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INBORN HEART DEFORMATION AS A CAUSE OF SUDDEN DEATH IN A 7-YEAR OLD GIRL

Abstract: We report the case of a 7-year old girl who is treated with antibiotics because of an angina tonsillaris. Five days later, vomiting and diarrhea develop and the child is admitted to a hospital. Shortly after admission, cardiac arrest occured, and rescuscitation attempts proved to be unsuccessful. Autopsy shows an inborn heart deformation of the right and also of the left, ventricle: basal trabecular hypertrophy and outflow problems such as a subvalvular pulmonary stenosis in the right ventricle. Additionally, hints for a chronic heart overload were seen in both ventricles. Histologically, multiple necrotic areas in the inner muscle layers in various stages of organization appear besides a multifocal, irregular course of hypertrophic muscle cells as well as fatty inclusions in the myocardial cells. The described heart deformation can be classified nearest as a primary cardiomyopathy with consecutive ventricular noncompaction. It remains questionable, how the girl could reach an age of almost eight years, even without showing signs of cardiac insuffiency.

Case

A 7 year old girl suffers from angina tonsillaris, and amoxicillin is prescribed. 5 days later, the child is admitted to a hospital as during the antibiotic therapy, repeated vomiting and diarrhoeas occurred. An antiemetic drug is prescribed, but the girl vomits repeatedly during the following night. The child is briefly unconscious and cerebral convulsions take place. Because of the possible diagnosis of volvulus/invagination, an abdominal sonography is performed. During sonography, the child suddenly shows tachyarrhythmia, generalized stretching cramps and finally a cardiac arrest. Immediate resuscitation measures are performed but are cancelled unsuccessfully after more than 1 hour. The sudden death is unexplainable. As now inappropriate medication as cause of death is assumed, a forensic autopsy is ordered.

Autopsy findings

Autopsy shows a heart [150g] deformation in both ventricles: In the lower half of the right ventricle partially over-crossing, clearly thickened trabecular muscles limit the full development of the ventricle strongly. The right blood flow course is narrowed

by a bulge-like trabecular muscle. Below the pulmonal valve, the endocardium is fibrotic. There is a strong dilatation of the basal ventricular half by general myocardial thickness. In the left ventricle, one mitral valve segment is split, the strong musculature overlaying itself partly. The endocardium is changed below the aortal valve fibrotically, the left atrium is moderately extended. The coronary arteries are inconspicuous. The environment of the tonsils does not exhibit any inflammatory changes. The suspected diagnosis volvulus / invagination cannot be confirmed.

Histology

The hypertrophied myocardial fibres are bizarrely over-crossing each other (Fig. 3b). Multiple necrotised areas in the subendocardium in different stages of organization predominate (Fig. 4a + 4b). The necrotic muscle cells present themselves in the immunohistochemical staining (C9) noticeably and can be differentiated from autolytic cells (Fig. 4b). There is a histological gradient of increasing necroses from outer to inner myocardium; arterialisation of the subendocardial myocardium is rarified. There are multiple, partly cord-like lined-up fat intracellular inclusions in some myocardial cells which partly dissolve the muscular continuity (Figure 4c).

Bacteriology / Virology

Microbiological investigations of the lungs show α-haemolysing Streptococci (physiological upper lung flora). In the blood culture, Clostridium species, Citrobacter freudii and Staphylococcus aureus can be proven. These findings contain no signs of sepsis. Viral DNA (Cytomegaly, Herpes simplex, Epstein-Barr, Varizella, Enterovirae, Adenovirae, Parvovirus B-19) was not detected.

Cause of Death

Acute heart failure by arrhythmia.

Discussion

The splitting of the left mitral valve muscle is no disease – here a large range of physiological variants exists [5]. The myocardial hypertrophy leads towards the diagnosis of primary cardiomyopathy (PC) with dilatative, hypertrophic, obstructive and obliterative components. The hypertrabeculation narrowing the right flow course can be added in terms of a subvalvular pulmonal stenosis to PC [7,8]. The subaortal endocardium fibrosis permits the conclusion that also in the left ventricle a developmental disturbance with in vivo muscular stenosis (during contraction) was present. Etiology of PC is various: hamartoma, genetic arrangement, myocardial metabolic disturbances, unnoticed hypertonia, etc. are discussed. With PC, a generalized

greasing of the myocardial cells and generalized fatty degeneration is known. The extent of the diffuse, fine-dropped fatty degenerations of myocardial cells with a gradient from the outside inward is too small pronounced and not generalized, so we interpret this as a hypoxia-caused degeneration of chronic myocardial ischemia [5]. The histological overall view is also not to be explained by the protracted frustrane resuscitation, as the necroses are already in different stages of organization. The prognosis of PC is bad, therapy attempts exist in the administration of ß-blocker and/or myectomia [8]. An already intrauterine developing PC may lead to ventricular noncompaction (VNC) and myocardial hypertrabeculation [2,3,6,9,10,11]: During embryogenesis, the loose network of myocardium fibres which are supplied via sinusoidal recessus directly by the ventricle clearing "compacts" and a connected muscle is formed [10]. A lack of this "compaction" leads to abolition of the myocardial texture in the inner muscle layer [1,2,3] and thus causes a morphologic hypertrabeculation with endocardium-covered sinusoids communicating with the ventricle clearing ("sponge heart"/"spongy myocardium"). VNC is considered to be a rare, innate and usually left-sided arising disturbance of the myocardial morphogenesis, but seldom cases report an affection of the right ventricle. Etiology is not enlightened yet. An underlying pathologic mechanism consists of a restricted flow course – in this case due to subvalvular pulmonal stenosis – with intrauterine intraventricular hypertonus. This may handicap the fusion of the loose myocardium fibres, the heart muscle is altogether unsatisfactorily consolidated. Such hypertonus also obstructs the angiogenesis in the ventricle, so that besides vascular rarifications also ischemic necroses and hypoxic fatty inclusions may result. As histological findings, these necrotic areas impress with minimal inflammatory reaction. Therapy is heart transplantation [4]; prognosis is bad, fatal are cardiac arrhythmias [1,2], heart failure and thrombembolic events [11]. It is remarkable that the girl never showed any signs of cardiac insufficiency. Typically, children concerned with heart deformations are in the last third of their age [7]. It must be assumed that in the described case sufficient compensation mechanisms were available, which possibly failed later in the context of an infection. Possibly a syncope could have taken place and was misinterpreted as cerebral convulsion. This case shows the fateful process of a non-diagnosed cardiomyopathy with consecutive VNC of the right ventricle.

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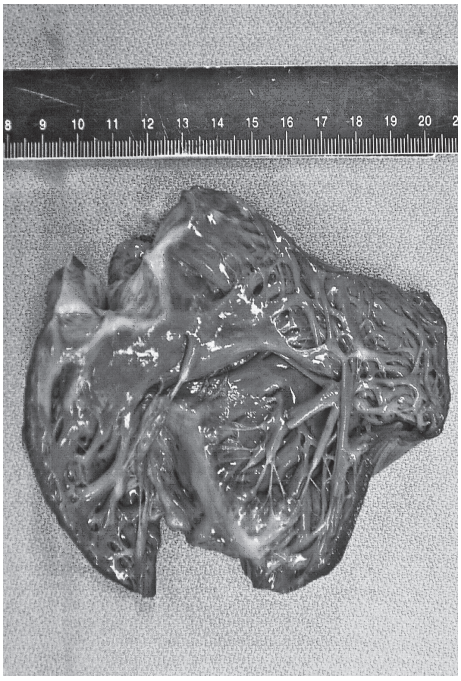

Figure 1 – Right heart: trabecular hypertrophy and endocardial fibrosis

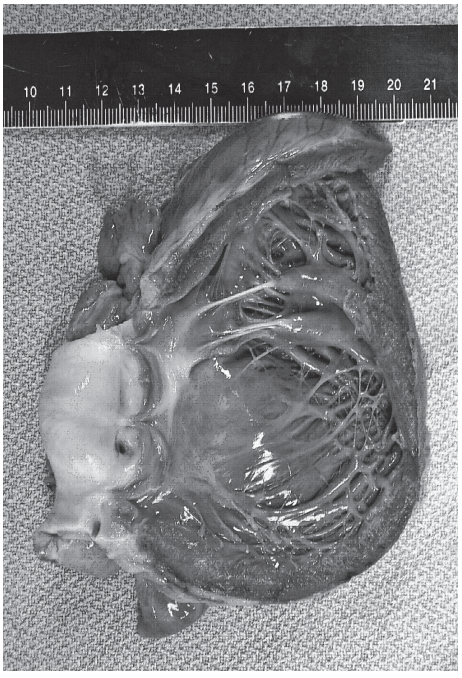

Figure 2 – Left heart: subaortal fibrotically changed endocardium and amplified trabeculation

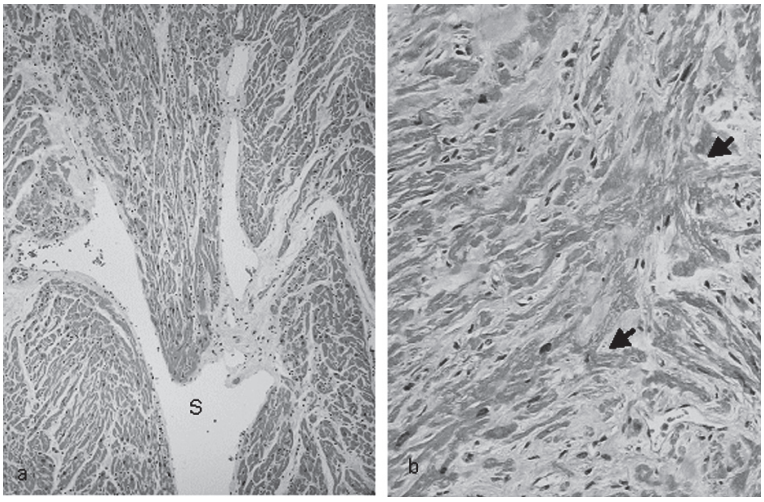

Figure 3 – shows the internal layers of the ventricle myocardium.
3a. VNC with sinusoids reaching deeply into the myocardium and communicating with the ventricle clearing on reduction of the normal vessel supply. HE x 25.
3b. Hypertrophic myocardium with hyperchromatic cells. Irregular, partly star-shaped myocyte arrangement (fiber disarray) [arrow]. HE x 50.

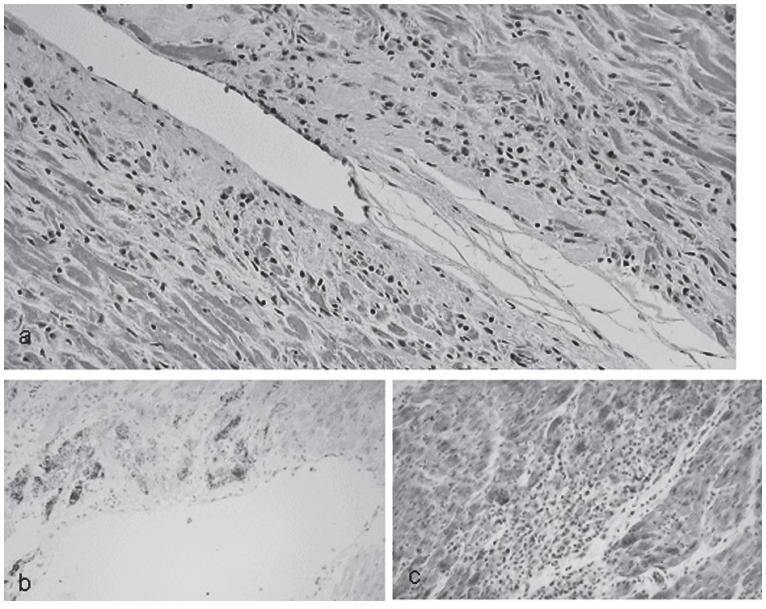

Figure 4 – shows the internal layers of the ventricle myocardium with small, oven-like necroses.
4a: Myocardial necroses with loose cellular clearing reaction. HE x 50.
4b: Brown colouration of necrotic myocardial cells; non-coloured vital myocardial cells in the right corner. Immunohistochemical on C9 x 50.
4c: Red colouration of fatty degenerated myocardial cells around a necrosis interspersed by absorptive cell infiltrates. Fat red 7 B x 50.