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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 study on Hereditary spastic paraplegia (SPG 48) with hearing loss and Azoospermia**
2. **A Case Report of a Mucormycosis patient with a Silent Cerebral Abscess.**
3. **A case report of paradoxical contralateral hemiparesis due to a spontaneous spinal epidural hematoma**
4. **A Case of Late-Onset Idiopathic Aqueductal Stenosis (LIAS) With Olfactory Dysfunction and Gait Disorder**

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 study on Hereditary spastic paraplegia (SPG 48) with hearing loss and azoospermia

Introduction:

Hereditary spastic paraplegia (HSP), a rare neurological condition, is spasticity in the lower extremities. The corticospinal pathways in the lateral region of the cervical and thoracic spinal cord frequently degenerate in HSP, which is classified based on genetics, clinical characteristics, and the process of molecular pathogenesis.[1] A rare genotype known as SPG48 is characterised by mutations in the gene AP5Z1, which is involved in intracellular membrane tracking.[2] The case of a 53-year-old male patient with SPG48 who had spastic paraplegia, infertility, hearing loss, cognitive impairments, and peripheral neuropathy is described in this paper.

Clinicians should be aware of non-neurological signs when there are suspected SGP48 cases because early diagnosis will reduce the need for unnecessary tests and treatments.

Case Presentation:

A 53-year-old man with distal weakness in his lower limbs and developing walking difficulties was hospitalised to our hospital in April 2023. He struggled academically in primary school and dropped out. At the age of 28, he got married. He was 35 years old when he was given the infertility diagnosis. When walking in 2013, the patient felt unsteady and somewhat disturbed. In 2018, when the patient reported having trouble rising from a sitting position, walking instability grew worse. The patient began using crutches to walk in 2020. Hearing ability has gradually declined in both ears since 2020 in both ears. Since 2021, the patient has no ear pain, infection, or tinnitus and can only recognise noises when spoken aloud. During the examination, researchers noted bilateral pyramidal symptoms, ankle clonuses (+), tendon hyperreflexia, and spastic gait on both sides. Romberg's test came up positive. There was also a slight spastic dysarthria to be seen. A sensory examination revealed diminished pinprick feelings in the lower limbs and lack of vibratory sensations at the great toes. The SPRS gave a score of 19 for spastic paraplegia. While orientation in time and space was normal. Both ears had hearing loss, and the Rinne tests on the left (+) and right (±) ears were positive. The right ear was favourably evaluated in the Weber test. The testis' appearance and size were both normal. Systemic hormone levels in the cerebrospinal fluid and blood tests were both normal. On fluid-attenuated inversion recovery (FLAIR) sequences, brain magnetic resonance imaging (MRI) revealed mild brain atrophy with periventricular white matter hyperintensities. A cervical and lumbar MRI revealed degenerative abnormalities in the lumbar spine and modest intervertebral disc distension, but no severe spinal cord compression or other aberrant signals. The left tibial nerve and the right common peroneal nerve both displayed lower amplitudes of compound muscle action potentials (CMAPs) throughout the electromyography test. Bilateral sural nerves showed a longer delay of sensory nerve action potentials (SNAPs), and both median and tibial nerves showed an increased latency of the F wave. The results of the audiometry

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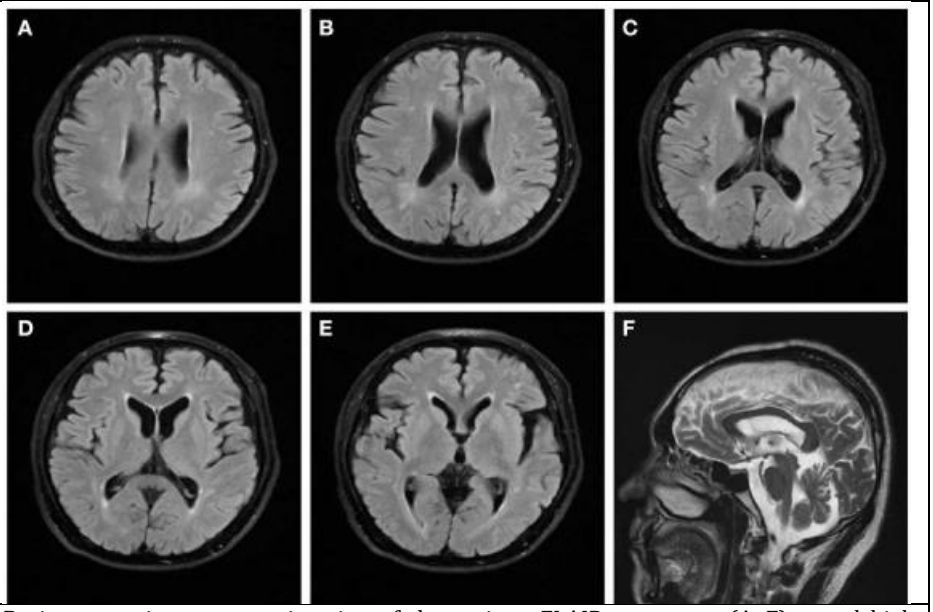
test revealed sensorineural deafness on the left side and mixed deafness on the right. An elevated binaural auditory threshold was indicated by the evoked potentials. Bilateral extended waves I and III on the auditory evoked potentials in the brain stem were discovered. Somatosensory evoked cortical potentials also revealed a longer left P40 latency. The scores of the cognitive tests were 18 on the Montreal Cognitive Assessment (MoCA) and 27 on the Mini Mental State Examination (MMSE). By using Sanger sequencing, we were able to pinpoint the exact position of the gene deletion. We discovered homozygous deletions in the exon 10 of the AP5Z1 gene, NM_014855.3, c.1133-345_1311 + 249del, p.G378Vfs 93X, in this region . Researchers discovered that the deletion results in the early end of amino acid production using the software Mutalyzer 2.0.35. The heterozygous mutations in the patient's asymptomatic brothers were discovered by fluorescence quantitative polymerase chain reaction (PCR). The standards and recommendations of the American College of Medical Genetics and Genomics classify this deletion mutation as a harmful variant. Researchers identified the patient as having SPG48 based on clinical characteristics, imaging results, and genetic anomalies. His symptoms improved by taking baclofen and tizanidine orally.



Brain magnetic resonance imaging of the patient. FLAIR sequences (A–E) reveal high signal changes in the periventricular white matter and mild brain atrophy. T2-weighted (F) sequence shows that the corpus callosum is normal.

Discussion:

This patient reported azoospermia and significant bilateral hearing loss, as well as spastic paraplegia, peripheral neuropathy, and minor cognitive impairment. Previous research indicated that HSP neurons had significantly reduced mitochondrial length and density, indicating that aberrant mitochondrial morphology may be a factor in axonal abnormalities.[3] In SPG11 and SPG48 neurons, axonal degeneration caused by poor mitochondrial dynamics led to shorter and less effective axons, according to a recent study. Human sperm morphology and motility are also correlated with mitochondrial function.[4] Deafness has also been linked to a reduction in mitochondrial function. Therefore, it is possible to hypothesise that AP5Z1 gene alterations may have an impact on both hearing and spermatogenesis.[2]

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Conclusion:

SPG48 has a significant negative effect on the central nervous system and exhibits complicated and variable radiological and clinical features. Clinicians should be mindful of non-neurological signs when there are suspected SGP48 cases present because early diagnosis will reduce the need for unnecessary tests and treatments. There is currently no specific treatment for SPG48, although symptomatic and supportive therapies like baclofen and tizanidine can help with urine incontinence and spastic paraplegia symptoms.

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2. A Case Report of a Mucormycosis patient with a Silent Cerebral Abscess

Introduction:

One of the most important public health crises of the century is acute respiratory syndrome, which is brought on by the SARSCoV-2 virus. Since the disease's emergence, numerous treatments have been proposed; nonetheless, prevention and symptomatic care are thought to be the primary forms of treatment.[1] Infection with mucormycosis is one of the severe issues brought on by this illness. It is crucial to diagnose and treat brain abscess as soon as possible because it may have a very terrible prognosis if it is linked to certain disorders.[2] In this paper, Researchers describe a middle-aged diabetic man who had rhino cerebral mucormycosis and required corticosteroid treatment because of COVID-19.

Brain abscess is a relatively uncommon but serious complication of mucormycosis that should be taken into account even if the patient shows no neurological symptoms.

Case Presentation:

A 48-year-old man with anosmia, palate paraesthesia, and signs of chronic rhinosinusitis was hospitalised to a tertiary referral hospital. The patient was diabetic, had hyperlipidemia for 12 years, and was taking glibenclamide in addition to metformin. After being admitted, he received insulin therapy. Other symptoms were proptosis and slight blurring of vision, but no chemosis was visible. He had had Covid-19 infection two

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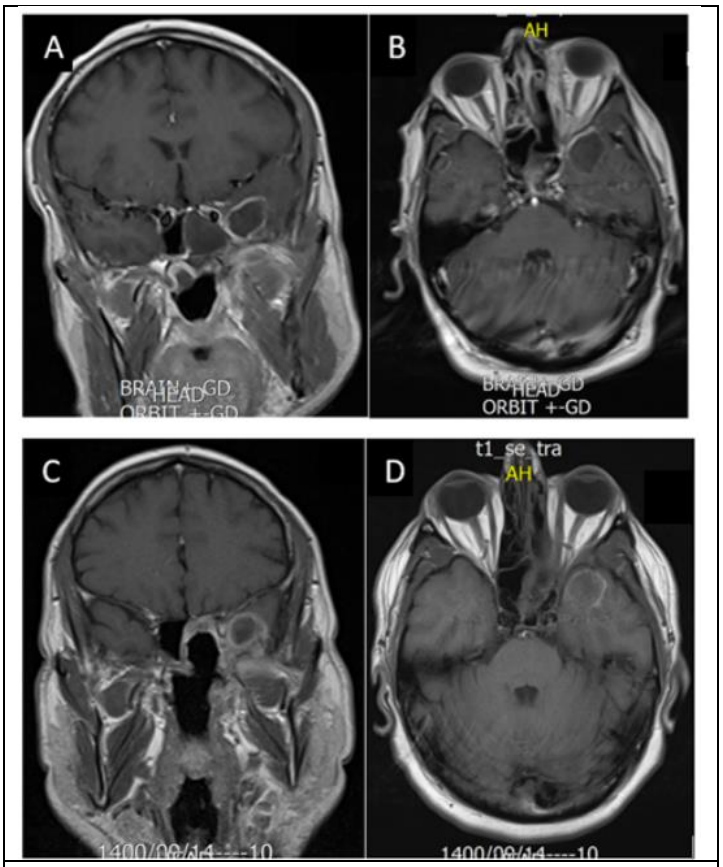
weeks prior, which was treated with dexamethasone and antiviral medications. The patient was conscious and had normal vital signs on the day of admission. Anterior rhinoscopy revealed mucopurulent fluids and necrotic tissue in the left middle concha. On the day of admission, the patient underwent endoscopic sinus surgery, and necrotic tissues were surgically debrided. Evidence of necrotic tissue with mucormycosis was found in the biopsy specimen report. The patient then underwent many endoscopic sinus surgeries to treat the illness. Additionally, because the patient's eyes were affected, the retrobulbar amphotericin was administered three times, on days 4, 7, and 9 following admission. An MRI of the brain was done because the eyes were involved, and the results revealed that the temporal lobe of the brain had an abscess. Vancomycin, cefepime, and metronidazole were therefore started for the patient. He was not a candidate for surgery, according to the neurosurgical consultation, because the abscess' size had reduced. After treatment, the patient's edoema and abscess size significantly decreased. Finally, three months after admission, the patient was discharged in good general health.



Low signal focal point in T1 with restriction and ring enhancement in anteromedial of left temporal lobe associated with vasogenic edema: A) coronal section before treatment, B) Axial section before treatment, C) Coronal section after treatment, and D)Axial section after treatment

Discussion:

A COVID-19 infection alone results in severe lung illness, interstitial alveolar destruction, and an elevated risk of invasive fungal infections. The COVID-19 virus can also interfere with immunological control, which includes a decrease in T cells, which impairs the immune system.[3] Due to the aggressive nature of mucormycosis, significant morbidity or fatality may readily result from delayed diagnosis and inadequate treatment. Early detection of mucormycosis is crucial for proper treatment and a better prognosis for the patient due to predisposing circumstances like those indicated above.[2]

Conclusion:

Although rare, the mucormycosis complication known as brain abscess has a significant mortality rate, hence patients with mucormycosis should be evaluated for it even if there are no neurological symptoms.

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3. A case report of Paradoxical Contralateral Hemiparesis due to a Spontaneous Spinal Epidural Hematoma

Introduction:

A spontaneous spinal epidural hematoma (SSEH) that is linked with hemiparesis typically develops ipsilateral to the hematoma.[1] According to the spinal levels and degree of cord or nerve root compression, the symptoms of spontaneous spinal epidural hematoma (SSEH) might range from mild neck or back pain to hemiparesis, total paraplegia, or quadriplegia.[1,2] Here, researchers describe an unusual SSEH case with sensory and motor impairments in the upper and lower limbs contralateral to the lesion.

Paradoxical contralateral hemiparesis is one of the potential presenting symptoms in patients with spontaneous spinal epidural hematoma.

Case Presentation:

A 70-year-old woman who had left hemiparesis and acute-onset neck pain arrived at the emergency ward. Without any apparent cause, these symptoms would occur when she was at rest, and turning her neck would make them worse. She had no recent trauma in her past. Her prescriptions contained antihypertensive drugs; antithrombotic drugs weren't listed. She presented with a blood pressure of 230/129 mmHg. She had left-sided hemiparesis (manual muscle testing results in upper and lower limbs of 3/5), left-side hypoesthesia and numbness, and no face involvement, according to the physical examination. Laboratory tests turned up nothing particularly noteworthy, including coagulopathy. To exclude stroke or vertebrobasilar artery dissection, brain MRI and magnetic resonance angiography were conducted. An epidural hematoma at the C2 to C3 level of the dorsolateral spinal cord was identified by cervical MRI. A well-defined crescent hematoma and lateral spinal cord displacement were seen on the right side, contralateral to the hemiparesis, using axial fluid-attenuated inversion recovery (FLAIR) imaging. Angiography of the spine did

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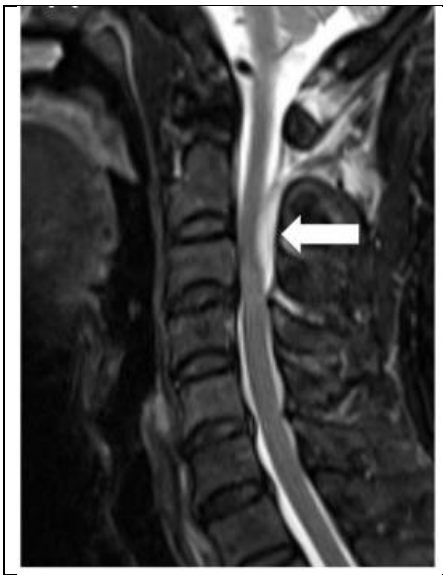
not show any unusual arteries. Researchers identified SSEH based on her clinical profile and MRI results. She had conservative treatment with a rigid cervical collar and hypertension medication, her upper and lower limb symptoms showed a significant improvement within three hours of the beginning. Her sensory impairments and left-side weakness were getting better every day. Six days later, a follow-up MRI revealed that the hematoma was reducing in size. Three weeks after the hematoma had totally disappeared the patient had fully recovered, and she had been released with no neurological sequelae.



Sagittal T2-weighted MRI shows a dorsolateral epidural hematoma compressing the spinal cord at the C2 to C3 level

Discussion:

Neurological symptoms from intracranial lesions, typically those that are supratentorial and developing, might occasionally arise. In the days before brain imaging, hemiparesis ipsilateral to an intracranial lesion, also known as paradoxical hemiparesis or false localising signs, sometimes resulted in wrong-site exploratory surgery.[1] Kernohan's notch phenomenon, which is the compression of the contralateral cerebral peduncle against the tentorial edge, vascular involvement of the contralateral hemisphere, or impaired functional activation of the contralateral hemisphere are all compelling candidate mechanisms for paradoxical hemiparesis caused by intracranial lesions.[3] The mechanism of paradoxical contralateral hemiparesis in this case involving a spinal lesion may be comparable to Kernohan's notch type damage.

Conclusion:

Patients with SSEH may present with contralateral hemiparesis as a symptom. In addition to the few prior reports of comparable contralateral hemiparesis linked to spinal compressive injuries, this case demonstrates the uncommon occurrence of direct (ipsilateral) and indirect (contralateral) compression mechanisms resulting in paradoxical hemiparesis.

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4. A Case of Late-Onset Idiopathic Aqueductal Stenosis (LIAS) With Olfactory Dysfunction and Gait Disorder

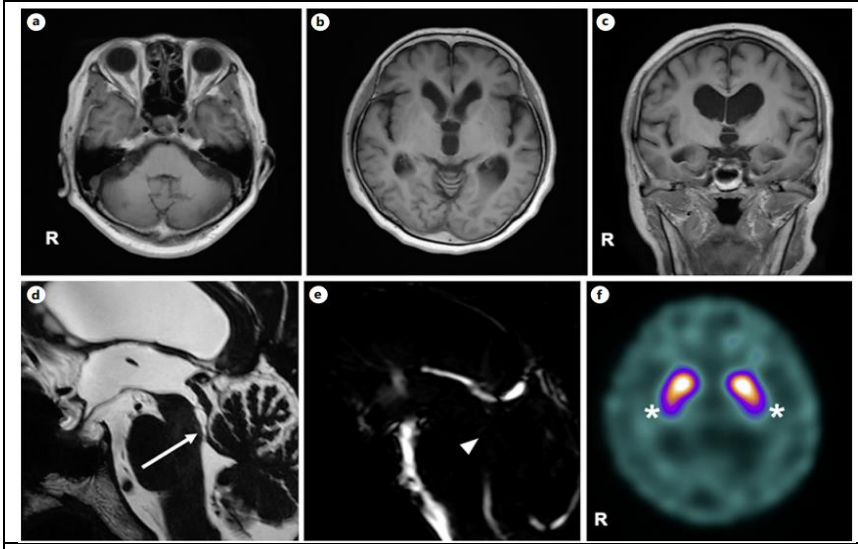
Introduction:

Olfactory dysfunction is characterised by a decreased sensitivity or a challenge in detecting odours. It has been suggested that olfactory function changes are early indicators of neurodegeneration. The two most prevalent neurodegenerative illnesses, Parkinson's disease (PD) and Alzheimer's disease (AD), are among the many that are age-related.[1] Patients who have experienced head trauma, intracranial tumours, or hydrocephalus may also experience olfactory dysfunction, some of which might get better with proper treatment of the underlying condition. Olfactory dysfunction is frequently masked by apparent motor symptoms in clinical practise since few patients complain of smell problems.[2] Researchers present a case of late-onset idiopathic aqueductal stenosis (LIAS) with olfactory dysfunction and gait disorder that significantly improved after neurosurgical treatment.

Olfactory dysfunction is a non-motor symptom of hydrocephalus that is treatable with surgery.

Case Presentation:

A 76-year-old right-handed woman's developing gait disturbance was her main complaint when she was admitted to the hospital. On admission, a neurological evaluation revealed a mildly broad-based, small-stepped gait, mild upper-limb rigidity, and postural instability. Scores on the neuropsychological battery were largely kept unchanged; for example, the mini-mental state test received 29 points, the Montreal cognitive assessment received 26 points, and the frontal assessment battery received 16 points. The odour stick identification test for Japanese (OSIT-J) score has decreased to 2 points (cut-off value 7) even though the patient was ignorant of hyposmia.The lateral and third ventricles of the brain were clearly enlarged on cranial magnetic resonance imaging (MRI), but neither



T1-weighted cranial MRI. a, b The axial images of MRI show an enlarged third and lateral ventricle with normal size of fourth ventricle. No evidence of atypicalor secondary parkinsonism. c Coronal T1-weighted imaging does not have a feature of DESH.d CISS imaging demonstrates the stenosis of the midbrain aqueduct(white arrowhead). e Phase-contrast cine MRI shows no cerebrospinal fluidflow in the same area (white arrow). f 123I-ioflupane single-photon emission computedtomography (DAT scan™) shows slightly reduced uptake in the bilateral, dorsal parts of the striatum (white asterisk

abnormally disproportionately enlarged subarachnoid-space hydrocephalus findings nor imaging indications of atypical/secondary parkinsonism were seen. Aqueductal stenosis caused by aberrant membranous structures was later confirmed by constructive interference in steady-state imaging, which was also accompanied by a nearly total lack of cerebrospinal fluid (CSF) flow revealed on cine MRI. Researchers were able to make the diagnosis of noncommunicating hydrocephalus caused by LIAS due to these distinctive imaging features. Further evidence for concomitant PD is shown by the presence of mild parkinsonism, olfactory impairment, and slightly reduced uptake of striatal dopamine transporter imaging. Endoscopic third ventriculostomy (ETV) was performed instead of ventricular CSF sampling after weighing the risks and benefits of doing the surgery with just one tap. Postoperative MRI demonstrated the third ventricle's floor had been successfully fenestrated, and cine MRI showed CSF flow in the same region. Following surgery, neurological testing revealed a wider stride, improvement in gait, and no evidence of parkinsonism; nonetheless, the patient's posture remained somewhat flexed. Unexpectedly, the surgical procedure improved the motor symptoms and brought the OSIT-J score back to normal (7 points).

Discussion:

Because both patients and doctors are mainly focused on motor and cognitive problems that significantly interfere with everyday living, hydrocephalus is one of the least discussed causes of central olfactory dysfunction.[2] Majority of reports of olfactory impairment linked with adult-onset hydrocephalus are associated with idiopathic normal pressure hydrocephalus, which is a frequent issue in many people.[3,4] The clinical observations, however, show that patients with cognitive decline frequently are not aware of their olfactory dysfunction, most likely as a result of pathological changes in the upper olfactory cortex and/or attention difficulties.[2]

Conclusion:

Hydrocephalus can cause olfactory dysfunction, a non-motor condition that can be treated surgically. Due to the lack of attention given by medical practitioners to hydrocephalus as a potential cause of central olfactory dysfunction, this non-motor symptom may go underdiagnosed. Olfactory function evaluation may be taken into consideration for the functional assessment both before and after the surgical treatment in patients with hydrocephalus, in addition to the motor and cognitive examination.

Olfactory dysfunction is a non-motor symptom of hydrocephalus that is treatable with surgery.

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