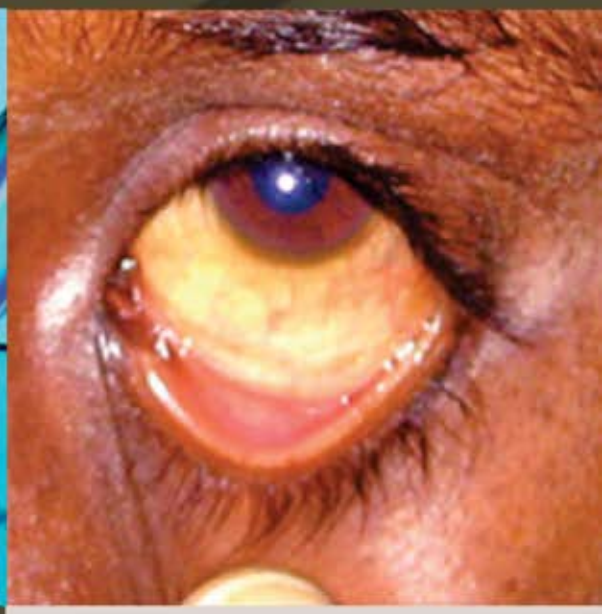
ur website **www.jaypeebrothers.com,** for detailed information on medicine books, visit our website **www.jaypeebrothers.com**







Short & Long Cases in CLINICAL MEDICINE



HN Sarker

JAYPEE

Contents

SECTION 1: SHORT CASES

Case 1: Respiratory System 23

- *Pleural Effusion 23*
- *Pneumothorax 24*
- *Pulmonary Fibrosis 26*
- *Collapse of Lung 27*
- *Consolidation of Lung 29*
- *Collapse/Consolidation of Lung (Mass Lesion) 31*
- *Lung Abscess 32*
- *Bronchiectasis 33*
- *Chronic Obstructive Pulmonary Disease 35*
- *Interstitial Lung Disease 37*

Case 2: Cardiovascular System 40

- *Mitral Stenosis (Case 1) 40*
- *Mitral Stenosis (Case 2) 41*
- *Mitral Regurgitation 42*
- *Mixed Mitral Valve Disease 44*
- *Aortic Stenosis 45*
- *Aortic Regurgitation 48*
- *Mixed Aortic Valve Disease 50*
- *Prosthetic Mitral Valve 51*
- *Prosthetic Aortic Valve 53*
- *Atrial Septal Defect 54*
- *Ventricular Septal Defect 57*
- *Patent Ductus Arteriosus 59*
- *Eisenmenger's Syndrome 61*
- *Tetralogy of Fallot 63*
- *Coarctation of Aorta 65*
- *Atrial Fibrillation 67*
- *Complete Heart Block 69*

Case 3: Abdomen 71

- *Chronic Liver Disease 71*
- *Ascites 72*
- *Hepatomegaly 73*
- *Splenomegaly 75*
- *Hepatosplenomegaly 77*
- *Abdominal Mass 80*
- *Primary Biliary Cirrhosis 89*

Case 4: Neurology 92

- *Spastic Paraplegia/Paraparesis 92*
- *Flaccid Paraplegia/Paraparesis 100*
- *Guillain–Barre Syndrome 100*
- *Monoplegia 101*
- *Hemiplegia 103*
- *Peripheral Neuropathy 106*
- *Charcot–Marie–Tooth Disease 110*
- *Cerebellar Syndrome 112*
- *Parkinson's Disease 113*
- *Myotonic Dystrophy 115*
- *Wasting of Small Muscles of Hands 117*
- *Ulnar Nerve Palsy 119*
- *Median Nerve Palsy 121*
- *Foot Drop 122*
- *7th Cranial Nerve Palsy 125*
- *6th Cranial Nerve Palsy 127*
- *3rd Cranial Nerve Palsy 128*

Case 5: Ophthalmology 130

- *Hypertensive Retinopathy 130*
- *Diabetic Retinopathy 132*
- *Papilledema 135*
- *Optic Atrophy 137*
- *Retinitis Pigmentosa 140*
- *Central Retinal Vein Occlusion 141*

Case 6: Endocrine System 143

- *Thyroid Swelling (Goiter) 143*
- *Assessment of Thyroid Status 148*
- *Hypothyroidism 148*
- *Hyperthyroidism 150*
- *Acromegaly 152*
- *Cushing's Syndrome 154*
- *Addison's Disease 156*
- *Gynecomastia 159*

Case 7: Locomotor System 161

- *Rheumatoid Arthritis 161*
- *Systemic Lupus Erythematosus 162*
- *Ankylosing Spondylitis 163*
- *Reactive Arthritis 164*
- *Osteoarthritis (Knees) 165*
- *Psoriatic Arthritis 167*
- *Gout 168*
- *Dermatomyositis 170*
- *Systemic Sclerosis 173*
- *Tall Stature (Marfan's Syndrome) 175*

Case 8: Skin 178

- *Psoriasis 178*
- *Scabies 180*
- *Herpes Zoster 181*
- *Bullous Lesions 183*
- *Clubbing 187*

SECTION 2: LONG CASES

Case 9: Abdominal Swelling (Ascites—Chronic Liver Disease) 195
Case 10: Jaundice (Acute Viral Hepatitis) 203
Case 11: Fever and Right Upper Abdominal Pain (Liver Abscess) 209
Case 12: A Painful Hard Irregular Mass in Upper Abdomen (Hepatocellular Carcinoma) 213
Case 13: Pruritus with Jaundice (Primary Biliary Cirrhosis) 216
Case 14: Epigastric Pain (Peptic Ulcer Disease) 219
Case 15: Chronic Diarrhea (Inflammatory Bowel Disease) 224
Case 16: Breathlessness and Wheeze (Bronchial Asthma) 228
Case 17: Cough and Breathlessness (Chronic Obstructive Pulmonary Disease) 233
Case 18: Cough, Breathlessness and Swelling of Legs (Cor Pulmonale) 238
Case 19: Heaviness of Right Side of Chest (Pleural Effusion) 241
Case 20: Cough, Fever and Hemoptysis (Pulmonary Tuberculosis) 244
Case 21: Cough, Hemoptysis and Left-sided Chest Pain (Bronchial Carcinoma) 250
Case 22: Swelling of Legs and Breathlessness (Congestive Cardiac Failure) 254
Case 23: Persistently Elevated Blood Pressure (Hypertension) 259
Case 24: Central Chest Pain on Exertion (Ischemic Heart Disease—Stable Angina) 263
Case 25: Prolonged Severe Central Chest Pain (Acute Coronary Syndrome) 267
Case 26: Recurrent Attacks of Exertional Dyspnea, Paroxysmal Nocturnal Dyspnea and Palpitation (Mitral Stenosis) 271
Case 27: Swelling of Face and Scanty High Color Urine (Acute Glomerulonephritis) 276
Case 28: Generalized Swelling of the Body (Nephrotic Syndrome) 280
Case 29: Anorexia, Nausea, Nocturia, and Swelling of Face and Feet (Chronic Renal Failure) 285
Case 30: Generalized Weakness and Pallor (Iron Deficiency Anemia) 290
Case 31: Anemia and Left Hypochondriac Lump (Thalassemia) 294
Case 32: Nodular Swellings in Neck and Armpits (Lymphoma) 300
Case 33: Purpuric Rash (Idiopathic Thrombocytopenic Purpura) 305
Case 34: Weakness, Fever and Gum Bleeding (Aplastic Anemia) 308
Case 35: Low Back Pain and Fatigue (Multiple Myeloma) 311
Case 36: Mass in Left Upper Abdomen and Weakness (Chronic Myeloid Leukemia) 315
Case 37: A Nodular Swelling in Neck (Tubercular Lymphadenitis) 318
Case 38: Prolonged Fever with Left Hypochondriac Lump (Kala Azar) 321
Case 39: Elevated Blood Sugar (Diabetes Mellitus) 325

Case 40: Weight Loss in Spite of Good Appetite (Thyrotoxicosis) 332
Case 41: Puffiness of Face and Weight Gain (Hypothyroidism) 336
Case 42: Multiple Joint Pain and Swelling (Rheumatoid Arthritis) 340
Case 43: Rashes and Painful Swelling of Small Joints (Systemic Lupus Erythematosus) 346
Case 44: Painful Swollen Right Knee (Monoarthritis—Reactive Arthritis) 351
Case 45: Back Pain and Stiffness (Ankylosing Spondylitis) 354
Case 46: Weakness and Stiffness of Both Lower Limbs (Spastic Paraparesis—Pott's Disease) 358
Case 47: Weak and Floppy Lower Limbs (Flaccid Paraplegia—Guillain–Barre Syndrome) 362
Case 48: Fasciculations, Cramps and Weakness of Limbs (Motor Neuron Disease) 366
Case 49: Weak, Stiff and Incoordinated Lower Limbs (Multiple Sclerosis) 370

Index *375*

CASE 4

Neurology

SPASTIC PARAPLEGIA/PARAPARESIS

Spinal Cord Compression

Scenario

Weakness of both lower limbs.

Instruction

Examine the lower limbs/neurological examination of lower limbs.

Proceed as Follows

1. Address the patient as your custom "Hi, Hello, Assalamu alaykum" and introduce yourself and tell your role.
2. Maintain 3Ps—Permission, Position and Proper exposure (preferably at supine position).
3. Look at the patient (from head to toe) and surroundings while positioning and exposing, and spend a few seconds to get any clue, e.g. walking stick, wheelchair, wasting of small muscles of hands (MND), and indwelling urinary catheter (sphincter involvement).
   Note: This patient has indwelling urinary catheter.
4. Inspect the limbs and look for wasting and fasciculations (MND).
5. Assess **bulk of muscle**. No need to measure by tape if there is no gross asymmetry and wasting.
6. Assess the **muscle tone** in each leg by passively moving it at the hip and knee joints (with the patient relaxed roll the leg sideways, backwards and forwards on the bed; lift the knee and let it drop, or bend the knee and partially straighten in an irregular and unexpected rhythm).
   Note: In this patient, there is hypertonia (spastic type).
   Causes of hypertonia—see Box 4.1.

Box 4.1: Causes of hypertonia

- Upper motor neuron (UMN) lesion (spastic, may be clasp knife)
- Extrapyramidal lesion (lead pipe or cogwheel regidity)
- HCR (rigidity continue to increase with more and more passive movement)
- Others—tetanus, catatonic state, strychnine poisoning

7. Test **muscle power** as a whole by asking the patient to raise his or her leg straight upward, then test power of individual group and grade muscle power.
   Note: In this patient, muscle power is 2/5 in both lower limbs.
   Medical Research Council scale for muscle power—see Box 4.2.

Box 4.2: Medical Research Council scale for muscle power

0	No muscle contraction visible
1	Flicker of contraction but no movement
2	Joint movement when effect of gravity eliminated
3	Movement against gravity but not against examiner's resistance
4	Movement against resistance but weaker than normal
5	Normal power

8. Elicit **jerks,** i.e knee (L3, 4) and ankle (S1, 2) jerks and also ankle and patellar clonus.
   Note: In this patient, knee and ankle jerks are exaggerated and ankle clonus is present (UMN lesion).

   Features of UMN lesion—see Box 4.3.

Box 4.3: Features of UMN lesion

- Muscle weakness
- Spasticity
- Increased deep tendon reflexes
- Depressed abdominal responses
- Extensor plantar response

9. Elicit **plantar response**.
   Note: Extensor plantar response in this patient.

   Causes of extensor plantar response—see Box 4.4.

Box 4.4: Causes of extensor plantar response

- UMN lesion
- First year of life (due to lack of myelination of pyramidal tract myelination occurs by 12 months)
- Deep coma (due to any cause)
- Sleep
- Postictal period

JAYPEE BROTHERS

10. Test **coordination** by using heel-shin test.
11. Test **sensation**—use cotton wool for light touch and pin prick for pain over dermatomes L2 to S1 on the legs. Ascertain the level of sensory loss by working up the legs. Test vibration and position sense also.
    Note: In this patient, sensory level is at T_{10}.
12. Test **gait** (check the patient can walk by asking for either his or the examiner's permission to examine the gait) and finally perform **Romberg's test.**
    Note: Not possible in this patient.
13. Examine the spine and look for tenderness, scars, and deformities.
    Note: There is gibbus deformity at T_7 spine.
14. Also ask for examination of upper limb.

Schematic Diagnosis

	Neurological examination
Inspection and muscle bulk	No wasting and fasciculation
	↓
Tone	Increased tone
	↓
Muscle power	Diminished (2/5) in both lower limbs
	↓
Jerks and plantar response	Exaggerated, ankle clonus present, extensor plantar response
	↓
	Spastic paraparesis
	↓
Sensation	Loss of all modalities up to T_{10}
	↓
Differential diagnosis	Spinal cord compression Acute transverse myelitis
	↓
Spine examination	Gibbus deformity
	↓
Final diagnosis	**Spinal cord compression**

Q. Please present your case or tell your findings.

A.
- There is bilateral weakness (power 2/5) with hypertonia, hyperreflexia, ankle clouns and extensor plantar responses but no wasting and fasciculation. Sensation is impaired in the lower limbs, with a sensory level at the level of T_{10}.
- There is gibbus deformity over the thoracic spine 7. The patient is on indwelling catheter.
- This patient has spastic paraparesis due to a lesion at the T_{10} level of the spinal cord.

Q. What would you think to be the underlying cause?

A. Spinal cord compression.

Q. What are the points in favor of your diagnosis?

A. Spastic paraparesis with definite sensory level and gibbus deformity of spine suggest spinal cord compression.

Q. What is your differential diagnosis?

A. Acute transverse myelitis (Table 4.1).

Discussion about spinal cord compression—Please read long case "Spastic Paraparesis—Pott's Disease".

Q. What are the causes of spastic paraparesis?

A. Causes of spastic paraparesis include:
- Spinal cord compression, i.e. tumors, trauma and tuberculosis, etc.
- Acute transverse myelitis
- Multiple sclerosis
- Motor neuron disease
- Subacute combined degeneration of spinal cord
- Friedreich's ataxia
- Syringomyelia
- Anterior spinal artery occlusion
- Hereditary spastic paraparesis
- Taboparesis
- Tropical spastic paraparesis
- Cervical myelopathy
- Parasagittal tumor.

Q. Which factors would help to identify the cause of spastic paraplegia?

A. The causes of spastic paraplegia include:
- Sensory signs
- Cerebellar signs (particulary in the upper limbs)
- Fasciculations
- Upper limbs involvement
- Wasting of the small muscles of the hands.

Q. What are the causes of spastic paraparesis with definite sensory level?

A. Causes include:
- Spinal cord compression
- Acute transverse myelitis
- Anterior spinal artery occlusion.

Q. What are the causes of spastic paraparesis without sensory loss?

A. Causes includes:
- Motor neuron disease
- Hereditary spastic paraparesis.

Q. What are the causes of spastic paraparesis with dorsal column loss?

A. Causes includes:
- Multiple sclerosis
- Friedreich's ataxia
- Cervical myelopathy

JAYPEE BROTHERS

Table 4.1: Examination findings in differential diagnoses of spastic paraparesis

Condition	*Motor signs*	*Sensory signs*	*Associated signs*
Spinal cord compression	Spastic paraplegia / paraparesis	Sensory level	Spincter involvement, +/– spine abnormality
Acute transverse myelitis	Spastic paraplegia	Sensory level	Spincter involvement, severe neck or back pain
Multiple sclerosis	Spastic paraparesis	Dorsal column signs/ variable sensory loss	Cerebellar signs (especially in the upper limbs); eye signs (optic atrophy, internuclear ophthalmoplegia)
Motor neuron disease	Spastic paraparesis	None	Wasting, fasciculations (limbs and tongue); bulbar involvement; wasting of small muscle of hands
Friedreich's ataxia	Spastic paraparesis	Dorsal column signs, peripheral neuropathy	Cerebellar signs; pes cavus; kyphoscoliosis
Cervical myelopathy	Spastic paraparesis	Dorsal column signs	Neck pain; Lhermitte's sign; inversion of bicep and suppinator reflexes
Subacute combined degeneration	Spastic paraparesis	Dorsal column signs, peripheral neuropathy	Absent ankle jerks and extensor plantars; anaemia; jaundice; glossitis
Syringomyelia	Spastic paraparesis	Dissociated loss (preserved posterior columns and loss of spinothalamic tract)	LMN weakness in upper limbs, wasting of small muscles of the hands, Horner's syndrome
Anterior spinal artery occlusion	Spastic paraplegia	Sensory level	May have irregular pulse (AF)
Hereditary spastic paraparesis	Spastic paraparesis	None	Upper limbs normal
Taboparesis	Spastic paraparesis	Dorsal column signs	Absent ankle jerks and extensor plantars; Argyll Robertson pupils

- Subacute combined degeneration of the cord
- Taboparesis.

Q. What are the causes of spastic paraparesis with spinothalamic column loss?

A. Causes are:
- Syringomyelia
- Anterior spinal artery occlusion.

Q. What are the causes of dissociated sensory loss?

A. Causes of dissociated sensory loss includes:
- Preserved spinothalamic and loss of posterior columns
  - Multiple sclerosis
  - Friedreich's ataxia
  - Cervical myelopathy
  - Subacute combined degeneration of the cord
  - Taboparesis.
- Preserved posterior columns and loss of spinothalamic tracts
  - Syringomyelia
  - Anterior spinal artery occlusion.

Acute Transverse Myelitis

Scenario

Sudden weakness of both lower limbs.

Instruction

Examine the lower limbs/neurological examination of lower limbs.

Examination

Follow the examination procedure as "spinal cord compression"

Schematic Diagnosis

Neurological examination

Inspection and muscle bulk — No wasting and fasciculation

↓

Tone — Increased tone

↓

Muscle power — Diminished (0/5) in both lower limbs

↓

Jerks and plantar response — Exaggerated, ankle clonus present, extensor plantar response

↓

Spastic paraplegia

↓

Sensation — Loss of all modalities up to T_8

↓

Differential diagnosis — Spinal cord compression
Acute transverse myelitis

↓

Spine examination — Normal and H/O preceeding RTI

↓

Diagnosis — **Acute transverse myelitis**

JAYPEE BROTHERS

Q. Please tell your findings

A. Neurological examination of lower limbs reveals:
- There is bilateral weakness (power 0/5) with hypertonia, hyperreflexia, ankle clouns and extensor plantar responses but no wasting and fasciculation. All modalities of sensation are lost in the lower limbs, with a sensory level at the level of T_8.
- There is no abnormality on spine examination. The patient is on indwelling catheter.
- This patient has spastic paraplegia with sensory level at the T_8 level of the spinal cord.

Q. What is your diagnosis?

A. Spastic paraplegia with definite sensory level and sphincter involvement, most probably due to "**acute transverse myelitis**".

Q. Why have you told this?

A. Spastic paraplegia with definite sensory level and sphincter involvement without any spinal abnormality suggest acute tranvrse myelitis, though spinal cord compression is not completely ruled out.

Q. What history would you like to ask this patient?

A. History of upper respiratory tract infection, trauma, surgery or vaccination and neck or back pain.

Q. How would you investigate this case?

A. Following investigations must be done:
- MRI of spine and spinal cord
- Lumbar puncture and CSF study.

Q. How would you treat this patient?

A. The treatment must include:
- Supportive measures—as for a paralysed patient (see long case—spastic paraplegia)
- Specific measure—high-dose methyl prednisolone—1g iv daily for 3 days or 500 mg daily for 5 days.

Q. What is the prognosis of acute transverse myelitis?

A. The outcome is variable: One-third have static deficit, one-third go on to develop MS and one-third recover with no subsequent relapse.

Multiple Sclerosis

Scenario

A 35-year-old lady presents with weakness of both lower limbs.

Instruction

Examine the lower limbs/neurological examination of lower limbs.

Examination

Follow the examination procedure as "spinal cord compression" and also examine upper limbs (especially cerebellar signs), speech (scanning), eyes (nystagmus, internuclear ophthalmoplegia) and fundoscopy (optic atrophy).

Schematic Diagnosis

	Neurological examination
Inspection and muscle bulk	No wasting and fasciculation
	↓
Tone	Increased tone
	↓
Muscle power	Diminished (3/5) in both lower limbs
	↓
Jerks and plantar response	Exaggerated, ankle clonus present, extensor plantar response
	↓
	Spastic paraparesis
	↓
Sensation	Impaired vibration and position sense
	↓
Differential diagnosis	Multiple sclerosis Freiderich's ataxia Subacute combined degeneration of cord
	↓
Coordination	Dyssynergia and impaired heel-shin testing
	↓
Differential diagnosis	Multiple sclerosis Friedreich's ataxia
	↓
Relevants	Staccato speech, cerebellar signs in right arm, nystagmus and left optic atrophy
	↓
Final diagnosis	**Multiple sclerosis**

Q. Please tell your findings?

A. Neurological examination reveals that there is bilateral weakness (power 3/5) with hypertonia, hyperreflexia, ankle clouns and extensor plantar responses but no wasting and fasciculation. Vibration and position sense are impaired in the feet. There is ataxia on the right, with dyssynergia and impaired heel–shin testing. On relevant examination, the speech is slurred with an explosive (staccato) character, cerebellar signs, such as intension tremor, past pointing and impaired finger-nose test are present on right upper arm, and nystagmus is present in eyes. On fundoscopy, the left optic disk is pale and has distinct margins (optic atrophy).

JAYPEE BROTHERS

Q. What is your diagnosis?

A. Multiple sclerosis.

Q. Why have you told this?

A. This patient displays pyramidal, dorsal column signs, eye, and cerebellar signs. The most likely unifying diagnosis is demyelinating disease, i.e. multiple sclerosis.

Q. Why is this not Friedreich's ataxia (FA)?

A. Age (FA before 20 years), unilateral cerebellar signs and optic atrophy exclude Friedreich's ataxia.

Q. Why is this not subacute combined degeneration of cord (SCD)?

A. Cerebellar signs are not found in SCD.

Discussion about "multiple sclerosis"—please read long case "Multiple Sclerosis".

Subacute Combined Degeneration of Spinal Cord (SCD)

Scenario

A 55-year-old man presents with weakness of both lower limbs and unsteadiness.

Instruction

Examine the lower limbs/neurological examination of lower limbs.

Examination

Follow the examination procedure as "spinal cord compression" and also see the relevants—upper limbs, anaemia, jaundice, glossitis, splenomegaly, and laparotomy scar.

Schematic Diagnosis

Neurological examination

Inspection and muscle bulk — Distal wasting and weakness

↓

Tone — Increased tone in thigh muscles but decreased in distal group

↓

Muscle power — Diminished (4/5) in both lower limbs

↓

Jerks and plantar response — Exaggerated knee, absent ankle jerk, extensor plantar response

↓

Sensation — Vibration and position sense lost, impaired light touch and pinprick in a stocking distribution

↓

Provisional diagnosis — Spastic paraparesis with dorsal column loss, and peripheral neuropathy

↓

Relevants — Impaired light touch and pinprick in a glove distribution in upper limbs, anaemia and glossitis

↓

Final diagnosis — **Subacute combined degeneration of spinal cord**.

Q. Please tell your findings?

A.
- On examination of the lower limbs, the skin has a shiny and dry appearance with loss of hair. There is distal wasting and weakness. There is hypertonia of thigh muscles; muscle power is 4/5. Ankle jerks are absent, knee jerks are exaggerated and extensor plantar response. There is loss of vibration and position sense, and bilateral symmetrical impaiment of light touch and pinprick in a stocking distribution.
- There is ataxia with a wide-based and high-steppage gait. Romberg's sign is positive.
- Relevant examination reveals impaiment of light touch and pinprick in a glove distribution in upper limbs, anemia and glossitis.

Causes of absent ankle jerk, but extensor plantar response—see Box 4.5.

Box 4.5: Causes of absent ankle jerk but extensor plantar response

- Subacute combined degeneration of spinal cord
- Friedreich's ataxia
- Multiple sclerosis
- Taboparesis
- Cervical myelopathy with peripheral neuropathy, e.g. diabetes mellitus

Q. What is your diagnosis?

A. The diagnosis is spastic paraparesis with dorsal column loss, peripheral neuropathy and anemia. The unifying diagnosis is **subacute combined degeneration of spinal cord**.

Q. What are the differential diagnoses?

A. Differential diagnosis includes:
- Friedreich's ataxia
- Multiple sclerosis
- Taboparesis.

Q. Why is it called combined degeneration?

A. As it involves both dorsal column and pyramidal tracts.

Q. What are the causes of both pyramidal and dorsal column lesion?

A. The causes includes:
- SCD
- Friedreich's ataxia

JAYPEE BROTHERS

- Multiple sclerosis
- Taboparesis
- Cervical myelopathy.

Q. What are the causes of dorsal column lesion?

A. The causes include:
- SCD
- Multiple sclerosis
- Friedreich's ataxia
- Tabes dorsalis
- Cervical myelopathy
- Brown–Sequard syndrome (ipsilateral).

Q. What is the cause of SCD?

A. Vitamin B_{12} deficiency.

Q. What are the neurological features of vitamin B_{12} deficiency?

A. The neurological features of vitamin B_{12} deficiency are
- Spinal cord disease (dorsal column lesion, pyramidal tract lesion)
- Peripheral sensory neuropathy
- Optic atrophy
- Dementia.

Q. What are the non-neurological features of vitamin B_{12} deficiency?

A. The non-neurological features of vitamin B_{12} deficiency are as follows:
- Anemia
- Glossitis (red beef tongue)
- Jaundice
- Splenomegaly.

Q. Which type of anaemia occurs in vitamin B_{12} deficiency?

A. Megaloblastic macrocytic anemia.

Q. What is the other cause of megaloblastic macrocytic anemia?

A. Folic acid deficiency.

Q. What are the causes of vitamin B_{12} deficiency?

A. The causes of vitamin B_{12} deficiency are:
- Pernicious anemia
- Partial or total gastrectomy
- Crohn's disease or ileal resection
- True vegetarian diet
- Blind loop syndrome
- Fish tapeworm (*Diphyllobothrium latum*)
- Pancreatic insufficiency (failure to transfer vitamin B_{12} from R protein to intrinsic factor)
- Chronic tropical sprue
- Congenital intrinsic factor deficiency.

Q. What is the daily requirement of vitamin B_{12}? How long does it take to develop vitamin B_{12} deficiency?

A. The daily requirement of vitamin B_{12} is 1–2 μg. To develop deficiency, it takes 3 years because liver stores up to 4 mg vitamin B_{12}.

Q. How would you investigate this patient?

A. Following investigations must be done:
- Complete blood count with PBF
- Bone marrow (to see megaloblast)
- Serum B_{12} assay
- Nerve conduction study
- Other investigations to find out cause, e.g. Schilling test, intrinsic factor assay.

Q. What are the expected findings in PBF?

A. The expected findings in PBF are as follows:
- Macrocytosis
- Hypersegmented neutrophil
- May be pancytopenia.

Q. How would you treat this patient?

A. The treatment involves the following:
- Injection vitamin B_{12}—1000 μg IM every alternate day or 2 days interval for 5 injections, then every 3 months to be continued for life long. Following therapy, hypokalemia and iron deficiency may occur. B_{12} orally 2 mg/day may be given. 1–2% is absorbed by diffusion without intrinsic factor. Sublingual B|9 may be effective
- Treatment of underlying cause.

Q. What is the response of neurological lesion to vitamin B_{12} therapy?

A. Response is variable. Neurological features may improve, remain unchanged or may even deteriorate. Sensory abnormalities improve more than the motor and peripheral neuropathy responses better than myelopathy.

Motor Neuron Disease (Fig. 4.1)

Scenario

A 45-year-old man presents with fasciculation and weakness of both lower limbs.

Instruction

Examine the lower limbs/neurological examination of lower limbs.

Examination

Follow the examination procedure as "spinal cord compression" and also see the relevants—upper limbs and tongue.

JAYPEE BROTHERS



Fig. 4.1: Motor neuron disease (MND)



Fig. 4.2: Pes cavus (Friedreich's ataxia)

Schematic Diagnosis

	Neurological examination
Inspection and muscle bulk	Wasting and fasciculation
	↓
Tone	Increased tone
	↓
Muscle power	Diminished (4/5) in both lower limbs
	↓
Jerks and plantar response	Exaggerated knee and ankle jerk, clonus present, extensor plantar response
	↓
Sensation	No sensory loss
	↓
Provisional diagnosis	Spastic paraparesis with wasting and fasciculations, no sensory loss
	↓
Relevants	Wasting of small muscles of hands, LMN features of upper limbs without sensory loss, bulbar palsy (wasted fasciculation of tongue), no cerebellar signs
	↓
Final diagnosis	**Motor neuron disease (MND)**

Q. Please tell your findings?

A.
- On examination of the lower limbs, there is wasting and fascicullations, hypertonia; muscle power is 4/5. Knee and ankle jerks are exaggerated; ankle clonus is present, and extensor plantar response. There is no sensory loss and coordination is intact.
- Relevant examination reveals wasting of small muscles of hands, LMN features of upper limbs without sensory loss, wasting and fasciculation of tongue (bulbar palsy) and no cerebellar signs.

Q. What is your diagnosis and why?

A. Motor neuron disease as wasted fasciculating limbs with exaggerated jerks without sensory involvement.
Note: Wasted fasciculating limbs with exaggerated jerks—diagnosis is MND unless otherwise proved.

Q. Which type of MND do you think and why?

A. Amyotrophic lateral sclerosis as there is predominantly UMN lesion in lower limbs and LMN lesion in upper limbs.

Discussion about MND—Please read long case—"MND".

Friedreich's Ataxia (Fig. 4.2)

Scenario

A 15-year-old boy presents with stiffnes of both lower limbs and unsteady gait.

Instruction

Examine the lower limbs/neurological examination of lower limbs and relevants.

Examination

Follow the examination procedure as "spinal cord compression" and also see the relevants—upper limbs, cerebellar sign, spine, palate and ophthalmoscopy.

Schematic Diagnosis

	Neurological examination
Inspection and muscle bulk	Pes cavus and distal wasting
	↓
Tone	Increased tone
	↓
Muscle power	Diminished (3/5) in both lower limbs
	↓

JAYPEE BROTHERS

Jerks and plantar response	Knee and ankle jerk absent, extensor plantar response
	↓
Sensation	Impaired vibration and position sense in the feet
	↓
Coordination	Impaired heel–shin test
	↓
Gait	Broad-based gait with positive Romberg's test
	↓
Relevants	Upper limb (cerebellar signs—intention tremor, dysdiadochokinesis, impaired finger-nose), eyes (nystagmus), fundoscopy (optic atrophy) and spines (kyphoscoliosis)
	↓
Final diagnosis	**Friedreich's ataxia**

Q. Please tell your findings?

A.
- On examination of the lower limbs of this young boy, there is pes cavus, distal wasting and weakness, muscle power is 3/5 and tone is increased. Knee and ankle jerks are absent and plantar response is extensor. There is impaired vibration and proprioception in the feet and impaired heel–shin test. There is ataxia and the patient walks with a broad-based gait with a tendency to fall to both sides and Romberg's test is positive.
- Relevant examination reveals cerebellar signs (intention tremor, dysdiadochokinesis, impaired finger-nose) in both upper limbs, kyphoscoliosis, nystagmus, and optic atrophy in fundoscopy.

Q. What is your diagnosis?

A. Friedreich's ataxia.

Q. What are the points in favor of your diagnosis?

A. This young patient has features of bilateral cerebellar syndrome, pyramidal weakness with loss of tendon reflexes with extensor plantar responses, dorsal column loss, pes cavus, kyphoscoliosis, and primary optic atrophy. All these would suggest a diagnosis of Friedreich's ataxia.

Q. What are the sites of lesions in Friedreich's ataxia?

A. There is progressive degeneration involving:
- Dorsal column (loss of vibration and position sense-sensory ataxia)
- Corticospinal tract
- Cerebellar lesion (lateral and ventral spinocerebellar tracts, Clarke column, dentate nucleus, superior vermis, and dentatorubral pathways)
- Eye (primary optic atrophy).

Q. What are the causes of combined cerebellar, pyramidal and dorsal column lesions?

A. The causes are as follows:
- Multiple sclerosis
- Friedreich's ataxia
- Spinocerebellar degeneration.

Q. Why is this case not multiple sclerosis?

A. This case is not multiple sclerosis as this patient is a young male and presence of pes cavus and kyphoscoliosis make multiple sclerosis less likely.

Q. What is Friedreich's ataxia?

A. Friedreich's ataxia is one of the hereditary ataxia, inherited as autosomal recessive (AR) (in some case inherited as AD).

Q. What do you know about the genetics of Friedreich's ataxia?

A. Fridreich's ataxia is an autosomal recessive condition, with the gene locus on chromosome 9q13. The mutation is a trinucleotide (GAA) repeat expansion in a gene encoding a protein called frataxin with anticipation.

Q. What are the features of Friedreich's ataxia?

A. Features of Friedreich's ataxia are as follows:
- Age of onset–typically in age 10–15 years, and almost always presents before 20 years of age
- Common presentations—progressive difficulty in walking (cerebellar ataxia, also sensory ataxia), weakness of lower limbs (pyramidal weakness)
- Associated features—pes cavus, clawing of toes, kyphoscoliosis, diabetes mellitus (in 10%), optic atrophy (in 25%), hypertrophic cardiomyopathy (in 50%), and sensorineural deafness (in 10%)
- Prognosis—usually slowly progressive disease, death occurs before 40 years of age (usually 20 years after the onset of symptoms).

Q. What are the causes of bilateral pes cavus?

A. The causes of bilateral pes cavus are as follows:
- Friedreich's ataxia
- Charcot-Marie-Tooth disease
- Hereditary spastic paraparesis
- Muscular dystrophies
- Spinal muscular atrophy
- Cerebral palsy.

Q. What are the different types of hereditary ataxia?

A. The different types of hereditary ataxia are has follows:
- Friedreich's ataxia
- Ataxia telangiectasia
- Olivoponto-cerebellar degeneration
- Hereditary spastic paraplegia
- Abetalipoproteinaemia (Bassen–Kornzweig syndrome)
- Refsum's disease.

JAYPEE BROTHERS

FLACCID PARAPLEGIA/PARAPARESIS

Scenario

Weakness of both lower limbs.

Instruction

Examine the lower limbs/neurological examination of lower limbs.

Proceed as Follows

1. Address the patient as your custom "Hi, Hello, Assalamu alaykum" and introduce yourself and tell your role.
2. Maintain 3Ps—Permission, Position and Proper exposure (preferably at supine position).
3. Look at the patient (from head to toe) and surroundings while positioning and exposing, and spend a few seconds to get any clue, e.g. walking stick, wheelchair, wasting of small muscles of hands (MND), and indwelling urinary catheter (sphincter involvement).
4. Inspect the limbs and look for wasting and fasciculations.
5. Assess **bulk of muscle**. No need to measure by tape if there is no gross asymmetry and wasting.
6. Assess the **muscle tone** in each leg by passively moving it at the hip and knee joints (with the patient relaxed roll the leg sideways, backwards and forwards on the bed; lift the knee and let it drop, or bend the knee and partially straighten in an irregular and unexpected rhythm).
   Note: In this patient, there is hypotonia (flaccid type).
   Causes of hypotonia—see Box 4.6.

Box 4.6: Causes of hypotonia

- Lower motor neuron (LMN) lesion
- Peripheral neuropathy
- Cerebellar syndrome
- Chorea
- Myopathy except myotonia
- Hypokalaemia or hyperkalemia.

7. Test **muscle power** as a whole by asking the patient to raise his or her leg straight upward, then test power of individual group and grade muscle power.
8. Elicit **jerks,** i.e knee (L3, 4) and ankle (S1, 2) jerks. Do reinforcement if jerks are absent.
   Note: In this patient, knee and ankle jerks are diminished or absent (LMN lesion).
   Features of LMN lesion—see Box 4.7.

Box 4.7: Features of LMN lesion

- Wasting and fasciculation
- Muscle weakness
- Hypotonia
- Diminished or absent deep tendon reflexes
- Plantar response normal or absent

- Elicit plantar response.
  Note: Normal or absent plantar response.
9. Test **coordination** by using heel-shin test.
10. Test **sensation**—Use cotton wool for light touch and pin prick for pain over dermatomes L2 to S1 on the legs. Ascertain the level of sensory loss by working up the legs. Test vibration and position sense also.
11. Test **gait** (check the patient can walk by asking for either his or the examiner's permission to examine the gait) and finally perform **Romberg's test** (if possible).
12. Examine the spine and look for tenderness, scars, and deformities.
13. Also ask for examination of upper limbs.

Diagnosis—Flaccid paraplegia or paraparesis
Causes of flaccid paraplegia or paraparesis—see Box 4.8.

Box 4.8: Causes of flaccid paraplegia or paraparesis

- Guillain–Barre syndrome
- Chronic inflammatory demyelinating polyradiculopathy (CIDP)
- Other motor or sensorymotor neuropathy
- Progressive muscular atrophy (one type of MND)
- Hypokalemia or hyperkalemia
- Poliomyelitis
- Acute spastic paraplegia with neuronal shock

GUILLAIN–BARRE SYNDROME (GBS)

Scenario

Progressive weakness of both lower limbs for three days.

Instruction

Examine the lower limbs/neurological examination of lower limbs.

Examination

Follow the examination procedure as "**Flaccid paraplegia**".

Schematic Diagnosis

	Neurological examination
Inspection and muscle bulk	Wasting and fasciculation
	↓
Tone	Decreased tone
	↓
Muscle power	Diminished (0/5) in both lower limbs
	↓

JAYPEE BROTHERS

Jerks and plantar response	Absent knee and ankle jerks, even after reinforcement, flexor plantar response
	↓
	Flaccid paraplegia
	↓
Sensation	No loss of sensation
	↓
Differential diagnosis	Guillain–Barre syndrome Hypokalemia

Q. Please tell your findings

A.
- Neurological examination of lower limbs reveals—there is wasting and fasciculation, muscle power is 0/5 with hypotonia, knee and ankle jerks are absent and flexor plantar responses. All modalities of sensation are intact in both lower limbs. Coordination and gait are not possible.
- This patient has flaccid paraplegia without sensory loss.

Q. What is your diagnosis?

A. Flaccid paraplegia without sensory loss, probably due to "**Guillain–Barre syndrome**".

Q. Why have you told this?

A. Flaccid paraplegia without sensory loss and sphincter involvement suggest Guillain–Barre syndrome, though hypokalaemia is not completely ruled out.

Q. What history would you like to ask this patient?

A. History of respiratory tract infection and bloody diarrhea.

Q. Why is this not CIDP?

A. As the presentation is of short duration, i.e. 3 days. CIDP typically presents with relapsing or progressive motor and sensory changes, evolving over more than 8 weeks.

Q. What are the causes of acute flaccid paralysis?

A. The causes includes:

a. **Infection**
   1. Viral:
      - Poliomyelitis
      - Tick paralysis
      - Japanese encephalitis
      - West Nile virus
      - Paralytic rabies.
   2. Bactrial:
      - Botulism
      - Diphtheria
      - Lyme disease

b. **Peripheral neuropathy**
   - Guillain–Barre syndrome
   - Acute intermittent porphyria
   - Lead poisoning.

c. **Spinal disease (due to spinal shock)**
   - Acute transverse myelitis
   - Spinal cord compression
   - Spinal cord infarction.

Discussion about Guillain–Barre Syndrome—Please read long case—"Guillain–Barre Syndrome".

MONOPLEGIA

Brown–Sequard Syndrome

Scenario

A 45-year-old man presents with weakness of left leg for 5 days.

Instruction

Examine the lower limbs/neurological examination of lower limbs.

Proceed as Follows

1. Address the patient as your custom "Hi, Hello, Assalamu alaykum" and introduce yourself and tell your role.
2. Maintain 3Ps—Permission, Position and Proper exposure (preferably at supine position).
3. Look at the patient (from head to toe) and surroundings while positioning and exposing, and spend a few seconds to get any clue, e.g. walking stick, wheelchair, wasting, and indwelling urinary catheter (sphincter involvement).
4. Inspect the limbs and look for wasting, fasciculation and atrophy.
5. Assess **bulk of muscle**. No need to measure by tape if there is no gross asymmetry and wasting.
6. Assess the **muscle tone** in each leg by passively moving it at the hip and knee joints (with the patient relaxed roll the leg sideways, backwards and forwards on the bed; lift the knee and let it drop, or bend the knee and partially straighten in an irregular and unexpected rhythm).
   Note: In this patient, there is hypertonia in left leg (spastic type) but normal in right leg.
7. Test **muscle power** as a whole by asking the patient to raise his or her leg straight upward, then test power of individual group and grade muscle power.
   Note: In this patient, muscle power is 0/5 in left leg and 5/5 in right leg.
8. Elicit **jerks,** i.e knee (L3, 4) and ankle (S1, 2) jerks and also ankle and patellar clonus.
   Note: In this patient, knee and ankle jerks are exaggerated and ankle clonus is present in left side and normal in right side.

JAYPEE BROTHERS

For detailed information on medicine books, visit our website **www.jaypeebrothers.com,** for detailed information on medici

Short & Long Cases in CLINICAL MEDICINE

Salient Features

- A unique text on the subject
- Presents 41 common long cases and 102 short cases
- Provides essential knowledge about history taking
- Elaborates examination procedures thoroughly
- Contains long cases as in examination settings
- Describes examination in short cases step by steps which helps the student to learn methodically
- Focuses on causes, differential diagnoses and schematic approach of the disease conditions
- Very helpful for both undergraduate and postgraduate students to prepare themselves for examination.



Harendra Nath Sarker MBBS FCPS (Med) MRCP (UK) MRCP (Neuro) FRCP (Edin) is Professor, Department of Medicine, Sher-e-Bangla Medical College, Barisal, Bangladesh. He obtained MBBS degree in 1984; FCPS in Internal Medicine in 2004 from Bangladesh College of Physicians and Surgeons of Bangladesh; MRCP in 2011 from the Royal Colleges of Physicians, UK, and FRCP (Edin) in 2013. He did also MRCP (Neurology) in 2016.

He has been serving as a teacher since 1995 in different medical colleges, as Registrar, Assistant Professor, Associate Professor and Professor. He is an examiner of undergraduate and postgraduate examinations. He is an author of five medical books. He has more than 50 publications to his credit in various national and international journals.

Available at all medical bookstores
or buy online at www.jaypeebrothers.com

JAYPEE BROTHERS
Medical Publishers (P) Ltd.
www.jaypeebrothers.com

Join us on facebook.com/JaypeeMedicalPublishers

Shelving Recommendation
MEDICINE

ISBN 978-93-86056-89-4