Gerbode defect: a rare congenital condition in the context of right ventricular heart failure and pulmonary hypertension

Jasna Čerkez Habek[1,2]*,
Ines Zadro Kordić[1],
Ante Petrović[1],
Jozica Šikić[1,3]

[1]University Hospital "Sveti Duh", Zagreb, Croatia
[2]Croatian Catholic University School of Medicine, Zagreb, Croatia
[3]University of Zagreb, School of Medicine, Zagreb, Croatia

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***ADDRESS FOR CORRESPONDENCE:** Jasna Čerkez Habek, Klinička bolnica Sveti Duh, Sveti Duh 64, HR-10000 Zagreb, Croatia. / Phone: +385-91-3712-966 / E-mail: jasna.habek@gmail.com

ORCID: Jasna Čerkez Habek, https://orcid.org/0000-0003-3177-3797 • Ines Zadro Kordić, https://orcid.org/0000-0002-0754-7194
Ante Petrović, https://orcid.org/0000-0003-0896-8430 • Jozica Šikić, https://orcid.org/0000-0003-4488-0559

Introduction: The congenital Gerbode defect is a rare cardiac anomaly characterized by an abnormal left ventricle–to–right atrium communication, resulting in a left-to-right shunt. This may lead to right heart volume overload, pulmonary hypertension, and eventual right heart failure if unrecognized. Due to its nonspecific clinical presentation, it is often misdiagnosed as tricuspid regurgitation[1,2].

Case report: 43-year-old male with no significant prior medical history other than long-term tobacco use was admitted with signs of right-sided heart failure. A computed tomography pulmonary angiography demonstrated a segmental pulmonary embolism and radiological features of pulmonary emphysema. Transthoracic echocardiography showed dilated right heart chambers with reduced systolic function, moderately impaired left ventricular systolic function due to abnormal septal motion, severe tricuspid regurgitation, and a high-velocity systolic jet from the left ventricle to right atrium at the membranous septum. Transesophageal echocardiography supported the suspicion of a shunt and cardiac MRI confirmed a Gerbode defect (direct, type 1). Coronary angiography excluded atherosclerosis of the epicardial coronary arteries. Right heart catheterization showed pulmonary hypertension with a pulmonary vascular resistance of 8.9 Wood units. The Qp/Qs ratio was 1.1:1 measured invasively and with MRI. Pulmonary function tests revealed severe obstructive ventilatory defect and reduced diffusing capacity, indicative of advanced chronic obstructive pulmonary disease. Because of markedly elevated pulmonary vascular resistance and pulmonary hypertension, he was treated with optimized therapy for heart failure, pulmonary hypertension, pulmonary embolism, and lung disease. Depending on the effect of the therapy, either surgical closure of the defect or heart and lung transplantation will be considered.

Conclusion: Determining the etiology of right-sided heart failure with pulmonary hypertension and severe tricuspid regurgitation is often challenging. Accurate diagnosis relies on multimodal imaging, echocardiography, cardiac MRI, and invasive diagnostic procedures. Despite comprehensive diagnostic workup and established diagnoses, the case continues to pose a therapeutic challenge.

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LITERATURE

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