POLISH HEART JOURNAL

Kardiologia Polska
The Official Peer-reviewed Journal of the Polish Cardiac Society since 1957

Online first
This is a provisional PDF only. Copyedited and fully formatted version will be made available soon

ISSN 0022–9032 e-ISSN 1897–4279

Heart failure in young adult: Cardiomyopathy or much more?

Authors: Aleksander Misiewicz, Krzysztof Badura, Maciej Nadel, Jarosław Drożdż

Article type: Clinical vignette

Received: July 18, 2024

Accepted: September 3, 2024

Early publication date: September 5, 2024

Heart failure in young adult: Cardiomyopathy or much more?

Short title: HF in young adult: CM or much more?

Aleksander Misiewicz[1], Krzysztof Badura[1], Maciej Nadel[1], Jarosław Drożdż[1]

[1]2nd Department of Cardiology, Chair of Cardiology, Cardiac Surgery and Vascular Diseases, Medical University of Lodz, Łódź, Poland

Correspondence to:
Aleksander Misiewicz MD,
2nd Department of Cardiology,
Chair of Cardiology, Cardiac Surgery and Vascular Diseases,
Medical University of Lodz,
Central Clinical Hospital,
Pomorska 251, 90–213 Łódź, Poland,
phone: + 48 42 201 43 08,
e-mail: aleksander.misiewicz@umed.lodz.pl

A 35-years-old otherwise healthy male with weakness, dyspnea on exertion, peripheral oedema and palpations which had been present for over 3 months was admitted to the Department of Cardiology due to symptoms exacerbation. Patient denied family history of cardiovascular disease, substance abuse and infections prior to symptoms onset. Laboratory tests revealed increased concentrations of N-terminal pro-B-type natriuretic peptide (20652 pg/ml) and cardiac troponin concentrations suggesting myocardial injury.

Transthoracic echocardiography revealed severe left ventricular (LV) systolic dysfunction with reduced ejection fraction of 28%, generalized LV hypokinesis with better contractility of apical segments, grade II diastolic dysfunction, concentric LV hypertrophy with ground-glass appearance (Figure 1A–E). LV strain assessment revealed global longitudinal strain of -4.2% with apical sparing (Figure 1F). Moreover, right ventricular (RV) hypertrophy and RV systolic dysfunction were present. Other echocardiographic findings included: left and right atrium enlargement and increased interatrial septum thickness.

At this point the patient was fulfilling criteria to diagnose hypertrophic cardiomyopathy as wall thickness in every myocardial segment was above 15 mm, however echocardiographic features suggested cardiac amyloidosis (CA) as a specific etiology. Further diagnosis revealed severe proteinuria and increased serum concentration of lambda-type free light chains. Cardiac involvement in potentially light-chain (AL) amyloidosis was confirmed via cardiac magnetic resonance which showed diffused subendocardial late gadolinium enhancement. After introducing treatment for heart failure (HF) composed of loop diuretic, empagliflozin and eplerenone sufficient reduction in symptoms was not achieved, due to persistent compensatory sinus tachycardia and proteinuria, thus bisoprolol and ramipril have been added. Bone marrow biopsy confirmed IgG lambda-type multiple myeloma (MM) and AL amyloidosis, thus targeted treatment for MM consisting of daratumumab, bortezomib, cyclophosphamide and dexamethasone has been introduced. Taking into consideration patient's young age, extensive subendocardial fibrosis, improvement of LVEF over 30%, patient’s preference, but also thrombosis of right subclavian and brachiocephalic veins and no evidence of atrioventricular blocks we implanted subcutaneous cardioverter-defibrillator in primary prevention of sudden cardiac death. Despite MM treatment and progressive improvement during the course of 6 months, patient suffered from rapid deterioration of HF that led to cardiac arrest in a form of pulseless electrical activity, which resuscitation proved ineffective and patient was declared dead. There was no evidence of prior subcutaneous cardioverter-defibrillator interventions.

CA is a rare condition that occurs in less than 5 cases per 10000 individuals, mostly affecting patients older than 65 years [1]. AL amyloidosis is associated with plasma cell dyscrasias such as MM, whereas cardiac involvement occurs in approximately 70% of patients with its systemic type [2, 3]. Echocardiographic features such as apical sparring and ground-glass myocardial appearance accompanied by increased thickness of interatrial septum, atrioventricular valve leaflets and RV free wall may suggest CA, whereas young patient’s age, massive concentric LV hypertrophy and global hypokinesia are less typical [4]. In our case, due to positive tests for free light chains we decided to follow invasive diagnostic pathway composed of transthoracic echocardiography, cardiac magnetic resonance and extracardiac biopsy, which allowed us to diagnose AL type of CA. Moreover, recently increasing survival rates in MM emphasize the need for prompt bone marrow biopsy to diagnose and implement novel therapies [5]. It should be noted that beta-blockers and angiotensin-converting enzyme inhibitors/angiotensin receptor blockers are not recommended in CA-associated HF treatment due to the risk of hypotension, however in this case they were crucial to achieve reduction in symptoms [2].

Article information

Conflict of interest: None declared.

Funding: None.

REFERENCES

1. Aimo A, Merlo M, Porcari A, et al. Redefining the epidemiology of cardiac amyloidosis. A systematic review and meta-analysis of screening studies. Eur J Heart Fail. 2022; 24(12): 2342–2351, doi: 10.1002/ejhf.2532, indexed in Pubmed: 35509173.
2. Garcia-Pavia P, Rapezzi C, Adler Y, et al. Diagnosis and treatment of cardiac amyloidosis: A position statement of the ESC Working Group on Myocardial and Pericardial Diseases. Eur Heart J. 2021; 42(16): 1554–1568, doi: 10.1093/eurheartj/ehab072, indexed in Pubmed: 33825853.
3. Grzybowski J, Podolec P, Holcman K, et al. Diagnosis and treatment of transthyretin amyloidosis cardiomyopathy: A position statement of the Polish Cardiac Society. Kardiol Pol. 2023; 81(11): 1167–1185, doi: 10.33963/v.kp.97648, indexed in Pubmed: 37768101.
4. Arbelo E, Protonotarios A, Gimeno JR, et al. 2023 ESC Guidelines for the management of cardiomyopathies. Eur Heart J. 2023; 44(37): 3503–3626, doi: 10.1093/eurheartj/ehad194, indexed in Pubmed: 37622657.
5. Durie BGM, Hoering A, Abidi MH, et al. Bortezomib with lenalidomide and dexamethasone versus lenalidomide and dexamethasone alone in patients with newly diagnosed myeloma without intent for immediate autologous stem-cell transplant (SWOG S0777): A randomised, open-label, phase 3 trial. Lancet. 2017; 389(10068): 519–527, doi: 10.1016/S0140-6736(16)31594-X, indexed in Pubmed: 28017406.

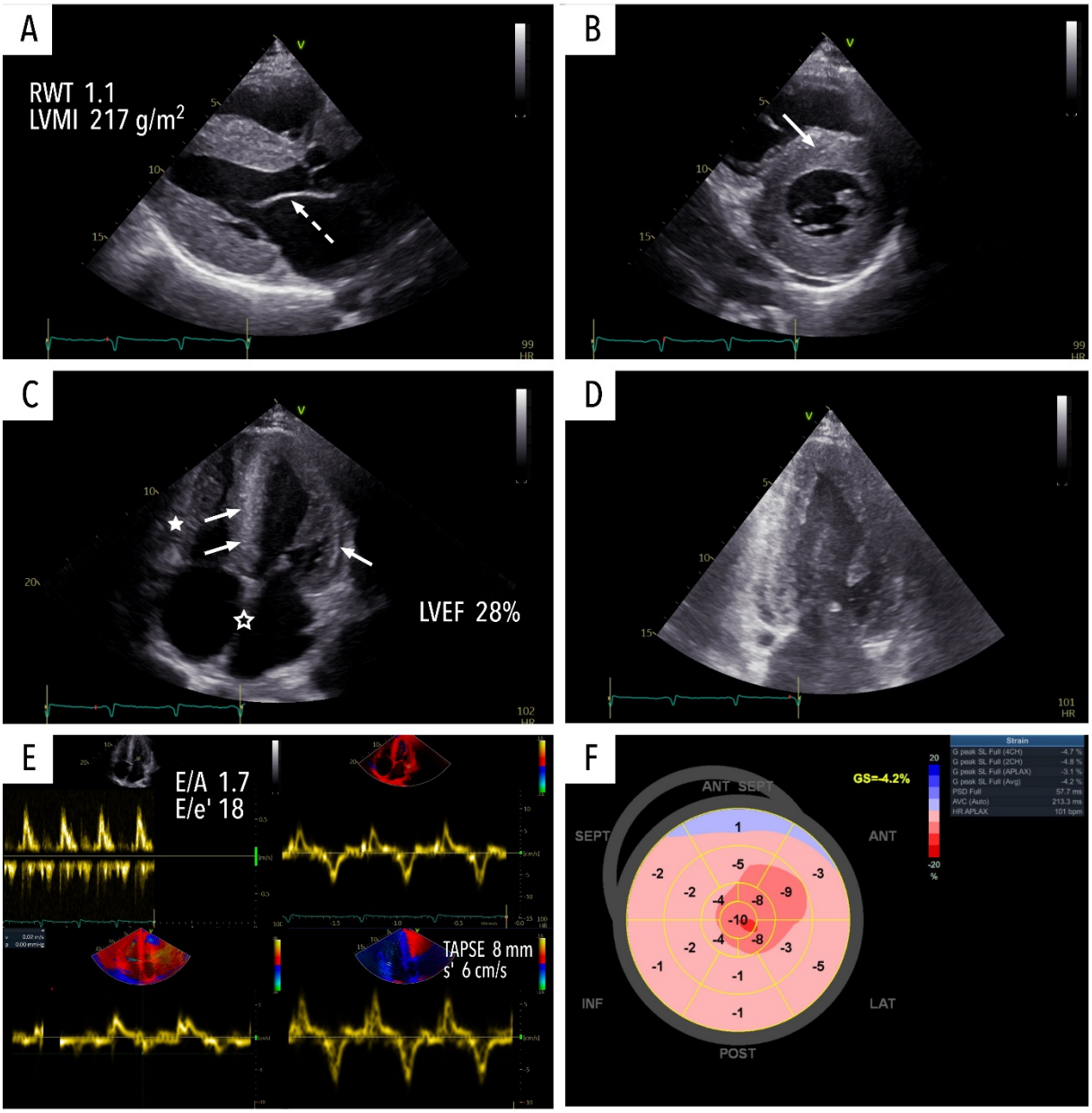

Figure 1. Findings in transthoracic echocardiography. Figures A-D showing increased thickness in all left ventricle segments, right ventricle hypertrophy (filled star), ground-glass myocardial wall appearance (simple arrow), left and right atrium enlargement. **A.** Parasternal long-axis view. Striped arrow indicates increased thickness of mitral valve. **B.** Parasternal short-axis view. **C.** Apical four-chamber view. A hollow star indicates increased thickness of interatrial septum. **D.** Apical two-chamber view with the focus on left ventricle. **E.** Pulse wave Doppler and tissue Doppler measurements showing 2. grade diastolic dysfunction and right ventricular systolic dysfunction. **F.** Bull's eye representation of global longitudinal strain with apical sparing

Abbreviations: A, late diastolic filling peak velocity of left ventricle; E, early diastolic filling peak velocity of left ventricle; e', mitral annular peak diastolic velocity; GS, global strain; LVEF, left ventricular ejection fraction; LVMI, left ventricular mass index; RWT, relative wall thickness; s', lateral tricuspid annular peak systolic velocity