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A case of reversible isolated cognition impairment in meningoencephalitis without abnormal MRI findings

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Most of meningoencephalitis can be showed fever, altered mentality, seizure, muscle weakness and abnormal MRI findings. We report a case of meningoencephalitis with reversible isolated cognition impairment without abnormal MRI findings. The patient showed only isolated cognition impairment without fever and there was no evidence of meningoencephalitis on the contrast enhanced MRI. Although the patient revealed CNS infection with unknown cause in CSF profile, it has some possibilities; 1) Asepticmeningoencephalitis with viral infection, 2)Bacterial meningoencephalitis with early stage, 3)Parasitic meningoencephalitis, 4) Autoimmune meningoencephalitis. Because of the negative findings in autoimmune antibody test, autoimmune meningoencephalitis was less likely. However, it was difficult to diagnose whether it was aseptic meningitis or bacterial meningitis with early stage. After treatment with acyclovir 0.6g for 7days, ceftriaxone 4g, vancomycin 2.5g and Ampicillin 8g for 10 days, confusion and mutism were improved and K-MMSE score was recovered to 18 points. Many reports have been published on meningoencephalitis accompanied by fever, altered mentality, seizure, muscle weakness and abnormal MRI findings respectively. However there have been few cases of meningoencephalitis with isolated cognition impairment and normal MRI simultaneously. If it is a change of consciousness and cognition impairment caused by meningoencephalitis, it can be reversible with proper treatment. Therefore, it is necessary to accurate diagnosis and prompt treatment. Although there is only sudden onset isolated CNS symptom with normal brain MR finding, it is essential to check CSF profile.

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Atypical Location of Diffuse Large B-Cell Lymphoma: A Case Report

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Introduction

Primary central nervous system lymphoma occurs in both immunocompetent and immunocompromised individuals, with diffuse large B-cell lymphoma (DLBCL) accounting for approximately 90% of cases. These lymphomas are commonly located in supratentorial regions, particularly the frontal lobe, thalamus, basal ganglia, and corpus callosum. Primary lymphoma of the skull base is a rare manifestation. Patients typically exhibit symptoms resulting from the compression of critical anatomical structures, such as headache, diplopia, and cranial nerve palsies.

Case presentation

A 42-yr-old female patient began with rightward tongue deviation, diplopia, and right eye esotropia. She was diagnosed with sixth cranial nerve palsy, and a brain MRI was requested. The imaging revealed an infiltrative lesion in the clivus with right-sided predominance, ventral and caudal extension, and involvement of the petrous apex and ipsilateral Meckel`s cave. A transsphenoidal resection of the clival lesion was performed, and histopathology reported DLBCL.

Discussion

Primary clivus lymphoma is an uncommon presentation of non-Hodgkin lymphoma affecting the skull base. This condition poses significant diagnostic challenges due to the overlap in symptoms and radiological findings with more common clival lesions, such as chordomas and meningiomas. Clinically, patients often present with progressive headache, diplopia, and sixth cranial nerve palsy, as observed in our case. Abducens nerve palsy serves as an early and crucial indicator, reflecting the anatomical proximity of the clivus to the nerve.

Conclusions

Our case highlights the importance of including lymphoma in the differential diagnosis of infiltrative skull base lesions, particularly when accompanied by cranial nerve involvement and findings on imaging studies.

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Adult-Onset Phenylketonuria Presenting with Neurological Symptoms: A Case Report

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Introduction

Phenylketonuria (PKU) is an autosomal recessive metabolic disorder caused by phenylalanine hydroxylase (PAH) deficiency, leading to phenylalanine accumulation and neurotoxicity. Early diagnosis through neonatal screening and dietary management significantly reduces neurological and systemic complications. While untreated PKU typically manifests in childhood with intellectual disability, seizures, and motor delays, some cases remain asymptomatic or mildly affected until adulthood, when late-onset neurological symptoms may develop. These include hyperreflexia, movement disorders, ataxia, cognitive decline, and behavioral changes.

Case report

We present a 24-year-old woman with a one-year history of progressive gait instability, cognitive impairment, and behavioral changes. Developmental milestones were normal, but academic performance was poor. Over the past year, short-term memory deficits and behavioral disturbances were reported alongside worsening gait impairment.

Brain MRI revealed diffuse, non-enhancing periventricular white matter lesions with restricted diffusion. This radiologic pattern suggested a metabolic leukodystrophy, with adult-onset metachromatic leukodystrophy and phenylketonuria considered as differential diagnoses. Genetic testing identified two heterozygous pathogenic variants in the PAH gene, confirming the diagnosis of autosomal recessive PKU.

Conclusion

Although PKU is predominantly a childhood-onset disorder, late-onset neurological symptoms can occur in untreated or poorly managed cases. MRI findings, biochemical testing, and genetic analysis are critical for diagnosis. This case highlights the importance of lifelong monitoring and management in individuals with PKU to prevent late-onset complications.

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Long term results for deep brain stimulation for tremor recurrence after focused ultrasound thalamotomy

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Background: MRI-guided focused ultrasound (MRgFUS) thalamotomy has demonstrated efficacy in tremor relief. However, tremor recurrence may occur. For tremor recurrence, deep brain stimulation (DBS) presents a viable alternative. This study reports our experience in preforming DBS following MRgFUS.

Methods: We evaluated four right-handed essential tremor (ET) patients with a mean age of 65 years (range 47-76 years); three of them males, with tremor recurrence 5±5.2 months following MRgFUS. DBS was performed 73.6±54.1 months following MRgFUS. Mean follow up time following DBS was 46±14 months. DBS electrodes were bilaterally implanted in the posterior subthalamic area (PSA) in 2 patients, the ventro-intermediate nucleus (VIM) in one patient and in the PSA traversing the VIM in one patient. Tremor was assessed using Clinical Rating Scale for Tremor questionaire (CRST) and hemi-CRST scores for the treated side. Quality of life was assessed using quality of life in ET questionnaire (QUEST).

Results: At last follow-up visit post-DBS (23.0±14.3), the mean total CRST score was non-significantly decreased compared with pre-DBS (37.0±10.4), and was significantly lower compared with mean pre-MRgFUS score (49.0±6.7, p.05). Hemi-CRST on MRgFUS treated side did not differ post-DBS. QUEST scores did not improve. Adverse events following DBS included persistent gait disturbance (n=3), dysarthria (n=2).

Conclusion: Tremor scores following DBS for tremor recurrence after MRgFUS were significantly improved as compared with pre-MRgFUS. We also found lower tremor scores following DBS, compared to the last pre-DBS follow-up but this did not reach statistical significance. The unfavorable adverse events profile should be considered in patient selection.

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Spinal Dural AVF: From Clinical Suspicion to Endovascular Cure - A Case Report

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Introduction: Spinal dural fistulas (SDAVFs) are rare vascular pathologies of the spinal cord, occurring in 5–10 per 1,000,000 individuals. Typically found in the thoracolumbar region (T6–L2), result in venous hypertension and medullary congestion, causing spinal cord dysfunction. Diagnosing SDAVFs is challenging due to their insidious onset and overlapping symptoms with other neurological conditions. Case presentation: A 61-year-old male presenting with progressive difficulty walking, lower limb numbness, and urinary retention. On neurological examination the Aminoff Logue scale was G4 U3. Spinal MRI showed medullary edema (T7–L1) and perimedullary vessels. Medullary angiography confirmed a right-sided dural arteriovenous fistula at L2. Endovascular embolization led to significant clinical improvement, with improvement in motor strength and resolution of urinary symptoms. Aminoff Logue scale G3 U1 on discharge. Discussion: Differential diagnosis included spinal stenosis, discopathy, and polyneuroradiculopathy, supported by elevated cerebrospinal fluid protein. However, the patient’s symptom progression, urinary retention, and MRI findings raised suspicion for SDAVF. Typical MRI features include medullary edema, tortuous perimedullary vessels, and hyperintensities on T2-weighted images, often spanning 5–7 vertebral levels. MR angiography can further refine the diagnosis, though catheter angiography remains the gold standard. Conclusion: Delayed diagnosis, averaging 12–44 months, underscores the importance of early suspicion and advanced imaging in progressive myelopathy cases. This case emphasizes the diagnostic utility of MRI and angiography and highlights the effectiveness of endovascular treatment in improving outcomes for patients with SDAVFs.

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"Carpal Tunnel Syndrome in Patients with End-Stage Renal Disease: A 24-Month Follow-Up Study of 24 Cases"

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Objective: This study aims to investigate the prevalence, clinical progression, and outcomes of Carpal Tunnel Syndrome (CTS) in patients with end-stage renal disease (ESRD) undergoing hemodialysis, over a 24-month period.Methods: A prospective cohort study was conducted involving 24 ESRD patients diagnosed with CTS. Diagnosis was confirmed through clinical assessment and nerve conduction studies. Patient demographics, duration of dialysis, severity of symptoms, and functional status were recorded at baseline. Follow-up assessments were performed at 6, 12, and 24 months, focusing on symptom severity, functional impact, and any interventional treatments received, including surgical release and dialysis access modifications.Results: Of the 24 patients, 16 (66.7%) were male, with an average age of 59 years. The average duration of dialysis prior to CTS diagnosis was 4.2 years. At baseline, 12 patients (50%) reported severe hand pain and functional limitations. Over the 24-month follow-up, 8 patients (33.3%) underwent carpal tunnel release surgery, which resulted in significant symptom improvement. Nerve conduction studies showed a progressive worsening in 6 patients (25%) who did not receive surgical intervention. A statistically significant correlation was found between the duration of dialysis and severity of CTS (p 0.05). No significant improvement was noted in patients managed conservatively without surgical intervention.Conclusion: CTS is a prevalent and progressively debilitating condition in patients with ESRD on long-term hemodialysis. Early diagnosis and intervention, particularly surgical release, are crucial in managing symptoms and improving quality of life. This study underscores the need for routine screening for CTS in this high-risk population and suggests that duration of dialysis is a significant risk factor for its severity.

Keywords: Carpal Tunnel Syndrome, End-Stage Renal Disease, Hemodialysis, Nerve Conduction Studies, Surgical Intervention.

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Diabetic Polyneuropathy and vitamin D, correlations to be set

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Introduction

Vitamin D hypovitaminosis has been recently at the center of large studies, especially in the normal functioning of peripheral nervous system. Correlations between low levels of vitamin D and nerve conduction velocities have been studied and discussed, although findings are not uniform. Painful diabetic neuropathy could be a subgroup deemed of scrutiny, while other parameters could serve as a background.

Methodology

Forty patients (20/20 men-women) with diabetic neuropathy of a certain duration (five years) in an age-controlled group (type II diabetic patients aging 50-60 years old) have been tested with electroneurography and responded to VAS (visual analogue scale) in an anonymized form. Values of plasmatic vitamin D were collected and data were correlated with VAS scores.

Results

Severe vitamin D deficiency (levels 4-13 ng/ml) strongly correlated with VAS scores of six points or more (r 0, 7) but not with the decrease of nerve conduction velocities (averaging NCVs of four motor nerves in the lower extremities). The decrease of NCVs was not uniformly distributed (more apparent in male patients) although VAS scores were highly similar in both gender subgroups.

Conclusions

While considering the severity of painful diabetic neuropathy, a diversity of parameters should be taken into account, such as comorbidities, therapeutic compliance, glycemic equilibrium, nutritional status here including any vitamin deficiency. A routine vitamin D check could be helpful and may be included in the diagnostic workup.

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POEMS - A case of plasma cell dyscrasia disguised as CIDP

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POEMS (polyradiculoneuropathy, organomegaly, endocrinopathy, M-protein, and skin changes) is a paraneoplastic syndrome associated with plasma cell disorders. The disease is rare, with an estimated prevalence of 0.3 per 100,000.

A 41 years old female patient, without known prior diseases, is admitted for symptoms progressing over a year consisting of weakness in the distal part of the limbs and painful paresthesias. On examination: symmetric flaccid tetraparesis more severe distally, tactile hypoesthesia, impaired proprioception and painful paresthesias in the lower limbs. On inspection erythema of the face and acrocyanosis were noted.
EMG revealed a severe axonal and demyelinating polyneuropathy with conduction blocks in all four limbs, predominantly in the legs.
Initial lab work revealed a high IgA and mild thrombocytosis. CSF analysis showed albuminocytologic dissociation. A course of IV immunoglobulin followed by rituximab showed no benefit. Extensive follow-up testing revealed monoclonal IgA bands and lambda chains, with highly elevated VEGF (2000 pg/mL). A medullary biopsy showed granulocytic hyperplasia and elevated plasmocytes, with an abnormal kappa/lambda ratio. Imaging revealed one osteosclerotic focal lesion in the right iliac crest and hepatomegaly
The diagnosis of POEMS was based on the following criteria: demyelinating polyneuropathy, IgA monoclonal gammopathy with lambda secretion, high VEGF, osteosclerotic lesion, acrocyanosis, and thrombocytosis.
A course of lenalidomide and dexamethasone was initiated, with clinical benefit yet to be evaluated.

Our case highlights the importance of considering alternative entities in cases of chronic polyneuropathy unresponsive to standard treatment and the need for continued reporting of similar cases to improve treatment strategies.

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Charcot-Marie-Tooth Disease Type 1A: A Case Study Highlighting Diagnostic Challenges and Genetic Implications in a Family with a History of Polyneuropathy

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Background:

Charcot-Marie-Tooth (CMT) disease, a hereditary motor and sensory neuropathy, presents a diagnostic challenge due to overlapping clinical features with acquired neuropathies and hereditary spastic paraplegia-plus syndromes (SPG-plus).In Georgia, there are only a few genetically established cases. Presented Familial case of CMT Type 1A illustrates the importance of genetic testing and early diagnosis in patients with progressive peripheral neuropathy and a family history of similar symptoms.

Case Description:

A 54-year-old male presented with a several-year history of weakness in the lower extremities, atrophy of tibial muscles, impaired tactile and pain sensation, poor fine motor coordination, and fatigue.

Neurological examination revealed spastic paraparesis, distal neuropathy, and sensory disturbance with pathological reflexes. In addition to progressive walking difficulty, the patient also experienced urinary incontinence. Chronic sensorimotor polyneuropathy was confirmed by EMG, alongside MRI findings of lacunar gliosis. Family history was significant, as both his sons reported numbness in their legs, confirmed by EMG revealing the same data as the father`s. Genetic neuropathy was suspected, prompting genetic testing for CMT.

Results confirmed a duplication in the PMP22 gene, diagnostic for CMT Type 1A. This clarified the underlying etiology of the patient’s and his sons’ progressive neuropathy.

Conclusion:

This case highlights the critical role of genetic analysis in diagnosing hereditary neuropathies like CMT, particularly in patients with family histories of polyneuropathy. Early diagnosis facilitates tailored management strategies, genetic counseling, and surveillance for disease progression.

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Clinical approach for diagnosing familial cortical myoclonic tremor with epilepsy - FCMTE

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Background

Familial cortical myoclonic tremor with epilepsy (FCMTE) is a rare disease with a benign course characterized by myoclonic tremor and rare epileptic seizures. It is often a diagnostic challenge because genetic studies are not fully established. Linkage analyses were performed with microsatellites encompassing the two known loci (8q 23.3-q24.1 and 2p11.1-q12.2). Besides this, it is not always possible to do genetic testing. Diagnosis is based on the clinical and electrophysiological findings.

Discussion

We report a 61-year-old woman with jerky movements and epileptic seizures that debuted six months before admittance. MRI and EEG were normal. Family history is positive as the patient's sister has a similar clinical phenotype. Involuntary movements were distributed in the upper body and head, also propriospinally. Jerks were fast, had high frequency and repetitive manner, and had some kind of rhythmicity. These jerks were expressed in resting positions including sitting and supine conditions, as well as standing and walking. Jerks were exaggerated by stress and postural changes. Intensity and frequency were rising while walking. The clinical scenario includes 3 episodes of losing consciousness with vocalizations and stiffness of limbs. Apart from myoclonus neurological exam revealed only mild intention tremor. The patient had a good response to levetiracetam for seizures and clonazepam for myoclonic tremor.

Conclusion

In the absence of genetic data, thorough history taking, neurological examination, and paraclinical data together with positive treatment response give us the opportunity to diagnose this rare disorder.

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Adolescence Onset Primary Coenzyme Q10 Deficiency - 4: A Case Report

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Introduction: Primary coenzyme deficiency - 4 (COQ10D4) is an autosomal recessive disorder characterized by onset of cerebellar ataxia and exercise intolerance. Some affected individuals develop seizures and have mild mental impairment, indicating variable severity.

Methods: A brief description of the Adolescence Onset Primary Coenzyme Q10 Deficiency - 4 in two brothers in North Macedonia.

Results: We report a case where the pathogen variant in the COQ8A gene is homozygous in two adult brothers. Autosomal recessive inheritance is consistent with the COQ10 deficiency transmission pattern in the families. In this case, genetic analysis showed the same variant was present in both parents. From a neurological perspective, cerebellar ataxia, head tremor, positive Gowers`s sign, proximal muscle weakness, and pseudohypertrophy of the calf muscles clarify the clinical framework. Both brain magnetic resonance imaging (loss of the white matter, cerebellar atrophy, and thinning of the corpus callosum) and electromyography confirmed the clinical diagnosis. As well, abnormal serum creatine kinase levels were monitored.

Conculsion: By highlighting the significance of early detection of potentially treatable COQ8A mutations, these case report overviews support healthcare professionals in better recognising the symptoms of this condition.

Kewords: adolescence onset; primary coenzyme Q10 deficiency - 4; neurological perspective; early detection; proximal muscle weakness.