

CASE REPORT

Congenital Absence of the Left Atrial Appendage Visualized by 3D Echocardiography in Two Adult Patients

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Congenital absence of left atrial appendage (LAA) is an extremely rare condition and its physiological consequences are unknown. We present two cases of incidental finding of a congenitally absent LAA in a 79-year-old male who presented for routine transesophageal echocardiogram (TEE) to rule out intracardiac thrombus prior to placement of biventricular implantable cardioverter-defibrillator and a 54-year old female who presented for TEE prior to radiofrequency ablation of atrial fibrillation. Characterization of patients with such an absence is important because congenitally absent LAA may be confused with flush thrombotic occlusion of the appendage. There are very few published reports of congenital absence of LAA. To our knowledge, our report is the first to demonstrate the congenital absence of LAA by 3D transesophageal echocardiography. (Echocardiography 2015;00:1–5)

Key words: left atrial appendage, congenital absence, 3D transesophageal echocardiography

Case report:

Case 1:

A 79-year old man with a history of hypertension, chronic atrial fibrillation (AF) (on long-term anticoagulation), severe nonischemic cardiomyopathy and worsening heart failure presented for a transesophageal echocardiogram (TEE) to rule out intracardiac thrombus prior to biventricular implantable cardioverter-defibrillator (ICD) insertion. He had no prior surgical or percutaneous procedures to close or exclude the left atrial appendage (LAA).

His prior workup included electrocardiogram (EKG), coronary angiogram, and transthoracic echocardiogram (TTE). EKG showed AF and a left bundle branch block with a QRS duration of 150 msec. Cardiac catheterization demonstrated mild nonobstructive coronary artery disease. TTE revealed severe global left ventricle systolic dysfunction (ejection fraction of $\leq 30\%$) and moderate mitral regurgitation.

Despite optimal medical therapy over the preceding 3 months, he continued to be symptomatic (New York Heart Association symptom class III).

Two-dimensional (2D) and three-dimensional (3D) TEE was performed using an X7-2 matrix probe and iE33 ultrasound system (Philips Medical Systems, Andover, MA, USA). 3DTEE images were then displayed in a custom layout used in our laboratory which differs from the layout recommended by the European Association of Echocardiography and the American Society of Echocardiography.¹

Spontaneous echo contrast (“smoke”) in the left atrium (LA) was present. There was no evidence for intracardiac thrombus in the atria or the ventricles.

Despite 2DTEE imaging at multiple acquisition angles, the LAA could not be visualized (Fig. 1 and movie clips S1, S2, S3, and S4). On spectral and color Doppler imaging, no tracings characteristic of LAA flow could be demonstrated. On 3DTEE imaging, en face views of the anatomic area where the LAA orifice is expected, only a small shallow pit was noted (Fig. 2 and movie clips S5 and S6). Cropping through 3D datasets revealed no body of the LAA. Overall, TEE findings were consistent with a congenitally absent LAA.

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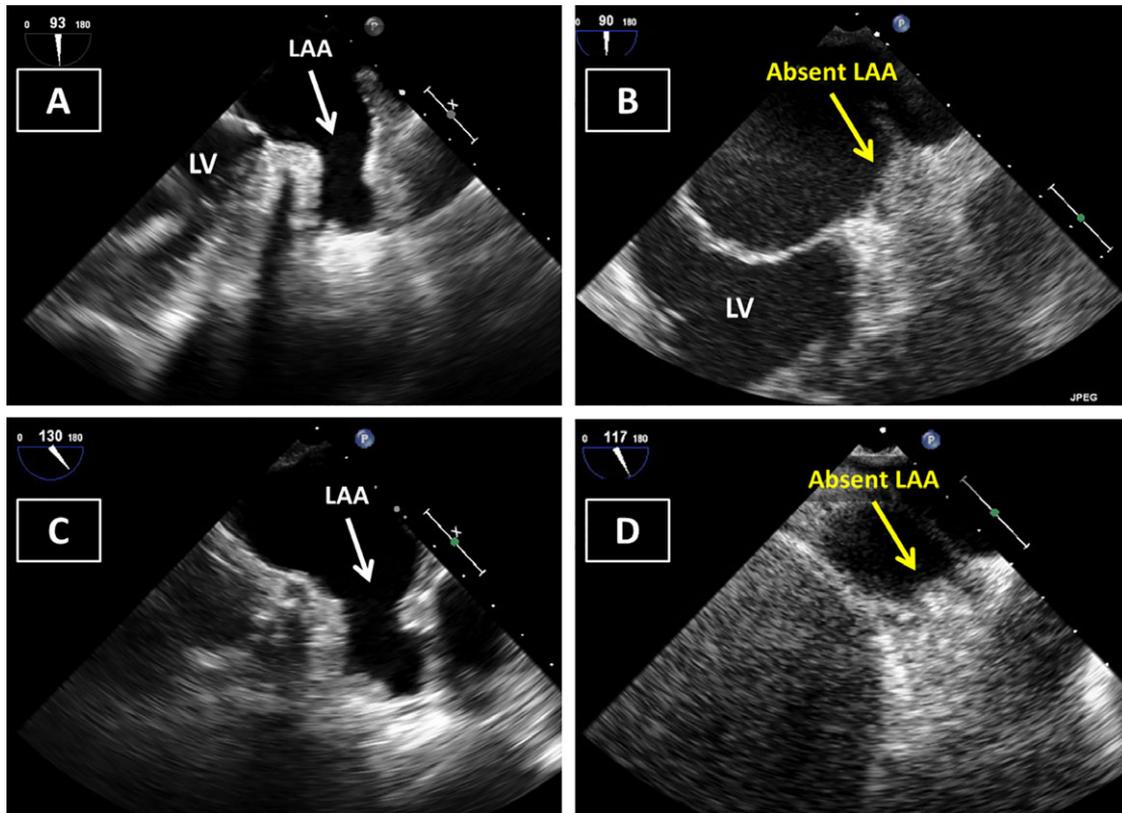


Figure 1. Absent versus present LAA on 2DTEE in case 1. **B, D.** Absent LAA on 2DTEE using typical acquisition angles of 90 and 117 degrees, respectively. **A, C.** Come from a different patient with a normal LAA morphology for comparison; they are acquired at equivalent angles. LAA = left atrial appendage; LV = left ventricle.

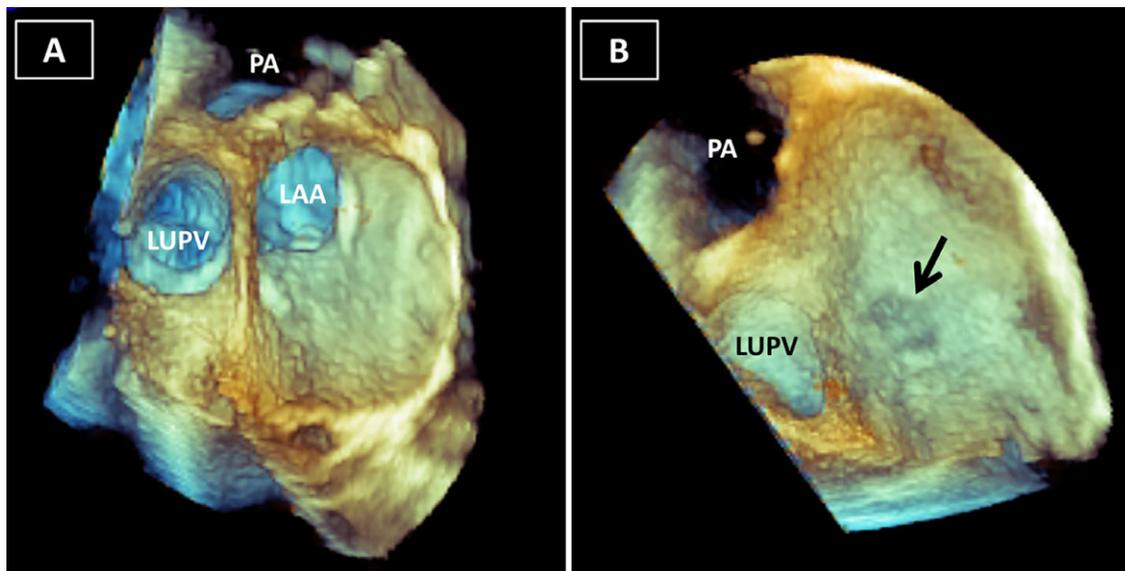


Figure 2. Absent versus present LAA on 3DTEE in case 1. **B.** Demonstrates absent LAA orifice (arrow) on 3DTEE image. For comparison, **A.** from a different patient with a normal LAA orifice is shown. LAA = left atrial appendage; LUPV = left upper pulmonary vein; PA = pulmonary artery.

Case 2:

A 54-year old woman with a history of myocardial infarction, congestive heart failure, and paroxysmal AF on long-term anticoagulation presented for a TEE prior to radiofrequency ablation for new, persistent symptomatic AF. She had a dual chamber ICD already in place.

Her workup included EKG, TEE, and computed tomography (CT) of the chest. TEE using the same ultrasound equipment described in Case 1 revealed moderate left ventricular systolic dysfunction (ejection fraction of 35%) and moderate mitral regurgitation. No LAA could be visualized on multiple 2D and 3DTEE views (Fig. 3 and movie clips S7, S8, and S9). TEE findings were supported by the chest CT findings (Fig. 4).

Discussion:

The LAA is derived from the left wall of the primary atrium and forms during the fourth week of

embryogenesis.² In fetal life, the LAA is a functional LA. The LAA is located within the pericardium,³ produces high levels of atrial natriuretic factor and contributes to the contractile functions of the LA.⁴⁻⁶

The shape and the size of the LAA vary significantly from patient to patient. LAA often has multiple crenellations and lobes, which are potential sites for thrombus formation in the setting of atrial arrhythmias and blood stasis.²

LAA is the most common site of thrombus formation in patients with AF especially in patients with nonvalvular AF. In such patients, 90% of all AF-related thrombi are located in the LAA.⁷ This is in contrast to patients with valvular AF in whom approximately 60% of all AF-related thrombi are located in the LAA. A multicenter study has found that patients with the so-called chicken wing LAA morphology are significantly less likely to have an embolic event compared to cactus, windsock, and cauliflower morphologies.⁸

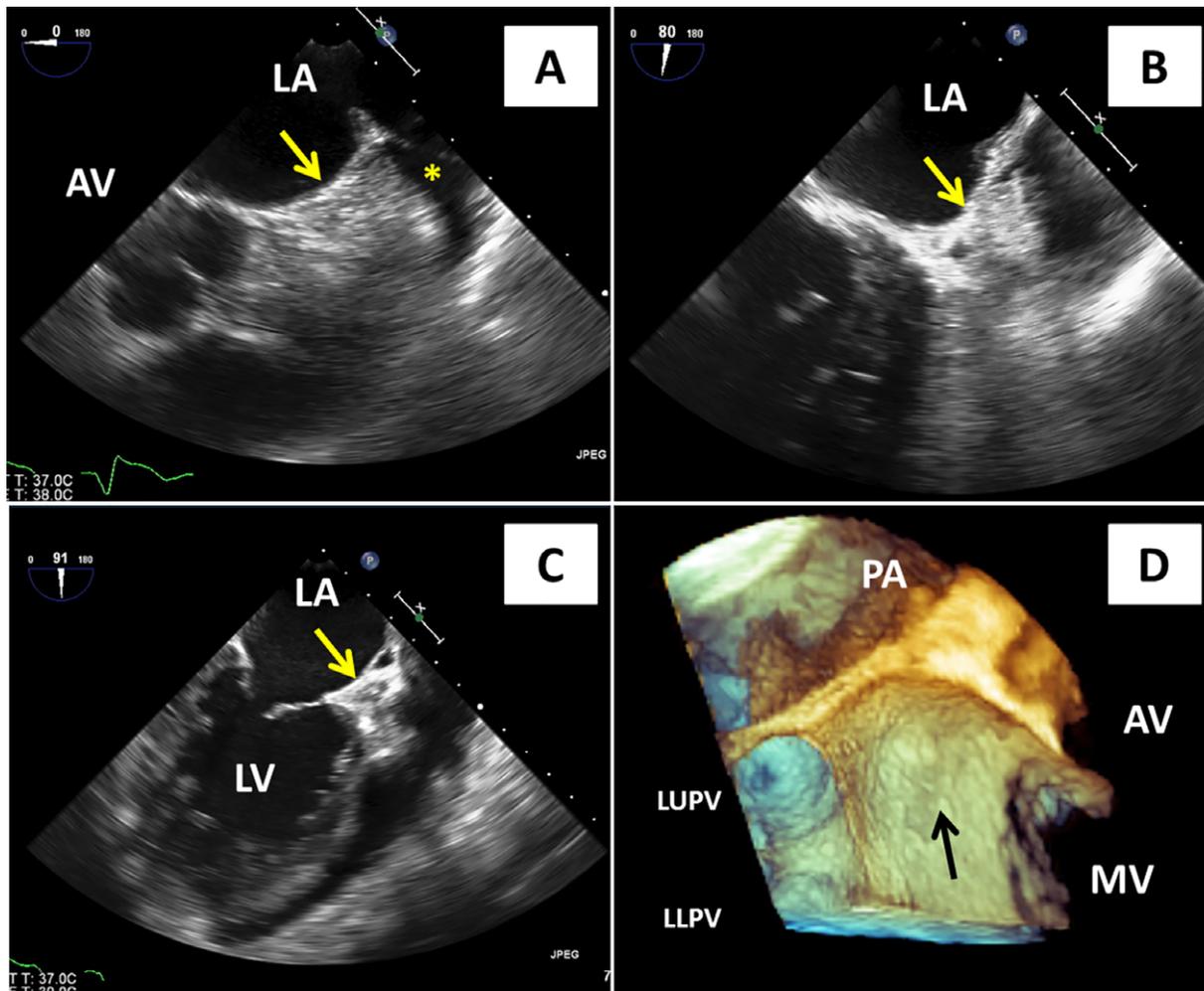
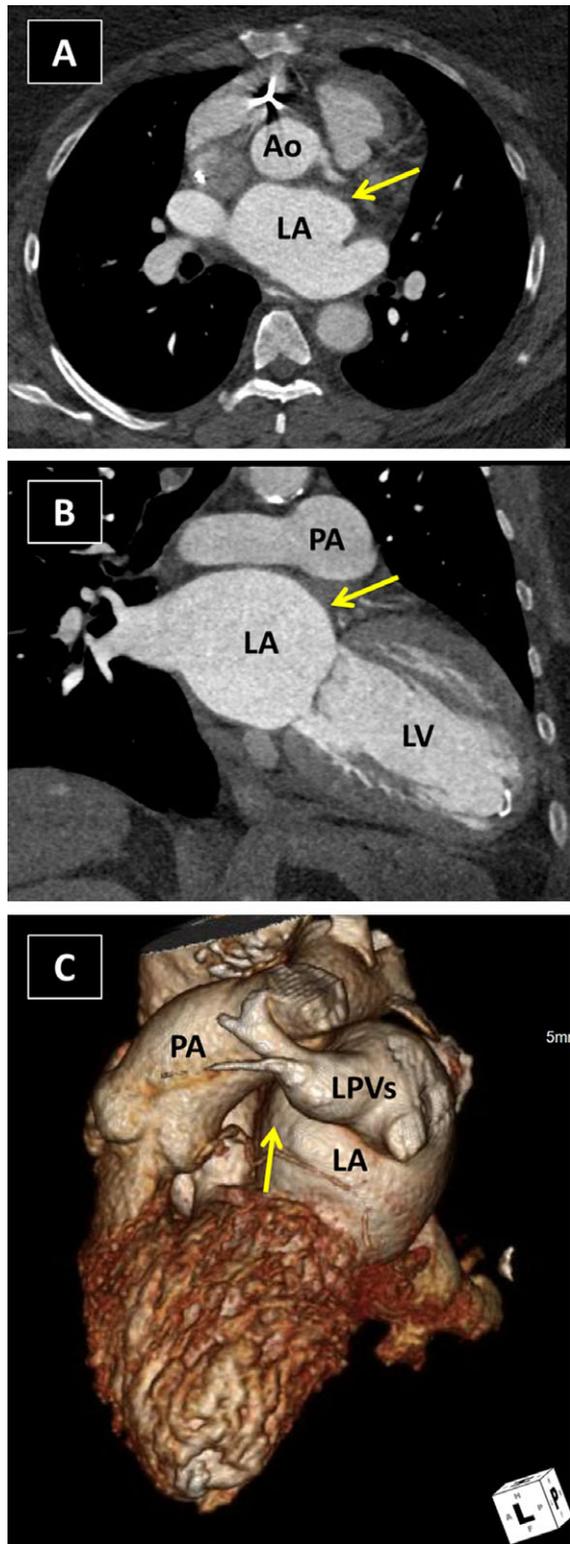


Figure 3. Congenital absence of left atrial appendage on TEE in case 2. **A–C.** Demonstrate 2DTEE while **D.** demonstrates 3DTEE imaging of a congenitally absent LAA (arrow). AV = aortic valve; LA = left atrium; LV = left ventricle; LUPV = left upper pulmonary vein; LLPV = left lower pulmonary vein; MV = mitral valve.



Here, we describe 2 patients with apparent congenital absence of LAA. The absence of LAA is demonstrated by TEE in both cases and corroborated by CT imaging in the second case. Differential diagnosis includes surgical or percutaneous

Figure 4. Congenital absence of left atrial appendage on CT in case 2. EKG-gated intravenous contrast-enhanced CT demonstrates the **A.** absence of LA appendage (arrow) on axial and **B.** two-chamber view. This is also shown on 3D volume rendered image in **C.** the arrow points to the area where LA appendage would have been normally located. Ao = ascending aorta; LA = left atrium; LV = left ventricle; LPVs = left pulmonary veins; PA = pulmonary artery.

closure of LAA, as well as a flush thrombotic occlusion of LAA. Our patients had no history of either surgical or percutaneous LAA intervention. We believe that multimodality imaging excludes the diagnosis of thrombotic LAA occlusion in our patients. In particular, the presence of a smooth endocardial surface in the region of expected LAA orifice on both TEE and CT in the setting of long-term anticoagulation supports the diagnosis of congenitally absent LAA. On CT, there is no appearance of a defined LAA. TEE has the advantage of providing dynamic, real time imaging of the LAA including flow measurements by Doppler. On the other hand, CT and MRI may provide more tomographic cuts and a larger field of view.

Given that AF-related thrombi form predominantly in the LAA, surgical and percutaneous procedures for LAA exclusion have been developed for patients who are not candidates for anticoagulation.^{9,10} While such procedures may eliminate the risk of thromboembolism, the long-term physiologic consequences of LAA exclusion are unknown.

Conclusion:

Congenital absence of the LAA appears to be rare and its physiologic consequences or an association with other congenital conditions are unknown. To our knowledge, only three other case reports of in vivo diagnosis by either 2DTEE or CT have been published thus far.^{3,11,12} Ours appears to be the first to demonstrate the 3DTEE appearance of congenitally absent LAA. Multimodality imaging may be necessary to fully differentiate a congenitally absent LAA from flush thrombosis, surgical ligation or percutaneous device closure of LAA. However, even with best imaging, flush thrombotic occlusion of LAA may not be fully excluded.

References

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Supporting Information

Additional Supporting Information may be found in the online version of this article:

Movie clip S1. For Figure 1A.

Movie clip S2. For Figure 1B.

Movie clip S3. For Figure 1C.

Movie clip S4. For Figure 1D.

Movie clip S5. For Figure 2A.

Movie clip S6. For Figure 2B.

Movie clip S7. For Figure 3B.

Movie clip S8. For Figure 3C.

Movie clip S9. For Figure 3D.

Mini-Abstract:

Congenital absence of left atrial appendage (LAA) is a rare condition with unknown physiological consequences. We present two cases of incidental finding of a congenitally absent LAA in two adult patients. Congenitally absent LAA may

be confused with flush thrombotic occlusion of the appendage. There are very few published reports of congenital absence of LAA, and our report is the first to demonstrate this absence by 3DTEE.