

## Isolated Left Ventricular Apical Hypoplasia: A Case Report and Literature Review

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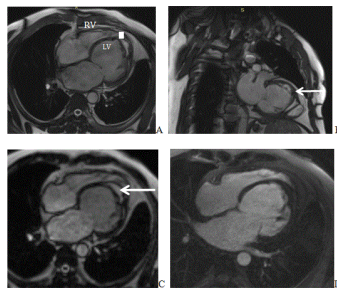
### Abstract

Isolated left ventricular apical hypoplasia (LVAH) is a newly recognized, rare cardiomyopathy which was first described in 2004. Until now, only 20 English-language literatures in relationship with Isolated LVAH were collected from PubMed by the author of this article. We report a case of 50-year-old man by health-examination without any symptom as this patient was first diagnosed in our hospital. This article includes two parts: case report and literature review.

**Keywords:** Isolated left ventricular apical hypoplasia; Echocardiography; Cardiomegaly

### Case Report

A 50-year-old man was examined by echocardiogram as a body-health examination which showed a ball-like enlarged left ventricle. He had no symptom and no past medical history. Thus he was suggested for cardiovascular magnetic resonance (CMR) after abnormal echocardiography as no reason was available to explain his asymptomatic cardiomegaly. CMR demonstrated a bizarre LV with spherical configuration, truncated apex, interventricular septum bulging to the right (Figure 1A), elongated RV wrapping around the deficient LV apex (Figure 1A), replacement of the LV apex with fatty material (Figure 1B and 1C).



**Figure 1:** A 50-year-old man with asymptomatic Isolated left ventricular apical hypoplasia. A: A bizarre LV with spherical configuration, truncated apex, interventricular septum bulging to the right; elongated RV wrapping around the deficient LV apex. B: Replacement of the LV apex with fatty material. C: Replacement of the LV apex with fatty material. D: Contrast imaging showed no thrombus and no late enhancement of these areas.

Contrast imaging showed no thrombus and no late enhancement of these areas (Figure 1D) The LV was shortened with apical fatty

replacement and the right ventricle wrapped around it. The CMR appearances were rare and not in accordance with dilated cardiomyopathy or any other common cardiomyopathy. The lack of scarring excludes several acquired conditions. Actually, the images are typical for the Isolated LVAH. He was the first patient for diagnosing this rare disease in our hospital. The patient left our hospital since no symptom could be found.

### Literature Review

Isolated left ventricular apical hypoplasia (LVAH) is a newly recognized, rare cardiomyopathy which was first described in 2004 in a case report by Fernandez-Valls et al. [1]. Until now, about 20 English-language literatures were reported. It was characterized by:

- a truncated and spherical LV configuration with rightward bulging of the interventricular septum,
- deficiency of the myocardium within the LV apex with adipose tissue infiltrating the apex,
- origin of the papillary muscle in the flattened anterior apex and
- elongation of the right ventricle wrapping around the deficient LV apex [2].

### Conclusion

This anomaly may occur as an isolated congenital anomaly or in conjunction with other congenital cardiac abnormalities, such as PDA [3]. The clinical presentation varies from asymptomatic [2-4] to lethal consequence [5]. Therefore, a close follow-up is needed even in asymptomatic patients. A systemic review of the literature was performed to identify all English-language case reports of Isolated LVAH. Eligible studies had to be consistent for patients' radiological findings in exception of their age, gender and so on. In this study 22 patients were reported in these literatures: **Age:** the minimum is 3-month-old [6], the maximum is 66-year-old [7], and the mean age is 29-year-old. **Gender:** Male: 9 [1,5,7-13]; Female: 11 [1,8-20]. **Not reported:** 2 [4]. **Initial Symptoms:** Atypical discomfort in the region of chest: 9 [2,4,8-15,17]; Arrhythmia: 2[9,18]; Murmur: 6 [6,10,12,14]; Dyspnea: 2 [9,13]; Heart failure: 3 [4,18,19]; Not reported: 1 [12].

**Prognosis:** Asymptomatic: 1 [4]; Atrial Fibrillation: 1 [4]; Medicine-controlled: 2 [15,19]; Die: 1 [5]; Not reported: 17 [3,8-14]. As to the etiology of Isolated LVAH, it remained unclear. However, one literature reported the mutation of the lamin A/C gene was associated with this rare disease [20], which would be beneficial for explore the etiology of this disease.

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