Circulation: Cardiovascular Imaging

CARDIOVASCULAR IMAGES

Isolated Left Ventricular Apical Hypoplasia: A Very Rare Congenital Anomaly Characterized by Multimodality Imaging and Invasive Testing

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A 58-year-old man presented to a clinical cardiologist following an episode of bradycardia and lightheadedness. He was told as a child that he had an abnormally rotated heart but experienced no symptoms until current presentation. Ten years earlier, he was referred by his primary care physician for cardiac magnetic resonance (CMR) imaging, which was reported as normal. He has no family history of heart disease or sudden death.

On physical exam, he was normotensive with a regular heart rate of 45 beat per minute. The remainder of the exam was unremarkable. An ECG demonstrated sinus bradycardia, borderline right axis deviation, low QRS voltages, and a mild interventricular conduction delay (Figure S1). Basic laboratory testing was normal.

A 2-dimensional (2D) transthoracic echocardiogram (TTE) was performed (Figure 1; Videos S1 and S2). The left ventricle (LV) was dilated (LV end-diastolic diameter=6.5 cm) with an abnormal spherical shape and truncated apex. The LV ejection fraction was 45%. The right ventricle (RV) was elongated, wrapped-around the deficient LV apex, and had normal systolic function. The interventricular (IV) septum bulged rightward. There was mild mitral regurgitation with a dysplastic mitral valve (MV) apparatus including hypertrophied papillary muscles in the flattened LV apex. Other cardiac structures were unremarkable with a notably normal aortic valve and aortic root. In summary, TTE findings were diagnostic of isolated LV apical hypoplasia (ILVAH).

A transesophageal echocardiogram further characterized the MV apparatus as having asymmetric attachment of the chordae to an abnormally hypertrophied anterolateral papillary muscle and 2 attachments to the posteromedial papillary muscles, and demonstrated similar LV morphology (Figure 2; Videos S3 and S4).

Hemodynamic measurements during cardiac catheterization found low-normal filling pressures with a cardiac index of 2.6 L/min. Coronary angiography showed a small left anterior descending system and a hyperdominant left circumflex artery that continued as a large posterior descending artery, which wrapped-around the cardiac apex. Otherwise, the coronaries had mild luminal irregularities without significant epicardial disease (Figure S2; Videos S5 through S7).

Repeat CMR imaging confirmed the anatomic and functional findings of ILVAH as previously seen on echocardiography. Fatty infiltrate was noted in the LV apex. Late gadolinium enhancement imaging demonstrated a midwall pattern of myocardial fibrosis in the basal and mid-anteroseptal walls. There was also evidence of subendocardial myocardial infarction in the mid anterior wall involving 25% to 49% of the transmural thickness (Figure 3; Figure S3; Video S8).

The patient now follows in our institution's adult congenital heart disease clinic. He is planned for annual surveillance TTE and is doing well.

Isolated left ventricular apical hypoplasia is a rare congenital malformation, and appropriate identification is crucial. The disease was first described in 2004, and there have been scant reports ever since.[1,2] Salient anatomic feature of ILVAH include the following: (1) truncated, spherical, and dysfunctional LV with IV septum bulging into the RV; (2) elongated RV wrapping-around the deficient LV apex; (3) MV apparatus dysplasia with a complex papillary network in

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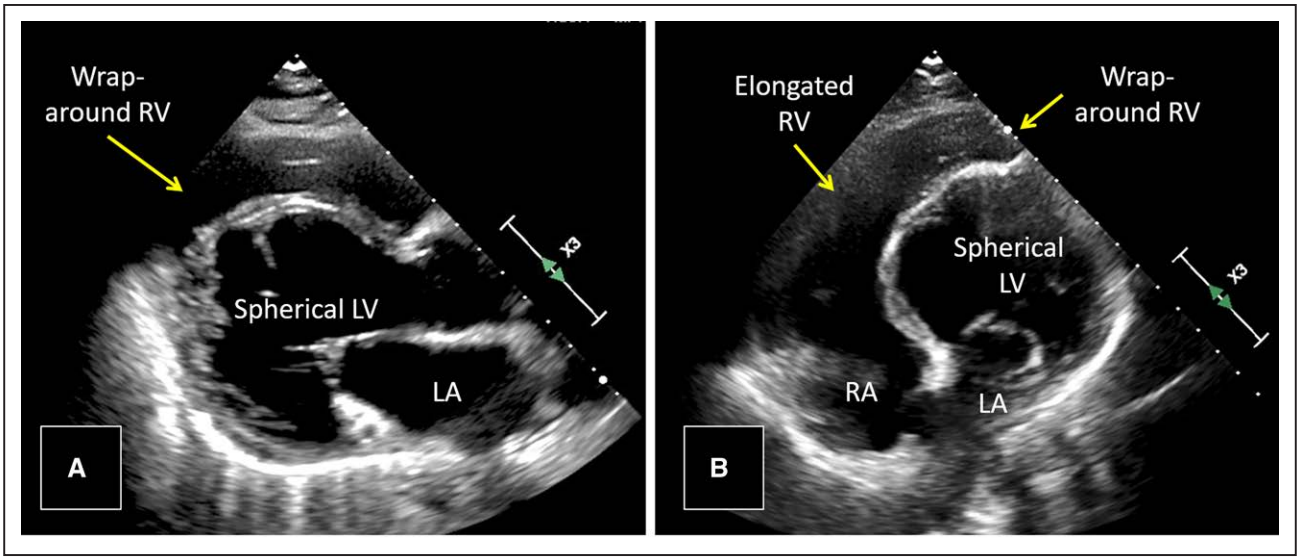

Figure 1. Transthoracic echocardiography characterization of isolated left ventricular (LV) apical hypoplasia.
Transthoracic echocardiography images from a parasternal long axis view (**A**) and an apical 4 chamber view (**B**) demonstrating a spherical LV as well as a wrap-around and elongated right ventricle (RV). This figure corresponds with Videos S1 and S2, respectively. LA indicates left atrium; and RA, right atrium.

the flattened anterior apex; and (4) fatty material infiltration near/within the deficient myocardium of the LV apex.[1,2] Patients present variably, with some completely asymptomatic and others in acute decompensated heart failure. In the presented case, ILVAH was first detected using 2D TTE after presenting with a bradyarrhythmia and ECG changes.

ILVAH is challenging to diagnose with imaging. Although our patient had a CMR done ten years prior, the diagnosis of ILVAH was missed until the characteristic anatomic findings associated with ILVAH were first fully visualized by 2D TTE. Even with 2D TTE, ILVAH can be overlooked given its rarity and unique morphology, which may be confused with endocardial dropout and/or apical foreshortening.[2] For this reason, CMR along with CT is the most commonly reported imaging modality when identifying the malformation, despite its own limitations in making an accurate diagnosis.[1,2] Cardiac imagers must be aware of the rare condition and utilize multiple modalities to identify ILVAH.

This case also raises questions regarding the exact underlying pathogenesis of ILVAH. The proposed theory includes erroneous LV and RV dilatation during partition, and an ongoing clinical trial is seeking to identify a genetic basis for the cardiomyopathy (REGISTRATION: URL: https://www.clinicaltrials.gov;

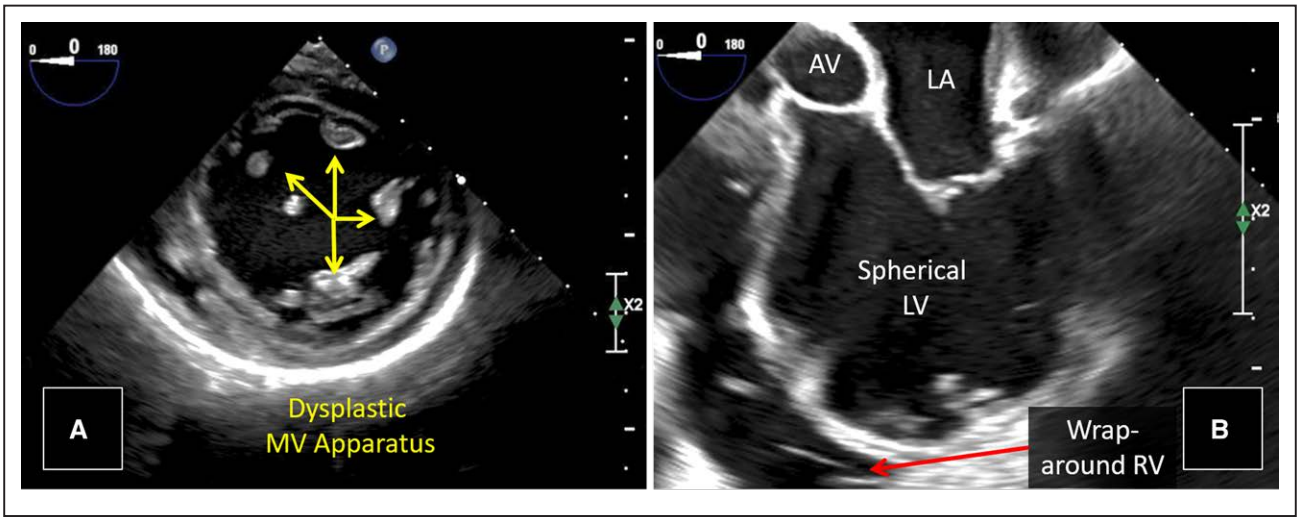

Figure 2. Transesophageal echocardiography characterization of isolated left ventricular (LV) apical hypoplasia.
Transesophageal echocardiography images. A transgastric short-axis view at papillary muscle level demonstrates a hypertrophied and dysplastic mitral valve (MV) apparatus (**A**). A midesophageal 5 chamber view shows a spherical LV with a wrap-around right ventricle (RV; **B**). This figure corresponds with Videos S3 and S4, respectively. AV indicates aortic valve; and LA, left atrium.

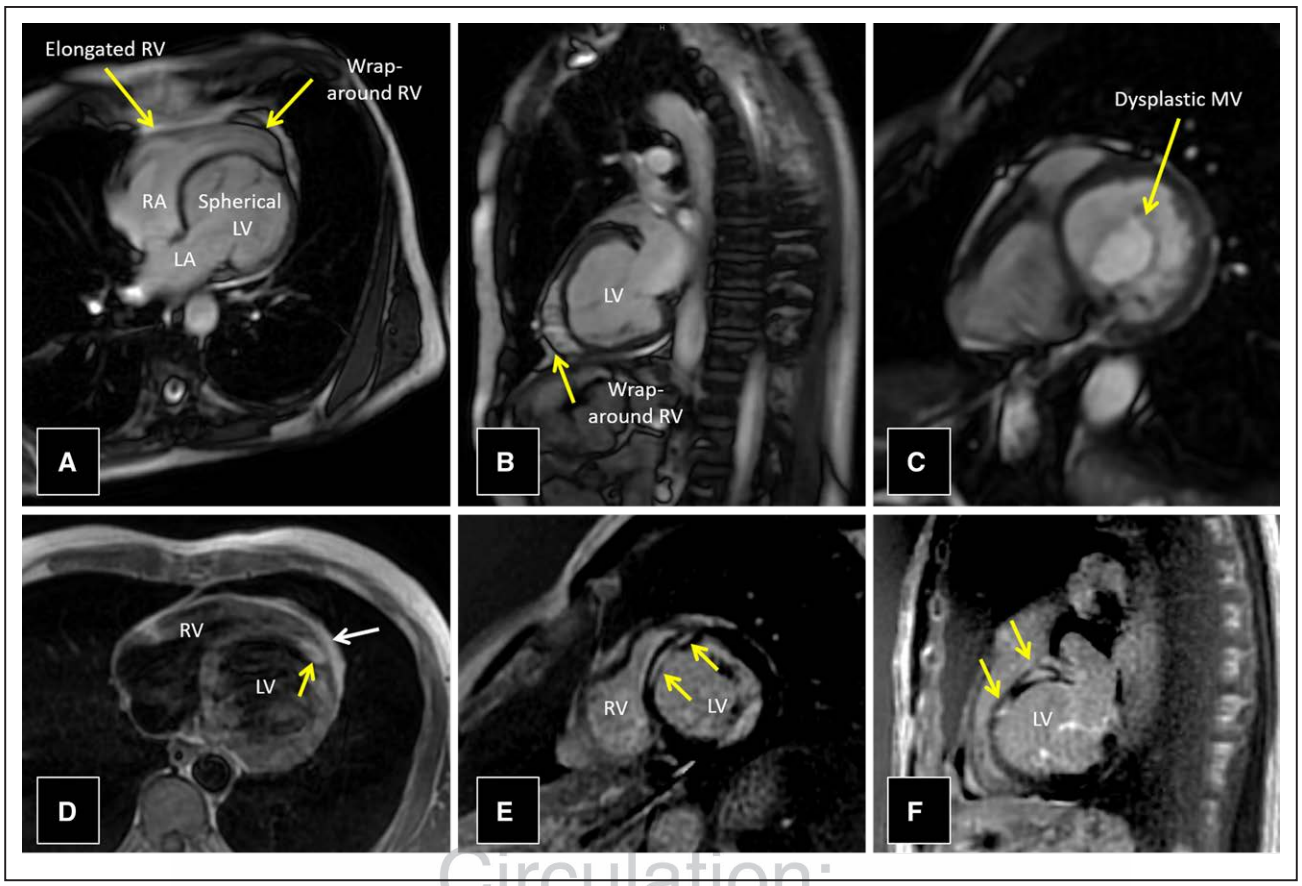

Figure 3. Cardiac magnetic resonance imaging characterization of isolated left ventricular (LV) apical hypoplasia. Cardiac magnetic resonance images. A T1-weighted transverse long axis 4-chamber view (**A**) and sagittal long axis 3-chamber view (**B**) highlight the spherical LV as well as a wrap-around and elongated right ventricle (RV). A T1-weighted transverse short axis 2 chamber view display a dysplastic mitral valve (MV) apparatus (**C**). A T2-weighted transverse short axis 2 chamber view demonstrates fatty infiltration within the myocardium (yellow arrow) as well as the pericardium (white arrow; **D**). Delayed contrast enhanced images show extensive subendocardial late gadolinium enhancement (LGE) (yellow arrows) in a midwall pattern of myocardial fibrosis in the mid-anteroseptal (**E**) and basal walls (**F**). This figure corresponds with Video S8. LA indicates left atrium; and RA, right atrium.

Unique identifier: NCT04339582) as it may be part of a spectrum of genetically linked congenital myocardial diseases, which in other patients manifests as hypertrophic cardiomyopathy or non-compaction, or inborn myofibril disarray.[1–3] Prior reports have also commented on a possibility of an acquired etiological component of the malformation.[4] One supporting clue for this etiology is myocardial scarring noted in reports of ILVAH. While original description of CMR findings in ILVAH did not have fibrosis on late gadolinium enhancement imaging, in the current era of higher quality CMR, more recent reports demonstrate fibrosis, signifying prior myocardial injury at an unknown timepoint.[1,4] In our patient, late gadolinium enhancement imaging revealed a midwall pattern of myocardial fibrosis in the basal- and mid-anteroseptal walls, and evidence of a subendocardial myocardial infarction in the mid-anterior wall. The differential diagnosis underlying these findings is broad, and they may be from an in-utero infection,[4] other teratogenic exposures, or dysfunction of the cardiac circulatory system during embryogenesis. Another possibility is that the late gadolinium enhancement findings are a consequence of the altered cardiac geometry, with areas of straining over time due to disruption of the helical structure of the heart resulting in scarring. Our patient's unique coronary anatomy further supports the possibility of circulatory compromise. Despite lacking significant epicardial disease, his atypical left hyper-dominant system with a posterior descending artery wrapping-around the cardiac apex suggests a relationship between coronary angiogenesis and ILVAH. However, it is impossible to discern whether his cardiac circulatory system resulted in ILVAH or if the malformation itself influenced an adaptation of the coronaries.

ILVAH is a rarely described malformation consisting of a truncated LV, an elongated RV, a dysmorphic MV, and apical fat infiltration. Given its paucity of cases, range of variability of clinical presentation, high possibility of underdiagnosis, and unknown cause, complete characterization of ILVAH is critical. The underlying mechanism of the condition remains unknown, and the results of

multimodality imaging presented in this report generate multiple hypotheses, including in-utero insults and dysfunctional angiogenesis.

ARTICLE INFORMATION

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None.

Supplemental Material

Figures S1–S3
Videos S1–S8

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