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CASE REPORT

Hypertrophic Cardiomyopathy with Massive Cardiomegaly in Indian Young Adult: Autopsy Case Report

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Abstract:

The sudden death of a bread winning adult in the family is devastating for any family. Even worse is when the adult dies in his young adulthood. Cardiovascular pathologies dominate the causes of sudden deaths in young adults more than the central nervous and respiratory systems. The enlarged heart or cardiomegaly at autopsy is the first indication of such cardiac pathology. Cardiomegaly may result from various pathologies such as hypertension, cardiomyopathies, rheumatic valvular diseases, etc. The resultant increase in the heart's weight due to the cardiac myocytes' adaptive response to meet the demand in the above conditions is known as cardiac hypertrophy. Although multiple reports on cardiomegaly were described from clinical settings based on imaging modalities like X-ray or echo cardiography, the data is lacking from forensic autopsies, especially from India. This is the first case report describing a young adult male of 30 years old who died due to massive cardiomegaly with a heart weight of 878 g from hypertrophic cardiomyopathy.

Keywords: Cardiomegaly; Sudden death; Hypertensive cardiomyopathy; Left ventricular hypertrophy; Forensic pathology

Introduction:

The sudden death of young adults poses devastating consequences for the surviving family. Cardiovascular diseases are the most common cause of premature death in young adults at autopsy.[1] While atherosclerotic coronary artery diseases cause more fatalities in adults more than 35 years, fatal arrhythmias from structural abnormalities of the heart that include cardiomyopathies or due to arrhythmogenic disorders such as long QT syndrome, Brugada syndrome, etc. result in more deaths among adults younger than 35-40 years.[1,2] However, it is predicted that the cause of death in about 30% of sudden death in the young still may remain unascertained after a complete autopsy.[2]

Heart weight is the first and fundamental evidence of cardiac pathology.[3] The average heart weight in men is about 300 g, and in women, about 250 g, which roughly corresponds to about 0.45 and 0.40%, respectively, of an individual's body weight in the Indian population.[4] However, the heart weight varies across different people in other countries and is primarily influenced by various factors such as age, race, body height, weight, BMI, etc. For example, the average heart weight in the USA is predicted to be 331 g,[5] remarkably higher than the Indian population.

Enlargement of the heart from hypertrophy or dilatation of the chamber is known as cardiomegaly.[6] It is predicted that a person having a heart weight of more than 420 g to 450 g is a candidate for sudden death irrespective of coronary artery stenosis.[4,7] This increase in heart weight most likely results from cardiac hypertrophy, an adaptive response to the increased demands of physiological or pathological stress on the cardiac tissue. It also serves as a pathological substrate for arrhythmias and enhances the risk for cardiac failure, resulting in sudden cardiac deaths.[8]

Various pathologies such as hypertension, cardiomyopathies (hypertrophic, dilated, restrictive), rheumatic heart disease, cor pulmonale, ischemic heart diseases, and obesity are implicated in the genesis of cardiomegaly.[6] Numerous reports in the medical literature describe cardiomegaly due to the above etiologies, extensively from clinical settings based on imaging modalities like X-ray or echo cardiography. However, the data is lacking on incidences of massive cardiomegaly found in forensic autopsies, especially from India. To the best of our knowledge, this is the first report describing massive cardiomegaly (Heart weight: 878 g) due to hypertrophic cardiomyopathy in an Indian young adult.

Case Report:

A 30-year-old male auto driver by profession was brought dead to the emergency of our tertiary care center with an alleged history of being found unconscious in his auto around 5 pm. His medical and surgical history was unremarkable. However, the family history suggested his father dying of some unknown ailments, probably of cardiac origin. On external examination, the body was of a young male with an average build. Blood-tinged mucoid fluid is present in and around both nostrils. The rigor mortis was developed throughout the body, and lividity was fixed and present over the dependent parts of the back except over pressure areas. There were no signs of putrefaction and no external injuries present on the body. All clothes were intact and free from any visible discharge or stain. On the internal examination, the brain was remarkable for cerebral edema and multiple petechial hemorrhages in the white matter of the bilateral cerebrum and

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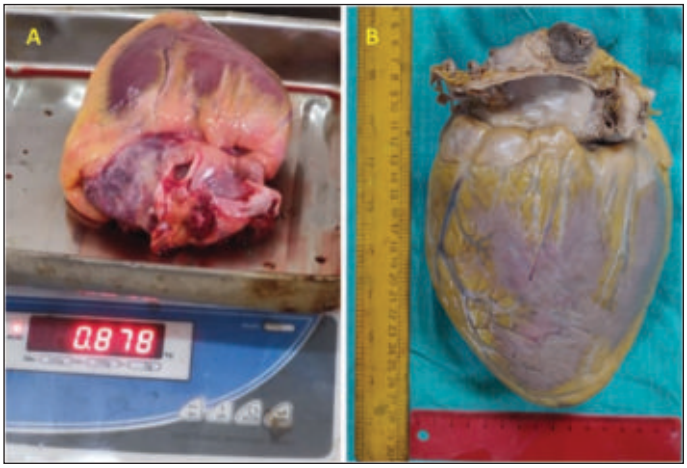

Figure 1. Shows weight of the heart measuring 778 g (1A) and grossly enlarged heart suggestive of massive cardiomegaly (1B).

Figure 2. demonstrates the asymmetric hypertrophy of the left ventricular wall (free wall>septum) on the cut section.

cerebellum.

The opening of the chest cavity has revealed striking enlargement of the heart (cardiomegaly), which weighed 878g after washing (Figure 1). The pericardial sac contained about 60 ml of serous fluid. The epicardial surface of the heart was unremarkable. Multiple atheromatous plaques were present in the lumen of the root of the aorta. There was no significant luminal narrowing present in the lumen of coronary arteries. Asymmetric hypertrophy of the left ventricle (Free wall >Septum) (Figure 2) was noted with the left ventricular wall thickness (LVWT) of 2.1 cm and the right ventricular wall thickness (RVWT) of 0.7 cm; both were measured 01 cm below the atrioventricular valves. Rest of the examination of the heart was unremarkable.

Both lungs were congested and edematous. On the cut section, blood-tinged frothy fluid was oozing out at places in both lungs. The stomach contained about 200g of semi-digested food material with no peculiar odor and mildly congested mucosa. The liver, spleen, and kidneys were congested, and the rest of the organs were unremarkable. Histopathological examination of the

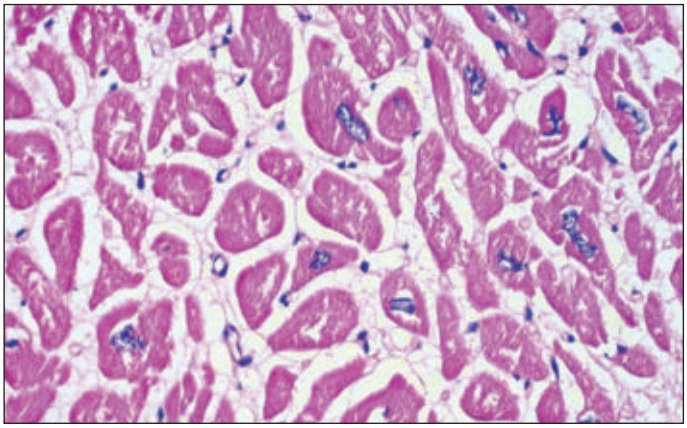

Figure 3. Exhibits photomicrograph of the cardiac muscle (H & E, 40X) exhibits enlarged, hyperchromatic nuclei indicative of cardiac muscle hypertrophy.

heart revealed cardiac muscle hypertrophy (Figure 3). The chemical analysis report was negative for any common poisons and intoxicants. Given the above findings, the cause of death was ascribed to hypertrophic cardiomyopathy and its complications which is a natural pathology.

Discussion:

Massive cardiomegaly with a heart weight of more than 800 to 850g is most often encountered in cardiomyopathies, valvular heart diseases, chronic atherosclerotic heart disease, congenital heart disease and morbid obesity.[8] Hypertrophic cardiomyopathy (HCM) is one of the most common causes of death among young adults. It is genetically inherited as an autosomal dominant disorder and frequently presents with sudden death as the first manifestation without any prior warning signs or symptoms. This is evident in the present case, who exhibited no signs and symptoms before death except for occasional mild chest discomfort. Clinically, age (less than 35 years), family history of sudden cardiac death in the first-degree relatives, maximum left ventricular wall thickness (LVWT), recent episode of unexplained syncope in the preceding six months, left ventricular outflow tract obstruction, LV systolic dysfunction, and LV apical aneurysm, etc. are attributed to be the indicators of HCM.[9] It is worth mentioning here that the patients with SCD of first-degree relatives are 20% more prone to die from SCD.[10] This is consistent with our case, whose father also had sudden premature death due to an unknown cardiac disease indicative of possible genetic inheritance.

Establishing the cause of death in SCD requires a detailed clinical history with special attention to the deceased's age and the circumstances of the death.[8] Diagnosis of HCM is challenging since it often results in no or minimal symptoms. Autopsy surgeons should preferably corroborate the findings of ECG and echocardiography together, if available, to arrive at a diagnosis of HCM. Because relying on either ECG or echocardiography, may result in poor judgment as both may indicate distinct risk information about LVH.[8]

Grossly, HCM is characterized by cardiomegaly and asymmetric hypertrophy of the septal wall. However, it should be stressed

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here that the free wall hypertrophy may occasionally supersede septal hypertrophy[3,8] as observed in the present case. There is no documented case of massive cardiomegaly from young adults at autopsy in India except a single case report by Hugar et al. from Bangalore, India.[11] The report described a case of coronary artery insufficiency resulting from massive cardiomegaly with a heart weight of 880 g in a 46-year-old male. In contrast, the present case was a young adult of 30 years with an average build (unlike obese as described in Hugar et al.) who died due to massive cardiomegaly in the setting of HCM.

Although cardiac myocyte hypertrophy, myocyte disarray, and interstitial fibrosis are frequently reported microscopical features of HCM, they are not pathognomic of HCM. For example, myocyte disarray may also present in congenital heart diseases and in normal hearts.[3] It is important to note that the sampling was done from left ventricular septal and free walls at different levels, but the myocyte disarray could not be elicited in the present case. This may be attributed to the absent myocardial disarray from the selected sites, given the possibility of its patchy involvement within the myocardium.[12]

HCM arises from over 8000 variants of genetic mutations involving more than 50 genes. The most common genes responsible for HCM are MYBPC[3] and MYH7 genes that encode cardiac myosin-binding protein C (cMyBP-C) and beta-myosin heavy chain (beta-MHC), respectively.[9] When the cause of death is difficult to be ascertained, especially in arrhythmogenic heart disorders, the genetic testing of DNA from the postmortem blood of the deceased may establish the diagnosis. This molecular autopsy has been increasingly practiced in developed countries in recent decades.[2] However, in the present case, the diagnosis of HCM was made from family history, gross and histological examination of the heart. Hence, the molecular autopsy was not done in the present case. However, the deceased's family members were counseled to undergo periodic checkups with the cardiologist to prevent fatal outcomes. The early screening and identification of the HCM may help reduce the associated mortality by opting for various life-saving treatment modalities, including implantable cardioverter-defibrillator.[10,13]

Conclusion:

Massive cardiomegaly with a heart weight measuring 878g resulting in sudden death in the Indian young adult is described in the report. This report highlights the importance of diagnosing the exact pathogenic cause (HCM) leading to cardiac hypertrophy. Because the proper identification may indicate genetic conditions like HCM running in the families and helps prevent the morbidity and mortality of the first-degree relatives of the victim in the future. Hence, the autopsy surgeons should exercise utmost diligence and involve a multi disciplinary team, including pathologists, cardiologists, geneticists, and genetic counselors, when encountering such cases in forensic practice.

Ethical Clearance: Taken from Institutional Ethical Committee, AIIMS Bhubaneswar. The identity of the deceased was not revealed in the manuscript.

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Conflict of Interest: None.

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