CASE REPORT

Adult cyanotic congenital heart disease: an unusual cause of stroke

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Abstract

A 33-year-old male patient with uncorrected tetralogy of Fallot was hospitalised for multiple peripheral arterial emboli. Bilateral above-knee amputation had been done after unsuccessful femoral embolectomy. A large thrombus was detected in the apical portion of the left ventricle which was the source of the embolus. The patient complained of mild frontal headache and progressive right-sided weakness shortly after an echocardiographic examination. A computed tomography (CT) scan revealed a left middle cerebral artery territory infarct. Patients with grown-up cyanotic congenital heart disease are at increased risk of thromboembolic cerebrovascular events. This report highlights the necessity for physicians to be alert for uncommon causes of acute stroke.

Keywords: Stroke, Tetralogy of Fallot.

Introduction

Stroke is one of the most common causes of permanent disability and death. The majority of strokes are ischaemic, and approximately 20% of them are related to cardiogenic cerebral embolism. Congenital heart diseases such as septal defects and patent foramen ovale could be potential causes of stroke in adults. Tetralogy of Fallot (TOF) is the most common form of cyanotic congenital heart disease (CHD) and consists of ventricular septal defect (VSD), right ventricular outflow tract obstruction, an overriding aorta and right ventricular hypertrophy. Although it is usually diagnosed and treated during infancy, only 3% of the patients reach the age of 40 without surgical correction. Venous, and less commonly arterial, in situ thrombus formation have been well documented in patients with TOF as the underlying mechanism of cerebrovascular event (CVE).[1-3] Here we report an intra-cardiac thrombus as the source of acute cardioembolic stroke in an adult with TOF.

Case Report

A 33-year-old male patient with uncorrected TOF was admitted to the hospital with complaints of sudden onset of pain and weakness in his legs. He was known to have TOF since the age of seven but surgical treatment was refused. He had been taking 0.125 mg digitalis and 40 mg furosemide daily. On physical examination, both his feet

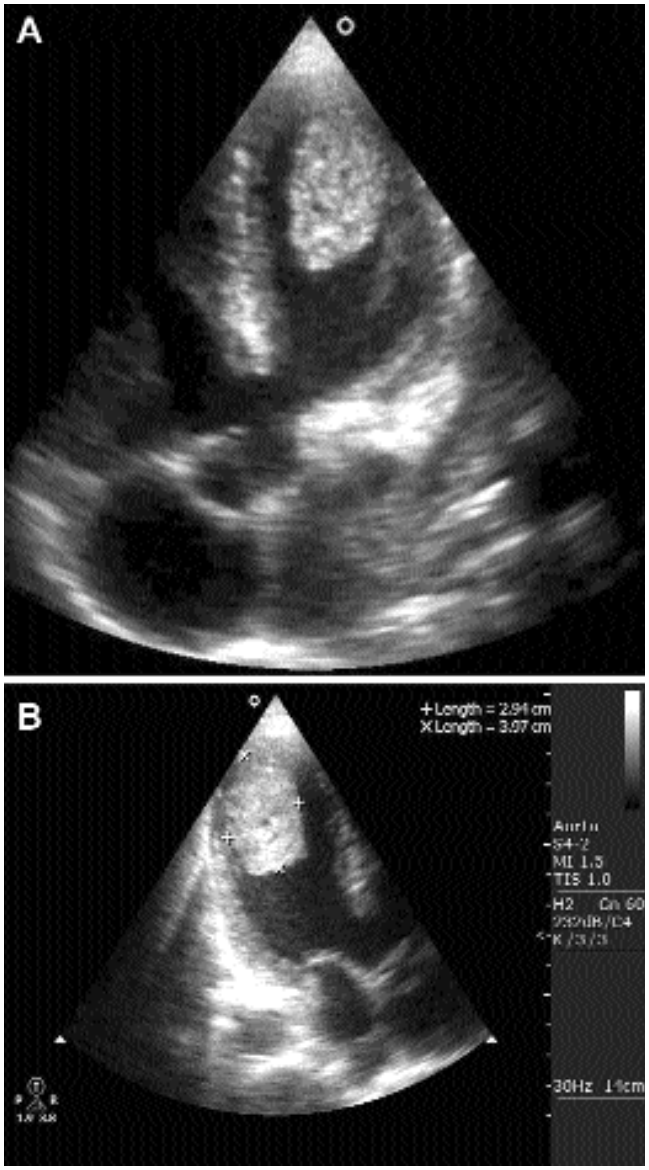

Figure-1A,B: Transthoracic echocardiogram in four chamber view showing a membranous ventricular septal defect, overriding aorta and thrombus in the apical portion of ventricle (A). Thrombus sized 4.0 x 3.0 cm in the apical portion of left ventricle (B).

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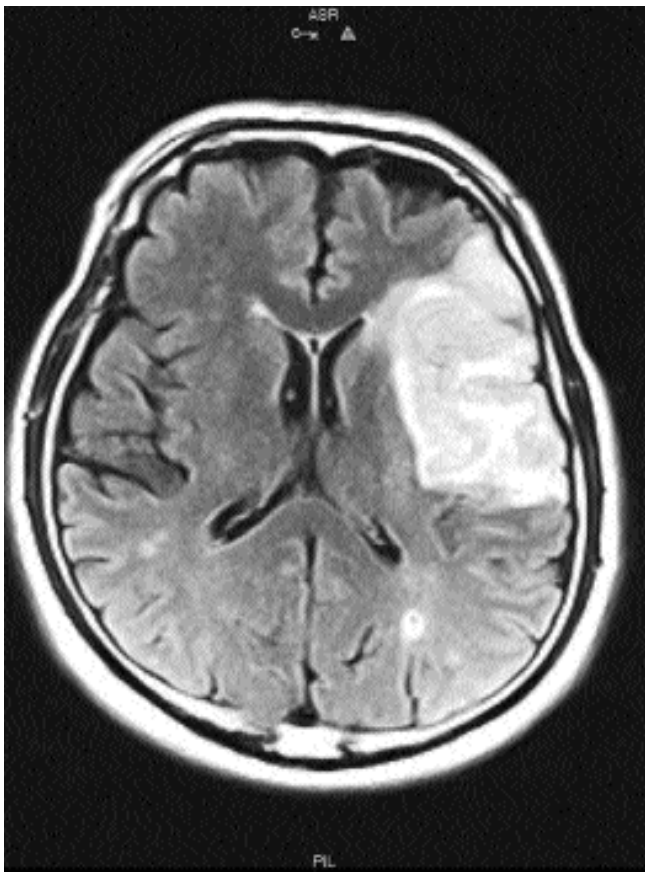

Figure-2: Magnetic resonance imaging (MRI) of brain showing a left middle cerebral artery territory infarct.

were viable but obviously cool and ischaemic. Livedo reticularis and petechiae were seen on his feet and legs. Motor and sensory functions were intact. Femoral pulses were bilaterally palpable but were non-detectable distally. The patient's haemoglobin was 19.2 g/dL (12.1-17.2 g/dL), the mean corpuscular volume was 90.5 fL (82.2-99 fL), haematocrit was 50.8% (36.1-50.3%) and the white cell count was 10.5 K/uL (4-10 K/uL) with 0.1% eosinophils. The electrocardiogram revealed sinus rhythm, right ventricular hypertrophy and right axis deviation.

Bilateral simultaneous femoral arterial embolus was detected with duplex ultrasonographic scanning. He was systemically heparinized and was taken to the operating room for femoral embolectomy. Bilateral above-knee amputation was done after unsuccessful femoral embolectomy. A transthoracic echocardiogram (TTE) was performed postoperatively to rule out a possible cardiac source of embolus. It demonstrated a membranous VSD, an overriding aortic valve and right ventricular hypertrophy (Figure-1A). A large thrombus sized 4.0 x 3.0 cm was seen in the apical portion of left ventricle (LV) (Figure-1B). After TTE, the patient complained of sudden onset mild frontal headache and progressive right-sided weakness. On neurological examination, he was drowsy, with speech disorder, gaze deviation and right hemiplegia with hemihyperacthesia (the muscle power of his right limbs according to Medical Research Council (MRC) motor power grading was grade 2/5 with upgoing right plantar reflex). Magnetic resonance imaging (MRI) revealed a left middle cerebral artery territory infarct (Figure-2).

Discussion

Adults with cyanotic CHD are also at high risk of stroke and thromboembolism.[4] Stroke is also a frequent complication in patients with TOF, the commonest form of grown-up CHD.[5] Congenital cardiac malformations could lead to formation of an intra-cardiac thrombus (in the right heart chambers and left atrium in patients with CHD and in left ventricle in patients with cardiomyopathy) as well as facilitate thrombogenicity in central nervous system (cerebral artery, cerebral vein, and cerebral sinus).[3] Alioglu et al. have reported intra-cardiac thrombosis (2 in right atrium, 1 in right ventricle) in three of the 9 children with TOF.[1] Ammash et al. reported two CVE among 8 patients with TOF in a seven-year period.[2] Ammash's study was the first to show the association between cyanotic CHD and CVE in adults. In contrast, Perloff et al. did not find an increased risk of stroke in 112 adult patients with cyanotic CHD during a 12-year follow-up.[6] In another study, four young adults suffered CVE among 118 patients subsequent to surgical repair of TOF.[7] Echocardiography showed no evidence of intra-cardiac thrombus in these patients and the prevalence of CVE was found to be higher than in normal individuals. To the best of our knowledge, this is the first report of a case with intra-cardiac thrombus as the source of embolus in an adult with CHD and CVE.

There have been several risk factors implicated for the pathogenesis of thrombosis in patients with CHD such as secondary erythrocytosis, hypoxia/hypoxaemia-induced activation of the pro-coagulant pathways, chronic acidosis, increased fibrin deposition, increased tissue factor expression and impaired fibrinolysis.[1] Adults with cyanotic CHD had an increase in red blood cell mass. This secondary erythrocytosis may increase blood viscosity, and thereby reduce cerebral blood flow. Venous thrombosis and, less commonly, arterial thrombosis is thought to be primarily related to erythrocytosis and iron deficiency anaemia in these patients, whereas Agapito et al. reported that a haematocrit level was not a risk factor for adverse CVE.[8] Chronic hypoxaemia activates neutrophils and mononuclear cells that release vasoactive

and chemotactic factors, which results in endothelial injury.[1] Interaction between platelets and endothelial cells induces platelets and enhances intravascular thrombus formation by thrombin, which activates the coagulation cascade.[4] In addition, impaired fibrinolytic system due to increased plasminogen activator-1 levels contributed to thrombogenicity.[1] All of these factors should be taken into account to explain the increased frequency of thromboses in these patients. We presumed that our patient had coagulation abnormalities related to cyanosis and secondary polycythaemia.

Despite the presence of significant coagulation disorders and increasing tendency for thrombosis in patients with cyanotic CHD, anticoagulation is not routinely recommended in the absence of other indications (mechanical valves, atrial fibrillation etc.).[9] Phlebotomy might be effective in improving hyperviscosity but its ability to prevent CVE is controversial.[6,8,10] The role of antiplatelet and antithrombotic therapies to prevent such complications is also questionable.

Today, patients with CHD have a dramatically improved survival and most of these patients reach adolescence and adult life, even those with complex defects, as a result of advances in medical and surgical management. The number of adults with CHD is, therefore, increasing worldwide. Specialized centres are recommended to manage the care of adults with complex and moderate CHD like TOF in collaboration with a specialist in regional centres ideally, but the establishment of these centres are restricted by the barriers of finance and bureaucracy. Moreover, there are very few cardiologists and surgeons trained in this subspecialty. Therefore, primary care physicians, general practitioners and internists have been providing the care and follow-up in most of these patients. Adult health physicians could be faced with these patients in daily practice with different clinical scenarios such as arrhythmias, bacterial endocarditis, cyanosis, polycythaemia, heart failure, pulmonary hypertension and thromboembolic complications. This interesting case report emphasised the necessity for physicians taking care of such patients to be on alert for various presentations of grown-up CHD. Also, uncommon but serious causes of stroke with possible underlying mechanisms should be reviewed.

Conclusion

Adult patients with cyanotic CHD are at increased risk of thromboembolism. Physicians dealing with these patients should keep in mind an intra-cardiac thrombus as a source of systemic embolism which might present with a CVE.

References

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