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A Case of a Pair of Siblings with Moyamoya Disease

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1. Abstract

A pair of siblings with Moyamoya disease (MMD) were admitted to our department and treated with superficial temporal artery-middle cerebral artery (STA-MCA) anastomosis and encephalo-myo- synangiosis (EMS). We evaluated the relevant literature to analyze the causes and treatments of MMD in similar patients. This case report illustrates that the genes of the MMD patient plays an important role in the pathogenesis of MMD, and the combined STA-MCA bypass with EMS is useful for both ischemic-type and hemorrhagic type MMD.

2. Case

The brother: A 35-year-old man with a chief complaint of "right-sided weakness after cerebral hemorrhage operation for 3 months". Three months prior to admission he suffered from sudden unconscious, accompanied with right-sided weakness. He got coma when found by his family. In the local hospital, the computed tomography (CT) scan showed hemorrhage in the left basal ganglia (Figure1 A1). The trepanation and drainage of intracranial hematoma was performed immediately. After operation, his consciousness got better, but he got aphasia, and had his right-sided weakness. Later, the patient underwent rehabilitation therapy in another hospital for a long time. Before admitted to our department, the patient is clear, trouble with speaking and disfunction of right-sided limb. The patient had no history of hypertension, diabetes, heart disease, infectious disease, surgery, or trauma. On November 7, 2018, the patient was diagnosed with MMD by Digital subtraction angiography (DSA) in local hospital. Physical examination revealed aware in patients with consciousness, trouble with speaking clearly. Skull defects were found on the left frontal region. Bilateral pupils were normal. The myodynamia of left limbs is normal, but the right one is lower. Normal muscle tension and physiology reflexes exist, and the right-sided Babinski's sign was positive. Some relevant examinations were performed before operation. Computed tomography angiography (CTA)of head showed bilateral MMD (Figure1 B1). The cerebral perfusion imaging showed that the perfusion of bilateral frontal lobe and the left temporal lobe decreased (Figure1 C1). The patient's family members provided written informed consent for surgery, which was performed with the patient under general anesthesia. During the operation, the left STA was located by touching on the scalp. A Y-shaped incision on the left frontotemporal was taken to expose the main trunk of the left STA and its branches. The anterior branch was temporarily blocked and the end of the anterior branch was dissected. After the left frontal bone window were formed, the dura mater was cut and fixed. The branches of the MCA were anatomized along with cerebral sulcus. The branches of the MCA were temporarily blocked at both ends. After repeated irrigation by heparin, the branches of the MCA were anastomosed with the anterior branches of the STA. The posterior branches of the STA were applied to the surface of brain. Twenty days after surgery, the head CTA was reviewed (Figure1 D1). The patient was followed up by telephone for 3 months after discharge. His condition was

stable.

The sister: A 40-year-old woman was admitted to our hospital with the complaint of intermittent headache for 1 month. The location of the headache was mainly on the right temporal, which could be relieved or disappeared by itself. Without dizzy, vomiting, fever, seizure, or movement disturbances. The CTA on local hospital showed that the occlusion of the right MCA and ACA. The patient came to our department for surgery on march 20th. Magnetic Resonance Imaging (MRI) of the head showed that she got MMD (Figure1 A2 and B2).

Perfusion weighted imaging (PWI) showed that the perfusion of the right frontal, temporal and parietal lobe and the left parietal lobe decreased significantly (Figure1 C2). The patient had a history of hypertension and once had a orthopedic surgery. She is the elder sister of the man above. Physical examination by a specialist revealed that the patient was conscious. Bilateral pupils were about 3 mm in diameter, and the light reflection was normal. The body moved freely, physiology reflex exist, and pathological reflex had not elicited. The procedure of operation was the same as that of the former. She discharged without any discomfort.

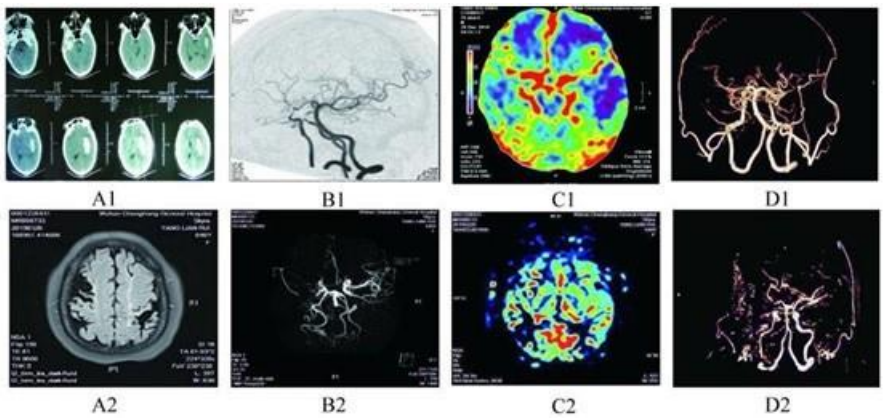

Figure 1: A1-D1 for the brother: **(A1)** hemorrhage in the left basal ganglia before operation **(B1)** Computed tomography angiography before operation **(C1)** the cerebral perfusion imaging before operation **(D1)** Computed tomography angiography afteroperation, **A2-D2** for the sister: **(A2)** and **(B2)** magnetic resonance imaging before operation **(C2)** perfusion weighted imaging before operation **(D2)** Computed tomography angiography after operation

3. Discussion

Moyamoya disease is a rare condition, primarily reported in 1957 by Takeuchi and Shimizi. This disease is a kind of chronic progressive occlusive cerebrovascular disease, also known as sub-cerebral vascular reticulosis [1]. The main manifestation is a steno-occlusive change at the end of the internal carotid artery (ICA), middle cerebral artery (MCA) and/or proximal anterior cerebral artery (ACA),which is accompanied by the formation of smoke-like abnormal blood vessels in the base of the skull [2, 3]. At present, the pathogenesis remains unknown. The clinical signs of this cerebrovascular entity can be roughly classified into two categories: brain ischemia (ischemic-type) and cerebral hemorrhage (hemorrhagic type). These two types of symptom differ in their distribution between pediatric and adult patients. The pediatric patients mainly presented progressive cerebral ischemia, including transient cerebral ischemic attack and cerebral infarction. In adults, intracranial hemorrhage is a common symptom, such as intracerebral hemorrhage, intraventricular hemorrhage, subarachnoid hemorrhage [4]. In recent years, large amounts of research on the aetiology and treatment of MMD have been performed.

According to the epidemiological studies in Japan,7% to 12% of MMD patients have family history. It is suggested that genetic factors may play an important role in the pathogenesis of MMD. The genetic pattern of a familial MMD is autosomal dominant inheritance, and the key gene maybe located on chromosome 17q25.3 [5, 6]. The results of genome-wide association analysis on 72 patients with MMD by Kamada et al [7] showed that the susceptibility gene RNF213 was highly correlated with familial MMD.A large number of studies have shown that MMD may be a hereditary disease.

Operation is the first-choice treatment for MMD. Surgical revascularization is the main method on operation. It includes three types: direct revascularization, indirect revascularization and combined revascularization. The most common method on direct revascularization is STA-MCA bypass. The indirect revascularization refer to using temporal muscle, dura mater and cerebral cortex to adhere and form new blood vessels, which often include encephalo-myo-synangiosis (EMS), encephalo-duro-arterio-synangiosis (EDAS), encephalo-duro-myo-arterio-synangiosis (EDMAS) and so on. Combined revascularization refers to the combination of direct and indirect revascularization or several different types of indirect revascularization. It can not only improve the local cerebral perfusion insufficiency, but also reduce the rate of re-bleeding. It is one of the more advocated treatment methods for MMD (Czabanka et al [8]). Divide thirty MMD patients into STA-MCA bypass with EMS treatment group and EMS treatment group, then

the DSA examination was performed after operation for 7 days, 6 months and 12 months to evaluate the blood flow reserve capacity of cerebrovascular. The results showed that STA-MCA bypass with EMS could improve cerebral perfusion more effectively than EMS alone.

In conclusion, although the genes of the above two patients had not been tested, genetic factors are associated with the incidence of MMD, so the pathogenesis of MMD should be explored further. The combined STA-MCA bypass with EMS is useful for both ischemic-type and hemorrhagic type MMD, and continuous optimization of surgical techniques should be done in order to improve the prognosis of MMD patients.

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