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CASE REPORT

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Sudden death associated with myxomatous transformation of the mitral valve in an 8-year-old boy

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Abstract An 8-year-old boy suffered a cardiac arrest while playing soccer. In contrast to a similar event at the age of 5 years, resuscitation was not successful. At autopsy, the cardiac findings were of a myxomatous transformation of the mitral valve with lacerations of the posterior cusp and the left vestibular endocardium and left ventricular hypertrophy. Sudden death due to arrhythmias in association with a myxomatous mitral valve is a rare event with only about 100 cases published world-wide. To our knowledge, the present case probably reports the youngest affected individual who died of this pathological condition.

Key words Sudden death · Myxomatous mitral valve · Arrhythmias

Introduction

The myxomatous transformation of the mitral valve is a relatively common condition in man, characterized by disintegration of the valvular connective tissue with accumulation of acid mucopolysaccharides in the connective tissue compartments of the valve [4]. According to Silver [14] it can be found in up to 10% of apparently healthy young adults in clinical studies. As normality merges into abnormality, the entity has been given a variety of names by diverse medical specialities, leading to a certain degree of confusion in the medical literature [1]. Generally, the prognosis is good and sudden cardiac death is a very rare complication [3]. From review articles on this topic only about 100 such cases have been published world-wide [11] and the most frequent cause of this event seems to be ventricular tachyarrhythmia [6, 17]. The pathogenesis is still unknown [10]. Persons afflicted with this condition and with known clinical signs during their lifetime are often subjected to routine post mortem pathology examination. The diagnosis of a lethal complication associated with a myxomatous mitral valve is therefore a rarity in forensic practice. Moreover, to our knowledge this case presents the youngest known victim of this pathological condition.

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Case report

An 8-year-old boy suddenly collapsed for no evident reason while playing soccer. A physician who was coincidentally present, diagnosed a cardiac arrest and immediately started cardiopulmonary resuscitation (CPR) which was continued extensively in the ambulance and in hospital. As it was not possible to re-establish a cardiac rhythm, CPR was terminated about 2 h after the incident.

The history revealed that the child had already had a cardiac arrest at the age of 5 years while tobogganing and CPR at that time had been successful. The extensive medical examinations which were subsequently carried out had not revealed any pathology findings. In particular, there was no evidence for cardiac conductivity disorder and the family case history showed no associated pathology.

Autopsy findings

- "Glassy", swollen aspect of the mitral valve margin and of the elongated chordae tendineae (Fig. 1)
- Both mitral cusps with sites of opaque, focal thickening with no apparent "hooding" or "ballooning" in the sense of a focal interchordal prolapse (Fig. 1)
- Thickening of the endocardium and dilatation of the left vestibule
- Left ventricular hypertrophy with a left ventricular wall thickness of 1.2 cm (Fig. 1) and pronounced enlargement of the heart (heart-weight 170 g, normal mean value 131.7 g) [15]
- Body weight and height age-appropriate [9]
- A tear in the posterior mitral cusp reaching from the closure line of the valve almost to the valve ring (Fig. 1)
- A superficial fissure of the left vestibular endocardium in very close proximity to the atrioventricular valve (Fig. 1)
- No other gross pathological findings in any of the intimal organs and blood vessels

Histologically, both mitral cusps of the mitral valve showed considerable focal myxomatous transformation with a thickened spongiosa layer encroaching on the atrialis and fibrosa layers with an

Fig. 1 Mitral valve and surrounding heart muscle tissue showing a vitrous swollen aspect of the mitral valve margin and of the seemingly elongated chordae tendineae as well as focal sites of opaque thickening (i.e. larger arrow) of the valve cusps. Furthermore the left ventricular hypertrophy and the lacerations (small arrows) of the posterior cusp and the vestibular endocardium can be noted

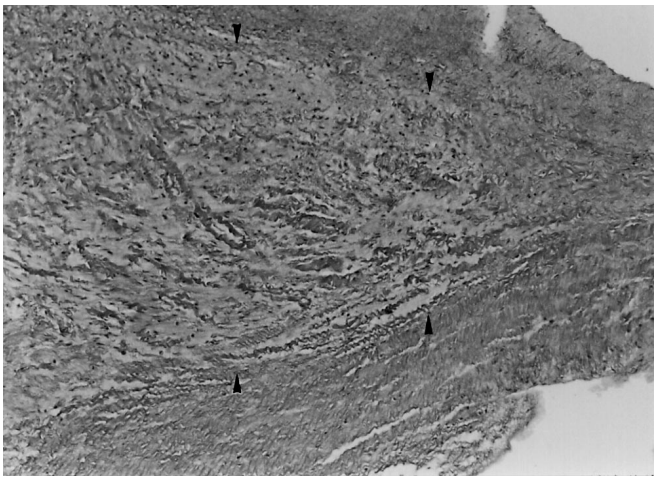

Fig. 2 Section of the mitral valve stained by Alzian blue and van Gieson method (× 95). Considerable focal myxomatous transformation with a thickened spongiosa layer encroaching on the atrialis and fibrosa layers. Area with accumulated proteoglycans marked by arrows

Alcian blue-positive accumulation of proteoglycans in the interstitial tissue (Fig. 2). There was no evidence of similar alterations in the other three cardiac valves. In accordance with the gross findings, microscopical examinations merely confirmed a high-grade left ventricular hypertrophy while the cardiac conduction system [19], as far as seen in the numerous heart sections, revealed no abnormalities. All other organs were histologically unremarkable. Toxicological examinations were negative.

Discussion

As already pointed out, a large variety of names exist in the medical literature for the entity of the myxomatous transformation of the mitral valve. To clarify the confusion it has been proposed that the term mitral valve prolapse (MVP) or mitral valve prolapse syndrome (MVPS) should be used [1]. The principal complications of this condition include systemic emboli, infective endocarditis, progression to severe chronic and acute mitral regurgitation, arrhythmias and as a consequence of these, rare and sudden death [2]. Malignant ventricular arrhythmias have clinically been detected before the fatal event [17], leading to ventricular fibrillation in most cases [12]. A possible autosomal dominant pattern of inheritance is meanwhile generally accepted [3, 13]. However, discussion still continues concerning the aetiology and pathogenesis of the condition.

Bearing the myxomatous mitral transformation and the absence of any other causes of death in mind, there principally seem to remain only two explanations for the lethal occurrence in the case reported.

The laceration of the posterior mitral cusp would indicate an acute mitral insufficiency. However as this lesion was in close proximity to a superficial fissure of the left vestibular endocardium it seems more likely that both findings are due to the extensive CPR carried out, given that higher fragility and brittleness of the myxomatously transformed tissue is more probable. This assumption is additionally affirmed by the reported sudden collapse of the boy, which would be an unusual manifestation of an acute mitral insufficiency due to valvular lesions of the described extent. Moreover, there is a complete lack of comparable descriptions of spontaneously ruptured valves in the available literature whereas there are publications of sudden death due to acute ruptures of the chordae tendineae of myxomatous mitral valves [5].

On the other hand sudden arrhythmias could be held responsible for the fatal outcome in this instance. As a pronounced left ventricular hypertrophy without evidence for aortic valve stenosis was found, it seems justified to postulate a mitral valve prolapse (MVP) along with a degree of insufficiency which must have been compensated, as there were no signs of chronic congestion. This does not contradict the fact that no “hooding” of the mitral cusps was found at necropsy as the phenomenon could have been functional and/or in an early, not yet macroscopically detectable stage although the chordae tendineae already seemed elongated. The fact that, according to the literature [7], significant mitral regurgitation is an important predisposing factor for sudden death in patients with myxomatous mitral valve transformation therefore indicates the assumption of arrhythmias. It is very likely that they were triggered or increased by the physical exercise (soccer in this case), causing an elevated sympathetic tone and higher blood concentrations of catecholamines [18]. The similar constellation at the age of 5 years, when the boy suffered a cardiac arrest while tobogganing, further underlines this reasoning. Theoretically it has to be admitted that arrhythmias leading to sudden death can of course develop independently of the described primary disease. Considering the case history, the gross and microscopical findings and the frequency of arrhythmias associated with myxomatous transformation of the mitral

valve reported in the literature [6, 8, 16, 18], this possibility can however be neglected. The cause of the dysrhythmias in subjects with a myxomatous mitral valve is still unclear. A variety of explanations are under discussion, the most favoured one appears to be traction on the papillary muscle as the mechanism for repolarization changes [3]. It remains obscure, why no cardiac abnormalities were detected in the course of the examinations after the first incident. This might perhaps be due to the assumption that the changes were so minute, that they were technically not detectable. Another possible explanation would be a lack of compliance during the examinations at the age of 5 years or that they were, especially concerning electrocardiography, carried out in a period of time without any arrhythmic potential at all.

As arrhythmias cannot be recognized morphologically an unambiguous cause of death could not be given in this case. In the absence of other reasons for this demise and in recognition of the pathology findings and the case history an arrhythmia, as a rare complication of a myxomatous transformed mitral valve, is believed to be responsible for the cardiac arrest and the death of the 8-year-old boy perhaps more of a diagnosis by exclusion.

References

1. Abrams J (1976) Mitral valve prolapse: a plea for unanimity. Am Heart J 92 (4):413–415
2. Barlow JB, Pocock WA (1988) Mitral valve billowing and prolapse: perspective at 25 years. Herz 13 (4):227–234
3. Chesler E, King RA, Edwards JE (1983) The myxomatous mitral valve and sudden death. Circulation 67 (3):632–639
4. Davies MJ (1980) Pathology of cardiac valves. Butterworths, London Boston, pp 62–92
5. Ferguson DW, Kiefaber RW, Ziegelman DS, Uphold RE, Jackson RS, Tabakin BS (1987) Acute rupture of myxomatous mitral valve presenting a refractory cardiopulmonary arrest. J Am Coll Cardiol 9 (1):215–220
6. Jeresaty RM (1976) Sudden death in the mitral valve prolapse-click syndrome. Am J Cardiol 37:317–318
7. Kligfield P, Hochreiter C, Kramer H, Devereux RB, Niles N, Kramer-Fox R, Borer JS (1985) Complex arrythmias in mitral regurgitation with and without mitral valve prolapse: contrast to arrythmias in mitral valve prolapse without mitral valve prolapse. Am J Cardiol 55:1545–1549
8. Kolibash AJ, Bush CA, Fontana MB, Ryan JM, Kilman J, Wooley CF (1983) Mitral valve prolapse syndrome: analysis of 62 patients aged 60 years and older. Am J Cardiol 52:534–539
9. Largo RH (1991) Wachstum und somatische Entwicklung. In: Betke K, Künzer W, Schaub J (ed) Lehrbuch der Kinderheilkunde. Thieme, Stuttgart New York, p 27
10. Marshall CE, Shappell SD (1974) Sudden death and the ballooning posterior leaflet syndrome. Arch Pathol 98:134–138
11. Penning R, Betz P, Werdan K (1991) Zur Häufigkeit eines plötzlichen Herztodes bei Mitralklappenprolaps-Syndrom. Versicherungsmedizin 43:83–88
12. Perloff JK, Child JS (1987) Clinical and epidemiological issues in mitral valve prolapse: overview and perspectives: Am Heart J 113:1324–1332
13. Shappell SD, Marshall CE, Brown RE, Bruce TA (1973) Sudden death and the familial occurrence of mid-systolic click, late systolic murmur syndrome. Circulation 48:1128–1134
14. Silver MD (1991) Cardiovascular pathology, vol 2, 2nd edn. Churchill Livingstone, New York Edinburgh London Melbourne Tokyo, p 963
15. Stocker JT, Dehner LP (1992) Pediatric Pathology, vol. 2. Lippington, Philadelphia, p 1317
16. Swartz MH, Teichholz LE, Donoso E (1977) Mitral valve prolapse: a review of associated arrhythmias. Am J Med 62:377–389
17. Vohra J, Sathe S, Warren R, Tatoulis J, Hunt D (1993) Malignant ventricular arrhythmias in patients with mitral valve prolapse and mild mitral regurgitation. Pace 16:387–393
18. Winkle RA, Lopes MG, Fitzgerald JW, Goodman DJ, Schroeder JS (1975) Arrhythmias in patients with mitral valve prolapse. Circulation 52:73–81
19. Zack F, Wegener R (1994) Zur Problematik der Diagnose „rhythmogener Herztod“ durch histologische Untersuchungen des Erregungsbildungs- und -leitungssystems. Rechtsmedizin 5:1–5