SCIENTIFIC LETTER

Isolated Left Ventricular Apical Hypoplasia in an Asymptomatic 45-Year-old Woman

Hipoplasia aislada del ápex del ventrículo izquierdo en una mujer de 45 años asintomática

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Isolated left ventricular apical hypoplasia (ILVAH) is a rare congenital heart disease that was first described in 2004. (1) Approximately 40–50 cases have been reported to date. It is characterized by a spheroidal left ventricle (LV) with apical truncation, abnormal papillary muscles, and an elongated right ventricle that partially surrounds the left apex. (2,3). The congenital heart defect is usually isolated but may be associated with other congenital heart diseases in approximately 16% of cases. The clinical manifestations are variable. Children may present with serious symptoms, while adult patients may be asymptomatic or present with heart failure, arrhythmias, or even cardiac sudden death. (4) We present the case of an asymptomatic adult female patient with ILVAH and an electrocardiographic pattern suggestive of structural heart disease, which prompted the diagnostic workup.

A 45-year-old female patient sought medical care for preoperative risk assessment before an endoscopic procedure. She had no history of cardiovascular risk factors, and the physical examination was normal.

The 12-lead electrocardiogram (Figure 1) showed deep S waves in the right precordial leads (V1–V3), low QRS voltage in the left precordial leads (V4–V6), and no signs of hypertrophy or ischemia. Due to these findings, additional imaging tests were required.

The chest X-ray showed a cardiothoracic index of 0.42, a rounded LV apex, slightly elevated above the left diaphragm, with normal lung fields.

A transthoracic Doppler echocardiogram revealed a spheroidal LV with apical truncation and an elongated right ventricle surrounding the apex. The LV ejection fraction was 52%. Both atria were normal, and there were no abnormalities in the heart valves (Figure 2).

Cardiac magnetic resonance imaging (CMRI) was performed using a 1.5 Tesla Philips device with the Compressed SENSE protocol. The acquisition protocol included survey sequences, SSFP short-axis cine images, 2-, 3-, and 4-chamber views, native T1 mapping, T2-weighted sequences with the STIR technique, and late gadolinium enhancement (LGE).Image analysis was performed using Segment software.

The LV had a spheroidal and truncated morphology with hypoplasia of the left ventricular apex that was partially occupied by the right ventricle. Wall thickness was preserved. The LV ejection fraction was 49% (Simpson-CMRI). Right chambers dimensions and function were preserved. There was abundant supepicardial fatty material in the apex and absence of significant late enhancement or myocardial edema. Conclusion: findings consistent with isolated apical hypoplasia of the left ventricle (Figures 3 and 4).

A 24-hour Holter monitoring showed sinus rhythm and no arrythmias.

The exercise stress test was submaximal and negative for ischemia, and without arrhythmias.

Isolated LVAH is a rare congenital heart defect in children and adults. Although the etiopathogenesis is unclear, abnormalities in the ventricular partitioning process and hypoplasia of the apical trabecular component have been proposed. Associated mutations in the LMNA gene (p.Arg644Cys) and NEXN gene have been described, linked to related myocardial phenotypes. (5)

The clinical presentation is highly variable. A systematic review conducted in 2022 identified 37 patients, mostly pediatric or young adults, with ventricular dysfunction and frequent arrhythmias. (4)

In our case, the 45-year-old patient represents an

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Fig. 1. 12-lead electrocardiogram: sinus rhythm, normal QRS axis, poor R wave progression and low QRS voltage in V4-V6.

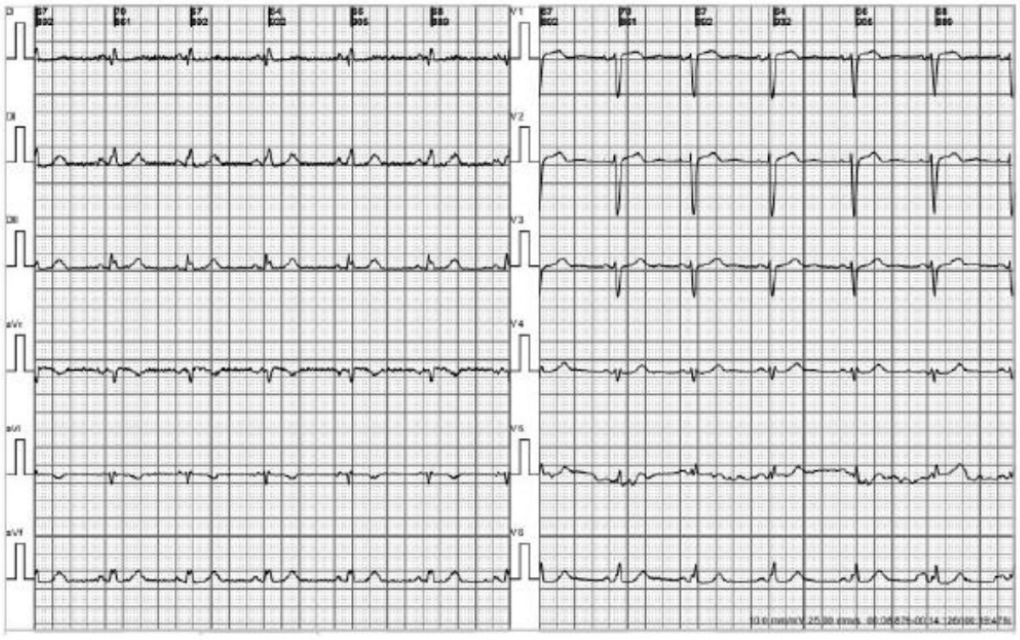


Fig. 2. Two-dimensional echocardiography, apical view

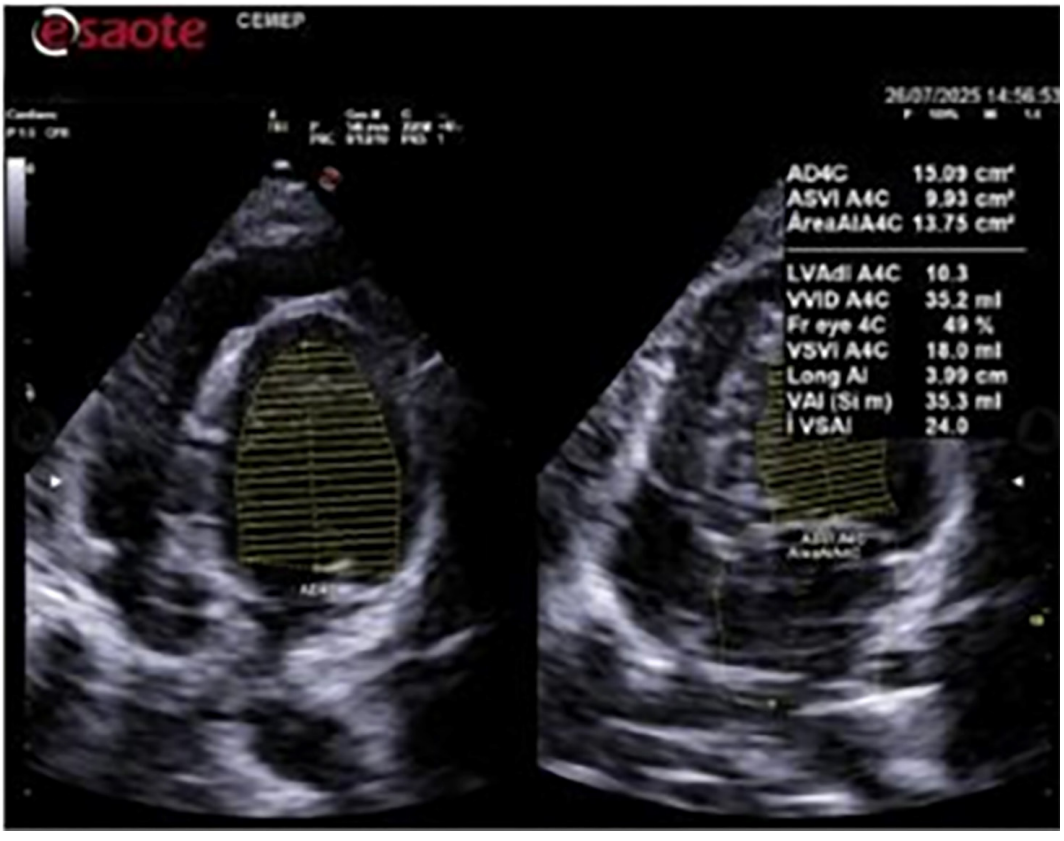


Fig. 3. Cardiac magnetic resonance imaging (4-chamber cine imaging): apical LV truncation with preserved contractility in basal and mid segments

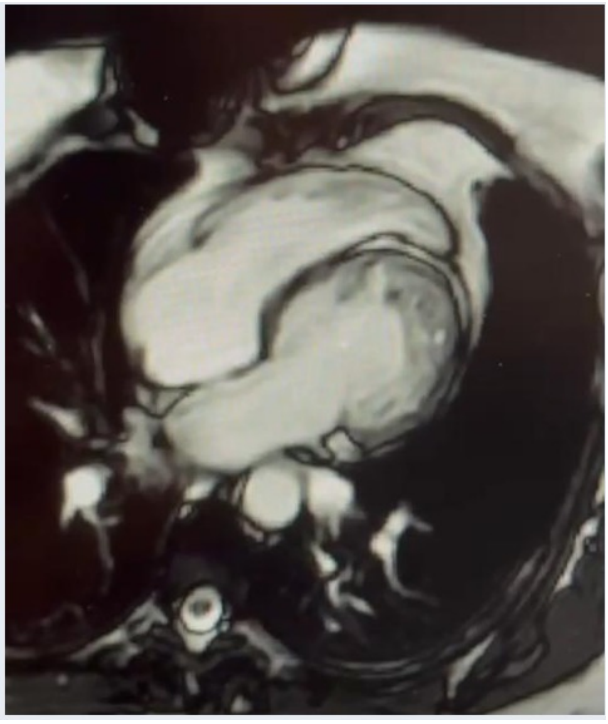
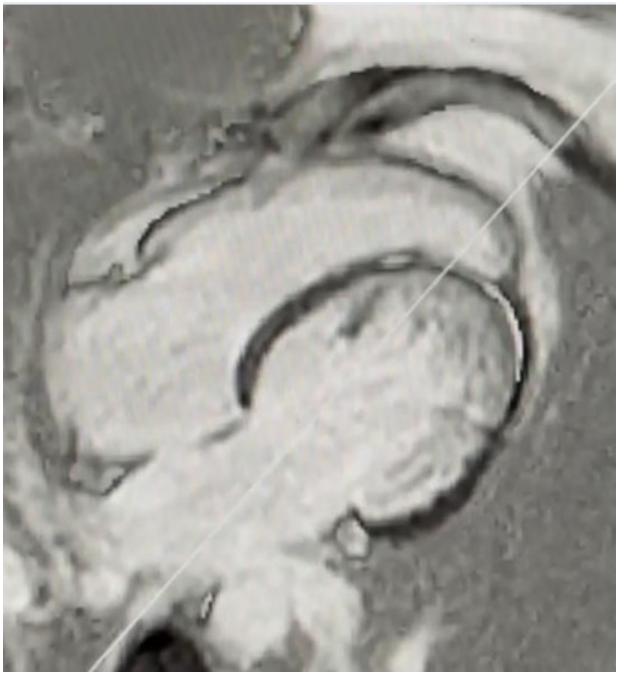


Fig. 4. Cardiac magnetic resonance imaging with absence of late gadolinium enhancement and myocardial fibrosis.



asymptomatic presentation in adults. Although there is no pathognomonic electrocardiographic pattern, the combination of deep S waves in the right precordial leads and low QRS voltage in the left precordial leads may prompt the search for structural abnormalities. Multimodality correlation between ECG, echocardiography, and cardiac magnetic resonance imaging is essential for establishing the diagnosis. This case report underscores the importance of integrating electrocardiographic findings with those of imaging tests for the identification of rare structural heart diseases.

Conflicts of interest

None declared.

(See conflicts of interest forms on the website).

Ethical considerations

Not applicable

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