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Hemi-Hydranencephaly: Living with Half a Brain

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ABSTRACT

Background: Hemi-hydranencephaly (HHE) is a rare brain anomaly with absent cerebral cortex, which gets replaced by a cerebrospinal fluid (CSF) filled membranous sac. Prenatal vascular disruption leads to unilateral carotid artery disease, resulting in an encephaloclastic process with cerebral resorption. Genetic and acquired causes can be implicated in the pathogenesis. Clinical features encompass a range of manifestations, including motor impairment, reduced mental development, seizures and ocular signs. Treatment targets the underlying deficit encountered by the patient and physical rehabilitation.

Case Presentation: We present a case of a 22-year-old female, with atonic seizures and a significant past history of generalized tonic clonic seizures, with development of right hemiparesis and facial asymmetry at 9 months of age. On examination, she had disuse atrophy with contractures of the right upper and lower limbs. Magnetic resonance imaging (MRI) suggested replacement of the left cerebral hemisphere with CSF and the MR angiography showed narrowing of the left internal carotid artery with its branches, confirming the diagnosis of HHE. Anti-epileptic treatment was initiated, and physical rehabilitation was started for the limb contractures.

Conclusion: Cognitive development in HHE is a natural experiment with special interest in long term language and neuropsychological development.

KEYWORDS: *Hemi-hydranencephaly, brain malformation, congenital anomaly*

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1. INTRODUCTION

HHE is an atypical brain condition with total or near-total, unilateral absence of the cerebral cortex. CSF fills the membranous sacs of the affected hemisphere, which may contain minimal remaining neurons, glial tissue and blood vessels [1]. The infratentorial regions including basal ganglia, pons, medulla, cerebellum, falx and meninges remain uninvolved [2]. Literature shows less than ten reported cases worldwide, which fulfil all the criteria for diagnosis, including unilateral cerebral hemisphere replaced by a CSF filled sac [1].

Unilateral carotid artery disease leading to ipsilateral cerebral hemisphere replacement with a CSF sac is thought to be the main pathogenesis. Lack of involvement of the vertebrobasilar system leads to preservation of the infratentorial structures [3]. The vascular disturbance during fetal development is thought to occur after neuronal migration, during fourth and sixth months of gestation [4]. Thus, initial formation occurs in the affected cerebral hemisphere but is resorbed by encephaloclastic processes. Theory suggests that during brain development, functional compensation should occur for the occluded vessel via the circle of Willis, the reason for it not occurring is unknown [1]. Patients with HHE carry a better prognosis than other destructive encephalopathies, despite a large parenchymal defect. This is attributed to neuronal plasticity of the developing brain [5].

Non-genetic pathways suspected in the pathogenesis include in utero vascular accidents and infections, caused by cytomegalovirus, herpes simplex, Toxoplasma gondii and necrotizing vasculitis. An arterial stroke from a thromboembolic event originating from fetal vessels, heart also placenta, occurring in the perinatal period can also be implicated. Another cause could be the relative hypercoagulability in pregnancy, with reduced levels of protein S, decreased activity of protein

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C pathway, increased thrombin generation and elevated fibrinogen levels [6].

Genetic pathways may also participate in multiple cellular processes and could be implicated in the pathogenesis of HHE. Somatic mutations in PI3K-Akt3-mTOR have been found responsible in neurodevelopmental disorders and could be a possible target site [7]. Corticospinal neurons may have the potential to regenerate disrupted pathways in the perinatal period, as exhibited by plasticity during axonal growth and synaptogenesis [6]. Further research into the pathogenesis pathway is required.

Clinical features show a variable spectrum in past reported cases. Motor impairment included hemiparesis with occasional facial involvement. Some patients had reduced mental development and IQ. Generalized tonic clonic seizures (GTCS) and status epilepticus were rarely seen with few patients showing poor language development. Ocular signs including strabismus, optic atrophy, nystagmus, asymmetric pupils, abducent paresis and blindness have also been reported [1]. Our patient had left hemiparesis with contractures and left facial nerve palsy. She had an unusual presentation with atonic seizures and was diagnosed at a later age than typical.

Establishment of diagnosis is primarily based on neurological examination with motor deficits, delayed mental development or ocular signs, and use of neuroradiological techniques, including computed tomography (CT) scan, MRI, and MRA. Diffusion tensor MR can be used to show reduced fractional anisotropy and higher diffusivity of contralateral hemispheric tracts. The corticospinal and optic tracts are completely absent on the affected side [1]. An MRI and MRA was conducted in our patient showing absent left hemisphere and narrowing of left side cranial vessels.

Treatment options address the underlying deficit encountered in the patient. Physical rehabilitation, botulin injections and when necessary, orthopaedic intervention can be used for spasticity. Logotherapy is beneficial in language dysfunction. Anticonvulsive therapy should only be used for seizure episodes and routine use is not recommended. Neurosurgery for hydrocephalus or a midline shift towards the normal hemisphere maybe required [4], [8]. Physical therapy was initiated in our patient and anticonvulsive treatment was initiated due to seizure episodes at presentation.

HHE is a natural experiment with potential implications for normal cognitive development. Long term follow-up of language development and other neuropsychological functions can be interesting [4]. We present the case of a 22-year-old female with seizures and right hemiplegia, diagnosed with HHE.

2. CASE REPORT

A 22-year-old female visited the emergency department with four episodes of atonic seizures occurring over the last one year. She was born at term, of non-consanguineous healthy parents, with no prenatal complications. She is the third child among three otherwise healthy children. The family history is unremarkable.

She was diagnosed with measles infection at nine months of age, for which supportive home care was initiated. During this period, she had one episode of generalized tonic clonic seizure and developed acute onset right sided hemiparesis with facial asymmetry. Medical management was done, for which records are not available. She is not on any medication currently and has no new or progressive deficits. There were no further restrictions in the sensorimotor and language developmental milestones. She had poor school performance and studied till the fifth grade. She continues to assist in all chores at home.

Her general and physical examination shows disuse atrophy and contractures of the right upper and lower limbs, demonstrated in figure 1. There was right upper motor neuron type of facial weakness, demonstrated in figure 2. Tone was increased in the right upper and lower limbs, with preserved power and extensor plantar on the right side.

Magnetic resonance imaging (MRI) was done which demonstrated replacement of the left cerebral hemisphere by CSF. There was a large cerebrospinal fluid space involving the left hemi-cranium and supra-tentorial brain parenchyma, communicating with the third ventricle and gliosis of the right cerebellar tonsil, probably due to vascular insult, demonstrated in figures 3 to 6. MR Angiography (MRA) showed narrowed left internal carotid artery (ICA), A1 segment of anterior carotid artery (ACA) and M1 segment of middle carotid artery (MCA), dipicted in figures 7 to 9. Routine laboratory examination, including coagulation assessment, electrocardiogram, fundus, and renal ultrasound, were normal.

We initiated anti-epileptic treatment with lamotrigine for the atonic seizures. Physical rehabilitation therapy was started for the spasticity and contractures of the right upper and lower limbs with an aim to improve mobility.

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REFERENCES

[1] P. Pavone *et al.*, "Hemihydranencephaly: living with half brain dysfunction," *Ital J Pediatr*, vol. 39, p. 3, 2013, doi: 10.1186/1824-7288-39-3.

[2] S. Ulmer, F. Moeller, M. A. Brockmann, J. P. Kuhtz-Buschbeck, U. Stephani, and O. Jansen, "Living a normal life with the nondominant hemisphere: magnetic resonance imaging findings and clinical outcome for a patient with left-hemispheric hydranencephaly," *Pediatrics*, vol. 116, no. 1, pp. 242–245, 2005, doi: 10.1542/PEDS.2004-0425.

[3] J. A. Eyre, S. Miller, G. J. Clowry, E. A. Conway, and C. Watts, "Functional corticospinal projections are established prenatally in the human foetus permitting involvement in the development of spinal motor centres," *Brain*, vol. 123 (Pt 1), no. 1, pp. 51–64, 2000, doi: 10.1093/BRAIN/123.1.51.

[4] F. Greco, M. Finocchiaro, P. Pavone, R. R. Trifiletti, and E. Parano, "Hemihydranencephaly: case report and literature review," *J Child Neurol*, vol. 16, no. 3, pp. 218–221, 2001, doi: 10.1177/088307380101600311.

[5] "(PDF) Hemihydranencephaly; a Case Report." Accessed: Jun. 27, 2025. [Online]. Available: https://www.researchgate.net/publication/26633534_Hemihydranencephaly_a_Case_Report

[6] S. M. A. Hassanein, Y. A. Abbas, A. M. Monib, and M. S. El Alfy, "Hemihydranencephaly syndrome: case report and review," *Dev Neurorehabil*, vol. 14, no. 5, pp. 323–329, Oct. 2011, doi: 10.3109/17518423.2011.593574.

[7] A. Poduri *et al.*, "Somatic Activation of AKT3 Causes Hemispheric Developmental Brain Malformations," *Neuron*, vol. 74, no. 1, p. 41, Apr. 2012, doi: 10.1016/J.NEURON.2012.03.010.

[8] R. P. Moser and E. L. Seljeskog, "Unilateral hydranencephaly: case report," *Neurosurgery*, vol. 9, no. 6, pp. 703–705, 1981, doi: 10.1227/00006123-198112000-00015.

Figure 1 – Disuse atrophy and contracture of right upper limb

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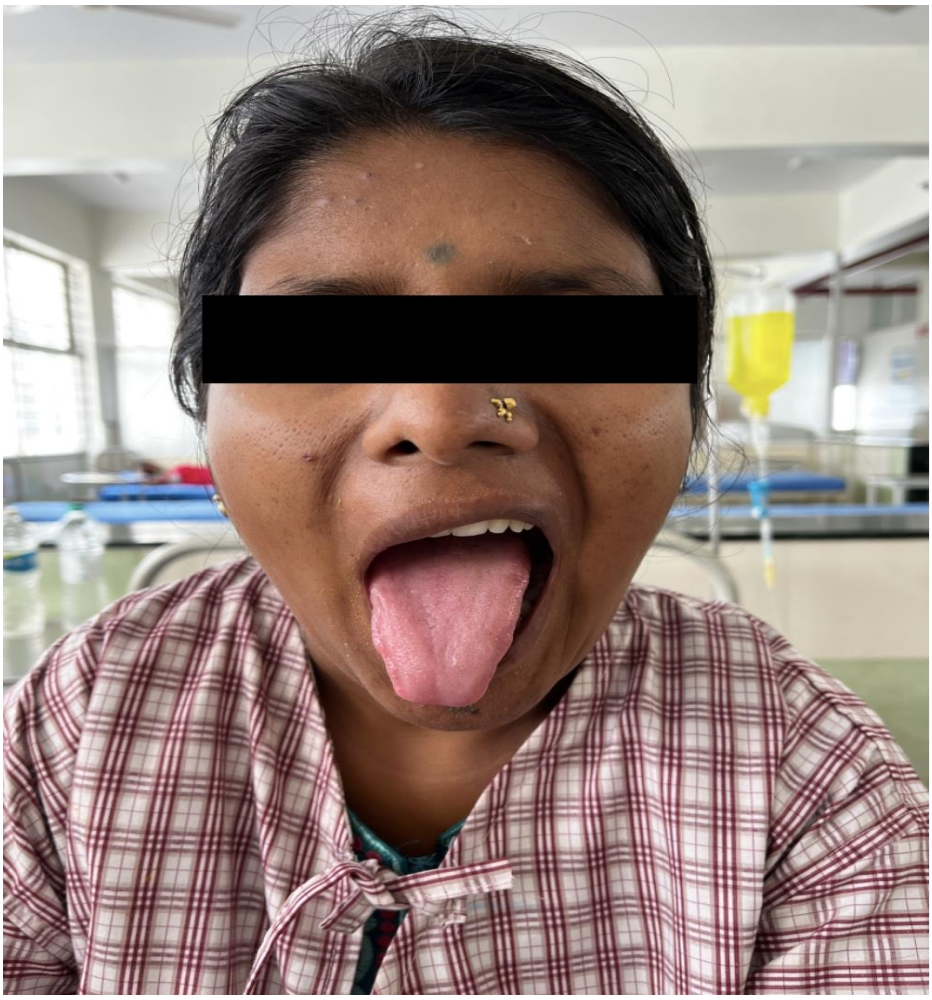

Figure 2 - Right upper motor neuron deficit with left sided deviation of face

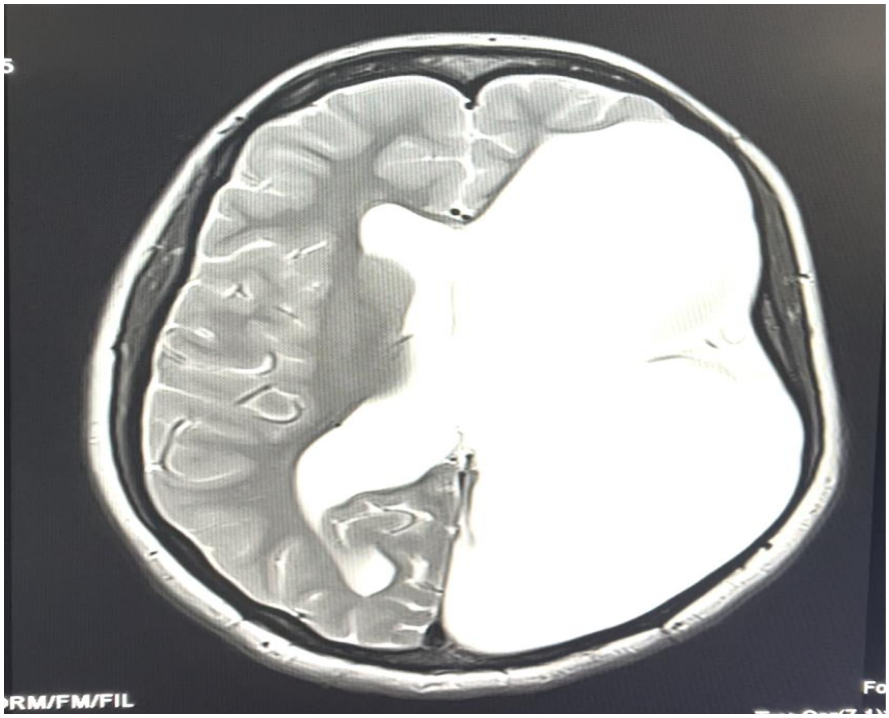

Figure 3 – T2W MRI

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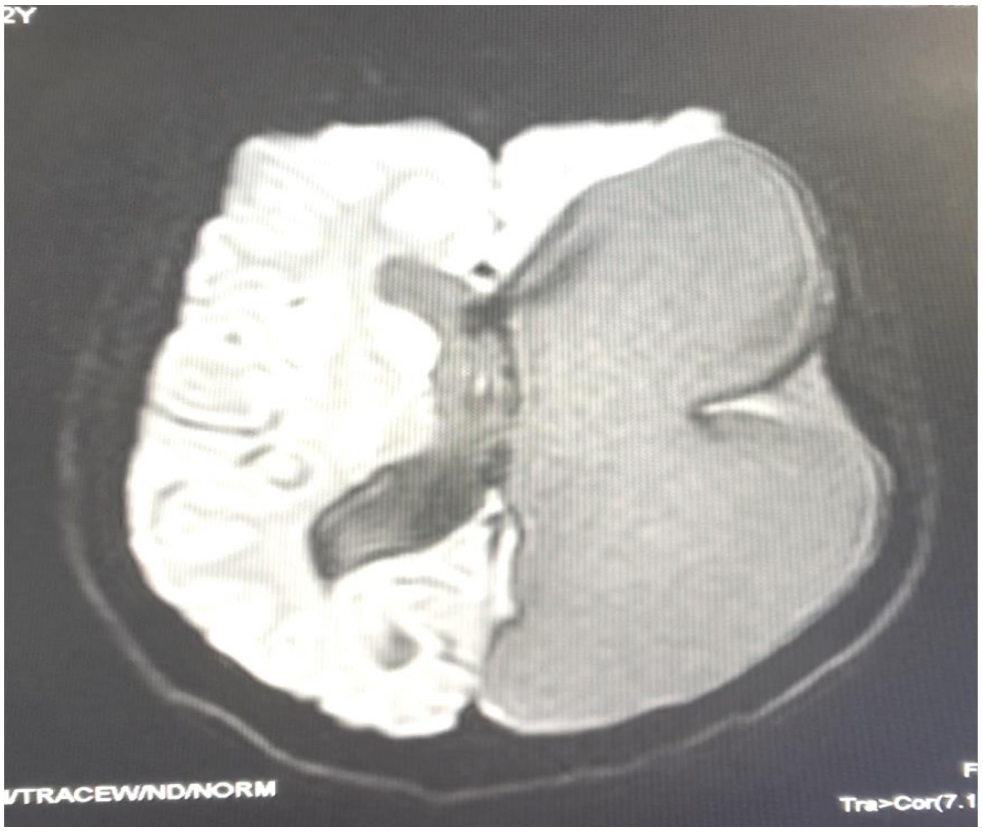

Figure 4 – T1W MRI

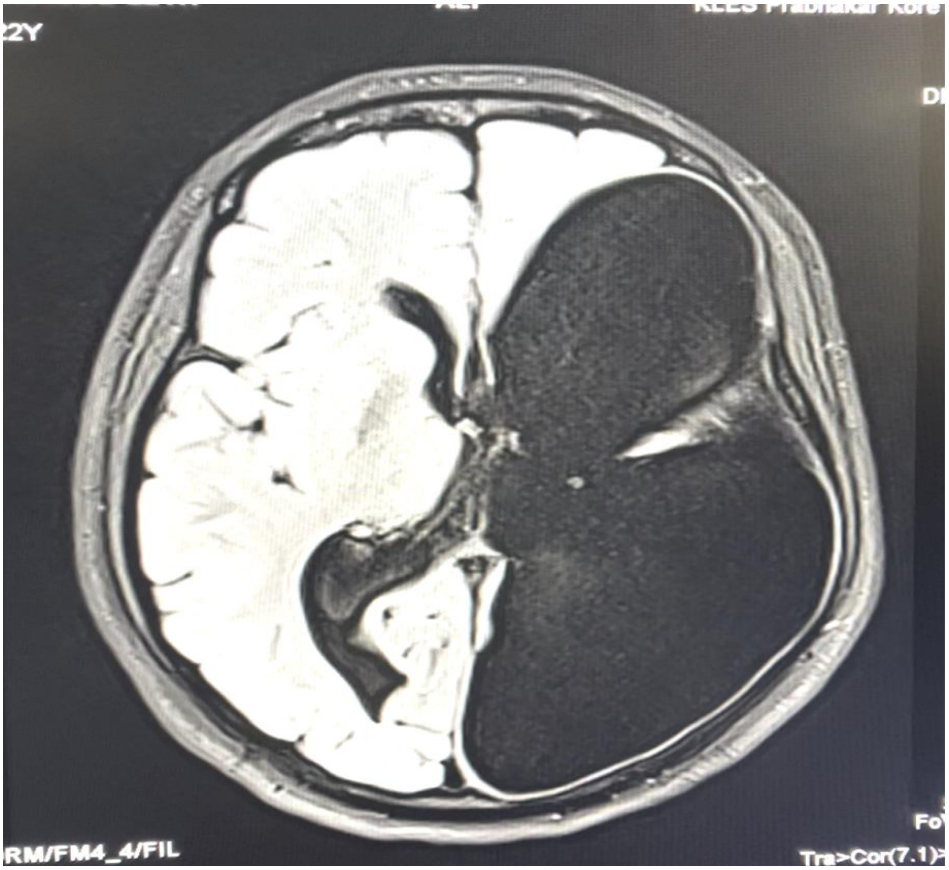

Figure 5 – MRI FLAIR

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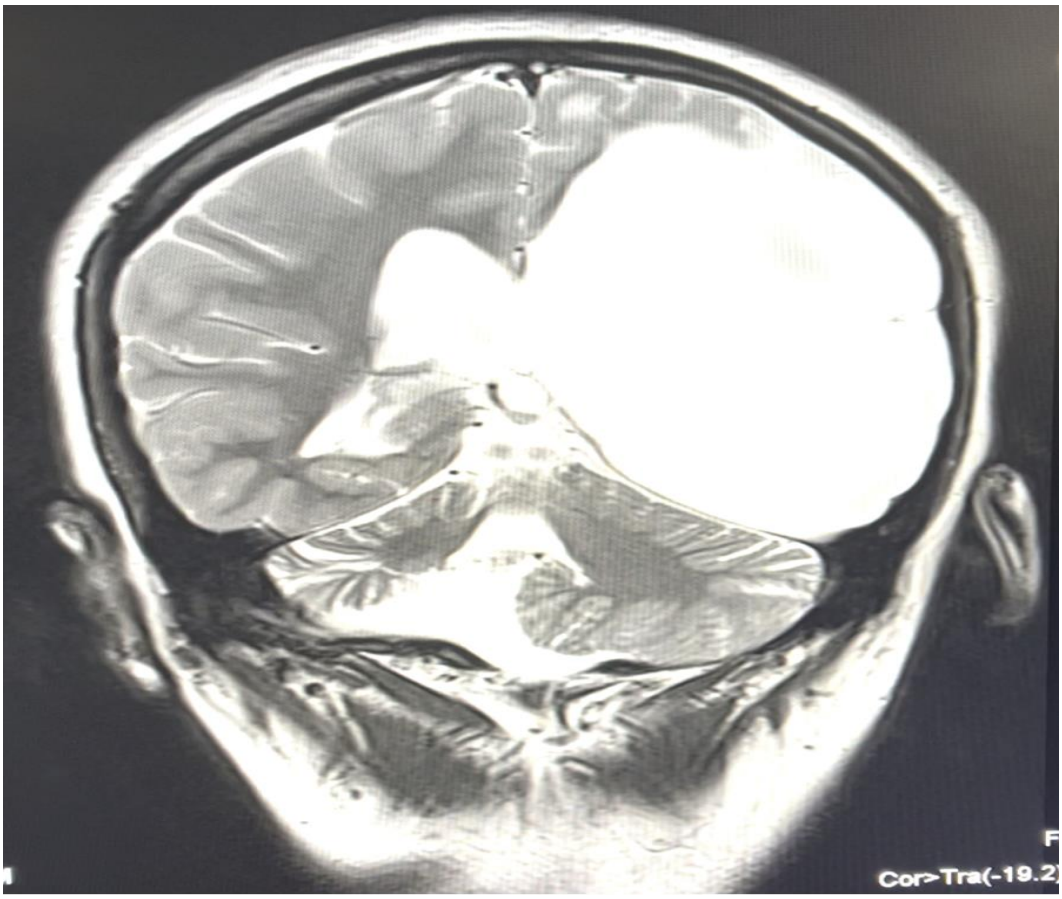

Figure 6 – Coronal view

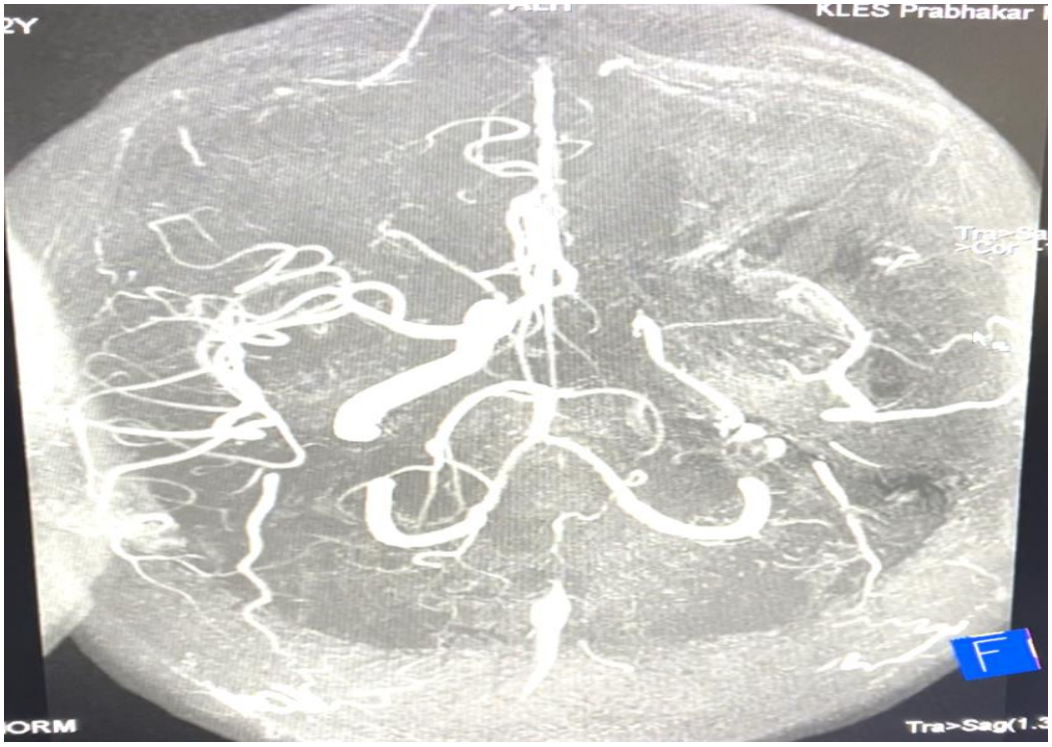

Figure 7 - MRA suggestive of narrowed left internal carotid artery (ICA), A1 segment of anterior carotid artery (ACA) and M1 segment of middle carotid artery (MCA)

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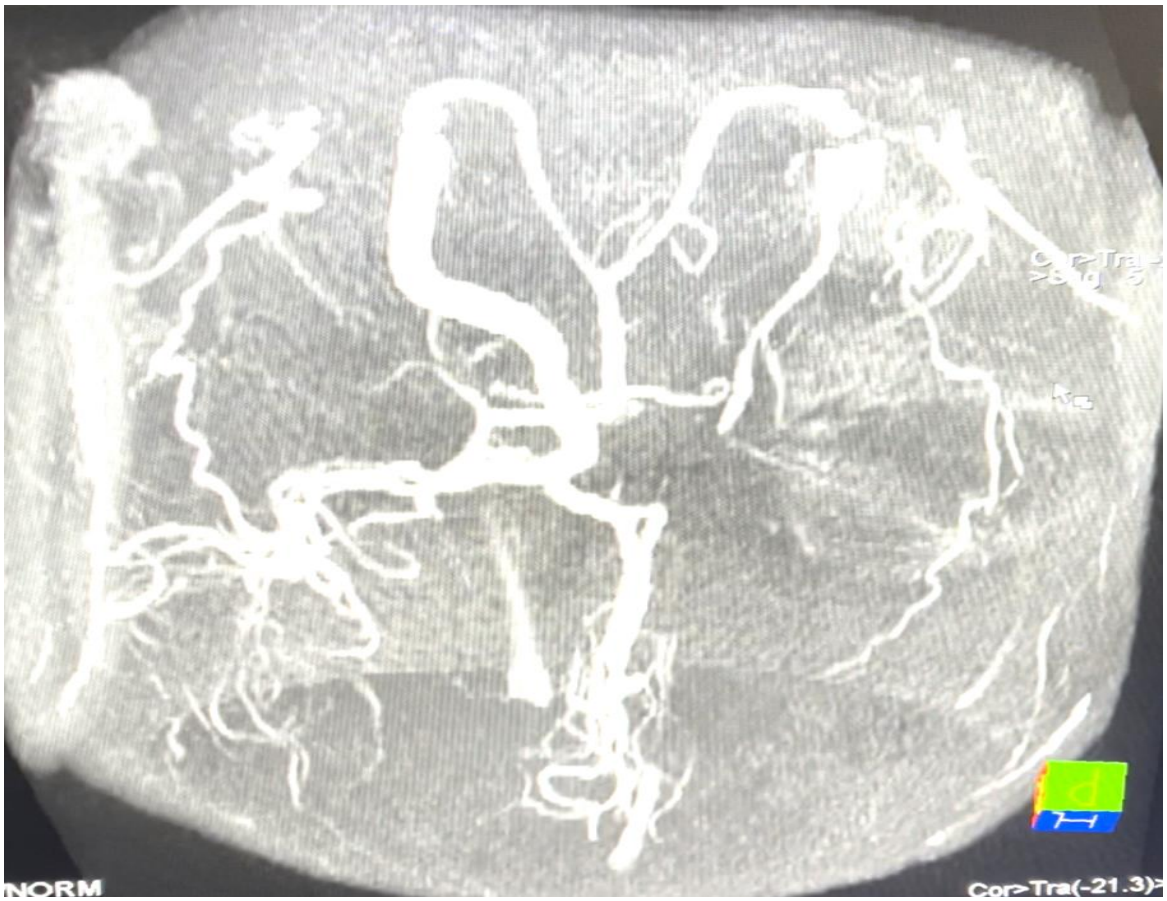

Figure 8 - MRA suggestive of narrowed left internal carotid artery (ICA), A1 segment of anterior carotid artery (ACA) and M1 segment of middle carotid artery (MCA)

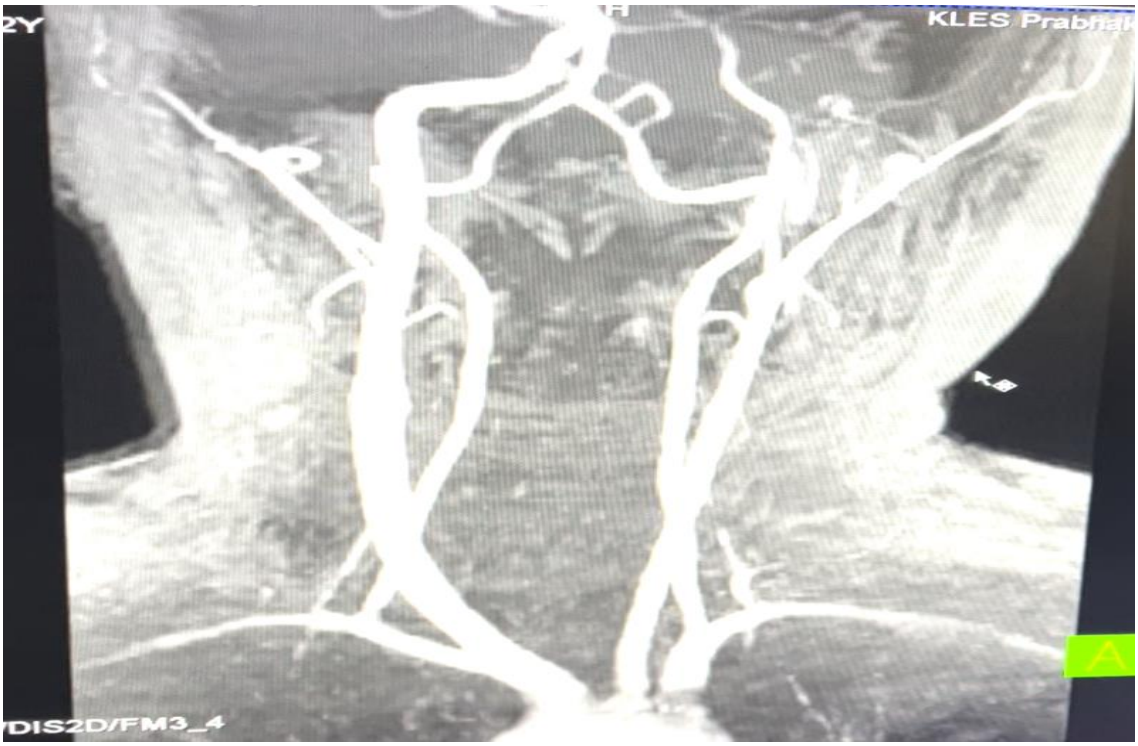

Figure 9 - MRA suggestive of narrowed left internal carotid artery (ICA), A1 segment of anterior carotid artery (ACA) and M1 segment of middle carotid artery (MCA)

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