



Asymptomatic patient with Holmes heart and compensatory polycythaemia

Piotr Jagodowski^{1,C-D}, Łukasz Gawlik^{2,C-D}, Michał Spałek^{3,4,A-C,E-F}, Jan Spałek^{5,B-C},
Marcin Gregorczyk^{6,A-B}, Azita Rezaei^{7,A-B}, Łukasz Wypchło^{3,A,C,E-F}, Ewa Stochmal^{3,A,E}

¹ Southern Hostpital, Warsaw, Poland

² Ludwik Rydygier Specialist Hospital, Kraków, Poland

³ Collegium Medicum, Jan Kochanowski University, Kielce, Poland

⁴ Department of Diagnostic Imaging, Świętokrzyskie Cancer Centre, Kielce, Poland

⁵ John Paul II Independent Public Healthcare Centre, Kielce, Poland

⁶ Health Care Complex, Koneskie, Poland

⁷ University Clinical Hospital, Poznań, Poland

A – Research concept and design, B – Collection and/or assembly of data, C – Data analysis and interpretation, D – Writing the article, E – Critical revision of the article, F – Final approval of the article

Jagodowski P, Gawlik Ł, Spałek M, Spałek J, Gregorczyk M, Rezaei A, Wypchło Ł, Stochmal. Asymptomatic patient with Holmes heart and compensatory polycythaemia. J Pre-Clin Clin Res. doi: 10.26444/jpccr/186934

Abstract

The presented case report details a rare variation of a congenital heart anomaly known as double-inlet single left ventricle (DILV) – Holmes heart. Unlike other forms of DILV, the Holmes heart variant is distinguished by the absence of transposition of the great vessels. In this particular case, the diagnosis of DILV was made shortly after birth, but due to the severe nature of the condition, corrective surgery to address the defect was not performed. After a long time, the patient sought a follow-up examination with a cardiologist. Despite the absence of reported anginal symptoms during the visit, abnormalities were detected in the blood morphology, indicating deviations from the normal range. Specifically, the blood morphology exhibited erythrocytosis, and a high haematocrit level. Consequently, an MRI scan was recommended during the visit, which subsequently confirmed and provided a detailed description of the Holmes heart anomaly.

Ke words

double inlet single ventricle (DILV), congenital heart defect (CHD), functionally univentricular heart (FUH), magnetic resonance imaging (MRI), polycythemia.

INTRODUCTION

Double Inlet Left Ventricle (DILV) represents a relatively uncommon form of congenital heart defect (CHD) that falls within the broader category of univentricular hearts. The term ‘univentricular heart’ is a topic of ongoing discourse, and the presently accepted term is ‘functionally univentricular heart’ (FUH). The concept of FUH encompasses not only the morphological characteristics but also the functional aspects of the heart chambers. The designation ‘univentricular’ arises from the fact that, despite the presence of two ventricles, only one ventricle exhibits proper functionality and the capacity to effectively pump blood into the circulatory system. The other ventricle is typically underdeveloped or rudimentary, rendering it non-haemodynamically functional [1, 2, 3]. DILV constitutes approximately 1% of all CHDs and accounts for 78% of FUH cases, thus emerging as the most prevalent form within the FUH subgroup [3]. FUH also encompasses other defects [1, 4]:

1. single ventricle;
2. common ventricle;
3. univentricular atrio-ventricular (AV) connection;
4. hypoplastic left heart syndrom (HLHS);
5. tricuspid atresia
6. unbalanced AV septal defect;

7. mitral atresia with normal aortic root;
8. heterotaxy syndromes with one functionig ventricle.

DILV exhibits a diverse range of configurations, with classification based on distinct morphological features. Several variants have been identified:

1. Ventricular topology: refers to the development of the left-right axis in embryogenesis. In most cases (normal development), the looping of the cardiac tube around its own axis occurs to the right, and ultimately the apex of the heart is positioned in the pericardial sac on the left side [5, 6].

- a. Right-sided (D-loop) – the ventricles are in their normal positions (*situs solitus*), with the residual right ventricle located to the right of the left ventricle.
- b. Left-sided (L-loop) – the ventricles are a ‘mirror image’ (*situs inversus*), with the residual right ventricle located to the left of the left ventricle.
- c. Intermediate [2].

2. Atrial position:

- a. normal;
- b. right isomerism;
- c. left isomerism;
- d. mirror image.

3. Atrioventricular (AV) connections:

1. two patent valves;

✉ Address for correspondence: Piotr Jagodowski, Southern Hostpital, Warsaw, Poland
E-mail: piotr.jagodowski@gmail.com

Received: 07.03.2024; accepted: 08.04.2024; first published: 17.04.2024

- i. with a dominant left-to-right connection (RA → LV);
- ii. without a dominant connection;
- b. one common valve;
- c. left valve atresia;
- d. right valve atresia.

In order to classify a connection as a common atrioventricular valve, it is necessary for more than 75% of the blood to flow through that particular valve [3]. Instead of employing the terms 'mitral' and 'tricuspid' valves, it is more appropriate to refer to them as the left and right valves, respectively, as their morphological characteristics frequently exhibit an intermediate nature [2, 3]. To establish a connection as dominant through the atrioventricular valve, it is required for more than 50% of the blood to flow into the associated ventricle [4].

4. Ventriculoarterial (VA) connections:

- a. concordant;
- b. discordant;
- c. double outlet from left ventricle (DOLV);
- d. double outlet from right ventricle (DORV);
- e. single outlet from right ventricle with pulmonary atresia (SORV with pulmonary atresia).

5. Relationship and course of arterial trunks [3]:

- a. Type I – normal arrangement, known as Holmes heart.
- b. Type II – aorta anterior and to the right.
- c. Type III – aorta anterior and to the left;
- d. Type IV – 'inverted' vessels, with a posterior and left position.

CASE REPORT

A 46-year-old patient visited the cardiology outpatient clinic for a routine health check-up, presenting the results of venous blood morphology tests. The patient did not report any symptoms at the time of the visit. The morphological examination, however, revealed elevated haemoglobin levels of 26.5 g/dL (normal range for men: 13–18 g/dL), a haematocrit of 78.4% (normal range for men: 40%–50%), and an erythrocyte count of 7.84 million/cu (normal range for men: 4.2–5.4 million/cu). The patient currently experiences rapid fatigue, but does not exhibit other symptoms such as chest pain, shortness of breath, or arrhythmias. Physical examination did not reveal signs of pulmonary congestion or peripheral oedema; however, central and peripheral cyanosis, as well as finger clubbing, were observed. The patient was diagnosed with double inlet left ventricle (DILV) since birth. Over time, in addition to symptoms of circulatory insufficiency, a notable increasing trend in haemoglobin levels was observed in the patient's morphological examinations, ranging from 11.6 g/dL at birth to 22 g/dL at the age of 18. Due to the defect, the patient underwent three palliative operations involving a systemic-to-pulmonary shunt and cardiac catheterization. The patient was deemed unsuitable for corrective heart surgery due to anatomical constraints. After turning 18, the patient was not under the care of any specialized outpatient clinic.

During the visit, an electrocardiogram (ECG) and echocardiogram were conducted and described as follows: the ECG examination revealed a normal sinus rhythm with regular extrasystoles, a heart rate of 65/min, signs of right

atrial enlargement (p-pulmonale), and features of ventricular hypertrophy. The transthoracic echocardiogram showed an ejection fraction (EF) of approximately 45%, a residual right ventricle, and an existing ventricular septal defect (VSD) facilitating communication between the ventricles. The tricuspid valve was positioned between both ventricles and connected to the right atrium, and moderate tricuspid regurgitation was observed.

Figure 1 shows the cardiac magnetic resonance imaging (MRI) scan, which revealed the inflow of both atria into the left ventricle, a residual right ventricle, and a large defect in the ventricular septum with intact middle and apical portions. The majority of the blood flow was directed towards the aorta, with a small portion flowing through the ventricular septal defect into the right ventricle, and subsequently into the pulmonary trunk. The right ventricle exhibited a narrow lumen. No transposition of the great vessels was observed in the MRI, indicating that the patient has a Holmes heart, which is one of the rarest forms of DILV. Flow measurements were also obtained during the MRI (additional flow measurement results should be provided to complete the scientific description). The results were as follows:

Qs – systemic flow measured at the level of the ascending aorta, approximately 7715 ml/min.

Qp – pulmonary flow measured at the level of the pulmonary trunk, approximately 47 ml/min.

A Qp to Qs ratio >1:1, unequivocally indicating a right-to-left shunt. (Normal: Qp/Qs (1:1) [8])

DISCUSSION

Double Inlet Left Ventricle (DILV) is an infrequent congenital heart defect characterized by cyanosis. Its prevalence is estimated at approximately 5–10 cases per 100,000 live births [9]. Diagnosis of DILV typically occurs either at birth or within the first few days after delivery. Management of single-ventricle hearts entails a 3-stage surgical procedure known as the Fontan procedure. The initial stage involves the Norwood procedure, which establishes a connection between the single heart chamber and the systemic circulation. This is achieved by separating the pulmonary trunk from the pulmonary arteries and connecting it to the aorta. Blood flow to the lungs is provided through the Blalock-Taussig shunt, which involves connecting the subclavian artery to the pulmonary artery. The second stage is the Glenn operation, redirecting blood from the superior vena cava to the lungs, while the third stage is the Fontan operation, redirecting blood from the inferior vena cava to the lungs [10].

These surgical interventions enable survival rates of 70–80% in patients. Without surgery, the prognosis for these individuals is unfavourable, with an average survival of only 14 years [9]. It is important to note that not all anatomical defects are amenable to the afore-mentioned procedures. Additionally, complications, such as arrhythmias, heart failure, liver fibrosis, and even liver cirrhosis, may arise. While liver biopsy is currently considered the gold standard for assessment, elastography is the preferred method for evaluating liver condition in these patients [10].

Moreover, it is important to acknowledge that DILV often coexists with other congenital heart defects. Among the most frequently encountered concurrent anomalies are atrial

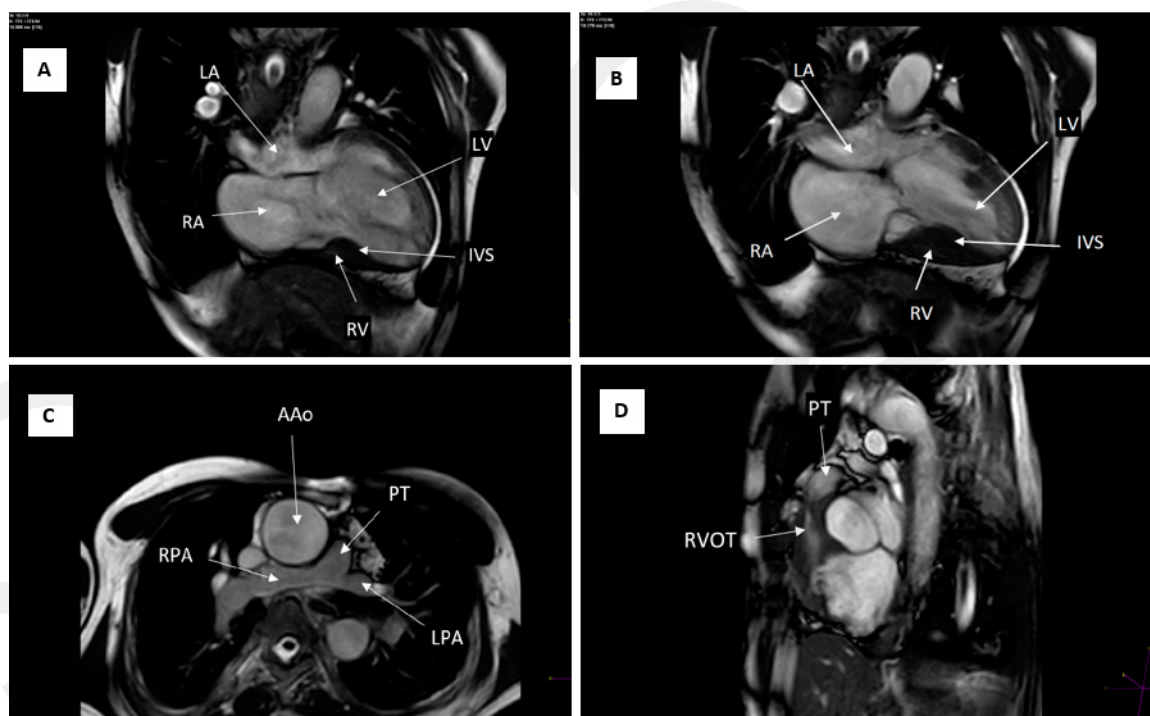


Figure 1. Holmes heart in cardiac magnetic resonance (CMR), steady-state free precession (SSFP) images: A) end-diastolic (ED) 4 chamber (4CH) view, B) end-systolic (ES) 4 chamber (4CH) view, C) *para-axial* oblique view, D) the right ventricular outflow tract (RVOT) view. LV – left ventricle, RV – right ventricle, LA – left atrium, RA – right atrium, IVS – interventricular septum, AAo – ascending aorta, PT – pulmonary trunk, RPA – right pulmonary artery, LPA – left pulmonary artery, RVOT – right ventricular outflow tract.

septal defect (ASD), patent ductus arteriosus (PDA), patent foramen ovale (PFO), and subaortic arterial cone [5]. Given this possibility, performing an MRI examination becomes crucial for accurately assessing the morphology of the defect, detecting any associated abnormalities, and guiding the selection of an appropriate treatment strategy. The utilization of MRI allows for a comprehensive evaluation of the heart's structure and aids in determining the optimal approach for management.

CONCLUSIONS

DILV is associated with heart failure and life-threatening arrhythmias that pose a risk of mortality to the affected individuals. Accurate determination and detailed description of the defect are crucial for appropriate management of the patient. According to the guidelines outlined by the American Heart Association (AHA) and Canadian Cardiovascular Society (CCA) in 2018, if there are no clinical indications for transthoracic echocardiography, magnetic resonance imaging (MRI) should be prioritized as the initial diagnostic investigation [11]. In the case presented, despite the absence of reported symptoms and haemodynamic stability, the patient exhibited secondary polycythaemia and elevated haematocrit levels in laboratory tests. These findings contribute to increased blood viscosity, impaired blood flow, and reduced tissue perfusion, thereby augmenting the risk of cardiovascular events such as stroke, embolism, or myocardial infarction [12].

These patients are not only at risk due to the inherent defect, but also due to the secondary consequences arising from compensatory mechanisms. Therefore, continuous cardiological monitoring and specialized care from facilities experienced in managing congenital heart defects are imperative for the well-being and optimal management of these patients.

REFERENCES

1. Rao PS. Single Ventricle-A Comprehensive Review. *Children*. 2021;8(6):441. <https://doi.org/10.3390/children8060441>
2. Kouchoukos NT, Blackstone EH, Hanley FL, Kirklin JK. Double Inlet Ventricle and Atrioventricular Valve. In: Kirklin/Barratt-Boyes, ed. *Cardiac Surgery*, Philadelphia; 2013. p. 2008–2033.
3. Khairy P, Poirier N, Mercier L-A. Univentricular heart. *Circulation*. 2007;115:800–812. <https://doi.org/10.1161/CIRCULATIONAHA.105.592378>
4. Earing MG, Hagler DJ, Edwards WD. Univentricular Atrioventricular Connection. *Thoracic Key Online* <https://thoracickey.com/univentricular-atrioventricular-connection/> (access: 2023.03.12)
5. Frescura C, Thiene G. The new concept of univentricular heart. *Frontiers Pediatrics*. 2014;2:62. *Frontiers Pediatrics Online*: <https://www.frontiersin.org/articles/10.3389/fped.2014.00062/full> (access: 2023.03.14)
6. *Fetal Development of the Heart and Great Vessels in Fetal Heart Defects – Diagnosis and Management*. pp. 10–12.
7. Rao PS. Double-Inlet Left Ventricle. *Children*. 2022;9:1274. <https://doi.org/10.3390/children9091274>
8. Sommer RJ, et al. Pathophysiology of congenital heart disease in the adult: part I: Shunt lesions. *Circulation*. 2008;117:1090–1099. *AHA Journals Online*: <https://www.ahajournals.org/doi/10.1161/CIRCULATIONAHA.107.714402> (access: 2023.03.12)
9. Abraham RR. Double Inlet Left Ventricle with Eisenmenger Syndrome in an Adult – A Case Report. *International Journal of Medical Students*. 2017;5,1:53–56. <https://doi.org/10.5195/ijms.2017.173>
10. Mareş R-C, Meliţ LE, Togănel R, Mareş R-G, Mărginean CO. Liver Disease Assessment in Children with Fontan and Glenn Surgeries for Univentricular Hearts-The Role of Elastography and Biochemical Fibrosis Markers. *Applied Sciences*. 2022;12(15):7481. <https://doi.org/10.3390/app12157481>
11. Stout KK, Daniels CJ, Aboulhosn JA, Bozkurt B, Broberg CS, et al. 2018 AHA/ACC Guideline for the Management of Adults With Congenital Heart Disease: Executive Summary: A Report of the American College of Cardiology/American Heart Association Task Force on Clinical Practice Guidelines. *Circulation*. 2019;139:698–800. *AHA Journals Online*: <https://www.ahajournals.org/doi/10.1161/CIR.0000000000000603> (access: 2023.03.15)
12. Rose SS, Shah AA, Hoover DR, Saidi P. Cyanotic congenital heart disease (CCHD) with symptomatic erythrocytosis. *Journal of General Internal Medicine*. 2007;22:1775–1777. <https://doi.org/10.1007/s11606-007-0356-4>