CASE REPORT

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Sudden Unexpected Death in Young Adult due to Right Ventricular Dysplasia

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ABSTRACT: This case report illustrates a rare familial cardiomyopathy first reported in the medical literature in 1982 known as right ventricular dysplasia (right ventricular cardiomyopathy). The patient is a young woman with a history of cardiac arrhythmias suspected to be associated with prolapsed mitral valve who presented to the Berks County Coroner's office as a sudden unexpected death in a young adult. It is important to recognize the illustrated classic cardiac pathology of this rare entity for clinical management, as an anatomic explanation of cause of sudden death and for the accumulation of statistics to establish frequency, conditions of predisposition, response to therapy and predicted outcome.

KEYWORDS: forensic science, forensic pathology, right ventricular dysplasia, (arrhythmogenic) right ventricular cardiomyopathy, lipomatous infiltration of the heart, sudden unexpected death, young adults

Sudden unexpected deaths in healthy young adults are associated with profound grief by family members and loved ones along with the additional perplexity demanding a medical explanation for this tragedy. The following case will serve to illustrate an infrequent but definite cause of death. A 33-year-old female underwent laparotomy with salpingectomy for ruptured ectopic tubal pregnancy with an uncomplicated postoperative clinical course. One month following surgery her lifeless body was discovered by her husband in the kitchen. Past medical history reveals an irregular heartbeat first noticed six years previously with medical evaluation resulting in the diagnosis of mitral valve prolapse. Symptoms abated following brief treatment with Lopressor. Four years later and two years before the fatal event cardiac palpitations again became symptomatic, cardiovascular evaluation revealed a non ejection click following a very short early systolic murmur, the electrocardiogram was interpreted as being within normal limits, and the diagnosis again was mitral valve prolapse. Treatment with Lopressor again relieved symptoms. She did not smoke, abuse alcohol, or use any other

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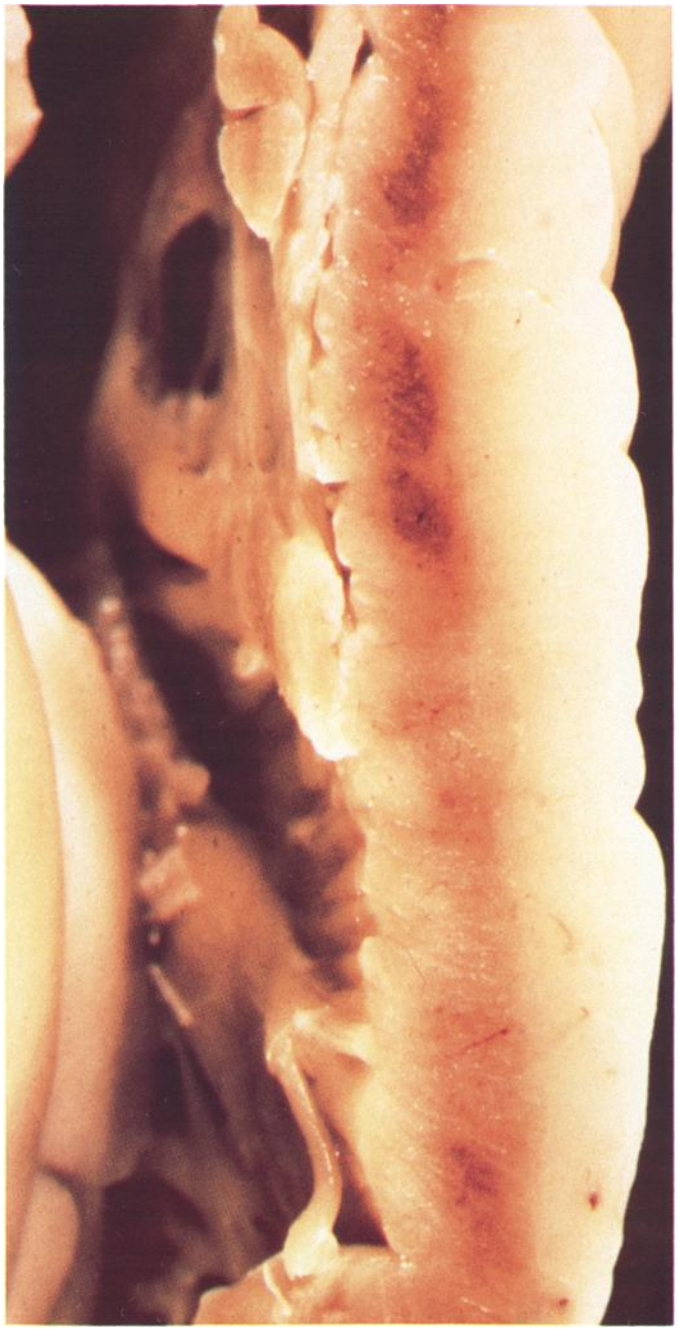

Fig. 1—*Right ventricular transmural myocardial replacement by mature adipose connective tissue.*

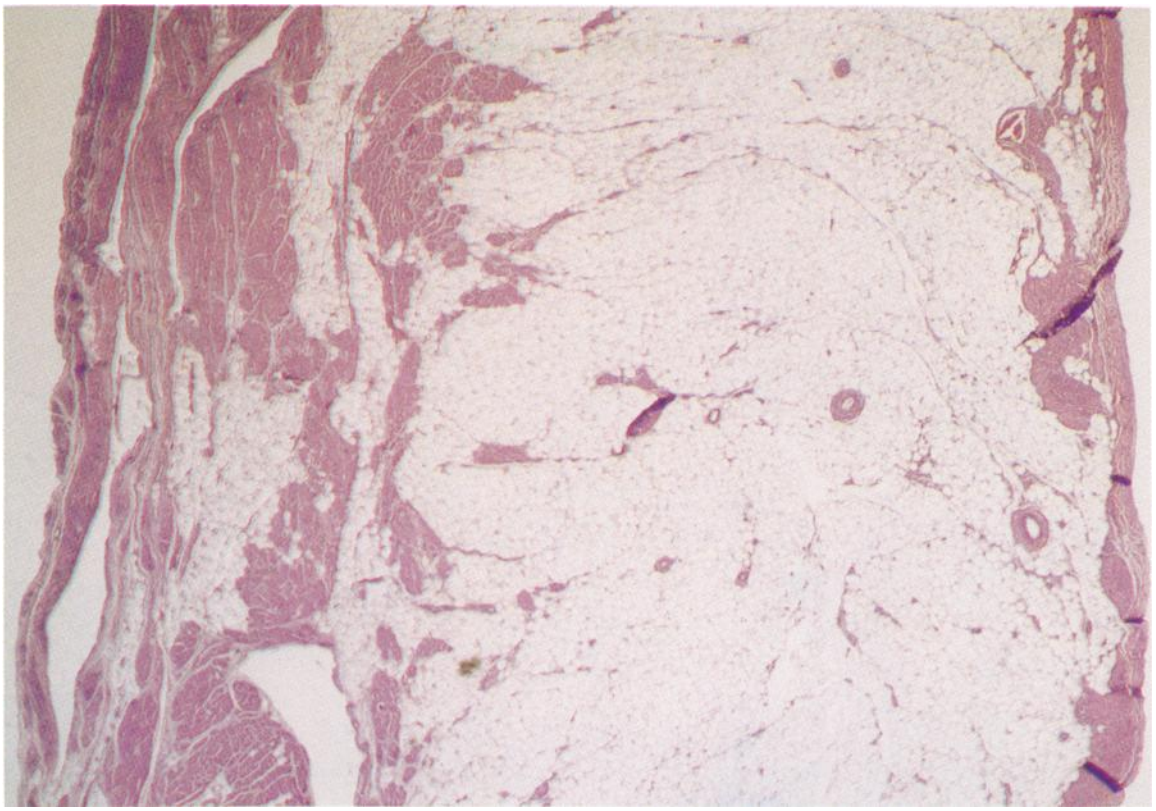

FIG. 2—*Epicardial and endocardial surfaces with transmural mature adipose connective tissue replacement. Few subendocardial myocardial fibers with no inflammation. (Hematoxylin & Eosin. ×20).*

prescription or nonprescription medications and was not a user of illicit drugs. Autopsy findings revealed a well-developed well-nourished female with two minor facial contusions. The well-healed recent transverse suprapubic surgical incision was uncomplicated. Significant findings were related exclusively to the 360 g heart which demonstrated a somewhat expanded rounded right ventricular contour, normal chambering with flattened right ventricular trabeculae carneae, and diffuse full thickness lipid infiltration of the right ventricular myocardium (Fig. 1). The interventricular septum was not involved, and the mitral valve leaflets, chordae tendineae, and papillary muscles appeared anatomically normal and physiologically competent. There was no atherosclerotic coronary artery disease. Both lungs were heavy with bulging edges due to acute passive congestion. Microscopic findings demonstrate presence of mature adipose connective tissue representing subtotal to total transmural right ventricular replacement from the epicardial to endocardial surfaces with focal presence of individual haphazardly arranged myocardial fibers or small fiber groups in some areas (Fig. 2). Areas of necrosis and chronic inflammation were not identified. Postmortem toxicology failed to demonstrate detectable levels of antiarrhythmic medication, other prescription and nonprescription drugs as well as illicit drugs of abuse.

Discussion

This case exhibits many of the features reported by Goodin et al. (1,2) in their review of six sudden deaths in young adults from the office of the Chief Medical Examiner in Baltimore, Maryland and the files of the Cardiovascular Branch of the Armed Forces Institute of Pathology in Washington, D.C. This excellent summary report lists the salient features of sudden cardiac deaths in young adults and emphasizes the rarity of right ventricular dysplasia accounting for less than 1% of these deaths. Although it is estimated that as many as 14% of young adult cardiac deaths will demonstrate an anatomically normal heart it is essential to be aware of the rare anatomic conditions which may be implicated. Absence of the right ventricular myocardium with paper thin wall and apposition of the epicardial and endocardial surfaces is described as Uhl's anomaly, whereas the myocardial absence with lipid replacement is more appropriately referred to as right ventricular dysplasia, right ventricular cardiomyopathy or arrhythmogenic right ventricular dysplasia (3,4).

Conclusions

Symptoms of RVD may be late onset and may initially respond dramatically to medication producing a false sense of security belying the seriousness of the condition. RVD may be misdiagnosed as some other cardiac pathology. Consideration of this condition along with appropriate radiologic studies and possibly endomyocardial biopsy should be able to establish the diagnosis. Although treatment with anti-arrhythmogenic medication may alleviate intermittent symptomatology lifelong therapy and monitoring are essential. Consideration for cardiac transplantation may be an appropriate approach in some clinical situations. The intrinsic value of autopsy pathology contribution to ascertaining cause of death as well as establishing societal statistical frequencies of pathological entities are emphatically demonstrated with this case.

References

1. Goodin JC, Farb A, Smialek JE, Field F, Virmani R. Right ventricular dysplasia associated with sudden death in young adults. Mod Path 1991;4:702–6.
2. Burke A, Virmani R. Atlas of tumor pathology, tumors of the heart and great vessels. Third Series, Fascicle 16. 1996;95–6.
3. Gerlis LM, Schmidt-Ott SC, Ho SY, Anderson RH. Dysplastic conditions of the right ventricular myocardium: Uhl's anomaly vs. arrhythmogenic right ventricular dysplasia. Br Heart 1993; 69(2):142–50.
4. Solenthaler M, Ritter M, Candinas R, Jenni R, Schneider J, Amann FW. Right ventricular dysplasia (right ventricular cardiomyopathy). Clinical aspects, diagnosis and course in 15 patients from the Zurich area. Schweiz Med Wochenschr [J Suisse Med] 1993;123(34): 1604–14.

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