REVIEW OF LITERATURE

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Comprehensive Physical and Systemic Examination in Neurological Disorders

ABSTRACT

Introduction: A comprehensive general physical and systemic examination is fundamental in evaluating patients with neurological disorders. It helps not only in localizing neurological dysfunction but also in identifying systemic conditions, such as diabetes, hypertension, and infections, that may contribute to or mimic neurological symptoms. A thorough examination is crucial for accurate diagnosis and effective management. **Key components of the physical examination:** This includes an assessment of the patient's general appearance, vital signs, head and neck, cardiovascular, respiratory, musculoskeletal, gastrointestinal and dermatological systems. Each of these aspects can reveal significant clinical findings, ranging from facial expressions and posture indicative of specific neurological disorders to skin manifestations and cardiovascular irregularities that provide clues for underlying conditions. The systemic examination's depth depends on the patient's history, yet a complete assessment is crucial for uncovering unexpected findings and ensuring accurate diagnosis and management of neurological conditions. **Conclusion:** By incorporating both targeted neurological and broader systemic assessments, this approach facilitates the holistic evaluation of patients helping in optimizing treatment strategies for neurological disorders.

Key words: Amyotrophic lateral sclerosis, Mucopolysaccharidosis, Transient ischemic attack, Arteriovenous malformation

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INTRODUCTION

General physical and systemic examinations are fundamental in assessing patients who present with neurological disorders. It not only aids in identifying and localizing site(s) of neurological dysfunction but also reveals systemic conditions that may contribute to or mimic neurological symptoms. For instance, systemic diseases such as diabetes, hypertension, and infections can have significant neurological manifestations, making a comprehensive examination essential for accurate diagnosis and management. The extent of the systemic examination in neurologic patients varies based on individual circumstances and in the real world, is guided by the history of the patient's complaints. Nevertheless, it must be thorough enough to uncover significant findings, some of which might be unexpected.

KEY COMPONENTS OF THE EXAMINATION

General appearance

Initial impression of a patient may help provide valuable clues about the diagnosis.

An emaciated patient with tell-tale signs of malnutrition would immediately raise suspicion of Whipple's disease, celiac disease, amyloidosis, and hyperthyroidism.

Body fat and hair distribution patterns and secondary sexual characteristics can help guide the diagnosis of endocrinopathies and disorders of the hypothalamus.[1] A diagnosis of idiopathic intracranial hypertension must be suspected in a young, overweight woman of reproductive age group presenting with headaches, nausea, vomiting, visual field defects, and double vision.

Stature is another important aspect that is evident in the initial observation. Patients suffering from conditions such as mitochondrial disorders, Refsum's disease, Andersen–Tawil syndrome, Niemann–Pick disease, and Turner and Noonan syndromes generally have short stature. A tall stature may suggest the diagnosis of Marfan's syndrome, homocystinuria, and Klinefelter's syndrome.

A person's posture can tell a lot about the underlying neurological condition. Patients with Parkinson's disease tend to have flexed neck, trunk, elbows, wrists, and knees, with

stooping, rigidity, mask-like facies, slowness of movement, and tremors.

Exaggerated lordosis, waddling gait, and calf hypertrophy may indicate myopathy (Figure 1) whereas a wrist or a foot drop with deformities like hammer toes, pes cavus, and claw hand can be encountered in patients suffering from peripheral nerve disorders (Figure 2).

Vital signs

Monitoring vitals such as blood pressure (BP), temperature, pulse rate, and respiratory rate is crucial in all patients including those suffering from a neurological ailment.

Abnormalities of pulse rate and character can help diagnose conditions like aortic regurgitation (bounding pulses) which may have neurological implications or arrhythmias such as atrial fibrillation as a cause of stroke.

In clinically relevant settings, the BP should be measured in both arms and various body positions (supine, seated, upright).

Significant discrepancies in BP values of both arms can indicate an extracranial cerebrovascular disease (important causes – atherosclerosis, fibromuscular dysplasia, cystic medial necrosis, arteritis, and dissection), which may be the cause of stroke or TIAs.

Dampening of radial pulse in a hyper-abducted and externally rotated arm with the head turned to the opposite direction can suggest a diagnosis of thoracic outlet syndrome.

Orthostatic hypotension as part of autonomic dysfunction may be the cause of pre-syncopal and syncopal symptoms. Autonomic dysfunction may be a result of neuropathy such as diabetic neuropathy or failure of central regulation in multisystem atrophy. In clinical practice, it is common to see orthostatic hypotension occurring due to anti-hypertensive therapy.

Elevated BP can signal raised intracranial pressure (ICP). It can also be in reaction to acute intracranial events such as stroke or subarachnoid hemorrhage.

Head and neck

Head examination involves careful assessment of shape, size, and symmetry which may reveal developmental anomalies such as macrocephaly, microcephaly, or premature cranial suture closure.

Abnormal head tilt can be indicative of cervical dystonia. Patients with craniovertebral junction anomalies have features such as head tilt, short neck, or webbed neck.

In head trauma patients, the presence of Battle's sign (ecchymosis over mastoid) or raccoon's eyes (ecchymosis around the eyes) is suggestive of basilar skull fracture.

Infants or children with raised ICP have bulging fontanelles. Scalp neurofibromas are present in von Recklinghausen's disease.

Tenderness and induration of superficial temporal arteries are seen in Giant cell arteritis.

Percussion of the skull may be important in a subdural hematoma or a tumor where the note may be dull on the lesion.

Macewan's sign or the cracked pot resonance refers to the tympanic note heard on percussion of the skull in children with raised ICP.[2]

Auscultation of the skull is helpful in relevant clinical scenarios. Cephalic bruits can be heard in angiomas, AV malformations, and neoplasms causing vascular compression or aneurysms and are best heard over the temporal region, mastoids, or eyeballs. Carotid bruits are transmitted over to the mastoid.

Signs of meningismus such as nuchal rigidity, Kernig's (pain and resistance during passive extension of knee with flexed hip), and Brudzinski's (involuntary flexion of legs at hips and knees when patient's head is flexed) signs suggest a diagnosis of meningitis.

Face

Facial expressions may aid in neurological diagnosis as many neurological conditions have characteristic facies.

Hypomimia or masked face is seen in Parkinson's disease whereas procerus sign is indicative of progressive supranuclear palsy (PSP). Ptosis and weakness of facial muscles can lead to a diagnosis of myopathies and myasthenia gravis. Pseudobulbar affect (uncontrolled laughter or crying not in line with the social context) may be seen in dementias such as Alzheimer's disease, extrapyramidal and cerebellar

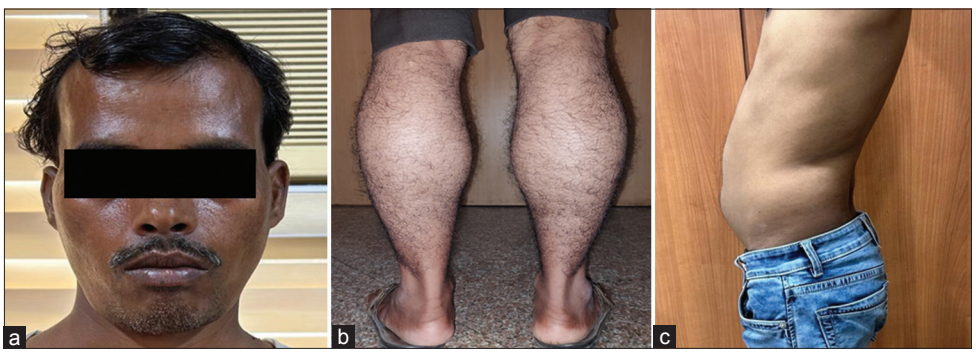

Figure 1: (a) 40-year-old with hatchet like facies with temporal wasting, elongated face, baldness classical of Myotonic Dystrophy; (b) 24-year-old boy with proximal motor weakness with calf hypertrophy diagnosed with Becker's Muscular Dystrophy (BMD); (c) Lumbar lordosis in a 21-year-old male with proximal limb girdle weakness with fatiguability diagnosed with Congenital Myasthenia Gravis (CMS)

disorders such as Parkinson's disease, PSP, MSA, and other neurological disorders such as multiple sclerosis and ALS.[3]

Ear, nose, and throat

Examination of the ears and ear canal may reveal a perforated tympanic membrane as a cause of vertigo or hearing loss.

The presence of vesicles in the case of herpes zoster and clear or bloody discharge in the case of cerebrospinal fluid (CSF) otorrhea are other important clues to diagnosis.

Similarly, leakage of clear watery fluid through the nose might be a clue for CSF rhinorrhea.

Nose examination can also reveal a saddle nose deformity in a case of congenital syphilis.

Patients with rhinocerebral mucormycosis may have redness and swelling of the nasal bridge with evidence of orbital and nasal cellulitis.

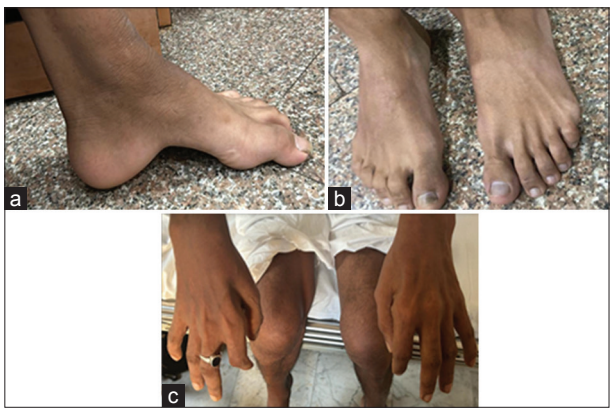

Figure 2: Skeletal deformities – (a) Foot deformity-Pes Cavus in a 19-year-old male with bilateral foot drop and areflexia diagnosed with Charcot Marie Tooth disease (CMT). (b) Foot Deformity-Hammer toes in a 15-year-old male with ataxia, areflexia, and impaired joint position sense in lower limbs diagnosed with Friedrich's Ataxia. (c) Hand deformity- Claw hand in a 48-year-old male with distal upper and lower limb sensorimotor weakness and areflexia diagnosed with Chronic Inflammatory Demyelinating Polyneuropathy (CIDP)

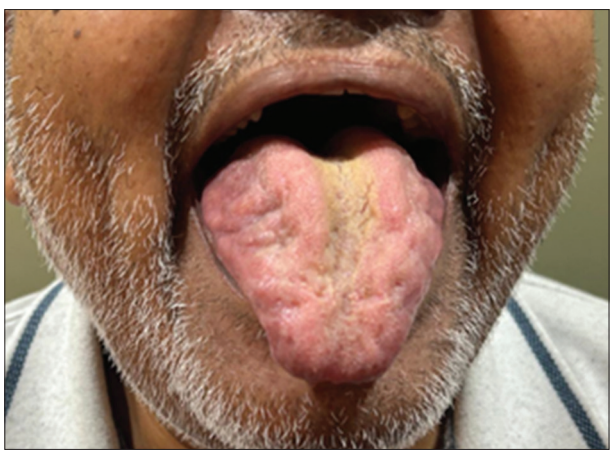

Figure 3: Macroglossia in a patient with systemic amyloidosis

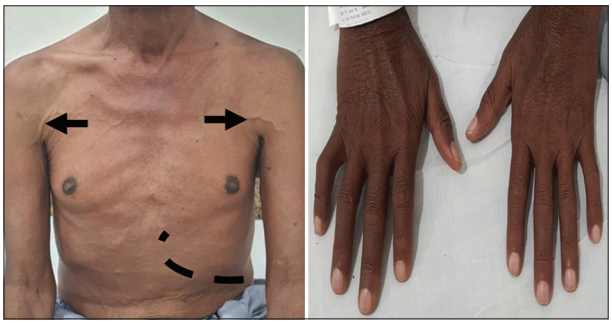

Figure 4: A 50-year-old male, with gradually progressive distal sensorimotor weakness with A) dilated veins on the chest and splenomegaly, B) Skin hyperpigmentation and clubbing, diagnosed with POEMS syndrome

A black eschar in nasal or palatine mucosa strongly points toward the diagnosis.[4]

An oral examination can aid in diagnosing several neurological disorders. Macroglossia can be seen in certain congenital disorders such as Down syndrome, Hunter syndrome (MPS2), Pompe disease, or in acquired conditions such as amyloidosis, acromegaly, and hypothyroidism (Figure 3).[5]

A fissured or plicated tongue (lingua plicata) is characteristic of Melkersson–Rosenthal syndrome. Triple furrowed tongue, although seen rarely can be a distinctive feature of anti-MuSK-positive myasthenia gravis.[6]

Tongue fasciculations are commonly encountered in patients with motor neuron disease.

Other significant findings include a dry mouth in Sjogren's syndrome and oral mucosal ulcerations in Behcet's disease.

Respiratory system

Neuromuscular disorders can cause respiratory dysfunction by causing impaired contractility of the inspiratory and expiratory groups of muscles.

Involvement of upper airways can present with swallowing dysfunction, failure to clear secretions, and impaired airway protection.

On examination, one may find the use of accessory muscles of respiration as a sign of impaired ventilation in patients with motor neuron disease such as ALS and patients with GBS.

Children with spinal muscular atrophy Type 1 may develop respiratory failure before the age of three while patients with SMA Type 2 tend to have a respiratory involvement later in life.[7]

Respiratory insufficiency may occur before the non-respiratory group of muscles in patients with Pompe's disease while respiratory failure in Duchenne muscular dystrophy usually occurs after the loss of ambulation.[8]

Cardiovascular examination

Measurement of BP, assessment of pulse rhythm by palpation as well as auscultation of the heart and neck vessels can provide

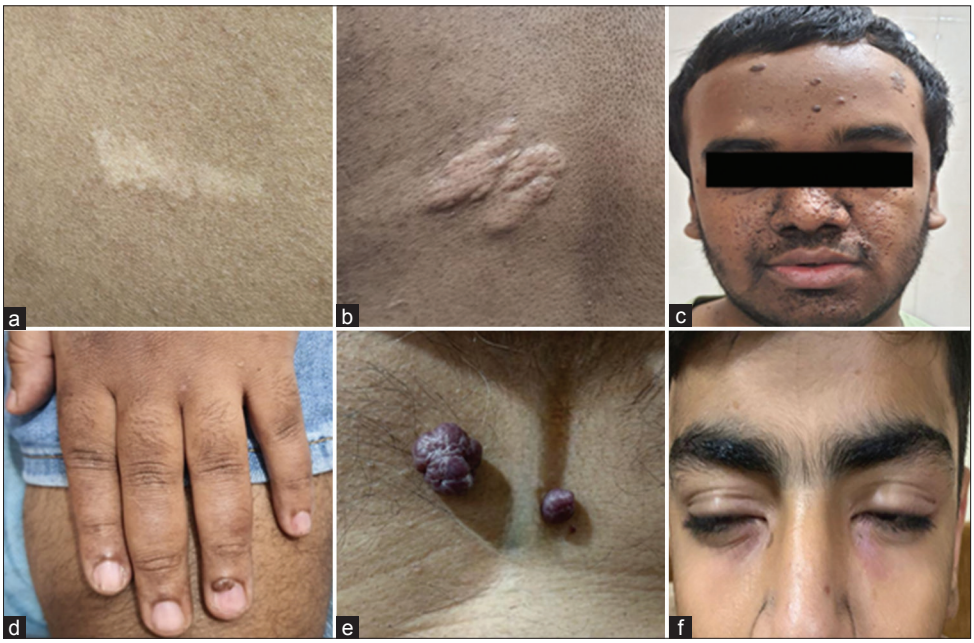

Figure 5: Skin manifestations: (a-d) 24-year-old male diagnosed with tuberous sclerosis with (a) Ash-leaf macule, (b) Shagreen patch, (c) Adenoma sebaceum, (d) Subungual fibroma; (e) 55-year-old male, presented with mild to moderate bilateral lower lim`bs weakness with multiple hemangiomas on the skin, diagnosed with hemangioma of the vertebral body causing mild compression on spine; (f) 19-year-old male with peri-orbital heliotrope rash in a case of inflammatory myopathy (anti-SRP antibody positive)

significant insights in patients with cerebrovascular disease, helping identify valvular disease, arrhythmias, endocarditis, and hypertension as a cause of neurological complications.

Chest and abdomen

Inspection and palpation of the chest for gynecomastia might be relevant when suspecting a diagnosis of Kennedy's disease, thyrotoxic hypokalemic periodic paralysis, and myofibrillar myopathies.[9]

Abdominal palpation may reveal hepatomegaly in patients with amyloidosis and POEMS syndrome (Figure 4); splenomegaly may indicate a metabolic disease such as Niemann–Pick disease Type C or a viral etiology such as Epstein–Barr virus or human immunodeficiency virus.

Ascites may point toward hepatic encephalopathy.

Musculoskeletal system

A detailed examination of the musculoskeletal system gives valuable clues to several neurological disorders including many neuromuscular diseases.

One must look for any obvious deformity, joint contractures, abnormal postures, muscle wasting or hypertrophy, or fasciculations.

Gibbus deformity can be due to tuberculosis or a neoplasm, while scoliosis is seen in patients with Friedreich's Ataxia.

Patients with Marfan's disease and Ehlers–Danlos syndrome have hyperextensible joints.

An elongated and narrow face with a high arched palate,

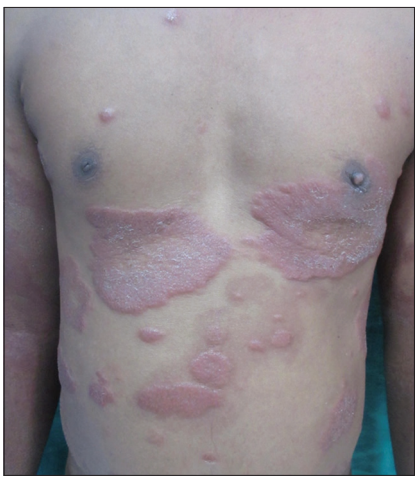

Figure 6: A 30-year-old man with multiple well-defined erythematous plaques with fine scaling with few of them showing central clearing on chest and abdomen in a patient diagnosed with borderline lepromatous leprosy

flat feet, and hyperextensible finger joints in a patient with intellectual disability should raise a suspicion of Fragile X syndrome.[10]

Increased joint laxity with atlantoaxial instability is a feature of Down's syndrome.

Examination of the foot for deformities such as hammer toes and pes cavus is important when dealing with an inherited neuropathy.

Other diseases causing neuropathic arthropathy include diabetes, syphilis, leprosy, and syringomyelia.[11]

Skin

A dermatological examination is crucial for identifying stigmata of various diseases causing neurological manifestations. A careful inspection of the skin can reveal various hallmarks of neurocutaneous disorders.

The presence of spider angiomas, palmar erythema, and dilated veins points toward alcoholic liver disease.

A patient with herpes zoster may initially present with abnormal skin sensations and pain in a dermatomal distribution, along with headache, malaise, and photophobia in the pre-eruptive stage without the presence of characteristic lesions. It is important to observe such patients for the development of macular rash which turns into painful vesicles, common sites being thoracic, cervical, trigeminal, and lumbosacral region.

Patients with leprosy can have a variety of skin lesions with loss of sensations including characteristic hypopigmented lesions which may be solitary or multiple as well as erythematous macules, papules, or nodules.[12]

Erythema chronicum migrans is a distinct feature of Lyme disease.

Neurocutaneous markers include café au lait spots, axillary freckling in NF1; port wine stains in Sturge–Weber syndrome; subungual fibromas, shagreen patch, and angiofibromas in tuberous sclerosis (Figure 5).

Telangiectasias can be seen in ataxia-telangiectasia and Osler–Weber–Rendu disease (where AVMs in the brain can be a cause of stroke and seizures).

Patients with POEMS syndrome can have several cutaneous manifestations including hyperpigmentation, hypertrichosis, erythema, and hemangiomas (Figure 6).

The presence of tufts of hair and skin dimpling on the lower back hints toward spinal dysraphism such as tethered cord syndrome or spina bifida (Figure 4).

When suspecting a diagnosis of dermatomyositis, one must look for Gottron's papules (erythematous or violaceous papules on dorsal metacarpophalangeal and interphalangeal joints) and heliotrope rash (violaceous and erythematous rash affecting the upper eyelids).

CONCLUSION

A detailed and thoughtful clinical examination incorporates general physical and targeted system examinations for proper diagnosis and management of patients. The relevant systemic examination should be performed in all neurological patients, guided by clinical suspicion.

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