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An unexpected result of a routine cardiac consultation in a patient with nephrological problems

Zaskakujący wynik rutynowej konsultacji kardiologicznej u pacjenta z problemami nefrologicznymi

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Abstract

A 17.5-year-old boy, previously treated in a nephrology clinic due to chronic proteinuria, was referred for a routine cardiology consultation before being transferred to an adult clinic for further care. Physical examination and echocardiography showed no circulatory abnormalities and normal blood pressure, while echocardiography revealed an abnormal tumour-like structure measuring 1.3 × 1.5 cm in the left atrium, remaining in contact with the interatrial septum. Continuous infusion of heparin was started, after which no change in tumour size was obtained. The diagnosis was extended to include computed tomography, which showed a soft 1.5 × 2.1 × 2.1 cm tissue structure connecting with the interatrial septum with uneven contours, and magnetic resonance imaging, which indicated that the left atrial structure corresponded to myxoma, and the presence of enhancement spoke against the suspicion of a thrombus, although the presence of a small thrombus on the tumour could not be clearly excluded. The boy was qualified for a cardiac surgery, during which the pathological structure was removed and then sent for histopathological analysis, which revealed a heart tumour with myxoma. After the surgery, the patient was transferred to the department of paediatric cardiology for further treatment, where he received enoxaparin sodium, antibiotics and acetylsalicylic acid. After a few days, an about 1 cm layer of fluid appeared in the pericardium, which regressed after the incorporation of ibuprofen and dehydrating agents. After 2 weeks, the boy was discharged home in good condition, with a recommendation to continue care at a nephrology, cardiology and genetic clinic due to *MTHFR* mutation, which may be associated with hereditary hypercoagulability, detected during hospital stay.

Keywords: heart tumour, myxoma, thrombus, magnetic resonance imaging

Streszczenie

Chłopiec w wieku 17,5 roku, leczony dotychczas w poradni nefrologicznej z powodu przewlekłego białkomoczu, został skierowany na rutynową konsultację kardiologiczną przed przekazaniem go pod dalszą opiekę w poradni dla dorosłych. W badaniu przedmiotowym oraz elektrokardiograficznym nie stwierdzono nieprawidłowości w układzie sercowo-naczyniowym, ciśnienie tętnicze było prawidłowe, natomiast w badaniu echokardiograficznym w obrębie lewego przedsionka uwidoczniono nieprawidłową strukturę guzowatą o wymiarach 1,3 × 1,5 cm, pozostającą w łączności z przegrodą międzyprzedsionkową. Włączono podawanie heparyny w ciągłym wlewie, po której nie uzyskano zmiany wielkości guza. Diagnostykę poszerzono o tomografię komputerową, w której w lewym przedsionku uwidoczniono miękkotkankową strukturę o nierównych obrysach, o wymiarach 1,5 × 2,1 × 2,1 cm, będącą w łączności z przegrodą międzyprzedsionkową, a następnie wykonano badanie rezonansu magnetycznego, które wskazało, że struktura w lewym przedsionku w pierwszej kolejności odpowiada śluzakowi, obecność wzmocnienia przemawiała zaś przeciwko podejrzeniu skrzepliny, chociaż nie można było jednoznacznie wykluczyć obecności drobnej skrzepliny na guzie. Chłopiec został zakwalifikowany do operacji kardiochirurgicznej, podczas której usunięto nieprawidłową strukturę, a następnie przesłano ją do analizy histopatologicznej, w której stwierdzono nowotwór serca

o utkaniu śluzaka. Po operacji pacjenta przekazano na oddział kardiologii dziecięcej w celu dalszego leczenia, gdzie chory otrzymywał enoksaparynę sodową, antybiotyki oraz kwas acetylosalicylowy. Po 10 dniach w osierdziu pojawiła się około 1-centymetrowa warstwa płynu, który uległ regresji po włączeniu ibuprofenu i leków odwadniających. Chłopiec został wypisany do domu w stanie dobrym z zaleceniem kontynuacji opieki w poradniach nefrologicznej, kardiologicznej oraz genetycznej, ze względu na wykrytą podczas hospitalizacji mutację w genie *MTHFR*, co może się wiązać z wrodzoną nadkrzepliwością.

Słowa kluczowe: guz serca, śluzak, skrzeplina, rezonans magnetyczny

INTRODUCTION

Primary cardiac tumours are very rare in paediatric patients, with a prevalence of about 0.02% in autopsy cases[1,2], and 2.5/1,000 among children referred for cardiology consultations[3]. Since patients show no typical symptoms, and echocardiography is performed in children most often due to a heart murmur or arrhythmia rather than a suspected cardiac malignancy, pre-mortem diagnosis is usually accidental[1,3,4]. About 90% of primary paediatric cardiac tumours are benign, while the remaining 10% are malignant.

Benign rhabdomyoma (approximately 70%), which has an unusual feature of spontaneously regressing with age, is the most common primary benign tumour in this age group[4,5]. Other, rarely diagnosed, tumours include fibromas, teratomas, myxomas and haemangiomas (about 20% in total). Other primary tumours are malignant and are most often represented by sarcomas: angiosarcoma, fibrosarcoma and lymphosarcoma. In adults, approximately 75% of all cardiac neoplasms are primary benign tumours, with myxoma being the most common (40%), while the remaining 25% are malignant[6,7].

Myxoma is a single tumour; however, patients with Carney complex may develop multiple tumours. Although benign, it may have a malignant nature in some cases due to its location, especially if present in the left atrium and causing mechanical obstruction of blood flow through the valve, suggesting mitral stenosis[4,8,9]. Yin et al. found blood flow-obstructing stenoses in 60–70% of adult patients with myxoma, with chest pain and loss of consciousness being the main clinical symptoms[10]. Other significant complications of myxoma include thrombus formation and venous embolization with tumour fragments, which occur in approximately 21.5% of cases and may lead to strokes or pulmonary embolism[11]. Diagnosis is mainly based on echocardiography (ECG), which allows to assess the location, number and size of tumours, as well as their impact on haemodynamics. Usually, computed tomography (CT) and magnetic resonance imaging (MRI) imaging are also necessary, but the final verification of the tumour type is based on histopathological findings[12,13].

The treatment of cardiac tumours depends on many factors, secondary haemodynamic symptoms in particular. In the case of rhabdomyoma, a wait-and-see strategy is most often employed due to the tendency of these tumours to spontaneously regress. However, a growing tumour that obstructs blood flow is an indication for cardiac surgery[6]. Children with malignant tumours, which are extremely rare in this age group, have the worst prognosis[3,10,14].

CASE REPORT

A 17.5-year-old patient under nephrological surveillance due to chronic proteinuria and urolithiasis was referred for a routine cardiological consultation before being referred to an adult nephrology clinic. Clinical examination revealed pectus carinatum, pale skin, a soft systolic murmur over the heart, normal peripheral pulse and blood pressure.

Electrocardiography showed normal axis and electrical predominance of the left ventricle, while echocardiography showed a 1.3 × 1.5 cm tumour connecting to the atrial septum (Fig. 1). In real-time two-dimensional (2DE) projection, the tumour ballotted within the left atrium, but did not impede blood flow through the mitral valve. The inflow from the pulmonary veins to the left atrium also remained unobstructed. No arrhythmias were recorded, the left ventricular ejection fraction was normal (LVEF = 70%), and the heart walls did not show signs of hypertrophy. No pericardial fluid was detected, although the pericardial laminae had a slightly increased echogenicity. The presence of left atrial thrombus, vegetation or tumour was considered.

The most dangerous diagnosis was first considered, i.e. a thrombus, which could directly threaten the boy's health and even life (despite normal laboratory work-up, including basic coagulation parameters, normal D-dimers, C-protein and S-protein), because once mobilised, it could lead to acute stroke. For this reason, a continuous infusion of heparin monitored by activated partial thromboplastin time (APTT) was started, but did not reduce the tumour mass.

The diagnosis was extended by a CT scan, which revealed a 1.5 × 2.1 × 2.1 cm soft-tissue structure with irregular contours, connected to the atrial septum (Fig. 2). Additionally, a rare abnormality of the right coronary artery, which ran through the right atrium (without haemodynamic consequences), was found. Although the CT image indicated a thrombus or myxoma, an MRI was suggested and performed the following day. A mobile roundish structure measuring 1.3 × 1.3 × 1.6 cm, which did not protrude into the mitral valve, was found. The lesion showed intermediate signal intensity in the T1-weighted sequence, high signal intensity in the T2+FS sequence, and early peripheral

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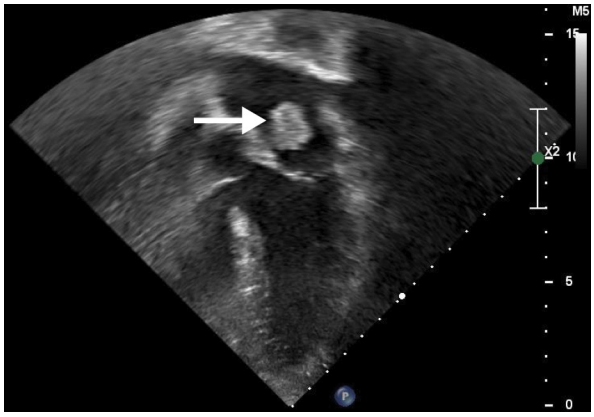

Fig. 1. Two-dimensional echocardiography (2DE), five-chamber projection. A 1.3 × 1.5 cm tumour is present in the left atrium, just above the mitral valve (arrow). It did not impede blood flow through the mitral valve during heart movement

enhancement after contrast injection. Late enhancement analysis showed a more uniform lesion enhancement with a 4-mm non-enhancing focus. The structure in the left atrium was first considered to be consistent with a myxoma. Although the suspicion of a thrombus was ruled out by enhancement, the presence of a small thrombus covering the tumour could not be unequivocally excluded. For this reason, the patient was qualified for surgical resection of the tumour under extracorporeal circulation. The resected 1.3 × 1.3 × 1.6 cm structure was sent for histopathological analysis, which confirmed myxoma.

The patient was postoperatively transferred to the department of paediatric cardiology for further treatment. On admission, the boy remained in a stable general condition, received enoxaparin sodium, antibiotics and acetylsalicylic acid. Skin paleness was noticeable, there was a faint systolic murmur over the heart, while laboratory tests showed a gradual normalisation of the previously elevated inflammatory markers and cardiac enzymes, as well as improved blood count. The results of genetic tests were also obtained, confirming the presence of *MTHFR* mutation, which may be associated with congenital hypercoagulability, and therefore a consultation at a genetic clinic was planned. A follow-up Holter ECG showed no arrhythmias or conduction disturbances, while echocardiography at 10 days postoperatively showed an approximately 1-cm layer of pericardial fluid, which regressed after inclusion of ibuprofen and dehydrating agents. Additionally, the boy periodically complained of chest pain, especially when changing body position, which was a typical symptom after cardiac surgery.

He also developed previously absent visual disturbances. For this reason, a neurological consultation was performed and no abnormalities were found. A very detailed medical history showed that the boy's mother suffered from migraine headaches and had demyelinating brain lesions, while his sister had recurrent epileptic seizures. The boy also admitted that he experienced visual disturbances with

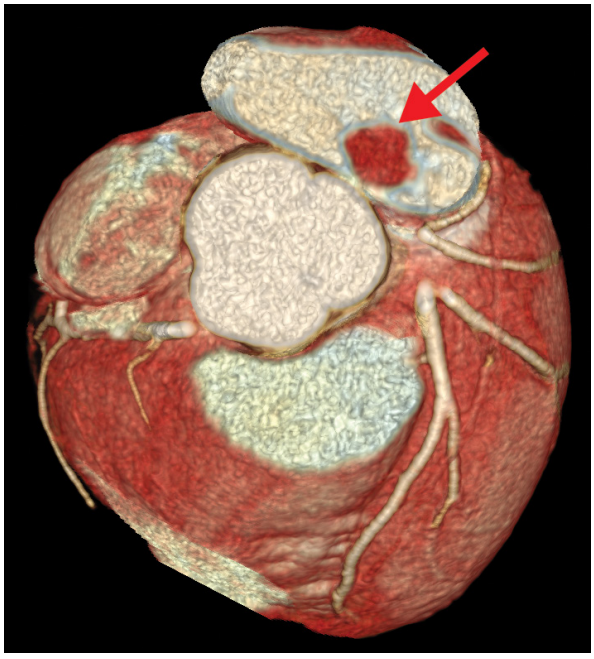

Fig. 2. Computed tomography of the heart. A 1.5 × 2.1 cm tumour measuring (arrow) in the left atrium, at the interatrial septum

reduced right-sided visual field a year earlier, during long-term work with a computer, which resolved spontaneously after rest. However, the consulting neurologist recommended further neurological and ophthalmological follow-up. After a few days, the boy was discharged home in good condition with recommendations to attend the arranged consultations.

DISCUSSION

Cardiac tumours are usually diagnosed in children accidentally, as they do not cause any specific symptoms, and the children are referred for cardiological consultations most often due to heart murmur or arrhythmia rather than a suspected heart malignancy[2]. They are mostly benign, do not metastasise, and some of them (e.g. rhabdomyoma) may even resolve spontaneously or grow very slowly. However, larger tumours may obstruct blood flow and therefore require surgical resection[10].

The described patient was diagnosed with left atrial myxoma. This tumour is diagnosed sporadically in children, unlike adults, in whom it is the most common[7]. A thrombus, cardiac tumour, or vegetation should be considered in the differential diagnosis of abnormal cardiac masses. Each of these structures may be a source of embolic material[9], with the thrombus characterised by the highest risk of mobilisation. Therefore, heparin was started in the described patient due to a suspected thrombus. Such management turned out to be reasonable, as the boy was later found to carry a *MTHFR* mutation, which promotes thrombus formation and, additionally, the MRI suggested that the myxoma was

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actually covered with thrombus. If the gene mutation had already been known at the time of first echocardiography, the initial suspicion of thrombus would be even more likely, and the use of heparin even more justified. However, the implemented heparin treatment failed to bring the expected improvement, therefore a heart tumour was contemplated, with suspicions directed at myxoma as the left atrium is the most common site for this tumour.

Although the tumour was large in size, it did not interfere with blood flow through the mitral orifice. Fainting or syncope incidents are always an alarming symptom in such patients as they may indicate mechanical blockage of blood flow inside the heart. Sudden deaths via the mechanism of low output syndrome in patients with myxoma have also been described. This was a very unexpected diagnosis, and if the boy had not been referred for a routine cardiological consultation in association with his nephrological problems before being transferred for further follow-up in an adult nephrology clinic, the cancer would have probably remained undetected for a long time. This could have dramatic, detrimental consequences for the boy's health and even life in the future. Although cardiac surgery was fully effective, myxomas may recur (not metastasize). The postoperative recurrence rate is estimated at around 3%; therefore further monitoring is needed after myxoma resection.

CONCLUSIONS

Primary cardiac tumours in children are mostly benign, diagnosed incidentally and generally do not require surgical treatment. Blood-flow obstructing tumours are an exception as they carry a high risk of sudden cardiac death. Echocardiography supported by CT and MRI is of primary importance in the diagnosis of cardiac tumours. Cardiac malignancies require differentiation from a thrombus and a vegetation.

Conflict of interest

The authors do not declare any financial or personal links to other persons or organisations that could adversely affect the content of this publication or claim rights thereto.

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