CASE REPORT

Isolated Left Ventricular Apical Hypoplasia with Right Ventricular Outflow Tract Obstruction: A Rare Combination

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Isolated left ventricular (LV) apical hypoplasia is a unusual and recently recognized congenital cardiac anomaly. A 19-year-old man was found to have an abnormal ECG and cardiac murmur identified during a routine health check since joining work. His ECG revealed normal sinus rhythm, right-axis deviation, poor R wave progression, and T wave abnormalities. On physical examination, a 2/6~3/6 systolic murmur was heard at the second intercostal space along the left sternal border. Subsequent echocardiography and cardiac magnetic resonance imaging confirmed the LV apical hypoplasia. Of note, we first found that LV apical hypoplasia was accompanied by RV outflow tract obstruction due to exaggerated rightward bulging of the basal-anterior septum during systole. A close follow-up was performed for the development of heart failure, pulmonary hypertension, and potentially tachyarrhythmia.

Ann Noninvasive Electrocardiol 2015;20(5):502–505

hypoplasia; right ventricular outflow tract obstruction, cardiomyopathy imaging

Isolated left ventricular (LV) apical hypoplasia is a recently described congenital cardiac anomaly. To date, there are only 22 previous cases reported.¹ Most reported individuals remained asymptomatic or mildly nonspecifically symptomatic with no other cardiac abnormalities.² We herein present a case of a 19-year-old male patient with isolated LV apical hypoplasia accompanied by RV outflow tract obstruction due to rightward bulging of the basal-anterior septum during systole. To our knowledge, this rare combination has not been reported in the English literature

CASE REPORT

A 19-year-old young man was found to have an abnormal 12-lead electrocardiogram (ECG) and cardiac murmur identified during a routine health check and then referred to our hospital for further medical evaluation in March 2013. He had experienced a normal, healthy childhood without any developmental concerns, and at the time

of evaluation, he was completely asymptomatic (no chest discomfort, fatigue, and shortness of breath, etc.) from a cardiac perspective. He had no history of congenital heart disease, and no family history of premature coronary artery disease, cardiomyopathy, or sudden cardiac death. His physical examination was normal except for a 2/6~3/6 systolic murmur at the second intercostal space along the left sternal border. All biochemical examination including myocardial enzymes and thyroid-stimulating hormone revealed no abnormalities.

Current ECG revealed normal sinus rhythm, right-axis deviation, poor R wave progression, and T wave abnormalities (Fig. 1). After admission, his 24-hour Holter monitoring showed a single premature atrial ectopy. A chest radiograph showed a spherically shaped heart and normal cardiothoracic ratio. Two-dimensional echocardiography revealed a truncated, spherical, and mildly dilated left ventricle (62 mm in diastole), with thin-walled (about 4.8 mm in apical segment

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Figure 1. Presenting ECG On admission showing sinus rhythm, right-axis deviation, poor R wave progression and precordial T wave abnormalities (T_{v1}(T_{v2}) > T_{v5}(T_{v6})).

Figure 2. Echocardiographic images of patient (A) Apical four-chamber view demonstrates spherically truncated LV with elongated RV wrapped around deficient LV apex. (B) Parasternal short-axis view taken during systole, showing the dyskinetic basal segment of anterior interventricular septum exaggeratedly bulged toward the RV outflow tract, which led to RV outflow tract obstruction with local internal diameter around 8.5 mm, pressure gradient 28 mmHg, and peak velocity 2.15 m/sec.

of interventricular septum during diastole) and akinetic apical portion. The LV papillary muscles abnormally originated from the deficient apex. The normally functioning, slightly dilated right ventricle (RV) formed the apex of the heart by means of an elongated segment, which wrapped around the LV. The overall LV systolic function was mildly depressed at 52%. Of note, besides the bulging of the interventricular septum toward RV during diastole, the dyskinetic basal segment of anterior interventricular septum exaggeratedly bulged toward the RV outflow tract during systole, which led to RV outflow tract obstruction with local internal diameter around 8.5mm, pressure

gradient 28 mmHg, and peak velocity 2.15 m/sec (Fig. 2).

He was referred for cardiovascular magnetic resonance imaging (MRI) after diagnostic coronary angiography revealed normal coronary arteries and left ventriculography showed truncated, spherical LV (Fig. 3). The MRI confirmed a spherical, modestly dilated LV (64 mm in diastole, instead of a normal conical LV) with apparent hypoplasia of the LV apex. The banana-shaped distal RV was again noted to wrap around the thin-walled, truncated, and akinetic distal LV. There was bulging of the interventricular septum toward the RV and anteroapical displacement of the papillary

Figure 3. Left ventriculography on the right anterior oblique 30° view showing a truncated, spherical LV during diastole.

Figure 4. Cardiac MRI findings of patient. Four-chamber cardiac MRI taken during diastole shows: a truncated, spherical LV with rightward bulging of interventricular septum and elongation of RV; papillary muscles originate from flattened anterior apex; and the high signal intensity in LV apex represents fatty infiltration (arrowhead).

muscle insertion points. There was an area of focal fatty infiltration noted in the LV apical wall that appeared to be contiguous with the epicardial fat (Fig. 4).

These echocardiographic and cardiac MRI anatomic features were consistent with the newly defined, rare type of cardiomyopathy, isolated LV apical hypoplasia.

Our patient was managed as a case of cardiac dysfunction and cardiomegaly. He was given

angiotensin converting enzyme (ACE) inhibitor and beta-blocker therapy. There was no recurrence of the tachyarrhythmia "atrial tachycardia, atrial fibrillation and ventricular tachycardia, ventricular fibrillation." during the hospital stay. He was stable with no evidence of arrhythmias in serial Holter monitoring and with no signs of heart failure during a 1-year follow-up, and remains on close clinical monitoring.

DISCUSSION

Isolated LV apical hypoplasia was first described in 2004 in a case report by Fernandez and Srichai.³ This unusual, newly recognized cardiomyopathy is characterized by: (1) a truncated and spherical LV configuration with rightward bulging of the interventricular septum; (2) deficiency of the myocardium within the LV apex with adipose tissue infiltrating the apex; (3) both mitral papillary muscles originating in the flattened anterior apex; and (4) elongation of the RV which wraps around the deficient LV apex.⁴⁻⁷

As of now there are only 22 previous cases (3 months to 63 years) with this entity.¹ The majority of reported cases of isolated LV apical hypoplasia were not accompanied by other cardiac abnormalities, with the exception of five cases among which the accompanying cardiac abnormalities were congenital patent duct arteriosus,^{4,8} complete transposition of the atrioventricular valves,⁹ and infundibular pulmonary and aortic stenosis². In one patient reported by Moon et al.,² RV outflow tract acceleration was caused by infundibular hypertrophy. However, our patient had RV outflow tract obstruction due to exaggerated bulging of the dyskinetic basal-anterior septum toward the RV during systole. The midwall fibrosis in the basal-anterior septum had been identified using late gadolinium enhancement MRI in one case report by Motwani et al.¹⁰ To our knowledge, this is the first case of LV apical hypoplasia accompanied by RV outflow tract obstruction due to rightward bulging of the basal interventricular septum during systole. The long-term outcome of this anatomical abnormality, i.e., whether it will cause RV dysfunction, needs to be further followed up.

The clinical presentation of isolated LV apical hypoplasia varies widely, with most individuals

remaining asymptomatic or mildly nonspecifically symptomatic (only with abnormal ECG or cardiac murmur),^{1,2,6,8} a minority presenting with congestive heart failure with systolic or diastolic dysfunction,⁵ pulmonary edema,¹¹ and pulmonary hypertension,⁴ or even malignant tachyarrhythmia.¹² According to review of the literature, the clinical manifestation of this entity may be associated with: (1) age of diagnosis. Patients diagnosed during adulthood usually had symptoms. However, patients diagnosed during childhood were usually asymptomatic, indicating the disease progression over time^{2,5}; (2) the severity of the combined cardiac malformations. Among five cases with the accompanying cardiac abnormalities, only one patient had no symptoms^{2,4,8,9}; and (3) the emergence of atrial tachyarrhythmia. The hemodynamics of isolated LV apical hypoplasia is similar to restrictive LV cardiomyopathy. Because of altered LV geometry, such patients are highly dependent on atrial contractile function. Cardiac decompensation may occur due to the emergence of atrial tachyarrhythmia, such as atrial fibrillation and atrial flutter.^{5,7,10,11} Although our patient presented with normal RV function and low normal LV function, given that LV and RV were mildly dilated, he was treated with standard therapy for a nonischemic dilated cardiomyopathy and was initiated on ACE-inhibitor and beta-blocker therapy, with a plan for close follow-up. Almost all of the reported patients responded well (the longest follow-up period up to 9 years⁸) to the standard heart failure treatment with the exception of one, who died of ventricular fibrillation and subsequent hemodynamic deterioration.¹²

The pathogenesis of LV apical hypoplasia is unknown; however, the proposed mechanism is an abnormal ventricular septation during fetal cardiac development. Defective LV apical development may be due to relatively inadequate LV to RV dilatation during ventricular partitioning in the fifth week, resulting in a spherical LV with elongated RV wrapping.³ Motwani et al.¹⁰ proposed the possibility of an acquired etiology such as in utero infection by a novel finding of midwall fibrosis in the basal-anterior septum.

CONCLUSIONS

Isolated LV apical hypoplasia is a recently recognized congenital cardiac anomaly that has a characteristic appearance on echocardiography and MRI. We herein presented a patient with RV outflow tract obstruction due to rightward bulging of the basal-anterior septum during systole. Our patient is currently on a regular follow-up for the development of heart failure, pulmonary hypertension, and potentially tachyarrhythmia. As more cases are recognized and patients are followed over time, the natural history and optimal treatment for this cardiomyopathy may be further elucidated.

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