

NEET SS 2024 Diploma Paediatrics Question Paper 2 with Solutions

Time Allowed :3 Hours	Maximum Marks :100	Total Questions :10
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General Instructions

Read the following instructions very carefully and strictly follow them:

1. The test is of 3 hours duration.
2. The question paper consists of 10 questions. The maximum marks are 100.
3. Each Question is of 10 marks.

Q1. Migraine:

a) Classification. [3]

Solution:

Step 1: Definition of Classification.

Migraine is classified based on the frequency, duration, and severity of attacks. The main classifications include:

- (1) Migraine without Aura - The most common type, characterized by a unilateral headache without preceding neurological symptoms.
- (2) Migraine with Aura - This type includes visual, sensory, or speech disturbances before the headache. Aura usually lasts less than 60 minutes.
- (3) Chronic Migraine - Defined as having more than 15 headache days per month for at least 3 months.
- (4) Hemiplegic Migraine - A rare type that includes temporary paralysis on one side of the body during the migraine attack.
- (5) Retinal Migraine - Characterized by temporary vision loss or visual disturbances in one eye during a migraine attack.

Quick Tip

Understanding the classification of migraines is essential for proper diagnosis and treatment, as treatment strategies vary based on the type.

Q1. b) Clinical manifestation. [3]

Solution:

Step 1: General Manifestations.

The clinical manifestations of migraine typically include:

- (1) Headache - Unilateral, throbbing pain, often aggravated by physical activity, and commonly lasting from 4 to 72 hours.
- (2) Nausea and Vomiting - Common symptoms that accompany the headache in many migraine sufferers.
- (3) Photophobia and Phonophobia - Sensitivity to light and sound, often leading patients to seek a dark, quiet place during attacks.
- (4) Aura (in some cases) - Visual disturbances like flashing lights, blind spots, or zigzag lines, and sensory disturbances like tingling or numbness, typically occurring before the headache.

Step 2: Additional Features.

- (5) Aura Symptoms - These can involve vision changes, sensory disturbances, or speech difficulties and usually last between 5 to 60 minutes.
- (6) Prodrome - Symptoms that can occur a day or two before the headache, including mood changes, food cravings, or neck stiffness.

Quick Tip

Recognizing the early signs and symptoms of a migraine, including prodromal and aura phases, can help in the timely management and prevention of attacks.

Q1. c) Treatment. [4]

Solution:**Step 1: Acute Treatment.**

Acute treatment aims to relieve symptoms during an active migraine attack. Common treatments include:

- (1) NSAIDs - Non-steroidal anti-inflammatory drugs (e.g., ibuprofen) for pain relief.
- (2) Triptans - Serotonin receptor agonists (e.g., sumatriptan) are highly effective for aborting an acute attack.
- (3) Antiemetics - Medications like metoclopramide can be used for nausea and vomiting associated with migraines.
- (4) Ergotamines - Older drugs (e.g., ergotamine) that can be effective but have more side effects.

Step 2: Preventive Treatment.

Preventive treatment is used for frequent migraines, and options include:

- (1) Beta-blockers - Drugs like propranolol help in reducing the frequency of attacks.
- (2) Antidepressants - Tricyclic antidepressants like amitriptyline are effective for chronic migraines.

- (3) Anticonvulsants - Medications like topiramate and valproic acid are commonly used for migraine prevention.
- (4) CGRP Inhibitors - Newer medications like erenumab that block the calcitonin gene-related peptide, involved in the migraine process.

Step 3: Non-Pharmacological Treatments.

- (1) Lifestyle Modifications - Identifying triggers (e.g., stress, sleep patterns, diet) and adopting strategies to avoid them.
- (2) Cognitive Behavioral Therapy (CBT) - Psychological therapies that can help manage stress and prevent migraines.

Quick Tip

The goal of treatment is to relieve acute symptoms and prevent future attacks. A combination of pharmacological and non-pharmacological treatments is often most effective.

Q2. Chronic leukemia in children:

a) Classification. [2]

Solution:

Step 1: Classification of Chronic Leukemia.

Chronic leukemia in children is generally classified into two types:

- (1) Chronic Lymphocytic Leukemia (CLL) - Rare in children, characterized by the accumulation of mature but functionally impaired lymphocytes.
- (2) Chronic Myelogenous Leukemia (CML) - A hematological malignancy where there is uncontrolled proliferation of myeloid cells.

Quick Tip

In children, chronic leukemias are less common than acute forms. CML is more frequent in childhood than CLL.

Q2. b) Immunophenotyping. [2]

Solution:

Step 1: Importance of Immunophenotyping.

Immunophenotyping is used to classify leukemia based on the presence of specific cell surface

markers using flow cytometry. This is essential for distinguishing between different types of leukemia and determining the appropriate treatment.

(1) Chronic Myelogenous Leukemia (CML): Cells typically express CD13, CD33, and CD15 markers.

(2) Chronic Lymphocytic Leukemia (CLL): Cells are usually positive for CD5, CD19, and CD23 markers, with low or absent expression of CD20.

Quick Tip

Immunophenotyping helps in identifying the leukemia subtype and tailoring therapy accordingly, especially in chronic leukemia cases.

Q2. c) Risk stratification. [2]

Solution:

Step 1: Importance of Risk Stratification.

Risk stratification in chronic leukemia helps to assess the prognosis and determine the intensity of treatment required. This includes evaluating genetic, molecular, and clinical features.

(1) For CML: The presence of the Philadelphia chromosome (BCR-ABL fusion gene) is a key factor. Patients with this chromosome generally have a higher risk of progression.

(2) For CLL: Factors like age, stage, and serum markers (e.g., ZAP-70, CD38) are considered to assess risk.

Quick Tip

Risk stratification allows clinicians to identify high-risk patients early and adapt treatment plans to prevent complications.

Q2. d) Management. [4]

Solution:

Step 1: Management of Chronic Leukemia.

Management of chronic leukemia in children varies depending on the type (CML or CLL) and the individual patient's characteristics.

For Chronic Myelogenous Leukemia (CML):

(1) Tyrosine Kinase Inhibitors (TKIs): The first-line treatment for CML is Imatinib, which

targets the BCR-ABL fusion gene. Other TKIs like Dasatinib and Nilotinib may be used in resistant cases.

(2) Stem Cell Transplantation: For patients who do not respond to TKIs, a bone marrow or stem cell transplant may be considered.

For Chronic Lymphocytic Leukemia (CLL):

(1) Chemotherapy: Common regimens include Fludarabine, Cyclophosphamide, and Rituximab.

(2) Immunotherapy: Rituximab, an anti-CD20 monoclonal antibody, is used to target malignant B-cells.

(3) Stem Cell Transplantation: This may be considered for high-risk cases or when chemotherapy is ineffective.

Step 2: Supportive Care.

(1) Blood Transfusions: To treat anemia and thrombocytopenia.

(2) Infection Control: As both CML and CLL can lead to an increased risk of infections, antibiotics, and vaccines may be used as part of supportive care.

Quick Tip

The treatment of chronic leukemia is primarily based on molecular and genetic factors, with TKIs being the cornerstone for CML and chemotherapy/immunotherapy for CLL.

Q3. Neuro-cutaneous markers:

a) Types and appearance. [4]

Solution:

Step 1: Definition of Neuro-cutaneous Markers.

Neurocutaneous markers refer to cutaneous manifestations that are indicative of underlying neurodevelopmental or neurological disorders. These markers are important in diagnosing neurocutaneous syndromes.

Step 2: Types of Neuro-cutaneous Markers.

(1) Café-au-lait Spots - Light brown macules with irregular borders, often seen in conditions like Neurofibromatosis type 1 (NF1).

(2) Ash-leaf Macules - Hypopigmented macules seen in Tuberous Sclerosis.

(3) Port-wine Stains - Vascular malformations that may be seen in Sturge-Weber Syndrome.

(4) Sebaceous Nevi - Yellowish, raised lesions that can be seen in Basal Cell Nevus Syndrome.

(5) Fibromas - Soft, flesh-colored tumors typically associated with Neurofibromatosis type 1.

Step 3: Appearance of Markers.

(1) Café-au-lait Spots: Irregular, light brown spots, often more than six in number in NF1.

(2) Ash-leaf Macules: Small, oval-shaped light-colored spots, typically appearing on the trunk

and limbs.

(3) Port-wine Stains: Red or purple discoloration of the skin, often involving the face or limbs.

(4) Sebaceous Nevi: Yellowish, raised patches usually found on the scalp or face.

Quick Tip

Neurocutaneous markers are helpful clues in diagnosing genetic syndromes. They often correlate with underlying systemic involvement.

Q3. b) Describe four important neurocutaneous syndromes in brief. [6]

Solution:

Step 1: Overview of Neurocutaneous Syndromes.

Neurocutaneous syndromes are a group of disorders that affect both the skin and nervous system. They are usually inherited and can have variable clinical manifestations.

Step 2: Important Neurocutaneous Syndromes.

(1) Neurofibromatosis Type 1 (NF1):

NF1 is characterized by the presence of multiple café-au-lait spots, neurofibromas (benign nerve tumors), and Lisch nodules (iris hamartomas). It can also cause learning disabilities, skeletal abnormalities, and increased risk of malignancies.

(2) Tuberous Sclerosis Complex (TSC):

TSC is marked by the presence of ash-leaf macules, subungual fibromas, and shagreen patches. It can cause seizures, intellectual disabilities, and organ involvement (kidneys, heart, and lungs).

(3) Sturge-Weber Syndrome:

Sturge-Weber syndrome involves a port-wine stain over the face, often on one side, and can be associated with neurological issues such as seizures, glaucoma, and developmental delay. The syndrome affects the brain and skin.

(4) Von Hippel-Lindau Disease:

This syndrome presents with hemangioblastomas (tumors) in the brain, retina, and spinal cord, as well as pheochromocytomas (tumors in the adrenal glands). It can cause vision loss, headaches, and high blood pressure.

Step 3: Conclusion.

These neurocutaneous syndromes share skin manifestations but have varying clinical presentations. Diagnosis is important for management, including surveillance for associated malignancies.

Quick Tip

Recognizing the neurocutaneous markers can help diagnose these syndromes early, which is crucial for appropriate management and surveillance.

Q4. Short stature:

a) Definition. [3]

Solution:

Step 1: Definition of Short Stature.

Short stature is defined as a height that is more than 2 standard deviations (SD) below the mean height for a given age and sex, typically less than the 3rd percentile on growth charts. It can result from a variety of genetic, nutritional, hormonal, or pathological factors.

Step 2: Diagnostic Criteria.

Short stature is not a diagnosis in itself, but a clinical finding that warrants investigation into the underlying cause. Growth charts are used to assess whether a child's height is significantly below the expected growth trajectory.

Quick Tip

Short stature should be assessed in relation to growth patterns, family history, and other health factors before making a diagnosis.

Q4. b) Types. [3]

Solution:

Step 1: Types of Short Stature.

Short stature can be classified into the following types:

- (1) Constitutional Growth Delay - A normal variation in growth where children grow at a slower pace but eventually catch up to normal growth by adulthood. These children typically have a family history of delayed growth.
- (2) Familial Short Stature - Inherited short stature, where the child's height is proportionately short due to family genetics, without any underlying pathological cause.
- (3) Pathological Short Stature - Resulting from medical conditions affecting growth, such as:
 - Endocrine Disorders: Hypothyroidism, growth hormone deficiency, or other hormonal imbalances.
 - Chronic Diseases: Malnutrition, chronic kidney disease, or other systemic illnesses.

- Genetic Syndromes: Turner syndrome, Down syndrome, or skeletal dysplasias.
- (4) Idiopathic Short Stature - When no clear cause for short stature is found despite investigation. It may be due to a variety of unknown factors.

Quick Tip

Differentiating between constitutional growth delay and pathological short stature is essential for appropriate management and prognosis.

Q4. c) Management. [4]

Solution:

Step 1: General Approach to Management.

Management of short stature depends on the underlying cause, and a thorough evaluation is required to identify the type of short stature.

Step 2: Management based on Cause.

(1) Constitutional Growth Delay:

No medical treatment is typically required. Monitoring growth patterns and regular follow-up visits to ensure normal development are advised. Reassurance is key for parents.

(2) Familial Short Stature:

Similar to constitutional growth delay, the primary approach is monitoring growth, and there is no specific medical treatment. These children typically reach their genetic potential in adulthood.

(3) Pathological Short Stature:

- Endocrine Disorders: Hormonal treatments such as growth hormone therapy (for growth hormone deficiency) or thyroid replacement therapy (for hypothyroidism) may be needed.
- Chronic Diseases: Treatment of the underlying disease (e.g., nutritional interventions for malnutrition, dialysis for kidney disease) is important to support growth.
- Genetic Syndromes: While growth hormone therapy may help, the treatment is largely supportive. For conditions like Turner syndrome, growth hormone therapy and estrogen therapy may be used to support growth and pubertal development.

(4) Idiopathic Short Stature:

Treatment may include growth hormone therapy in cases where the short stature is significantly affecting quality of life or when it is associated with psychological or social difficulties.

Step 3: Monitoring and Support.

For all types of short stature, regular follow-up with growth assessments, lab tests (e.g., thyroid

function, growth hormone levels), and addressing psychological impacts is essential.

Quick Tip

Early intervention and appropriate treatment of underlying conditions can significantly improve growth outcomes in children with short stature.

Q5. Hypertension in children:

a) Definition. [2]

Solution:

Step 1: Definition of Hypertension in Children.

Hypertension in children is defined as a sustained elevated blood pressure that is above the 95th percentile for age, sex, and height, measured on at least three different occasions. Blood pressure measurements are categorized as follows:

- (1) Normal: Systolic BP \leq 90th percentile
- (2) Pre-hypertension: Systolic BP between 90th and 95th percentile
- (3) Hypertension: Systolic BP \geq 95th percentile for age, sex, and height.

Quick Tip

For diagnosing hypertension in children, repeated measurements and proper technique are essential to avoid false readings.

Q5. b) Aetiology. [2]

Solution:

Step 1: Aetiology of Hypertension in Children.

The causes of hypertension in children can be classified into primary and secondary causes:

- (1) Primary Hypertension: Often due to genetic and environmental factors, such as obesity, lack of physical activity, or high salt intake. It is more common in adolescents and is similar to adult hypertension.
- (2) Secondary Hypertension: Caused by an underlying medical condition, which includes:
 - Renal Disease (e.g., chronic kidney disease, glomerulonephritis)
 - Endocrine Disorders (e.g., hyperthyroidism, Cushing's syndrome, pheochromocytoma)
 - Cardiovascular Diseases (e.g., coarctation of the aorta)

- Medications (e.g., corticosteroids, birth control pills)
- Obstructive Sleep Apnea (OSA).

Quick Tip

Secondary hypertension should be suspected in children with no family history of hypertension or those with unusual presentations or younger age of onset.

Q5. c) Investigation. [2]

Solution:

Step 1: Investigations for Hypertension in Children.

The investigation of hypertension in children involves identifying the underlying cause, confirming the diagnosis, and assessing for organ damage. Common investigations include:

- (1) Repeated Blood Pressure Measurements: To confirm sustained hypertension on at least three separate occasions.
- (2) Urinalysis and Kidney Function Tests: To check for renal causes such as proteinuria or kidney damage.
- (3) Electrolyte and Renal Panel: To assess renal function and detect disorders like hyperaldosteronism.
- (4) Echocardiogram: To assess for heart-related causes or damage from hypertension.
- (5) Endocrine Tests: To screen for conditions like hyperthyroidism, pheochromocytoma, or Cushing's syndrome.
- (6) Fundoscopy: To evaluate for signs of target organ damage, such as retinopathy.

Quick Tip

Early diagnosis and investigation of secondary causes of hypertension is crucial to prevent long-term complications.

Q5. d) Management. [4]

Solution:

Step 1: General Approach to Management.

Management of hypertension in children involves lifestyle changes, pharmacological therapy, and treatment of underlying causes.

Step 2: Non-pharmacological Management.

(1) Lifestyle Modifications:

- Diet: Reducing sodium intake, promoting a balanced diet, and ensuring adequate potassium intake.
- Exercise: Encouraging regular physical activity to reduce weight and improve cardiovascular health.
- Weight Reduction: In obese children, reducing weight can have a significant impact on blood pressure.
- Limiting Caffeine and Alcohol Intake: In adolescents, reducing the intake of stimulants and alcohol.

Step 3: Pharmacological Management.

(1) Angiotensin-Converting Enzyme (ACE) Inhibitors: Commonly used to lower blood pressure and protect kidney function.

(2) Angiotensin Receptor Blockers (ARBs): Another class of drugs used for controlling blood pressure.

(3) Calcium Channel Blockers (CCBs): Often used in cases where ACE inhibitors are not sufficient or tolerated.

(4) Diuretics: To reduce fluid volume and control blood pressure, especially in cases with fluid retention.

(5) Beta-blockers: May be used for children with secondary hypertension or those with heart involvement.

Step 4: Management of Secondary Hypertension.

If secondary causes are identified, treatment should focus on managing the underlying condition (e.g., surgery for coarctation of the aorta, pharmacological treatment for endocrine disorders).

Quick Tip

A combination of lifestyle changes and medications is often required for effective management of hypertension in children, especially in cases with secondary causes.

Q6. Disseminated intravascular coagulation:

a) Etiology. [3]

Solution:

Step 1: Etiology of Disseminated Intravascular Coagulation (DIC).

DIC is a complex and systemic disorder characterized by widespread activation of the coagulation system, leading to the formation of microthrombi and consumption of clotting factors. The etiology of DIC includes:

(1) Infections: Particularly sepsis, which is the most common cause of DIC, especially with gram-negative bacteria and viral infections.

- (2) Trauma and Burns: Major trauma or burns can cause tissue factor release, triggering DIC.
- (3) Malignancies: Certain cancers, especially solid tumors (e.g., pancreatic cancer) and hematological malignancies (e.g., acute leukemia), can lead to DIC.
- (4) Obstetric Complications: Conditions such as placental abruption, amniotic fluid embolism, or severe preeclampsia/eclampsia.
- (5) Liver Disease: Severe liver failure impairs the synthesis of clotting factors, contributing to DIC.
- (6) Massive Transfusion: Over-transfusion of blood products can lead to DIC due to dilution of clotting factors and platelet dysfunction.

Quick Tip

DIC is often secondary to an underlying condition, and addressing the root cause is key to treatment and management.

Q6. b) Diagnosis. [3]

Solution:

Step 1: Diagnosis of Disseminated Intravascular Coagulation (DIC).

The diagnosis of DIC is based on clinical findings, laboratory tests, and identification of underlying causes. Key diagnostic features include:

- (1) Clinical Features: Symptoms such as bleeding (e.g., petechiae, ecchymoses), organ dysfunction (e.g., renal failure), and thrombotic events (e.g., deep vein thrombosis, pulmonary embolism).
- (2) Laboratory Tests:
 - Platelet Count: Typically low due to consumption in thrombi formation.
 - Prothrombin Time (PT) and Activated Partial Thromboplastin Time (aPTT): Prolonged, reflecting clotting factor consumption.
 - Fibrinogen Levels: Decreased due to fibrinogen consumption in clot formation.
 - D-dimer: Elevated, as D-dimers are produced during fibrin breakdown.
 - Schistocytes on Peripheral Blood Smear: Fragmented red blood cells, indicating microangiopathic hemolysis.
- (3) Identifying Underlying Causes: Diagnosis also includes identifying the underlying condition causing DIC, such as infections, trauma, or malignancy.

Quick Tip

DIC is a clinical diagnosis supported by laboratory findings. Early detection and intervention are critical to prevent severe complications.

Q6. c) Treatment. [4]

Solution:

Step 1: General Approach to Treatment of DIC.

Treatment of DIC focuses on managing the underlying cause, controlling bleeding, and supporting organ function.

Step 2: Management of Underlying Cause.

- (1) Infections: Appropriate antibiotics or antiviral therapy should be started immediately. In cases of sepsis, early source control (e.g., drainage of abscesses) is crucial.
- (2) Obstetric Causes: Management of obstetric emergencies like placental abruption, amniotic fluid embolism, or eclampsia is essential.
- (3) Malignancy: Treating the underlying malignancy, such as chemotherapy or surgery for tumor resection, is important.

Step 3: Supportive Treatment.

- (1) Blood Product Replacement: Transfusions of fresh frozen plasma (FFP), platelets, and cryoprecipitate to replace consumed clotting factors and restore normal blood clotting.
- (2) Antithrombotic Therapy: In some cases, heparin may be used to prevent further clotting, especially in patients with thrombotic manifestations. However, heparin use is controversial and must be carefully monitored, as it can exacerbate bleeding in some patients.
- (3) Renal Support: In cases of renal failure, dialysis may be necessary.
- (4) Organ Support: Intensive care support to manage multi-organ dysfunction, including respiratory support (e.g., mechanical ventilation) and circulatory support (e.g., vasopressors for hypotension).

Step 4: Monitoring and Follow-Up.

Frequent monitoring of coagulation parameters, platelet counts, and clinical status is necessary to adjust treatment and prevent complications.

Quick Tip

The treatment of DIC is multifaceted, with a focus on managing the underlying cause and providing supportive care to prevent complications such as bleeding and organ failure.

Q7. Paediatric SLE:

a) Clinical manifestation. [2]

Solution:

Step 1: Clinical Manifestations of Pediatric SLE.

Systemic lupus erythematosus (SLE) in children often presents with a wide range of symptoms,

which can vary in severity. Common clinical manifestations include:

- (1) Malar Rash: A butterfly-shaped rash across the cheeks and nose, often triggered by sun exposure.
- (2) Discoid Rash: Round, scaly, erythematous plaques that may scar and leave pigmentation changes.
- (3) Arthritis: Joint pain and swelling, especially in small joints, resembling juvenile arthritis.
- (4) Renal Involvement: Proteinuria and hematuria, which can lead to lupus nephritis.
- (5) Photosensitivity: Increased sensitivity to sunlight, leading to rashes and exacerbation of symptoms.
- (6) Oral Ulcers: Painful sores inside the mouth, often a characteristic sign.
- (7) Systemic Symptoms: Fatigue, fever, weight loss, and lymphadenopathy.
- (8) Neurological Symptoms: Seizures, psychosis, and cognitive dysfunction.

Quick Tip

In pediatric SLE, early recognition of characteristic signs like the malar rash and renal involvement is crucial for prompt diagnosis and treatment.

Q7. b) Investigation. [2]

Solution:

Step 1: Investigations for Pediatric SLE.

The diagnosis of SLE is supported by clinical findings and laboratory investigations. Common investigations include:

- (1) Complete Blood Count (CBC): To assess for anemia, leukopenia, or thrombocytopenia, which are common in SLE.
- (2) Urinalysis: To detect proteinuria, hematuria, and casts, which are signs of lupus nephritis.
- (3) Antinuclear Antibodies (ANA): Positive in most cases of SLE, though not specific.
- (4) Anti-dsDNA and Anti-Smith Antibodies: Highly specific for SLE and help in confirming the diagnosis.
- (5) Complement Levels (C3, C4): Low levels of complement are commonly seen during active disease.
- (6) Renal Function Tests: To monitor kidney involvement, including serum creatinine and urinalysis.
- (7) Erythrocyte Sedimentation Rate (ESR) and C-reactive Protein (CRP): Inflammatory markers that may be elevated during disease flares.

Quick Tip

Renal involvement is common in pediatric SLE, so regular urinalysis and renal function tests are essential for monitoring the disease.

Q7. c) Diagnostic criteria. [2]

Solution:

Step 1: Diagnostic Criteria for Pediatric SLE.

The diagnosis of pediatric SLE is based on the classification criteria established by the American College of Rheumatology (ACR) and the Systemic Lupus International Collaborating Clinics (SLICC). Common criteria include:

- (1) Malar Rash – A butterfly-shaped rash across the cheeks and nose.
- (2) Discoid Rash – Round, scaly, erythematous lesions with scarring.
- (3) Photosensitivity – Exacerbation of rashes or symptoms due to sun exposure.
- (4) Oral Ulcers – Painless sores inside the mouth.
- (5) Arthritis – Swelling and pain in two or more joints.
- (6) Serositis – Inflammation of the lining of the heart or lungs, often presenting as pleuritis or pericarditis.
- (7) Renal Involvement – Proteinuria or hematuria, often a hallmark of lupus nephritis.
- (8) Positive ANA – Found in the majority of patients.
- (9) Anti-dsDNA or Anti-Smith Antibodies – Highly specific for SLE.

The diagnosis of pediatric SLE is often made when at least 4 of the above criteria are met, with at least one being clinical (e.g., rash, arthritis) and one being immunological (e.g., positive ANA, anti-dsDNA).

Quick Tip

The classification criteria help in confirming the diagnosis of pediatric SLE but clinical judgment remains essential, especially in early disease.

Q7. d) Treatment. [4]

Solution:

Step 1: General Management of Pediatric SLE.

The treatment of pediatric SLE involves addressing the underlying disease, controlling inflammation, and preventing organ damage.

Step 2: Pharmacological Treatment.

- (1) Nonsteroidal Anti-inflammatory Drugs (NSAIDs): Used for pain and inflammation, especially in cases with mild arthritis or pleuritis.
- (2) Hydroxychloroquine (Plaquenil): An antimalarial drug commonly used in SLE to reduce disease activity and prevent flares.
- (3) Corticosteroids: Prednisone or other steroids are used to manage active disease, particularly in cases of severe flare-ups. The dose depends on the severity of organ involvement (e.g., kidney or CNS).
- (4) Immunosuppressive Agents: For patients with severe disease or organ involvement, drugs such as azathioprine, cyclophosphamide, or mycophenolate mofetil are used to suppress the immune system and prevent damage.
- (5) Biologic Agents: Belimumab (a monoclonal antibody) is used in certain cases of active SLE that do not respond to standard treatments.

Step 3: Supportive Management.

- (1) Renal Support: In cases of lupus nephritis, medications such as mycophenolate mofetil or cyclophosphamide may be used, and dialysis may be required in severe cases.
- (2) Vaccinations: Regular vaccinations are important as children with SLE are at increased risk of infections.
- (3) Monitoring: Regular monitoring of kidney function, blood counts, and disease activity through laboratory tests.

Step 4: Long-term Care.

Long-term management involves preventing flares, managing chronic symptoms, and providing emotional and psychological support for the child and family.

Quick Tip

Pediatric SLE treatment aims to control inflammation, prevent damage to organs like the kidneys, and reduce flare-ups with appropriate immunosuppressive therapy.

Q8. Acute disseminated encephalomyelitis (ADEM):

a) Clinical presentation. [3]

Solution:

Step 1: Clinical Presentation of ADEM.

Acute disseminated encephalomyelitis (ADEM) is a rare, autoimmune, inflammatory demyelinating disorder of the central nervous system, often following a viral infection or vaccination. The clinical presentation includes:

- (1) Acute Onset of Neurological Symptoms: The disease typically presents with rapid onset of symptoms, often days to weeks following an infection or vaccination.

- (2) Fever: Commonly seen at the onset of the illness.
- (3) Neurological Deficits: These include confusion, irritability, headache, and altered consciousness.
- (4) Motor Symptoms: Hemiparesis, quadriparesis, and ataxia are common.
- (5) Visual Disturbances: Blurred vision or optic neuritis can occur.
- (6) Seizures: These may develop, particularly in more severe cases.
- (7) Cerebellar Signs: Ataxia and dysmetria are common, especially in children.

Quick Tip

ADEM can mimic other neurological conditions, so it is important to consider recent infections or vaccinations in the history.

Q8. b) Diagnosis. [3]

Solution:

Step 1: Diagnosis of ADEM.

The diagnosis of ADEM is primarily clinical, supported by imaging and laboratory findings. Key steps in diagnosis include:

- (1) Clinical History: Recent history of viral infection (e.g., measles, mumps, or varicella) or vaccination (e.g., MMR, influenza) preceding neurological symptoms.
- (2) Neurological Examination: The physical examination often reveals multifocal neurological signs, including motor deficits, sensory changes, and cerebellar involvement.
- (3) Magnetic Resonance Imaging (MRI): The gold standard for diagnosing ADEM. MRI typically shows multiple, asymmetric, hyperintense lesions on T2-weighted images, often involving the white matter of the brainstem, cerebellum, and periventricular areas.
- (4) Cerebrospinal Fluid (CSF) Analysis: Mild pleocytosis (increased white blood cells) and elevated protein levels may be found, but the CSF typically remains normal.
- (5) Exclusion of Other Conditions: ADEM is a diagnosis of exclusion, so other conditions like multiple sclerosis, encephalitis, or acute viral infections should be ruled out.

Quick Tip

MRI with contrast is crucial in diagnosing ADEM, as it reveals the characteristic demyelinating lesions that help differentiate it from other conditions.

Q8. c) Investigation and treatment. [4]

Solution:

Step 1: Investigations for ADEM.

Investigations for ADEM focus on confirming the diagnosis and excluding other conditions. Common investigations include:

- (1) MRI of the Brain and Spine: To identify the characteristic demyelinating lesions, which are typically multiple, asymmetrical, and involve the brainstem, basal ganglia, and periventricular areas.
- (2) CSF Analysis: Mild pleocytosis with an elevated protein level may be observed. The CSF usually lacks oligoclonal bands, which helps differentiate ADEM from multiple sclerosis.
- (3) Blood Tests: To rule out infections, autoimmune conditions, or metabolic causes. Tests for viruses like Epstein-Barr virus (EBV), cytomegalovirus (CMV), and other infectious agents may be performed.
- (4) Electroencephalogram (EEG): In cases with seizures, EEG can help assess for abnormal brain activity.

Step 2: Treatment of ADEM.

The treatment of ADEM focuses on controlling inflammation and managing symptoms. Key treatment options include:

- (1) Corticosteroids: High-dose intravenous methylprednisolone is the first-line treatment. This helps to reduce inflammation and prevent further demyelination. Typically administered for 3-5 days, followed by oral prednisone tapering.
- (2) Plasmapheresis: In severe or refractory cases, plasmapheresis may be considered to remove circulating antibodies and cytokines that contribute to the inflammatory process.
- (3) Symptomatic Treatment: Antiepileptic drugs for seizure control, analgesics for headache, and physical therapy to address motor deficits and improve mobility.
- (4) Supportive Care: Management in an intensive care unit (ICU) may be required for patients with severe symptoms or respiratory compromise.

Step 3: Prognosis and Follow-up.

Most children recover fully or with minimal residual neurological deficits within a few months. However, some may experience long-term neurological sequelae such as motor deficits or cognitive impairment. Regular follow-up is needed to monitor for recovery and address any ongoing symptoms.

Quick Tip

Early treatment with steroids is key in improving outcomes in ADEM, and most patients show good recovery, but early intervention is essential to prevent long-term disability.

Q9. Autism spectrum disorder:

a) Clinical feature. [3]

Solution:

Step 1: Clinical Features of Autism Spectrum Disorder.

Autism spectrum disorder (ASD) is a neurodevelopmental disorder characterized by a range of symptoms. The clinical features typically include:

- (1) Impaired Social Interaction: Difficulty in understanding social cues, maintaining eye contact, and forming relationships. Children with ASD often prefer solitary activities and have difficulty engaging in peer interactions.
- (2) Communication Challenges: Delayed speech and language development, or a complete lack of speech. Some individuals may use repetitive or abnormal speech patterns.
- (3) Repetitive Behaviors: Engaging in repetitive movements (e.g., hand-flapping, rocking) or rituals. A strong need for sameness and routine is common.
- (4) Sensory Sensitivities: Overreacting or underreacting to sensory stimuli, such as lights, sounds, textures, or smells.
- (5) Restricted Interests: Intense focus on specific topics or activities, often to the exclusion of everything else.

Quick Tip

The clinical presentation of ASD can vary widely in severity, from individuals who are nonverbal to those with highly developed intellectual and communication skills.

Q9. b) Diagnosis. [3]

Solution:

Step 1: Diagnosis of Autism Spectrum Disorder.

The diagnosis of ASD is primarily clinical, based on behavioral assessments and developmental history. Key steps in diagnosis include:

- (1) Developmental Screening: Early screening during routine pediatric visits using tools such as the Modified Checklist for Autism in Toddlers (M-CHAT) or Autism Spectrum Rating Scales (ASRS).
- (2) Comprehensive Diagnostic Evaluation: This includes detailed assessments by a team of professionals (e.g., pediatricians, psychologists, speech therapists) to assess developmental milestones, behavior patterns, and family history.
- (3) Observational Tools: Standardized tests such as the Autism Diagnostic Observation Schedule (ADOS) or Autism Diagnostic Interview-Revised (ADI-R) are used to observe and score behavior indicative of autism.
- (4) Exclusion of Other Conditions: ASD is diagnosed by exclusion, so other conditions such as hearing loss, intellectual disability, or speech delays need to be ruled out.

(5) Genetic Testing: Although not routinely required, genetic testing may be considered, especially in cases of unusual presentations or if there is a suspicion of a genetic syndrome.

Quick Tip

A comprehensive evaluation by a multidisciplinary team, including developmental history and observation, is essential for an accurate diagnosis of ASD.

Q9. c) Management. [4]

Solution:

Step 1: General Approach to Management of Autism Spectrum Disorder.

Management of ASD involves a combination of behavioral, educational, and therapeutic interventions. The focus is on improving communication, social skills, and daily functioning.

Step 2: Behavioral Interventions.

- (1) Applied Behavior Analysis (ABA): A widely used therapeutic approach to improve specific behaviors, communication, and social skills through reinforcement and structured learning.
- (2) Speech and Language Therapy: To help with communication skills, including improving speech, nonverbal communication, and social interactions.
- (3) Social Skills Training: Targeted programs designed to help children with ASD improve their ability to interact with peers and understand social cues.

Step 3: Educational Interventions.

- (1) Individualized Education Plan (IEP): Tailored educational plans that address the child's specific learning needs, often including one-on-one instruction or specialized programs.
- (2) Specialized Schools and Programs: In some cases, placement in a specialized school for children with ASD may be recommended. These schools provide tailored curricula and specialized teaching techniques.

Step 4: Pharmacological Treatment.

- (1) Medications for Comorbid Symptoms: While no medication cures ASD, certain medications may be used to manage symptoms such as anxiety, irritability, or hyperactivity. Common medications include:
 - Antipsychotic Medications: e.g., Risperidone or Aripiprazole to reduce irritability and aggression.
 - Selective Serotonin Reuptake Inhibitors (SSRIs): For anxiety and obsessive-compulsive behaviors.
 - Stimulants: Used in cases of ADHD symptoms to improve focus and impulse control.
- (2) Melatonin: To help manage sleep disturbances commonly seen in children with ASD.

Step 5: Family Support and Counseling.

Support for families, including counseling and parent training, is important to help caregivers

manage challenges and provide better care.

Quick Tip

Early intervention is crucial for improving long-term outcomes in children with ASD, and a multidisciplinary approach is often most effective.

Q10. Extrahepatic portal hypertension:

a) Pathophysiology. [3]

Solution:

Step 1: Pathophysiology of Extrahepatic Portal Hypertension.

Extrahepatic portal hypertension (EHPH) is a condition where there is increased pressure within the portal venous system due to obstruction or narrowing of the extrahepatic portal vein. The pathophysiology includes:

- (1) **Obstruction of the Portal Vein:** The most common cause is thrombosis or fibrosis of the extrahepatic portal vein. This leads to impaired blood flow and increased pressure in the portal system.
- (2) **Development of Collateral Circulation:** As a result of the obstructed portal flow, collateral veins (e.g., esophageal, gastric, or rectal veins) dilate to bypass the obstruction, leading to varices and other complications.
- (3) **Increased Portal Pressure:** The backup of blood increases pressure in the spleen and gastrointestinal tract, leading to splenomegaly and variceal bleeding.
- (4) **Liver Function:** In EHPH, the liver is usually not involved in the obstructive process, unlike cirrhosis. However, portal pressure rises as a result of the obstruction or thrombosis of the extrahepatic portal vein.

Quick Tip

EHPH is typically caused by a blockage of the portal vein or its branches, and the key feature is the development of collateral circulation to bypass the blockage.

Q10. b) Clinical feature. [3]

Solution:

Step 1: Clinical Features of Extrahepatic Portal Hypertension.

The clinical presentation of extrahepatic portal hypertension is often related to the complications of increased portal venous pressure and collateral circulation. Common features include:

- (1) Splenomegaly: Enlargement of the spleen due to increased pressure in the portal system, often the first sign of EHPH.
- (2) Variceal Bleeding: The development of dilated varices in the esophagus, stomach, or rectum can lead to life-threatening bleeding. This is a common and severe complication of EHPH.
- (3) Ascites: The accumulation of fluid in the abdomen due to increased portal pressure can cause abdominal distention.
- (4) Thrombocytopenia: A reduced platelet count due to sequestration in the enlarged spleen, leading to a risk of bleeding.
- (5) Jaundice: Though not typical in EHPH, jaundice may occur if there is associated liver involvement or if the condition leads to hepatic dysfunction.
- (6) Abdominal Pain and Discomfort: Due to the enlarged spleen or variceal bleeding.

Quick Tip

The most common clinical feature of EHPH is splenomegaly, often accompanied by thrombocytopenia and variceal bleeding, which can be life-threatening.

Q10. c) Management. [4]

Solution:

Step 1: General Approach to Management of Extrahepatic Portal Hypertension.

The management of EHPH focuses on addressing the underlying cause, controlling complications, and preventing variceal bleeding.

Step 2: Management of Complications.

- (1) Variceal Bleeding:
 - Endoscopic Therapy: Endoscopic variceal ligation (EVL) or sclerotherapy is performed to control bleeding from esophageal or gastric varices.
 - Beta-blockers: Non-selective beta-blockers (e.g., propranolol) can reduce portal pressure and prevent variceal bleeding.
 - Vasoactive Drugs: Drugs like octreotide may be used to control acute bleeding episodes.
- (2) Splenomegaly and Thrombocytopenia:
 - Splenectomy: In severe cases of splenomegaly or thrombocytopenia causing bleeding, a splenectomy may be considered.
- (3) Ascites Management:
 - Diuretics: Loop diuretics (e.g., furosemide) and potassium-sparing diuretics (e.g., spironolactone) are used to manage ascites.

- Paracentesis: Removal of excess fluid from the abdominal cavity if ascites becomes severe.

Step 3: Long-term Management.

- (1) Shunt Surgery or Transjugular Intrahepatic Portosystemic Shunt (TIPS): In some cases, surgical procedures such as a portosystemic shunt or TIPS may be performed to bypass the portal obstruction and reduce portal hypertension.
- (2) Management of Underlying Causes: If EHPH is secondary to another condition (e.g., thrombophilia), treatment of the underlying cause may help prevent further complications.
- (3) Liver Transplantation: In cases where there is progressive liver damage or failure associated with EHPH, liver transplantation may be considered.

Step 4: Follow-up and Monitoring.

Regular monitoring for variceal bleeding and ascites management is important. Ongoing surveillance with imaging and endoscopy may be necessary to assess the condition and manage complications.

Quick Tip

The primary goals in the management of EHPH are to control variceal bleeding, manage splenomegaly, and prevent complications such as ascites.