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"Pediatric Presentation of Moya-Moya Disease: A Genetic Insight"

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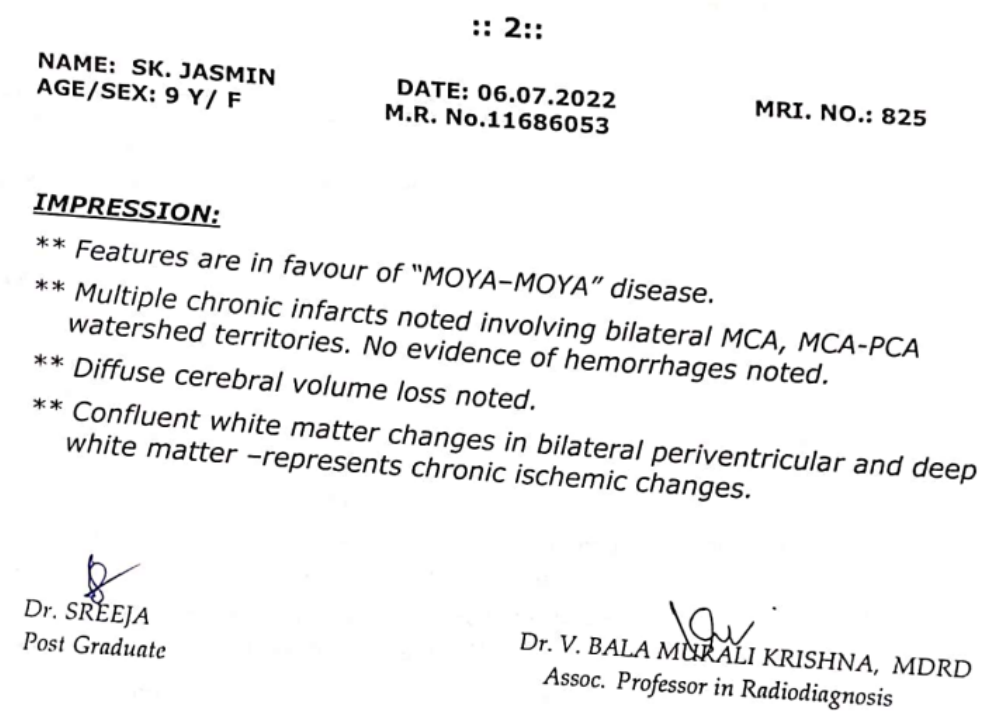
ABSTRACT:

This abstract summarizes a case of Moya-Moya disease in a 9-year-old female patient with symptoms resembling those of her elder sister. Laboratory and imaging diagnostics confirmed the diagnosis, revealing characteristic findings of parenchymal and vascular calcifications, chronic infarct changes, and post-operative alterations. The familial occurrence of symptoms suggests a genetic component, emphasizing the importance of genetic counseling. Additionally, autoimmune markers were detected, indicating potential autoimmune associations with the disease. This case underscores the necessity of a multidisciplinary approach for early detection, personalized management, and improved outcomes in Moya-Moya disease.

KEYWORDS: Moya-Moya, Genetic disorder

CASE REPORT:

A 9-year-old female patient presented to the emergency department with complaints of abnormal gait, pain in all four limbs, and a 4-month history of urinary incontinence. These symptoms are reminiscent of those experienced by her elder sister, who suspects a genetic disorder. Laboratory diagnostics confirmed the suspicion, revealing CT findings indicative of parenchymal and vascular calcifications in the bilateral high parietal lobes and left frontal lobe, chronic infarct changes in the left high parietal lobe, and post-operative changes consistent with encephalo-myo-synangiosis. MRI further supported the diagnosis, showing features consistent with Moya-Moya disease, including multiple chronic infarcts involving bilateral MCA and MCA-PCA watershed territories, as well as diffuse cerebral volume loss. The ANA-17 profile revealed traces of dsDNA and SmD1, as well as traces of PCNA and positivity for SS-B/La.



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NAME: SK. JASMIN
AGE/SEX: 9 Y/ F

DATE: 06.07.2022
M.R. No.11686053

MRI. NO.: 825

IMPRESSION:

** Features are in favour of "MOYA–MOYA" disease.
** Multiple chronic infarcts noted involving bilateral MCA, MCA-PCA watershed territories. No evidence of hemorrhages noted.
** Diffuse cerebral volume loss noted.
** Confluent white matter changes in bilateral periventricular and deep white matter –represents chronic ischemic changes.

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Post Graduate

Dr. V. BALA MURALI KRISHNA, MDRD
Assoc. Professor in Radiodiagnosis

MRI OF BRAIN WITH M R A

Clinical H/o: 2-3 episodes of seizures at the age of 3 years. Present complaint of weakness in right upper and lower limbs.

Technique: T1W, T2W, FLAIR, Gradient, DWI : AXIALS T1W :AXIALS ,SAGITTALS
3D TOF – MIP Images: Radial ranges, T2W Axial & DWI: BRAIN

FINDINGS: movement artifacts noted in most of the sequences limiting the evaluation of study.

MRI BRAIN

- No evidence of restriction of diffusion seen in the present scan.
- Diffuse cerebral volume loss noted which is evident by prominent sulci, cisternal spaces and prominent ventricular system.
- Evidence of peripherally placed wedge shaped T2 hyperintensities and FLAIR hypointensities noted in left frontal and bilateral parietal, occipital cortex and white matter – represents chronic infarcts.
- T2 and FLAIR hyperintensities noted in bilateral periventricular and deep white matter areas – represents chronic ischemic changes.
- Chronic lacunar infarcts noted involving head of caudate nucleus bilaterally, left lentiform nucleus and thalamus on right side.
- There are numerous small curvilinear flow voids seen in basal cisterns, surrounding brain stem and also in bilateral thalami and capsulo-ganglionic region – these represent multiple collateral vessels.
- No evidence of susceptibility blooming noted in SWI.
- Mucosal thickening noted in frontal sinus on right side.

M R ANGIOGRAM:

- There is significant narrowing seen at supraclinoid segment of bilateral internal carotid arteries.
- A1 segment of bilateral anterior cerebral arteries are narrowed in caliber. A2 and A3 segments of ACA are normal.
- M1 segment of bilateral middle cerebral arteries are narrowed in caliber. Cortical branches of MCA appear to be maintained.

Asst. Professor

Report Date : 25-Jul-22 09:55 am

ANA PROFILE - 17P

Parmeter	Result Va	Units	Bilological Reference	Method
ds DNA	TRACES (+/-)			Immunoblot
Nucleosome	NEGATIVE			Immunoblot
Histone	NEGATIVE			Immunoblot
SmD1	TRACES (+/-)			Immunoblot
PCNA	TRACES (+/-)			Immunoblot
PO	NEGATIVE			Immunoblot
SS-A/Ro 60 kD	NEGATIVE			Immunoblot
SS - A/Ro 52 kD	NEGATIVE			Immunoblot
SS - B / La	POSITIVE (+)			Immunoblot
CENP - B	NEGATIVE			Immunoblot
Scl 70	NEGATIVE			Immunoblot
UI - snRNP	NEGATIVE			Immunoblot
AMA M2	NEGATIVE			Immunoblot
Jo - 1	NEGATIVE			Immunoblot
PM - Scl	NEGATIVE			Immunoblot
Mi - 2	NEGATIVE			Immunoblot
Ku	NEGATIVE			Immunoblot

*** End Of Report ***

INTRODUCTION :

Moya-Moya disease is a rare condition characterized by the narrowing of the blood vessels, particularly the internal carotid arteries, which supply blood to the brain. This narrowing restricts blood flow to the brain, increasing the risk of stroke.[1]

INCIDENCE :

No community-based studies from India are available regarding the prevalence of Moya-Moya disease. However, its occurrence is estimated to range from 3.2 to 10.5 cases per 100,000 population. Generally, the disease is observed to have a higher prevalence among individuals of Asian descent. The precise cause of this disease remains unknown. Approximately 57% of affected individuals are of Asian descent, and 71% are female. While Moya-Moya disease can manifest across all age groups, it is more frequently observed in individuals aged 5-15 years and 30-40 years. A familial history is noted in about 10%-15% of cases.[2]

SIGNS & SYMPTOMS :

- Headache
- Seizures
- Vision problems
- Involuntary movements
- Weakness, numbness, or paralysis in your face, arm, or leg
- Cognitive or developmental dealys [3]

RISK FACTORS :

Moyamoya disease exhibits a higher prevalence among individuals of East Asian descent, particularly in countries such as Korea, Japan, and China. This elevated occurrence is also observed in Asian populations residing in Western countries, indicating a potential genetic predisposition within these ethnic groups.

A family history of moyamoya disease significantly elevates the risk of developing the condition, with individuals having a 30 to 40 times higher likelihood compared to the general population. This familial clustering strongly implicates a genetic basis for the disease.

Moyamoya syndrome can occur in conjunction with various other medical conditions, including neurofibromatosis type 1, sickle cell disease, and Down syndrome, among others, further complicating the clinical landscape of the disease.

While moyamoya disease affects both genders, females exhibit a slightly higher incidence compared to males.

Although moyamoya disease can affect individuals of all ages, it predominantly manifests in children under the age of 15, highlighting the vulnerability of this age group to the condition.[3]

DISCUSSION :

The presentation of a female patient with symptoms mirroring those of her elder sister, alongside confirmatory laboratory and imaging findings indicative of Moya-Moya disease, underscores the significance of recognizing both the clinical and genetic aspects of this condition.

Moya-Moya disease is a rare cerebrovascular disorder characterized by progressive narrowing of the internal carotid arteries, leading to compromised blood flow to the brain. This reduction in cerebral perfusion can manifest in various neurological symptoms, such as abnormal gait, limb pain, and urinary incontinence, as observed in the patient.

The familial occurrence of symptoms strongly suggests a genetic predisposition to Moya-Moya disease, as supported by the presence of the condition in both siblings. While the precise genetic mechanisms underlying Moya-Moya disease remain incompletely understood, studies have implicated various genetic factors contributing to its pathogenesis. Understanding the genetic basis of Moya-Moya disease not only aids in elucidating disease mechanisms but also facilitates genetic counseling and screening in affected families to identify at-risk individuals and guide personalized management strategies.

Laboratory investigations, including CT and MRI imaging, play a pivotal role in the diagnosis of Moya-Moya disease. In this case, CT findings revealed characteristic calcifications and chronic infarct changes in specific brain regions, alongside post-operative alterations consistent with surgical interventions aimed at improving cerebral blood flow. MRI further supported the diagnosis by highlighting features typical of Moya-Moya disease, including multiple chronic infarcts involving critical cerebral artery territories and diffuse cerebral volume loss.

The presence of autoimmune markers in the ANA-17 profile adds an additional layer of complexity to the patient's presentation. While autoimmune associations with Moya-Moya disease have been reported, the significance of these specific autoantibodies in disease pathogenesis warrants further investigation.

Overall, this case underscores the importance of a multidisciplinary approach to the diagnosis and management of Moya-Moya disease, integrating clinical evaluation, imaging studies, and laboratory investigations. Furthermore, the familial occurrence of the condition highlights the need for heightened awareness of its genetic component and emphasizes the importance of genetic counseling and screening in affected families. Early recognition and intervention are crucial for optimizing outcomes and improving the quality of life for individuals affected by Moya-Moya disease.

CONCLUSION :

The case highlights the intricate interplay between clinical presentation, genetic predisposition, and diagnostic modalities in Moya-Moya disease. Familial clustering underscores the importance of genetic factors in disease pathogenesis, necessitating genetic counseling for at-risk families.

Advanced imaging techniques like CT and MRI play a crucial role in confirming the diagnosis and guiding management decisions. The presence of autoimmune markers adds complexity to the disease spectrum, warranting further research into potential autoimmune associations. A multidisciplinary approach is essential for early detection, personalized management, and improved outcomes in Moya-Moya disease.

Statement of Ethics: An informed consent form was taken from the patient

Conflict of Interest: The authors declared no conflicts of interest

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