

Moyamoya Disease with Ebstein Anomaly: an Unusual Co-occurrence

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Abstract Moyamoya (meaning a hazy puff of smoke) disease is a rare, idiopathic, persistent, occlusive cerebrovascular disease involving bilateral progressive stenosis or occlusion of a terminal portion of the internal carotid artery, or a proximal portion of the anterior cerebral arteries and the middle cerebral arteries. There are irregular perforating vascular networks (moyamoya vessels), seen in the base of the brain, which produce magnetic resonance images of this ‘puff of smoke’ condition. The association of moyamoya disease and congenital heart disease such as Coarctation of aorta, ventricular septal defect and tetralogy of fallot has been previously reported. Here we report a case of 15 yr old female child presented with seizure episode and was diagnosed as having moyamoya disease with ebstein anomaly. To the best of our knowledge this is the first case report of this co-occurrence of moyamoya disease with ebstein anomaly.

Keywords: moyamoya disease, ebstein anomaly

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1. Introduction

Moyamoya disease is a chronic cerebral vasculopathy first described in 1957 by Taceuchi and Shimizu from Japan, who named the disease in 1969. [1] In Japanese Moyamoya means ‘hazy’. The disease derived its unusual name from angiographic appearance of cerebral vessels that resembles a ‘puff of smoke’ [2]. Moyamoya disease is characterized by a slowly progressive stenosis and obliteration of the large vessels at the base of the brain, affecting mainly the supraclinoid segment of the internal carotid artery and the initial portion of the anterior or middle cerebral arteries and the posterior cerebral arteries [3]. Due to the slow progression of the disease and in response to progressive cerebral ischemia, a large network of collateral vessels is formed from the external carotid arteries, the vertebra-basilar system and other vessels [4]. The idiopathic or primary form of Moyamoya disease, which is sometimes familial, has to be distinguished from the secondary form, referred to as Moyamoya syndrome, which can be associated with certain systemic conditions such as sickle cell disease, chronic basilar meningitidis, neurofibromatosis, X-ray irradiation, homocystinuria, the syndromes of Down, Turner, Alagille and Williams and congenital heart defects. The association of Moyamoya disease and congenital heart defect such as coarctation of the aorta, ventricular septal defect and tetralogy of fallot has been infrequently reported. [5] To the best of our knowledge, this is the first case report of the association of Moyamoya disease with Ebstein anomaly.

We report a case with Moyamoya disease with ebstein anomaly presented with acute convulsions.

2. Case Report

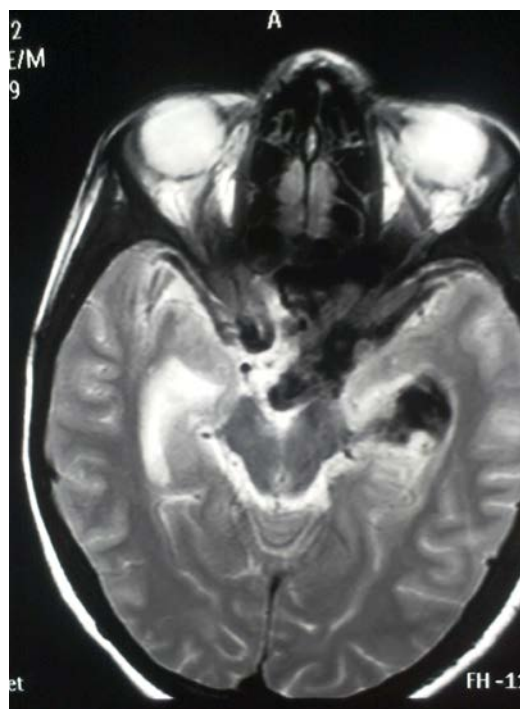


Figure 1. MRI brain (T2W) showing chronic hematoma involving temporal horn of left lateral ventricle

The patient is a 15 ye-old female child who was admitted to medical ward at our institute with complaints of two episodes of seizures one day back. She had previous history of repeated headache which was throbbing in nature and having history of intermittent bluish discoloration of lips mainly during exertion. There was no history of fever, head trauma and ear discharge. There was no history of delayed mile stones, no neurocutaneous markers or asymmetry of face. She had no history of inherited or cerebrovascular diseases. Physical examination revealed no focal neurological deficit but on cardiac evaluation there was splitting of first heart sound and a systolic murmur at left parasternal area. Routine blood investigations were within normal limit. Based on the history and physical examination further neurological and cardiac evaluation was planned. MRI brain (T2W) showed chronic hematoma involving temporal horn of left lateral ventricle (Figure 1) and infarct in left basal ganglia (Figure 2). MR angiography showed stenosis of both distal Internal carotid arteries in suprasellar region with aneurysmal dilatation involving both middle cerebral arteries and multiple collaterals seen peripherally, suggestive of Moyamoya disease (Figure 3). 2-D echocardiography (apical four chamber view) showed a 33 mm apical displacement of the septal tricuspid leaflet and dilatation of the right atrium and right ventricle. There was also a patent foramen of ovale with right to left shunt (confirmed by contrast echo) which was responsible for the intermittent cyanosis (Figure 4). All these echocardiographic findings were suggestive of ebstein anomaly. Patient was managed conservatively on antiepileptics and refer to neurosurgery department for further management for moyamoya disease. Some case reports in the literature where moyamoya disease was associated with congenital cardiac defects but this is the first case report where moyamoya disease presented in association with Ebstein anomaly.

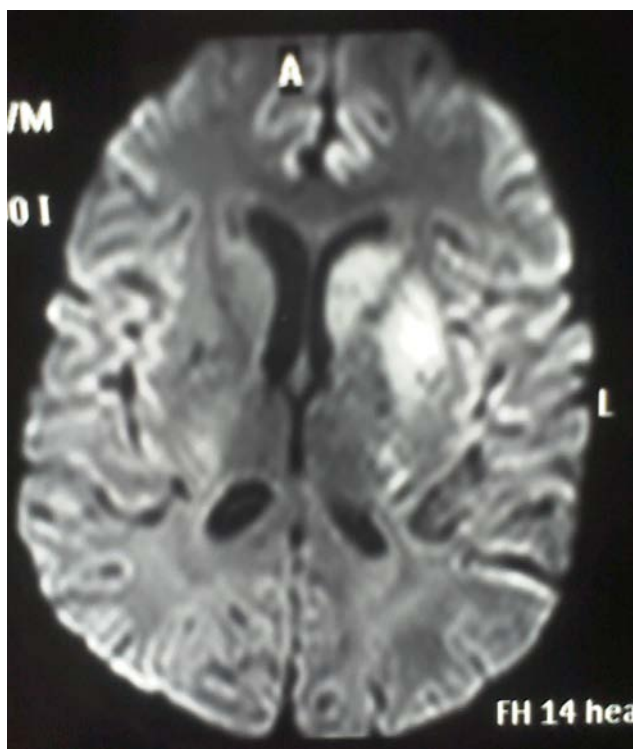


Figure 2. MRI brain (FLAIR) image showing infarct in left basal ganglia



Figure 3. MR angiography of brain showing stenosis of both distal Internal carotid arteries in suprasellar region with aneurysmal dilatation involving both middle cerebral arteries and multiple collaterals seen peripherally, suggestive of Moyamoya disease

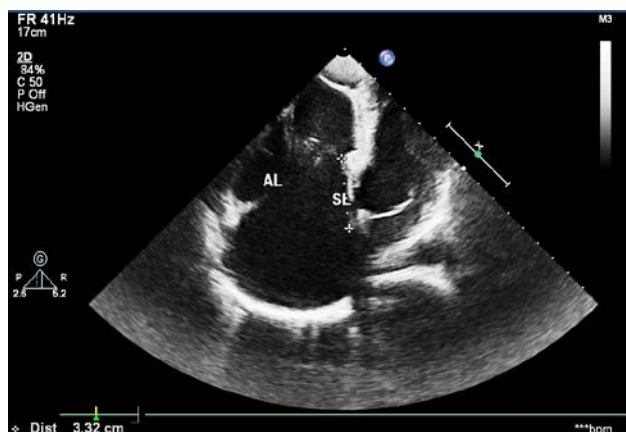


Figure 4. Transthoracic echocardiography (apical 4 chamber view) showing a 33 mm apical displacement of the septal tricuspid leaflet and dilatation of the right atrium and right ventricle. There was also a patent foramen of ovale with right to left shunt (confirmed by contrast echo)

3. Discussion

Moyamoya disease was described initially in Japan in the sixties as (waving puff of smoke). Moyamoya disease is a rare disease characterized by multiple occlusions of the cerebral circulation with an unusual net like system of collaterals. In Japanese, Moyamoya means “hazy”. The disease derives its peculiar name from the angiographic appearance of cerebral vessels in the disease that resembles a “puff of smoke” due to collateral formation, as in our patient (Figure 3). In children, the most common presentation is that of recurrent episodes of headache, cerebral ischemia manifesting clinically as focal deficits, paresthesiae, and seizures [6]. Moyamoya is accepted as primary if it is isolated (Moyamoya disease),

or as secondary (Moyamoya syndrome) if the anomaly is associated with an acquired condition or congenital disorder. The process of narrowing of cerebral vessels seems to be a reaction of brain blood vessels to a wide variety of external stimuli, injuries, or genetic defects. Conditions such as sickle cell anemia, neurofibromatosis-1, Down's syndrome, congenital heart defects, have been found to be associated with Moya Moya disease in the literature [5]. Lutterman et al described five patients of Moyamoya syndrome with congenital heart disease. In their study, coarctation of the aorta was in three patients, in association with a ventricular septal defect (1 patient), aortic and mitral valve stenoses (1 patient), and tetralogy of Fallot (1 patient). Tetralogy of Fallot and a large perimembranous ventricular septal defect were found in the other 2 patients. [5]. An association of Ebstein anomaly and Moyamoya disease has not been described previously, to our knowledge our case is the first report. There may be different explanations for this association. One explanation is that this combination is incidental co-occurrence, as our patient was not so symptomatic at this age from cardiac point of view (Ebstein anomaly). Another explanation is that both findings could be the expression of a systemic congenital malformation involving cardiovascular system.

In conclusion, careful evaluation of children with congenital heart disease and symptoms of cerebral ischemia is warranted to detect the presence of Moyamoya

disease. Prompt diagnosis and treatment of Moyamoya disease is important in these children to prevent progressive neurological deterioration.

Statement of Competing Interests

Authors have no competing interests.

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