

CASE IMAGE

Familial cryptogenic stroke

Hiroya Takafuji  | Tetsuya Kobayashi | Nahoko Kato | Kotaro Obunai

Department of Cardiology, Tokyo Bay Urayasu Ichikawa Medical Center, Urayasu, Japan

Correspondence

Department of Cardiology, Tokyo Bay Urayasu Ichikawa Medical Center, Hiroya Takafuji, 3-4-32, Todaijima, Urayasu-city, Chiba 279-0001, Japan. Email: hiroyat@jadecom.jp

Key Clinical Message

Familial cryptogenic stroke associated with atrial septal defect and patent foramen ovale is rare. The presence of a family history of cryptogenic stroke may lead to the requirement for careful follow-up for younger family members.

KEYWORDS

atrial septal defect, cryptogenic stroke, familial, patent foramen ovale

1 | CASE DESCRIPTION

The majority of ischemic strokes are due to large artery atherosclerotic stenosis, lacunar stroke, cardiogenic embolism, or other unusual mechanisms. However, many ischemic strokes have unknown etiology and are labeled as cryptogenic strokes. Cryptogenic stroke comprises about 25% of all ischemic strokes. It is essential to determine the possible culprit because this will improve secondary stroke prevention strategies.¹ There are several possible mechanisms implicated in cryptogenic stroke, including occult paroxysmal atrial fibrillation, aortic arch

atherosclerosis, patent foramen ovale (PFO), atrial septal defect (ASD). This case is a very rare Familial cryptogenic stroke associated with ASD and PFO.

Here, the father, a 63-year-old man, traveled and spent the night in his car. The next morning, he awakened to incomplete paralysis and expressive dysphasia. He was transferred to a hospital and diagnosed with an acute ischemic stroke by emergency magnetic resonance imaging (MRI) (Figure 1A). Electrocardiography (ECG) during hospitalization revealed a normal sinus rhythm. Echocardiography revealed no significant plaques in the carotid artery. Laboratory tests showed no hypercoagulable state; Protein

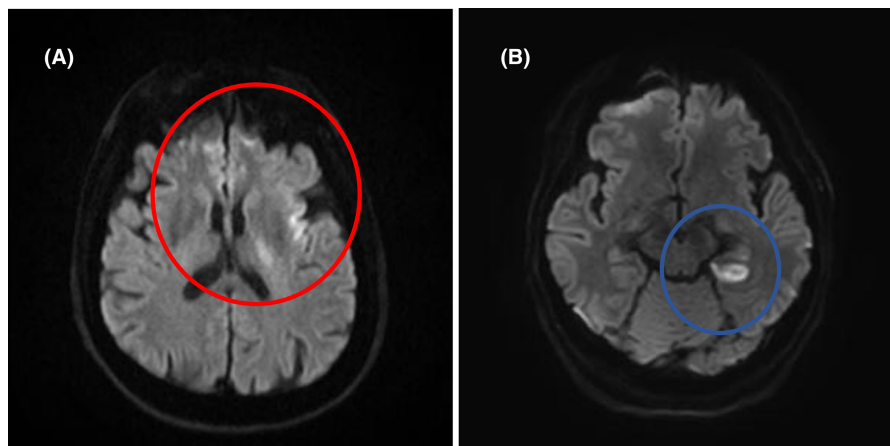


FIGURE 1 Magnetic resonance imaging showing ischemic stroke (red circle and blue circle) (A: father; B: son).

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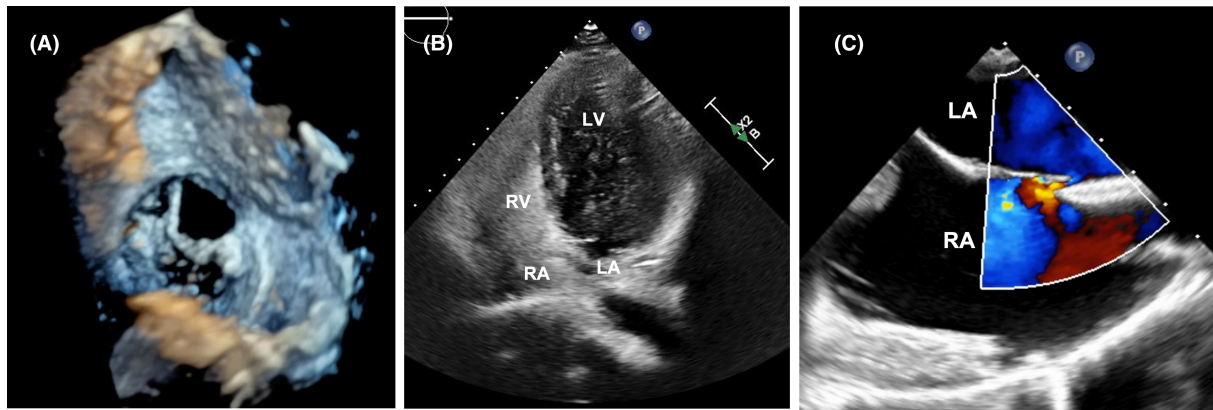


FIGURE 2 Echocardiography showing atrial abnormalities (A: father; B, C: son). LA, left atrium; LV, left ventricle; RA, right atrium; RV, right ventricle.

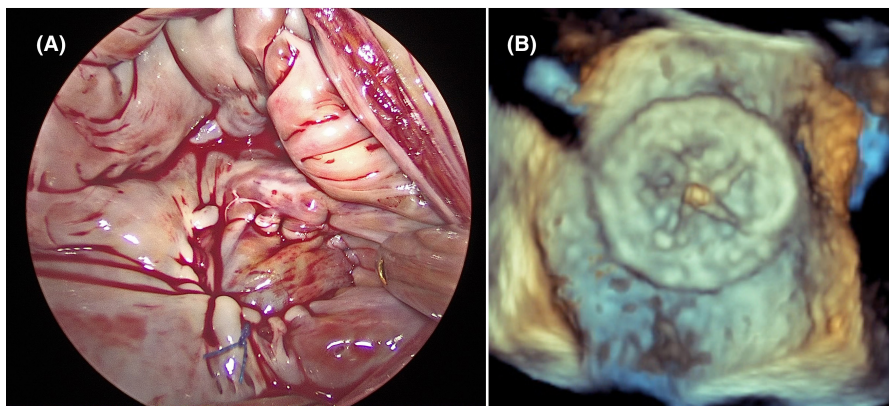


FIGURE 3 Performing closure (A: father; B: son).

S, Protein C, antithrombin III, antinuclear antibody, anti-cardiolipin antibody, and lupus anticoagulant levels were within the normal ranges. Consequently, he was diagnosed with a cryptogenic stroke. Transesophageal echocardiography (TEE) showed multiple defects including an ASD (Figure 2A). Surgical repair was performed to prevent recurrent stroke (Figure 3A).

The son, a 37-year-old man, played on a trampoline. He suddenly felt dysphasia and tinnitus on landing. MRI showed an acute ischemic stroke (Figure 1B). ECG revealed a normal sinus rhythm. Laboratory tests showed no hypercoagulable state. He was also diagnosed with a cryptogenic stroke. Transthoracic echocardiography with a bubble study showed a large right-to-left shunt (Figure 2B). TEE showed persistent color flow through a PFO at rest (Figure 2C). Percutaneous PFO closure using a Gore Cardioform Septal Occluder (W.L. Gore and Associates) was performed to prevent recurrent stroke (Figure 3B).

Familial cryptogenic stroke associated with ASD and PFO is rare.² However, the presence of a family history of cryptogenic stroke may lead to the requirement for careful follow-up for younger family members.

AUTHOR CONTRIBUTIONS

Hiroya Takafuji: Conceptualization; data curation; writing – original draft; writing – review and editing. **Tetsuya Kobayashi:** Writing – original draft; writing – review and editing. **Nahoko Kato:** Writing – original draft; writing – review and editing. **Kotaro Obunai:** Writing – original draft; writing – review and editing.

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CONFLICT OF INTEREST STATEMENT

All authors have reported that they have no relationships relevant to the contents of this paper to disclose.

DATA AVAILABILITY STATEMENT

The data that support the findings of this study are available from the corresponding author upon reasonable request.

CONSENT

We obtained written informed consent for publication from the patient.

ORCID

Hiroya Takafuji  <https://orcid.org/0000-0002-3448-0992>

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