Case Report

Headache as unusual presentation in moyamoya disease

Pathik Shah[1], Harsh Gandhi[2*]

[1]Department of Internal Medicine, GMERS Medical College, Gotri, Vadodara, Gujarat, INDIA.
[2]Department of Internal Medicine, Baroda Medical College, Vadodara, Gujarat, INDIA.
Email: harshgandhi345@gmail.com

Abstract

Our case highlights basics of clinical presentations, causes, risk factors, epidemiology, physiology, and pathophysiology in Moyamoya diseases, along with introduction to terminologies, prospect of evaluation and various modalities available for diagnosis. Moyamoya angiopathy is characterized by a progressive stenosis of the terminal portion of the internal carotid arteries and the development of a network of abnormal collateral vessels. This chronic cerebral angiopathy is observed in children and adults. It mainly leads to brain ischemic events in children, and to ischemic and hemorrhagic events in adults. This is a rare condition, with a marked prevalence gradient between Asian countries and Western countries. Two main nosological entities are identified. On the one hand, moyamoya disease corresponds to isolated moyamoya angiopathy, defined as being "idiopathic" according to the Guidelines of the Research Committee on the Pathology and Treatment of Spontaneous Occlusion of the Circle of Willis. This entity is probably multifactorial and polygenic in most patients. On the other hand, moyamoya syndrome is a moyamoya angiopathy associated with an underlying condition and forms a very heterogeneous group with various clinical presentations, various modes of inheritance, and a variable penetrance of the cerebrovascular phenotype. Diagnostic and evaluation techniques rely on magnetic resonance imaging (MRI), magnetic resonance angiography (MRA) conventional angiography, and cerebral hemodynamics measurements. Revascularization surgery can be indicated, with several techniques. Characteristics of genetic moyamoya syndromes are presented, with a focus on recently reported mutations in *BRCC3/MTCP1* and *GUCY1A3* genes. Identification of the genes involved in moyamoya disease and several monogenic moyamoya syndromes unravelled different pathways involved in the development of this angiopathy. Studying genes and pathways involved in monogenic moyamoya syndromes may help to give insights into pathophysiological models and discover potential candidates for medical treatment strategies.

Key Word: moyamoya.

[*]**Address for Correspondence:**
Dr. Harsh Gandhi, Department of Internal Medicine, Baroda Medical College, Vadodara, Gujarat, INDIA.
Email: harshgandhi345@gmail.com

Received Date: 22/05/2019 Revised Date: 16/06/2019 Accepted Date: 09/07/2019
DOI: https://doi.org/10.26611/102111212

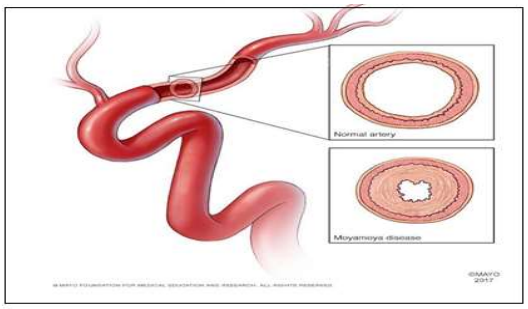
Access this article online	
Quick Response Code:	Website: www.medpulse.in
	Accessed Date: 14 August 2019

INTRODUCTION

Moyamoya is a disease in which certain arteries in the brain are constricted. Blood flow is blocked by the constriction, and also by blood clots (thrombosis)[1]. A collateral circulation develops around the blocked vessels to compensate for the blockage, but the collateral vessels are small, weak, and prone to bleeding, aneurysm and thrombosis. On conventional MR angiography, these collateral vessels have the appearance of a "puff of smoke" (described as "(moyamoya)" in Japanese). Moyamoya syndrome is unilateral arterial constriction, or occurs when one of the several specified conditions is also present[2] Patients usually present with TIA, ischemic/hemorrhagic stroke, or seizure.[3] The age distribution is bimodal being either young adolescence or mid-forties[4] Moyamoya disease can be either congenital or acquired. Patients with Down syndrome, sickle cell anemia, neurofibromatosis type1, congenital heart disease, fibromuscular dysplasia, activated protein C resistance, or head trauma can

How to cite this article: Pathik Shah Harsh Gandhi. Headache as unusual presentation in moyamoya disease. *MedPulse International Journal of Medicine.* August 2019; 11(2): 115-118. https://www.medpulse.in/Medicine/

develop moyamoya malformations.[5] It is more common in women than in men, although about a third of those affected are male[6].



CASE PRESENTATION

18 years old female presented to outpatient department with generalized headache since 4 days. Headache was acute in onset, severe in intensity, thunderclap type, associated with difficulty in performing daily routine activities, no associated aggravating or reliving factors or any variation. No complain of nausea, vomiting, vision abnormalities, trauma, any focal neurological deficit, fever, giddiness, convulsion or altered sensorium. Patient had similar episodes of headache in the past and was given over the counter medication. No past history of hypertension or blood transfusion. Birth History is born at term, normal vaginal delivery, cried immediately after birth. Patient was vitally stable and neurological examination was under normal limit. Patient was admitted for further evaluation and management. During hospital stay various routine investigation was done (table 1), along with CT–Scan and MRI brain. CT–Scan Head reviled hemorrhage in fornix and lateral ventricle along with ill defined hypodensity in splenium of corpus callosum. (Figure 1) MRI Brain with Angiography reveled acute Intraventricular hemorrhage in bilateral lateral ventricles (left > right). A small lesion of cytotoxic edema seen in splenium of corpus callosum. Focal moderate to severe short segmental luminal narrowing at the bifurcation of bilateral middle cerebral arteries. Multiple small caliber collateral arteries in bilateral basal cisterns and bilateral basal ganglia with flow related enhancement. (Figure 2,3,4).

Table 1:

Test	Result	Reference range
HAEMOGLOBIN	9.2	12 - 16 gm%
Blood Group	A Positive	
RBC Count	5.21	3.8 - 4.8 million/cmm
HCT	32	36 - 48%
MCV	62	83 - 101 fl
MCH	17.8	26.4 -33.2 Pg
MCHC	28.8	31.8 – 35.9%
RDW-CV	16.7	11.6 – 14.0%
Total WBC Count	5780	4000 - 10000/cumm
Platelet Count	381	150 - 450 thou/cumm
Smear Examination	Microscopic hypochromic RBC	
PS for MP	Not detected	
ESR	12	00-15 mm/hr
Total Bilirubin	1.0	0/1 – 20 mg/dl
Direct Bilirubin	0.4	0 – 0.4 mg/dl
Indirect Bilirubin	0.6	0 – 1.08 mg/dl
APTT	Test - 27.0, Control – 29.5	25.0 – 33.0 Seconds
Prothrombin Time	Test – 13.9, MNPT- 12.6	11.5 – 14.1 Seconds
INR	1.13	Non Therapeutic: upto 1.2 Therapeutic Range: 2.0 – 3.0
Random Glucose	96	<140 mg/dl
S. Creatinine	0.89	0.52 – 1.04 mg/dl
HIV 1 and 2	0.07	<1.0 S/Co : Non-Reactive >=1.0 S/Co: Reactive
Sickling	Negative	

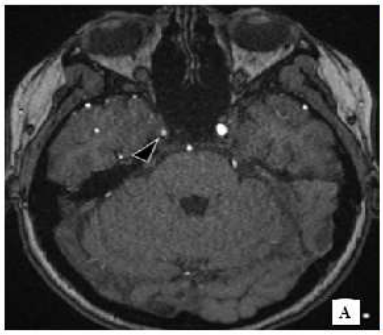


Figure 1

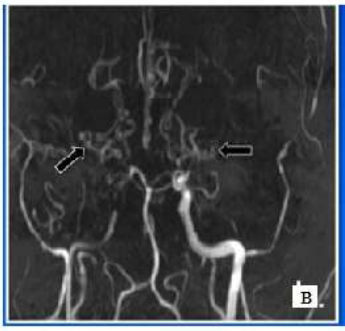


Figure 2

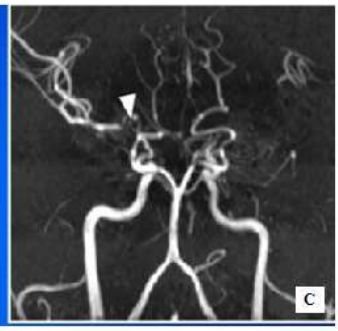


Figure 3

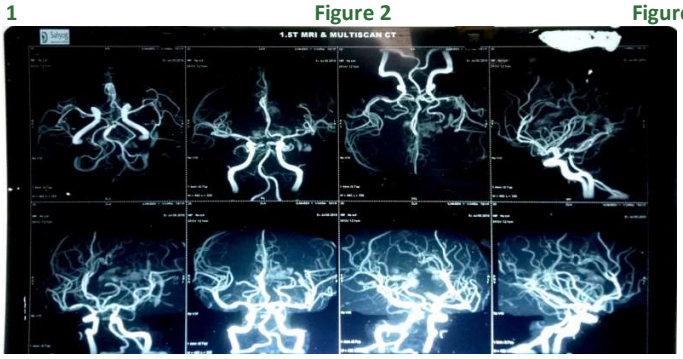


Figure 4

DISCUSSION

Moyamoya disease may occur at any age, though symptoms most commonly occur between 5 and 10 years of age in children and between 30 and 50 years of age in adults. The first symptom of moyamoya disease is usually stroke or recurrent transient ischemic attacks (TIAs), especially in children. Adults may also experience these symptoms but more often experience bleeding in the brain (hemorrhagic stroke) from abnormal brain vessels.

Accompanying signs and symptoms of moyamoya disease related to reduce blood flow to the brain include:

- Headache
- Seizures
- Weakness, numbness or paralysis in your face, arm or leg, typically on one side of your body
- Visual disturbances
- Difficulties with speaking or understanding others (aphasia)
- Developmental delays
- Involuntary movements
- Cognitive decline

These symptoms can be triggered by exercise, crying, coughing, straining or fever.

Causes: The exact cause of moyamoya disease is unknown. Moyamoya disease is more common in Japan, Korea and China, but it also occurs in other parts of the world. Researchers believe the higher concentration of moyamoya disease in these Asian countries strongly suggests the disease may have genetic causes. Moyamoya is also associated with certain conditions, such as Down syndrome, sickle cell anemia, neurofibromatosis type 1 and hyperthyroidism.

Risk factors: Though the cause of moyamoya disease is unknown, certain factors may increase your risk of having the condition, including:

- **Being of Asian descent.** Moyamoya disease is found all over the world, but it's more common in East Asian countries, especially Korea, Japan and China. This may possibly be due to certain genetic factors in those populations. Higher rates of moyamoya disease have been documented among Asians living in western countries.
- **Having a family history of moyamoya disease.** If you have a family member with moyamoya disease, your risk of having the condition is 30 to 40 times higher than the general population — a factor that strongly suggests a genetic component to the disease and may justify screening of family members.
- **Having a certain medical condition.** Moyamoya disease sometimes occurs in association with another disorder, including neurofibromatosis type 1, sickle cell anemia and Down syndrome, among others.
- **Being female.** Females have a slightly higher incidence of moyamoya disease.
- **Being young.** Though adults can have moyamoya disease, children younger than 15 years old are most commonly affected.

Complications

Most complications from moyamoya disease are associated with the effects of stroke, such as:

- **Vision problems.** As a result of stroke, some people with moyamoya disease experience visual disturbances.
- **Weakness (hemiparesis).**
- **Language disturbance (aphasia).**
- **Movement disorders.** Though rare, some people with moyamoya disease experience involuntary movement of certain muscles.
- **Learning or developmental issues.** Following a stroke, a child may have problems with mental processing, which can affect schoolwork as well as cause emotional difficulties and low self-esteem. Adults may experience memory decline as well as challenges with other areas of cognitive function.
- **Seizures.**

CONCLUSION

This case highlights the importance of considering Moya Moya Disease one of the main etiologies of Acute Ischemic Stroke in children from Asian population. It also emphasizes the rare presentation such as headache among the population and the use of neurovascular imaging techniques to facilitate diagnosis of Moya Moya Disease. Final treatment is revascularization surgery. However, if detected early patient can be managed symptomatically and subjected to treatment to avoid life threatening complication and death.

REFERENCES

1. Moyamoya Disease and Moyamoya Syndrome, R. Michael Scott and Edward R. Smith, New England Journal of Medicine, 360:1226-1237, March 19, 2009
2. Jump up to:[a] [b] 13. Ganesan, V., and Smith, E. R. (2015). Moyamoya: defining current knowledge gaps. Developmental Medicine and Child Neurology, 57(9), 786-787. doi:10.1111/dmcn.12708
3. Kleinloog, R (May 2012). "Regional differences in incidence and patient characteristics of moyamoya disease: a systematic review". J Neurol Neurosurg Psychiatry. 83(5):531-6.
4. Duan, L (October 2011). "Moyamoya disease in China: its clinical features and outcomes". Stroke.
5. Janda, Paul; Bellew, Jonathan; Veerappan, Venkatachalam (2009). "Moyamoya disease: case report and literature review". The Journal of the American Osteopathic Association. 109 (10): 547–553. PMID 19861596.
6. Kuriyama S, Kusaka Y, Fujimura M, *et al*. (2008). "Prevalence and clinicoepidemiological features of moyamoya disease in Japan: findings from a nationwide epidemiological survey". Stroke. 39 (1): 42–7. doi:10.1161/STROKEAHA.107.490714. PMID 18048855

Source of Support: None Declared
Conflict of Interest: None Declared