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Neurology Case Series

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Dear Doctor,

Greetings from Micro Labs Limited.

We are happy to bring you "Neurology case series", which contains latest and interesting cases in the field of Neurology.

This issue contains

1. **A Case on Bilateral Moyamoya Disease with a Long-Lasting Unruptured Large Intracranial Aneurysm.**
2. **A Rare Case report on Bitter Vertigo.**
3. **A Case Report on the Effectiveness of Swallowing Pressure Assessment in the Diagnosis of Mild Pharyngeal Weakness in Myasthenia Gravis.**
4. **A case report on moyamoya and Turner syndrome in a patient with type 2 spinocerebellar ataxia.**

Should you require any specific article or medical information, please feel free to contact us at medicalservices@microlabs.in

Looking forward to your valuable feedback

Best Regards,

Medical Team,

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1. A Case on Bilateral Moyamoya Disease with a Long-Lasting Unruptured Large Intracranial Aneurysm

Introduction:

An isolated chronic vasculopathy with an unknown cause, moyamoya disease typically affects both sides of the body. Internal carotid artery (ICA) and circle of Willis terminal intracranial section gradual narrowing are its defining features.[1] Patients with moyamoya disease have a greater risk of intracranial aneurysm rupture than the general population.[2] This example of confirmed moyamoya disease features bilateral middle cerebral artery (MCA) blockage with a large, persistent aneurysm.

Case presentation:

This is a case of a patient who had moyamoya disease with a large aneurysm, but aneurysmal rupture did not occur even after 7 years

A 71-year-old woman who had an intracranial aneurysm presented. After experiencing paralysis in the left upper and lower extremities two months prior, the patient was initially identified as having an acute ischemic stroke in the right periventricular white matter and centrum semiovale at another hospital and was given aspirin. Mild dysarthria and Grade IV (Medical Research Council) left upper and lower extremities muscle weakness were seen upon admission. A big 11×11mm nonruptured cerebral aneurysm was found in the A3 segment of the left anterior cerebral artery and total blockage in the bilateral proximal MCA (M1) were both seen on the brain magnetic resonance imaging (MRI) scan (ACA). On the cerebral perfusion MRI, perfusion delay was seen in both MCA regions. The patient was found to have Suzuki grade VI moyamoya disease on transfemoral cerebral angiography, with bilateral MCA blockage. A brain MRI was carried out to monitor the cerebral aneurysm after 7 years. The aneurysm got bigger, measuring 17×13mm, but it didn't rupture.

Discussion:

The pathophysiological similarities between intracranial aneurysm and moyamoya illness may explain the high frequency of intracranial aneurysm in moyamoya patients. The intracranial aneurysm has pathological characteristics such as decreased mesothelial thickness and structural abnormalities. The vascular endothelium is destroyed and the lumen is constricted in moyamoya disease, which is caused by hyperproliferative vascular smooth muscle cells that infiltrate the endothelium.[2] The internal elastic lamina is destroyed by aneurysms and moyamoya disease, which weakens the vascular mesothelium and causes it to become fibrous. Recently, the fragility caused by wall shear stress was further exacerbated by moyamoya disease's dysregulation of the peri-endothelial matrix.[3]

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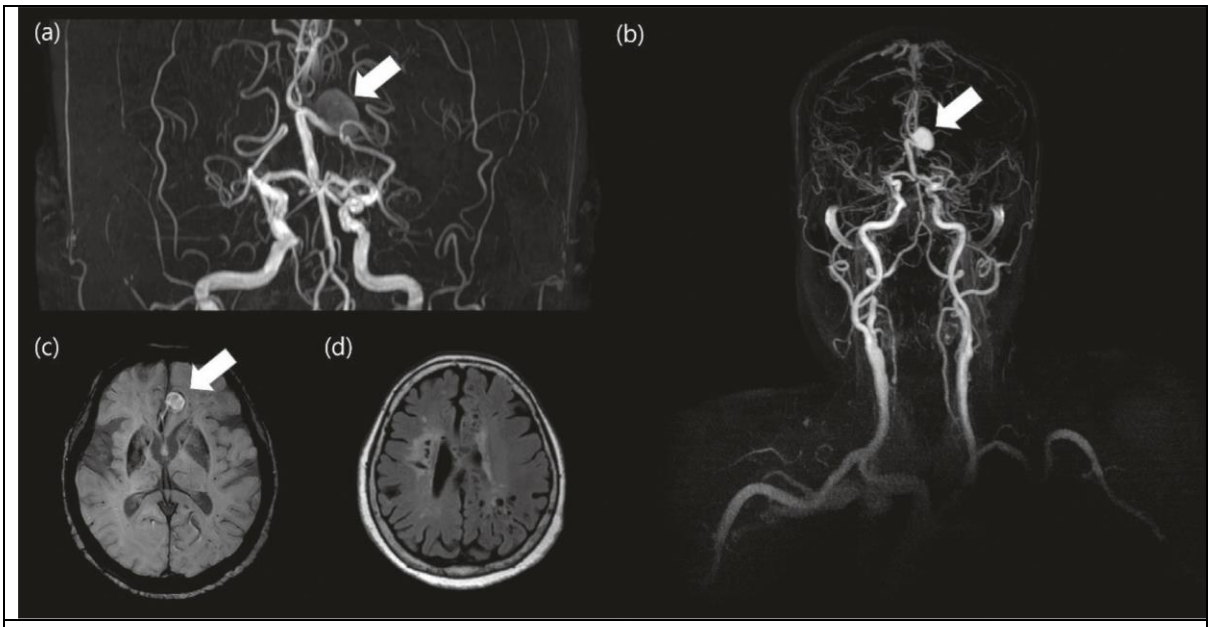

The brain MR images in moyamoya disease patients with a large intracranial aneurysm. (a) Brain TOF-MRA presenting a bilateral middle cerebral artery occlusion with a large aneurysm (arrow). (b) Brain MRA with gadolinium enhancement in the same patient with a large aneurysm (arrow). (c) SWI image presenting with large intracranial aneurysms (arrow). (d) FLAIR image showing multiple old ischemic lesions in the parenchyma.

Conclusion:

Despite having a large aneurysm, the patient with moyamoya disease experienced no aneurysm rupture for a considerable amount of time. Numerous factors may be involved in aneurysm rupture in moyamoya patients, according to earlier studies. This research will aid in understanding how brain aneurysms form and burst in moyamoya patients.

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2. A rare case report on Bitter Vertigo

Introduction:

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Vertigo episodes can be caused by a wide variety of factors, while episodes of taste changes can be brought on by just a small number of factors.[1] Changes in taste can be caused by a number of frequent factors, such as chemotherapy or infections, but paroxysmal dysgeusia has rarely been documented. In this case, the patient experienced recurring short-lasting vertigo attacks along with concurrent episodes of recurrent paroxysmal dysgeusia and abnormal feeling on the left side of the face.[2] The symptoms were brought on by compression of the facial and vestibulocochlear nerves as a result of basilar artery dolichoectasia.[2]

In this instance, the vestibulocochlear and facial nerves were compressed by an elongated basilar artery, resulting in a rare triad of vestibular, unilateral facial sensory, and gustatory symptoms.

Case Presentation:

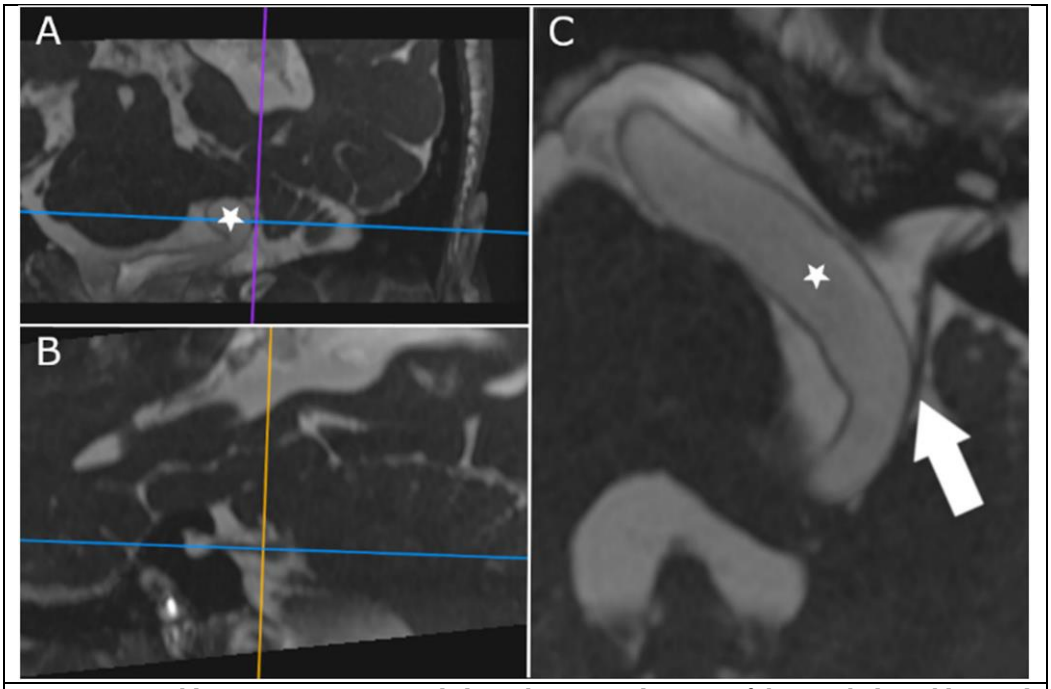
A 71-year-old patient arrived at the hospital with a two-year history of recurrent, admittedly short episodes of spinning vertigo. Each episode began with a change in the left side of the face's sensation, then a bitter taste on the left half of the tongue, and finally up to 10 to 15 seconds of vertigo. He could have 80 attacks a day, which was a high attack frequency.

The patient visited the neurological outpatient clinic as part of the regular clinical practise. He performed a thorough neurological, neuro-otological, and neuro-ophthalmological evaluation, which included a video head impulse test, caloric irrigation, ocular- and vestibular-evoked myogenic potentials, acoustic-evoked potentials, a neuroorthoptic examination, cranial MRI, and MR angiography. A right-sided horizontal headshaking nystagmus was discovered during the neurological evaluation. Between the attacks, there were no indications of any lingering sensory, motor, or taste abnormalities. The vestibulo-ocular reflex (VOR) gain was shown to have decreased in the video head impulse test, and the caloric testing revealed lower excitability on the left side. On the left side, both the amplitudes of the cervical vestibular-evoked myogenic potentials and the amplitudes of the ocular vestibular-evoked myogenic potentials were decreased. Acoustic evoked potentials appeared to be normal. An elongated basilar artery leading to an indentation of the facial and vestibulocochlear nerves on the left side of the brain was visible on an MRI scan. The trigeminal and glossopharyngeal nerves and the elongated basilar artery were not visible in contact on the MRI. Over the course of approximately 5 months, the treatment with lacosamide, initially 50 mg in the evening and thereafter 200 mg orally each day, resulted in a considerable and steady decrease in the attack intensity and frequency from 80 to 1–2 attacks each day. After the six-month follow-up visit, the patient requested that the drug be withdrawn. After four years, a follow-up appointment revealed 5–10 attacks each day without medication. The caloric testing demonstrated progressive reduced excitability on the left side and a lower gain of the VOR during the follow-up after 4 years in the video head impulse test. Both sides had low ocular vestibular-evoked myogenic potential amplitudes, and the left side's cervical vestibular-evoked myogenic potentials were also diminished.

Discussion:

The prevalence of basilar artery dolichoectasia ranges from 0.8 to 4% , with greater values seen in elderly males, as in this instance. [3] With an incidence of up to 17% in patients with posterior circulation stroke and those over 45, intracranial arterial dolichoectasia in those with stroke is well-known. In addition to hemifacial spasm, trigeminal neuralgia, vestibular paroxysmia, or a

combination of these symptoms, a dolichoectatic artery can cause compression of cranial nerves.[2] Here, researchers describe a seemingly uncommon trio of unilateral facial sensory, gustatory, and vestibular symptoms brought on by compression of thetwo cranial nerves, facial and vestibulocochlear nerves.



CISS-MRI in oblique axis reconstructed along the cisternal course of the vestibulocochlear and facial nerves (the horizontal line in (A,B) depicts the axis of (C); vertical lines delineate the axis of the other planes, (A,B), respectively; lines are centered on contact point of the vestibulocochlear nerve and the basilar artery). The dilated and elongated basilar artery (star) displaces the nerves, and direct contact is shown (arrow)

Conclusion:

The vestibulocochlear and facial nerves are compressed by an enlarged basilar artery in a patient who exhibits a rare trio of vestibular, unilateral facial sensory, and gustatory symptoms in recurrent short episodes. Treatment with lacosamide 200 mg per day p.o. resulted in a significantly lower frequency and intensity of attacks in this patient, which is consistent with the theory on ephatic discharges. This case report suggests that sodium channel blockers may also be a novel therapeutic choice for individuals with paroxysmal dysgeusia.

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3.A Case Report on the Effectiveness of Swallowing Pressure Assessment in the Diagnosis of Mild Pharyngeal Weakness in Myasthenia Gravis

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Introduction:

The most prevalent condition affecting the neuromuscular junction (NMJ) of the skeletal muscles is myasthenia gravis (MG). Weakness is the usual presentation. Usually, the muscles in the eyes, throat, and extremities are involved. Muscle weakness results from the diminished electrical impulse transmission across the neuromuscular junction caused by the development of autoantibodies against the particular postsynaptic membrane proteins.[1] Pharyngeal pressure is precisely measured using high-resolution manometry (HRM). Researchers proposed that the use of HRM during the edrophonium chloride (EC) test to measure swallowing pressure could detect mild bulbar symptoms in the absence of abnormalities on videofluorographic (VF) and videoendoscopic (VE) examinations of swallowing.[2] In this case report, a patient with mild MG dysphagia is shown how pharyngeal pressure fluctuates before and after receiving an EC injection using HRM.

Swallowing pressure assessment during the edrophonium chloride test may facilitate identification of very mild bulbar symptoms in patients with myasthenia gravis.

Case Presentation:

The diagnosis of ocular MG in a woman aged 72 pharyngeal discomfort that lasted for three months eight years ago. Manual muscle testing revealed a grade 4 level of neck flexor strength. There were no signs of vision issues or weakness in the extremities. Being administered pyridostigmine, the patient's condition was stable. A higher amount (22.0 nmol/L) of antiacetylcholine receptor antibodies were observed. Salivary pooling was not visible on VE. In spite of the fact that VF was conducted, no unusual findings—such as pharyngeal residue or aspiration—were found. Clinical Classification IIb for Myasthenia Gravis by the American Myasthenia Gravis Foundation. An HRM was used to measure the pressure felt when swallowing along the pharynx and upper esophageal sphincter (UES) (Unisensor AG). The velopharyngeal contractile integral (VPCI), meso-hypopharyngeal contractile integral (MHPCI), relaxation time in the UES, and nadir pressure in the UES were examined as HRM parameters. A parameter used to assess the pharyngeal swallowing pressure is the pharyngeal contractile integral (CI) (mm Hgcms). The amplitude of the contraction, its duration, and its length were multiplied by 20 mm Hg to determine the CI. The "vigor" of pharyngeal contraction is measured by this parameter. For three swallows of 3 mL of water, the HRM parameters were evaluated. These assessments were carried out before and after the intravenous administration of normal saline, 2 mg EC, and 8 mg EC. After injections of normal saline, the VPCI and MHPCI did not rise, but their mean values did after injections of EC. Both the VPCI (78.0±5.4 vs. 134.7±1.3 mm Hg cms) and the MHPCI (130.6±1.5 vs. 284.2±11.9 mm Hg cms) values were greater after the 8 mg EC injection. After the EC injection, neither the UES relaxation time nor the UES nadir pressure changed. A thymoma that had not been seen during earlier examinations was discovered during a chest CT scan. Researchers identified thymoma-associated MG as the cause of the transition from ocular MG to generalised MG. The

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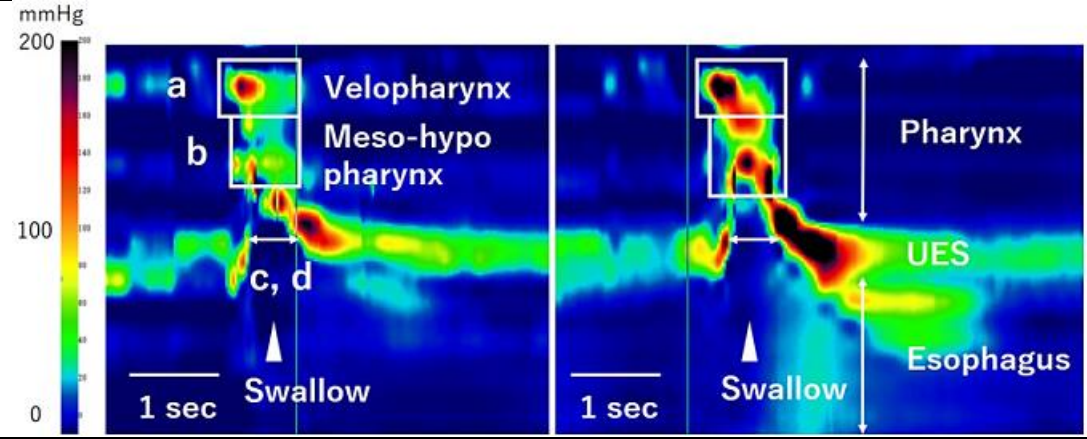
slight dysphagia and cervical muscular weakness were addressed by intravenous immunoglobulin therapy, and a thymectomy was done. Pathological analysis of the lesion (size: 4.0×1.4×3) revealed a type B2 thymoma.



Pressure topography of the present case of a patient with dysphagia before (left) and after (right) the EC test. Spatiotemporal plots of liquid swallows. y-axis, catheter position; x-axis, time. Pressure is indicated by the color scale; black or red for high pressure and blue for low pressure. Velopharyngeal pressure and meso-hypopharyngeal pressure were measured at the velopharynx (a) and meso-hypopharynx (b), respectively. UES relaxation duration and UES nadir pressure were evaluated at (c, d)

Discussion:

This is the first case of MG with mild bulbar symptoms discovered as a result of elevated pharyngeal pressure during the EC test utilising HRM. The only bulbar symptom in this case was a very minor sensation of discomfort in the pharynx.[2] Lower pharyngeal clearance is found in MG patients who have trouble swallowing. Clinically speaking, bulbar MG is difficult to diagnose.[3] Mild pharyngeal weakness, which is challenging to diagnose with traditional standard assessments, such as VE and VF, may be identified by measuring swallowing pressure during an EC test with an HRM.

Conclusion:

The study's findings suggest that measuring swallowing pressure during the edrophonium chloride test may help doctors identify very mild bulbar symptoms in Myasthenia gravis patients.

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4. A case report on moyamoya and Turner syndrome in a patient with type 2 spinocerebellar ataxia

A fully or partially missing X chromosome is the cause of Turner syndrome (TS), a rare disorder that affects 1 in 2500 girls. It is characterised by hypergonadotropic hypogonadism, infertility, short stature, endocrine and metabolic abnormalities, as well as other diseases.[1] TS raises the risk of autoimmune diseases. The most prevalent are autoimmune thyroid conditions like Graves' disease (GD), which is characterised by circulating autoantibodies to the thyroid-stimulating hormone receptor and results in a hyperthyroid condition. Internal carotid artery (ICA) and circle of Willis terminal intracranial portion progressive narrowing characterise Moyamoya Disease(MMD), an isolated chronic, typically bilateral, vasculopathy of unknown cause.[2] A CAG repeat expansion in ATXN2 is the cause of type 2 spinocerebellar ataxia (SCA2), an autosomal dominant cerebellar ataxia. The patient in this case had previously been diagnosed with TS and GD, tested positive for SCA2, and had imaging results that were consistent with an overlap of SCA2 and Moyamoya.

Due to the increased accessibility of genetic testing techniques, doctors must be aware of the possibility that a single patient may have two or more rare diseases.

Case Presentation:

A 43-year-old woman presented with a 2-year history of mild gait imbalance. She was able to walk alone and without assistance, and she didn't record any falls. On neurological examination, there was ataxic gait, difficulty to assume tandem stance, and bilateral proptosis with ophthalmoparesis. Hoffmann and Babinski signs were present, and bilateral deep tendon reflexes were overactive.The horizontal and vertical saccades were slow and dysmetric, and the gaze-evoked nystagmus was mild. Three years prior, she had received diagnoses for Graves disease, premature ovarian failure, and anxiety disorder in addition to her Turner Syndrome (45,X) diagnosis. Spinocerebellar ataxia type 2 (SCA2) was present in her family history. Based on clinical signs and family history, SCA2 was considered to be the cause, thus a brain MRI and polymerase chain reaction(PCR) genetic tests for SCA2 were requested . Brain MRI results showed atrophy of the pons, middle cerebellar peduncles, and cerebellum, as well as severe bilateral frontal and parietal cystic encephalomalacia in watershed zones. A 36 CAG (normal 32) repeat expanded allele in ATXN2 was discovered through molecular analysis, supporting the diagnosis of SCA2. Researchers believed that SCA2 may explain cerebellar and brainstem atrophy, but they did not think that the bilateral chronic watershed infarcts were related to this illness, therefore requested magnetic resonance angiography (MRA) to look into the pathophysiology of the supposedly vascular lesions. The MRA revealed a diagnosis for the

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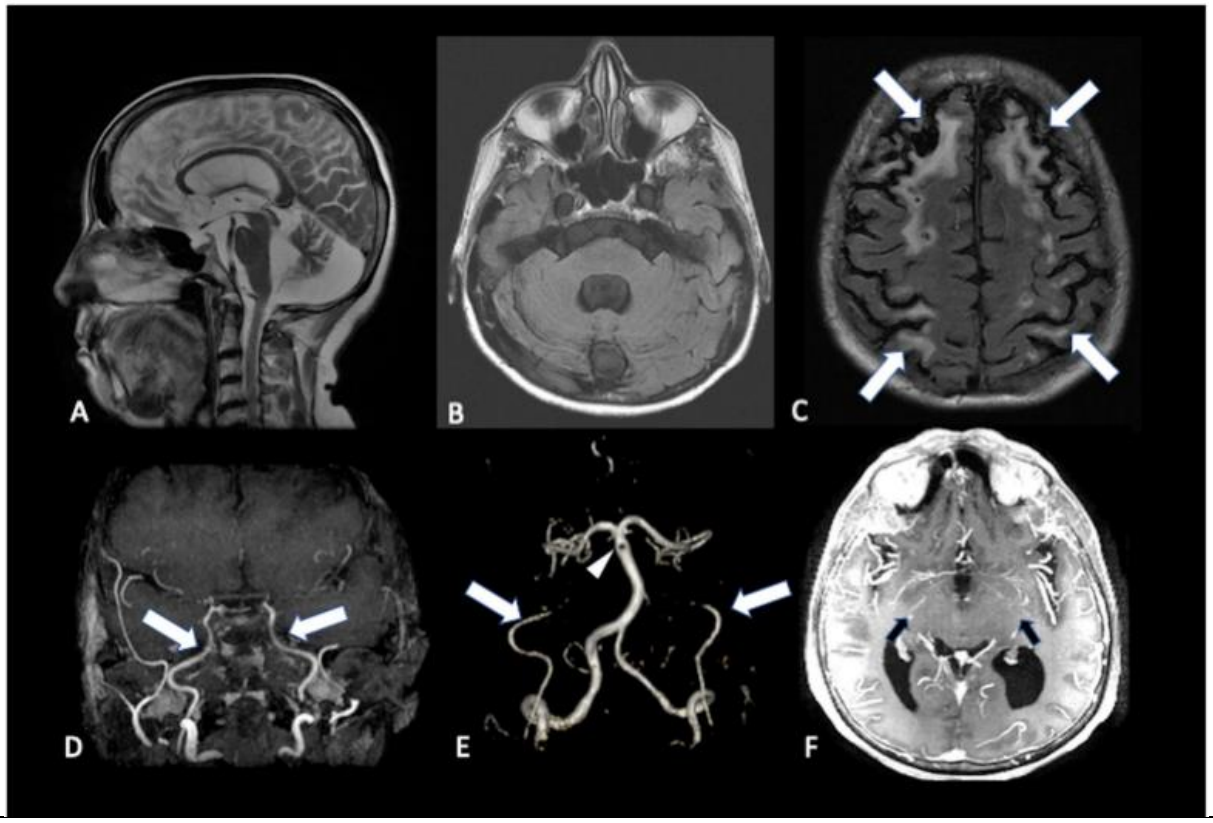
bilateral watershed infarcts: increasing stenosis of both internal carotid arteries with lenticulostriate collaterals, suggestive of Moyamoya disease.

Discussion:

TS, GD, Moyamoya, and SCA2 are four rare disorders that have never been reported together. This patient had four uncommon disorders at the same time. SCA2 prevalence has been calculated to be 1.5/100,000, Turner syndrome incidence to be 5.5/100.000 live births, and Moyamoya incidence to be 0.035/100.000.[3] The annual incidence of Graves disease is 20–50/100,000, which is higher in TS patients.[1] However, it would be extremely uncommon for these four diseases to co-occur independently in the same patient. It is likely that a common physiopathological mechanism connects at least some of them.[1]



Sagittal T2 (A) and axial FLAIR (B) images showing cerebellar atrophy and (C) chronic watershed zone infarcts (arrows). MR angiography reveals progressive bilateral intracranial carotid narrowing, followed by occlusion in supraclinoid segments (D), absence of fow signal in both supraclinoid carotid segments (E) and abnormal net-like vessels resembling a "puf of smoke" (F)

Conclusion:

With the increased accessibility of genetic diagnosis tools, such as commercial spinocerebellar ataxia panels and Next-Generation Sequencing (NGS), doctors must be aware of the possibility

that a single patient may have two or more rare diseases, which may or may not share the same mutation, as was the case in this patient.

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