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A previously asymptomatic two month-old infant with unique presentation of cor triatriatum sinister, right partial anomalous pulmonary venous return, and critical supravalvular stenosis of mitral valve

Dotychczas bezobjawowe dwumiesięczne niemowlę z unikatową prezentacją trójprzedsionkowego lewego serca, prawego częściowego nieprawidłowego spływu żył płucnych oraz krytycznej nadzastawkowej stenozy mitralnej

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Abstract

Major developments in diagnostic techniques in pre- and neonatal care have significantly reduced the rate of undetected congenital heart defects (CHD). Despite such advances, several patients with critical congenital heart defects are discharged annually from neonatal units with no proper diagnosis or treatment.

We present the case of a two month-old originally asymptomatic girl who represents the perfect example of such a situation. The infant was diagnosed just after pulmonary and cardiovascular decompensation with critical, complex CHD. The absence of disease symptoms of occurred due to a rare and specific morphology of pulmonary vessels and intracardiac membranes.

Key words: congenital heart defect, cor triatriatum sinister, anomalous pulmonary venous return, mitral stenosis, supravalvular, echocardiography, diagnosis, intervention, surgery, screening

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Case report

The patient was born as a eutrophic neonate at the 38th week of gestation vaginally, reaching 10 in the Apgar score. The mother claimed it was her first pregnancy and she had received all recommended obstetrician ultrasonography (USG) examinations on time. The second trimester USG was repeated at the mother's request. However, no defects were described in any of the prenatal examinations. The girl had no symptoms of cardiovascular disease. There was a negative result from the patients with critical congenital heart defects (CCHD) protocol screening pulse oximetry test. The first signs of a cardiac problem appeared in the second month of life, a few days after a scheduled vaccination. Slight fatigue and lack of appetite were noted, however no signs of infection or a gastrointestinal system problem were observed. The cautious mother decided to take the infant to see their general practitioner. Medical examination revealed no murmurs or objective signs of heart disease. Eventually, before leaving the doctor's surgery, the child developed sudden peripheral cyanosis, consciousness disorders, and heavy dyspnoea. The patient was transported

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to the Mother and Child Institute, Warsaw where severe cardiopulmonary failure was diagnosed. First transthoracic echocardiography (ECHO) suggested anomalous pulmonary venous return and additional structures in the left atrium (LA). It was decided to airlift her to the Paediatric Cardiology and Cardiosurgery Unit in the Polish Mothers' Memorial Hospital Research Institute in Lodz. The girl was admitted in a critical condition. The next ECHO revealed an additional vertical structure in the left atrium imitating intraatrial septum and an additional horizontal structure critically obstructing mitral inflow, and left pulmonary venous return located physiologically. Moreover, right pulmonary veins were returning directly to the right atrium.

The patient was primarily diagnosed with cor triatriatum sinister (CTS) and partial anomalous pulmonary venous return (PAPVR). Due to the exacerbation of the patient's condition, angiographic intervention was decided upon, which confirmed pathological right pulmonary venous return and displayed enlarged right chambers. Measurements of pulmonary circulation pressures allowed a diagnosis of massive pressure overload of the right heart. Further examination revealed the left atrium without any pulmonary inflow. This revelation suggested an explanation of the rapid progression of cardiopulmonary failure to cardiac shock due to a unique presentation of pathologies acting similarly to total anomalous pulmonary venous return (TAPVR). A life-saving Rashkind-like procedure was performed on the additional structures of the left atrium, resulting in stabilisation of the patient's haemodynamic condition.

Shortly after, the patient underwent full surgical correction of pulmonary venous connections in conditions of extracorporeal circulation and deep hypothermia. The earlier diagnosis was finally confirmed intraoperatively as proper left pulmonary venous return was restricted by intraatrial structures blocking the mitral valve inflow. Additional membranes in the left atrium were resected with simultaneous restoration of the atrial septum with a pericardial patch. The overall condition of the patient soon after surgery was stable. Unfortunately, several post-operative complications occurred. The patient developed end-stage kidney failure, thus treatment with peritoneal dialysis was necessary as well as prolonged mechanical ventilation due to an inability to breathe unaided. Furthermore, various bacterial species were found in the blood tests, resulting in administration of a wide spectrum of antibiotics. A few days later, fungi were discovered in the patient's urine which necessitated antifungal therapy. Due to these multiple health problems, total hospitalisation time was significantly prolonged and lasted up to two months after surgery.

Nevertheless, the patient survived and conquered all her challenges. At discharge, the girl was in an excellent condition and had no signs of her previous illnesses. ECHO revealed normalisation of chamber sizes and an optimal haemodynamic result of the surgical intervention. The six-month follow-up proved cardiologic management of the defect to be working, as the patient was developing correctly without any disabilities (Figures 1–3).

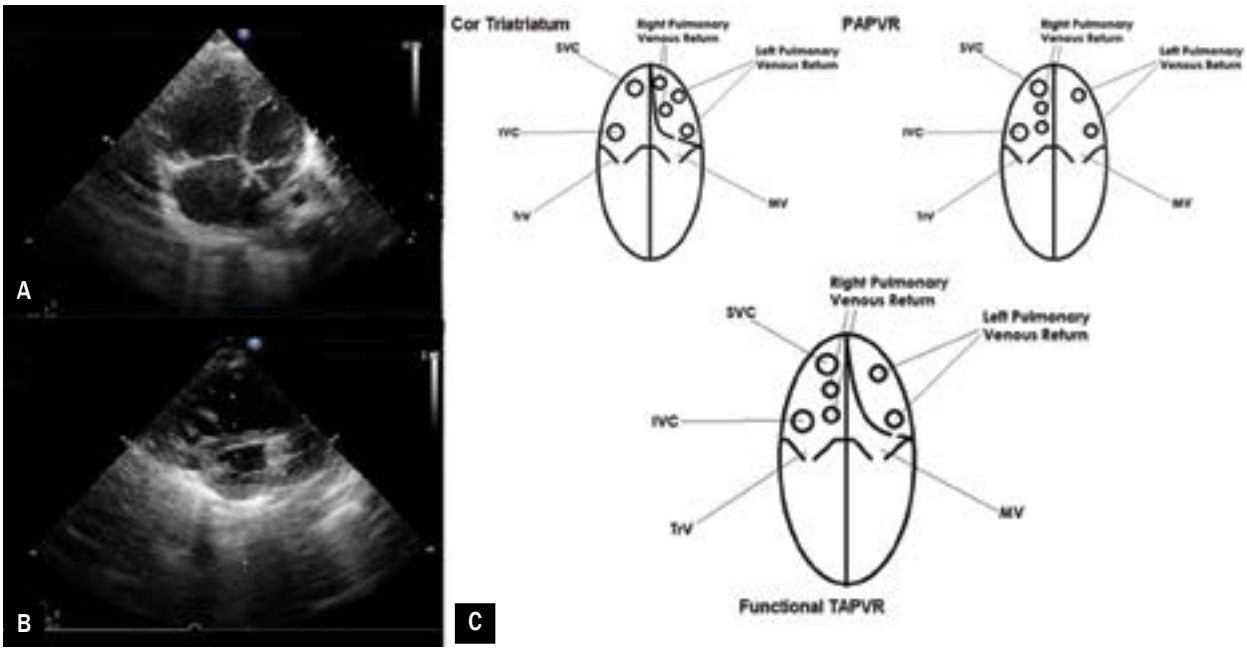


Figure 1. Preoperative state: **A**, **B.** Observable pressure and volume overload of right atrium and ventricle; **C.** Schematic presentation of concomitant defects

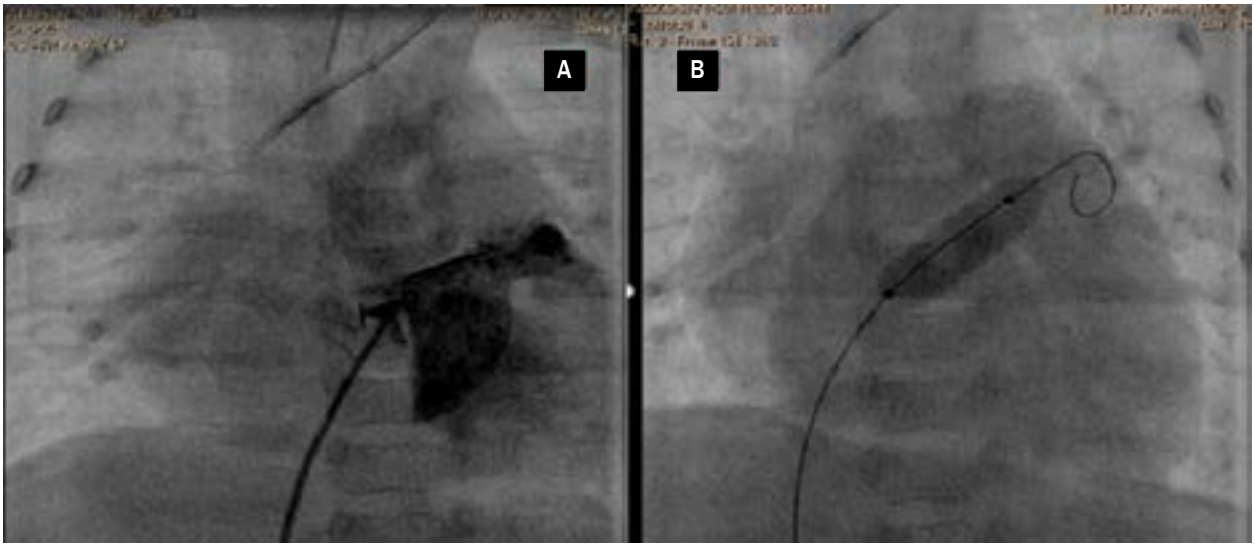
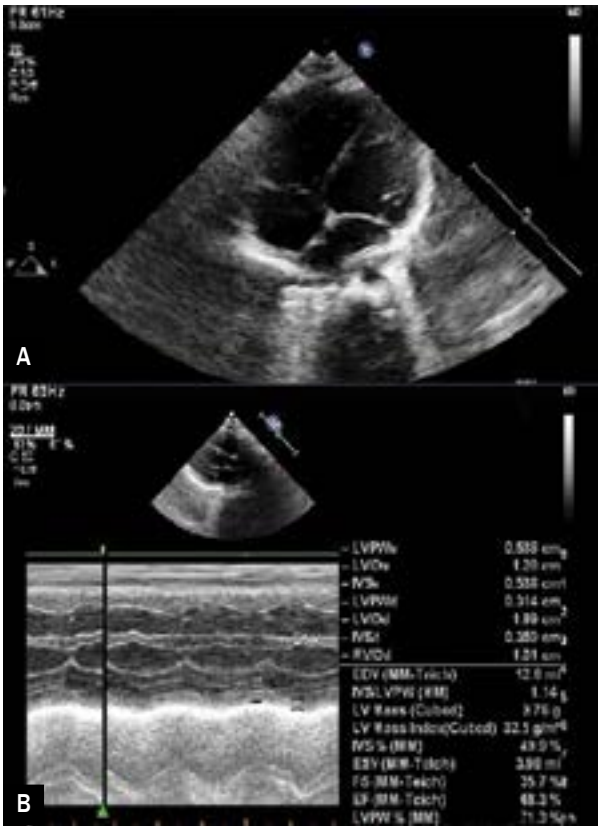


Figure 2. Catheterisation: **A.** Left atrium with anomalous pulmonary venous return and obstructed outflow due to membrane over mitral valve; **B.** Rashkind procedure

Discussion

The presented case describes a very rare situation wherein a coexistence of several factors led to the misdiagnosis of a severely ill child. Early detection of CCHD is crucial as any delay in diagnosis significantly affects morbidity and mortality. Such early detection is possible in the antenatal period and is now common due to the implementation of the pulse oximetry screening test [1]. The examination result is based on the observable decreased blood saturation level in neonates with CCHD. The test is highly sensitive and specific. However, several reports of undetected serious cardiac defects appear annually [2]. The main limitation of the method is the differing time of transformation of the foetal circulation into a post-natal one, especially when a unique heart morphology co-exists. If the pulse oximetry test is performed before shunt closure, it can give a false negative result. Such a situation probably occurred in the presented case. The patient screening test went negatively despite the life-threatening anomaly. The presentation of symptoms in such a situation is typically a matter of time. Prolonged shutting of physiological shunts and enlarging obstruction of inflow usually leads to haemodynamic decompensation in infancy. The sudden aggravation of this patient's condition appeared only after a rapid decrease in the amount of blood flowing into the left atrium, resulting in cardiac shock.

Nonetheless, standard post-natal pulse oximetry screening is not the only method to determine whether a child is in the high risk group for CHD existence. Obstetrician USG pre-natal screening is popular world-wide and provides moderately sensitive and specific data about the foetal heart and vessels [3]. The most common objection



Figures 3A, B. Post-operative state: observable normalisation of chamber sizes

made against this method is that the variation in skill levels among USG operators determines the diagnostic outcome of the examination. If performed incautiously, or by a person not trained in foetal heart visualisation, CHD can easily be

missed [4]. Even so, rare or not prominent CHD may be described as a variant of a normal developing heart. This is probably true of the presented patient because double second trimester USGs (by two different operators) revealed no red flag symptoms requiring referral to the prenatal cardiology unit.

The difficulty level of diagnosing the presented patient was also increased due to a very uncommon and specific concomitance of several heart pathologies which may not give any symptoms in the first months of life. The prevalence of cor triatriatum sinister is estimated to be only 0.4% of all CHDs. It is best described as left atrium division into two compartments by an additional septum — the first with pulmonary venous return and the second with mitral inflow [5]. The presented patient was also complicated by horizontal configuration of abnormal membranes in the left atrium with trans-membranous obstructed flow resulting in severe supravalvular mitral stenosis (MS). Such a situation has only been described in a few cases [6]. Partial anomalous pulmonary venous return is a condition defined as abnormal connection of one-sided pulmonary veins to the incorrect atrial chamber or vessels. It is found in approximately 0.4–0.7% of all CHDs [7]. The case described here is cardiac type right PAPVR. However, in the first two months of the girl's life, the small connection between the atrial chambers and the foramen ovale was probably persistent. Unfortunately, the coexistence of all anomalies (CTS, MS and PAPVR), as well as heart growth with restricting flows, resulted in a very rare clinical presentation imitating total anomalous pulmonary venous return that affected the patient's condition dramatically due to the critical limitation of inflow to the left ventricle and the massive overload of pulmonary circulation.

The essence of this case is a long chain of rare pathophysiologic events leading to a lack of diagnosis in the pre- and postnatal periods causing a life-threatening situation. It also shows the limitations of screening techniques, and thus emphasises the role of proper training for USG operators and the significance of a cautious neonatal examination based on experience and additional tests.

Streszczenie

Ogromny rozwój technik diagnostycznych w opiece przed- i okołoporodowej znacząco zmniejszył odsetek pacjentów z przeoczoną wrodzoną wadą serca. Mimo tego postępu, corocznie kilku pacjentów z krytyczną anomalią układu sercowo-naczyniowego jest wypisywanych bez odpowiedniej diagnozy i leczenia. Przedstawiony przypadek 2-miesięcznej, pierwotnie niewykazującej objawów dziewczynki jest doskonałym przykładem takiej sytuacji. Niemowlę zdiagnozowano tuż po ostrej dekompensacji krążeniowo-oddechowej. Początkowy brak objawów wiązał się z rzadką i nietypową morfologią współwystępujących anomalii żył płucnych i dodatkowych błon wewnątrzsercowych.

Słowa kluczowe: wrodzona wada serca, lewe serce trójprzedsionkowe, nieprawidłowy spływ żył płucnych, stenoza mitralna, nadzastawkowa, echokardiografia, diagnoza, interwencja, operacja, badanie przesiewowe

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Komentarz

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Autorzy przedstawili interesujący raport z leczenia pacjentki z rzadką wrodzoną wadą serca o przebiegu i obrazie klinicznym mogącym sugerować całkowity nieprawidłowy spływ żył płucnych [1]. Wady nie rozpoznano w badaniach prenatalnych ani we wczesnym okresie noworodkowym, a wykonywane sumiennie przesiewowe badania przedmiotowe oraz ocena czynnościowa noworodka wraz z testem pulsoksymetrycznym okazały się zawodne. Pełnoobjawowo wada ujawniła się u dziecka w okresie późniejszym, co ostatecznie doprowadziło do rozpoznania częściowego nieprawidłowego spływu prawych żył płucnych oraz nietypowej morfologii lewego serca trójprzedsionkowego, dającego objawy nadzastawkowej stenozy mitralnej [1].

Na uwagę zasługuje bardzo rzetelne przedstawienie problemów i pułapek diagnostycznych towarzyszących tej, mogącej powodować trudności, wrodzonej wady serca o asymptomatycznym wczesnym przebiegu u niemowlęcia. Nasilone objawy pod postacią napadowej duszności z sinicą wystąpiły dopiero w 2. miesiącu życia. Na podstawie wstępnej oceny w przezklatkowym badaniu echokardiograficznym (TTE, *transthoracic echocardiography*) rozpoznano nieprawidłowy spływ żył płucnych. Szczegółową ocenę dziecka z nasilającymi się objawami wstrząsu kardiogennego w badaniu angiograficznym uzupełniono o ratunkową procedurę balonowego poszerzenia dodatkowych struktur błoniastych w świetle lewego przedsionka (LA, *left atrium*), utrudniających napływ do zastawki mitralnej, co doraźnie poprawiło stan pacjentki. Ostatecznie dziewczynkę operowano w trybie pilnym; w trakcie zabiegu uwolniono nadzastawkowe błoniaste zwężenia w świetle LA oraz wszczepiono kierunkową łatę osierdziową tunelizującą spływ prawych żył płucnych do LA.

Podstawą diagnostyki większości wad wrodzonych, w tym także krytycznych wad serca i naczyń u noworodków i niemowląt, jest obecnie TTE [2]. W przypadku niejednoznacznych obrazów przezklatkowych kolejny krok to wykonanie przezprzełykowego badania echokardiograficznego (TEE, *transesophageal echocardiography*). Metodą niejednokrotnie rozstrzygającą o właściwym rozpoznaniu bywa diagnostyka inwazyjna — cewnikowanie serca z angiografią, a także tomografia komputerowa. Ostatecznej weryfikacji dokonuje kardiochirurg w śródoperacyjnej ocenie u pacjentów zakwalifikowanych do zabiegu.

W dostępnym piśmiennictwie opisy wad o podobnej manifestacji z dynamicznie narastającą niewydolnością krążeniowo-oddechową u niemowląt mają charakter kazuistyczny [2]. Diagnostyka i leczenie pacjenta z taką koegzystencją wad wymagają współpracy wielospecjalistycznej, z wykorzystaniem potencjału interwencji przezskórnych, oraz zaawansowanych metod intensywnej terapii, w tym wspomagania krążeniowo-oddechowego metodą ciągłego pozaustrojowego natleniania krwi (ECMO, *extracorporeal membrane oxygenation*). Szczególnie mylące w opinii wielu autorów bywają objawy przeciążenia i niewydolności prawej komory [3].

Pragniemy pogratulować Autorom dobrej pracy i zachęcamy do jej lektury, zważywszy na wysoką wartość edukacyjną tego opracowania. Zdecydowanie, zgodnie z opinią Autorów, podzielamy wątpliwości dotyczące przesiewowego testu pulsoksymetrycznego, którego wynik negatywny nie wyklucza zasadności wykonywania przesiewowych badań echokardiograficznych w diagnostyce krytycznych wrodzonych wad serca i naczyń u noworodków [4].

Piśmiennictwo

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