# **Case report**

# **Gerbode ventricular septal defect – a rare cardiac anomaly**

Smajlović E.¹, Smajlagić M.², Karahasan M.²

Pediatrics department, Cantonal hospital Zenica

**Background:** Left ventricular-right atrial shunts (LV-RA) were first described in 1958. by Gerbode et al. These shunts can be categorized anatomically into Type 1 (indirect or infravalvular) defects, which are a combination of a membranous interventricular septal defect and tricuspid septal leaflet perforation, and Type 2 (direct or supravalvular) defects which are a direct connection from the LV to RA through the superior part of the membranous interventricular septum. Congenital LV-RA shunt are rare, accounting for < 1% of all congenital heart defects, while aquired defects are incrisingly recognized after bacterial endocarditis, cardiac surgical procedures or myocardial infarction.

**Purpose:** Aim of our case presentation was to remind cardiologists about this rare heart defect which inexperienced eye could easily misdiagnose, and to point echocardiographic findings in these defects.

**Case presentation**: We report the case of 13 year old girl with previous diagnosis of a restrictive perimembranous VSD as a toddler. Physical examination was unremarkable except for a 4/6 Levine pansystolic murmur heard at the left sternal border. The presence of an aneurismatic perimembranous ventricular septal defect was initially confirmed, but a careful color Doppler flow mapping analysis showed high velocity flows in the right atrium. The short axis and four chamber views pointed to jet originating from the membranous part of ventricular septum above tricuspid septal leaflet into right atrium (jet velocity 3m/s). Another jet in right atrium was of the tricuspid regurgitation (jet velocity 2m/s). The echocardiogram detected no right atrium or ventricule enlargement.

**Discussion:** Transthoracic echocardiographic examination is the most frequently used diagnostic tool for identification of the defect although an actual communication is often extremely difficult to visualise. The large systolic pressure gradient between the left ventricle and the right atrium resulted in a high velocity systolic Doppler flow signal in right atrium. The diagnosis of this LV-RA shunt was quite challenging, especially in the context of coexisting abnormality as tricuspid regurgitation. Jet velocity of the shunt was different from that of the tricuspid regurgitation, with the shunt generating a significant higher jet velocity than tricuspid regurgitation. Increased pressure and blood volume within the right atrium and right ventricle could result in dilation of both chambers, which in our case was not established, but demanded yearly follow up.

**Conclusion:** This rare form of interventricular septal defect should be suspected during the performance of an echocardiogram, when there is interventricular septal defect with absent high velocity flow in right ventricule, present high velocity flow in right atrium, or unusually dilated right atrium.

**Key words:** Gerbode, ventricular septal defect

**Prikaz slučaja
Gerbode ventrikularni septalni sefekt – rijetka srčana anomalija**

Smajlović E.¹, Smajlagić M.², Karahasan M.²
Pedijatrijski odjel, Kantonalna bolnica Zenica

**Uvod:** Lijevi ventrikularno-desni atrijski šantovi (LV-DA) prvi put su opisani 1958. godine od strane autora Gerbode i saradnika. Ovi šantovi mogu se anatomski kategorizirati u defekte Tipa 1 (indirektne ili infravalvularne), koji su kombinacija membranskog interventrikularnog septalnog defekta i perforacije listića trikuspidalnog septuma, i defekte Tipa 2 (direktne ili supravalvularne) koji su direktna veza iz LV do RA kroz superiorni dio membranoznog interventrikularnog septuma. Kongenitalni LV-DA šantovi su rijetki, obzirom da se ubrajaju u < 1% svih urođenih srčanih defekata, dok stečeni defekti se sve više prepoznaju nakon bakterijskog endokarditisa, kardiohirurških procedura ili infarkta miokarda.

**Cilj** prikaza našeg slučaja bio je podsjetiti kardiologe na ovaj rijetki srčani defekt koji se u neiskusnom oku može lako pogrešno dijagnosticirati, kao i ukazati na ehokardiografske nalaz ovih defekata.

**Diskusija:** Transtorakalni ehokardiografski pregled je najčešće korišteni dijagnostički alat za identifikaciju defekta iako stvarnu komunikaciju je često iznimno teško vizualizirati. Veliki sistolni gradijent pritiska između lijevog ventrikula i desne pretkomore rezultirao je sistolnim Dopplerovim protokom signala velike brzine u desnom atriju. Dijagnoza ovog LV-RA šanta bila je prilično izazovna, posebno u kontekstu istovremene abnormalnosti kao što je trikuspidna regurgitacija. Brzina mlaza šanta bila je drugačija od trikuspidne regurgitacije, pri čemu je shunt generirao značajno veću brzinu mlaza od trikuspidne regurgitacije. Povećanje pritiska i volumena krvi unutar desnog atrija i desnog ventrikula moglo bi rezultirati dilatacijom obiju komora, što u našem slučaju nije utvrđeno,ali zahtjeva godišnje praćenje.

**Zaključak:** Na ovaj rijedak oblik defekta interventrikularnog septuma treba posumnjati tokom izvođenja ehokardiograma, kada postoji defekt interventrikularnog septuma s odsutnošću protoka velike brzine u desnom ventrikulu, prisutnim protokom velike brzine u desnom atriju ili neuobičajeno proširenim desnim atrijem.

**Ključne riječi:** Gerbode, defekt ventrikularnog septuma