autopsy play a vital role in assessment / screening of first degree family members by a cardiologist followed by genecist if necessary was recommended. To exclude a family member or to take necessary treatment / life style modifications if found to have inherited the disease.

This report highlights a rare condition which is rarely reported by a forensic pathologist, which confirms that HCM is one of the types of cardiomyopathy causing sudden death during recreational physical activity in an elite young individual.

Conflict of interest

Nil (No sponsorship)

Acknowledgement

We are thankful to the Department of Pathology SDM College of Medical Sciences & Hospital for their help.

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