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CASE REPORT

Sudden cardiac death with triple pathologies: A case report

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Abstract Sudden cardiac death in young adults may be associated with rare cardiomyopathies such as left ventricular noncompaction (LVNC) and arrhythmogenic right ventricular (ARVC) cardiomyopathies. LVNC is characterised by hypertrabeculations and deep recesses of the left ventricle. ARVC presents with thin myocardium as a result of extensive fibro-fatty infiltrations. In both conditions, death may be due to arrhythmia, thromboembolic events or heart failure. We report a case of a 21-year old athletic young man who collapsed at the futsal court right after the game. He was resuscitated but expired at the hospital after a brief admission. A week earlier, he had a similar episode of syncope and revived through cardio-pulmonary resuscitation at the site. Post mortem examination showed extensive acute myocardial infarction (AMI) involving the papillary muscles and the left ventricular wall. Features of LVNC were also observed. On top of that, the right ventricle showed patchy thin myocardium as the wall was largely comprised of fat. Histology examination confirmed the presence of AMI and massive fibro-fatty infiltrations of the right ventricle. This unfortunate young man had co-existing cardiomyopathies which is rare indeed. As he succumbed to AMI, this mechanism of death is also uncommonly associated with neither LVNC nor ARVC. In conclusion, young and physically active individuals may not be spared of sudden cardiac death. Mild and non-specific symptoms should not be taken lightly as it may be the subtle signs of cardiomyopathies.

1. Introduction

Sudden cardiac death (SCD) is defined as an unexpected death resulting from a cardiovascular cause in an individual with or without pre-existing heart disease.[1] The causes of SCD in the group of young adults of less than 35 years old include myocarditis, valvular heart disease, congenital coronary artery anomaly and cardiomyopathies such as left ventricular non-

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compaction cardiomyopathy (LVNC) and arrhythmogenic right ventricular cardiomyopathy (ARVC).[2,3]

LVNC or 'spongiform cardiomyopathy' is a rare congenital cardiomyopathy which arises from arrested endomyocardial compaction during fifth to eighth week of embryogenesis.[4–6] It is characterised by prominent left ventricular trabeculations of the endocardial layer, deep inter-trabecular recesses and thin compacted layer.[6] The morphological defects may cause clinical manifestations such as syncope, thrombo-embolic events and death may result from heart failure or arrhythmias.[7,8]

Another important type of cardiomyopathy which is associated with SCD in young athletic adult is ARVC. The prevalence of ARVC is reported to be 1/1000 in general population.[3] It is a heritable myocardial disease as a result of mutations in genes encoding cardiac desmosomal proteins.[3,9] At autopsy, ARVC is characterised by markedly thin wall of the right ventricle as it is largely replaced by fibro-fatty tissue.[3] Microscopically, the fibro-fatty infiltrations with chronic inflammatory cells may be seen affecting both the right and left ventricles.[3,10] Common presentations may include palpitations, ventricular tachycardia (VT) and syncope, but many patients are initially asymptomatic.[10]

We present a case of a 21-year-old gentleman who suddenly collapsed at the futsal court right after the game. He had no known medical illness and led an active lifestyle. Autopsy examination showed an extensive acute myocardial infarction (AMI) of the left ventricle. Apart from that, the right and left ventricles revealed interesting pathologies of co-existing LVNC and ARVC.

2. Case report

A 21-year-old Malay gentleman was noted to be having shortness of breath and jerky movements right after playing futsal with his group of friends. A similar episode took place a week ago following the same event; however, he regained consciousness after a cardiopulmonary resuscitation (CPR) procedure performed by his friend. He did not proceed to seek any medical treatment as he believed he was healthy and physically active. During this episode, he became unconscious, unresponsive and immediately brought to the nearest hospital. Spontaneous circulation has returned upon successful resuscitation. He was subsequently referred to our centre for further management. At the Emergency Department, he was haemodynamically unstable despite being on two inotropic agents. He had another episode of cardiac arrest and revived after 12 min of CPR. Bedside echocardiogram that was performed revealed thickened left ventricular wall with increased trabeculations and poor cardiac contractility (Fig. 1). Arterial blood gases showed pH of 6.7 and other parameters were in keeping with severe metabolic and respiratory acidosis. In spite of the maximum medical supports given he eventually succumbed after nearly three hours of admission. A police report was lodged and an order was issued for medico-legal autopsy examination to determine the cause of death.

2.1. Autopsy findings

Autopsy examination showed a muscular, medium-built young adult male measuring 166 cm in height and 76 kg in weight.

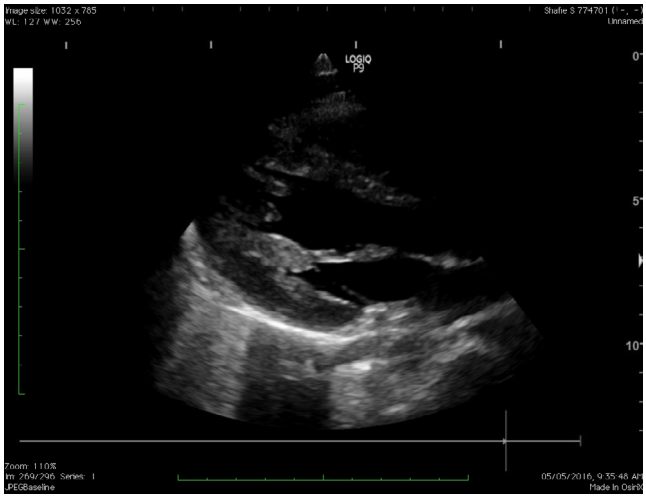

Fig. 1 Bedside echocardiogram showing thickened left ventricular wall and increased trabeculations.

Bluish discolouration was noted on the nail beds, indicating cyanosis. There was no significant injury noted on the body. Internal examination revealed a mild cardiomegaly as the heart weighed 350 gm. The right and left ventricular wall thicknesses measured 2 mm and 15 mm respectively. The left ventricle showed evidence of acute myocardial infarction (AMI) as demonstrated by the haemorrhagic myocardium vividly seen on the papillary muscles (Fig. 2a). The cut surfaces also showed areas of whitish discolouration, hyperaemia and soft parenchyma, consistent with AMI. On top of the ischaemic changes, the left ventricular wall also revealed excessive trabeculations with deep recesses (Fig. 2b). Coarse trabeculae resembling multiple papillary muscles were also observed. These features were consistent with LVNC. On the other hand, examination of the right ventricle also showed a remarkable finding. The free wall of the ventricle was thinner than normal and fatty tissues could be seen displacing the normal myocardium (Fig. 3a). These features were consistent with ARVC.

Other internal organs appeared grossly normal. Histopathology examination of the heart shows large areas of fibrosis, haemorrhage and neutrophilic infiltrations in the

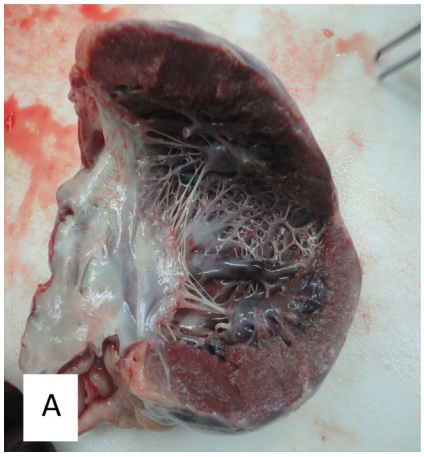

Fig. 2a Haemorrhagic papillary muscles.

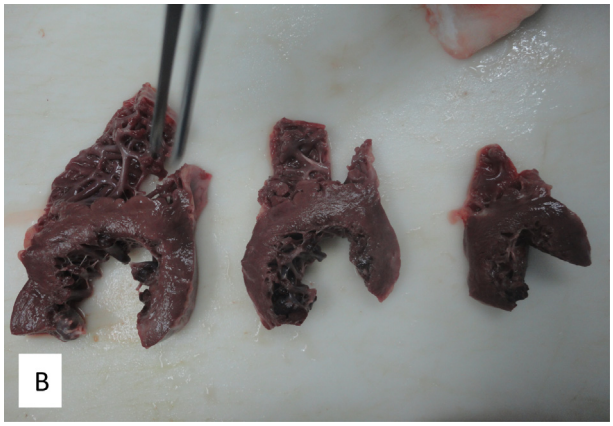

Fig. 2b The left ventricle shows excessive trabeculations and deep recesses. The cut surfaces also show areas of hyperaemia and pallor consistent with AMI.

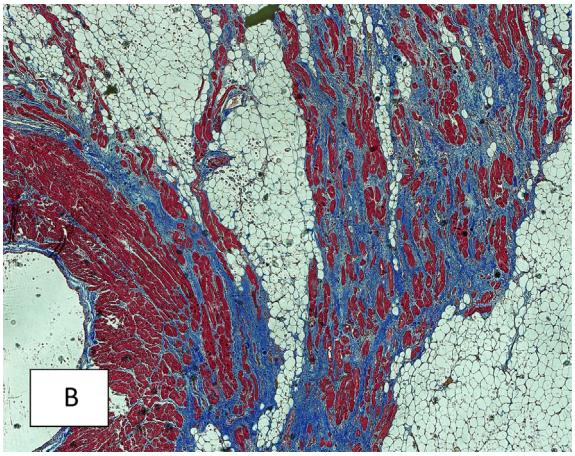

Fig. 3b Histology section using Masson trichrome stain showing extensive fibro-fatty infiltrations replacing the cardiomyocytes.

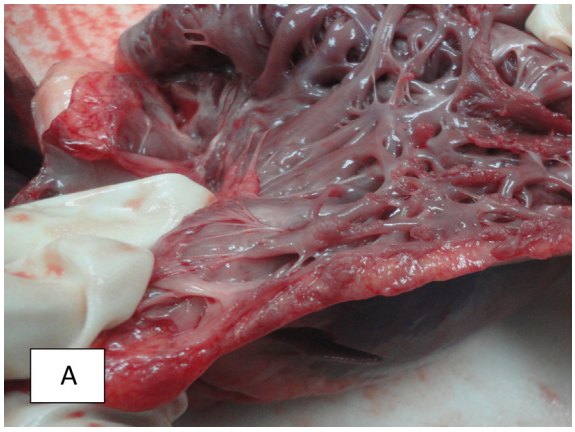

Fig. 3a The free wall of the right ventricle showing markedly thin myocardium.

left ventricle, in keeping with myocardial infarction. Samples from the right ventricle show scattered chronic inflammatory cells, patchy myocardial atrophy and massive fibro-fatty infiltrations displacing the myocytes. Masson trichrome stain aptly highlights the increase of fibrosis in the right ventricle (Fig. 3b). These features are in keeping with ARVC. In view of the gross and microscopic findings, the cause of death was given as acute myocardial infarction due to left ventricular noncompaction cardiomyopathy with arrhythmogenic right ventricular cardiomyopathy.

3. Discussion

Young and athletic individuals are thought to have lesser risk for cardiovascular diseases. Hence, SCD in this population is indeed rare. Ironically, quiescent congenital cardiovascular abnormalities may actually pose a greater risk for SCD up to 2.8 times higher compared to non-athletic counterparts.[3] The mechanism of death is usually ventricular arrhythmia as a result of exercise induced catecholamine surge acting on arrhythmogenic substrate. Other theories include dehydration, hyperpyrexia and electrolytes imbalance associated with the physical activity.[3] Our case highlights SCD in young individual. The young man was physically active and regularly engaged in sporting games such as futsal and football with his friends up to twice a week. The underlying biventricular cardiac anomalies coupled with regular strenuous activities have brought an untimely death as a result of myocardial infarction.

In the last week of his life, between the two episodes of syncope, he did not complain of any chest discomfort or pain. He insisted that he was healthy and calls to go to a healthcare facility simply fell on deaf ears. However, he did mention about feeling very lethargic even prior to the game, but insisted to play for just a short while. In retrospective, his mother described him as a healthy child and always engaged in sporting activities. There was also no history of sudden natural death of first and second degree relatives which could suggest the cardiomyopathies having familial occurrence.

The autopsy findings revealed what some of the famous athletes died of in the field, which was an underlying cardiomyopathy. However, this unfortunate young man did not just have one, but two types of cardiomyopathies affecting both the right and left ventricles. Several studies have shown that LVNC is a distinct cardiomyopathy which does not share its morphology as well as genetic basis with other forms of cardiomyopathies. American Heart Association classifies LVNC as a genetic cardiomyopathy and a few genes have been identified.[6] Mutations of Z-band alternatively spliced PDZ-motif protein (ZASP), α-dystrobrevin (DTNA), tafazzin (TAZ-G4.5) and genes encoding sarcomeric proteins have been established in LVNC.[11] In ARVC, mutations in desmosomal genes such as desmoglein-2 (DSG2), desmocollin-2 (DSC2), plakophilin-2 (PKP2), desmoplakin (DSP) and plakoglobin (JUP) were identified.[12] Apparently, the genetic bases for these two types of cardiomyopathies are not related, further emphasising the fact that they are of separate and distinctive entities. Understandably, it would be of utmost importance to investigate the possible fundamental genetic mutations in this case. Unfortunately, we were unable to get to the molecular level as the post mortem samples were subjected to fiscal issues. The next-of-kin were counselled on the findings as well as the possible familial nature of the disease right after the

autopsy. They were subsequently referred for genetic screening. To our dismay, the efforts were futile.

For both types of cardiomyopathies, neither LVNC nor ARVC usually present with AMI. In fact, common presentations for both pathologies are heart failure and arrhythmia. Acute myocardial infarction (AMI) is defined as irreversible myocardial necrosis caused by ischaemia.[13] Various causes could attribute to the myocardial injury with the top on the list is coronary artery disease (CAD). Other causes include hypertrophic cardiomyopathy, valvular heart disease, organ failure and sepsis.[13] In the case of SCD involving an athletic young adult, arrhythmia is most likely to be the mechanism of death. As AMI was prominent at gross and microscopic examinations of the heart, the authors postulate that combined cardiomyopathies which he had in the background, the heavy physical exertion during a futsal game had most probably triggered an acute episode of severe ischaemia leading to AMI. As he ignored both the syncopal attack and the lethargy, and decided to continue to play the game in the following week, it did not give time for the injury to heal. Another wave of ischaemia on top of the injured myocardium had just proved to be lethal.

In conclusion, physically active young individuals may not be spared of cardiovascular diseases and sudden death. Nonspecific and mild symptoms such as syncope and lethargy should serve as a warning sign for a possible underlying congenital cardiac abnormality in this population. Indifference may prove to be fatal.

Conflict of Interest

The authors declare that they have no competing interests.

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Ethical approval

None.

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