Short cases in paediatrics

Cardiovascular system

Examination scheme

Remember that most diagnosis in paediatrics is done based on clever observation of the child and the surroundings.

- Ask the age of the child
- Look at the child and get a general impression – Is the child ill looking or well looking? Is the child adequately grown for his or her age, is the child dyspnoeic?
- Look actively for dysmorphic features – especially suggestive of Down syndrome
- General examination
    - Warm to touch/ not
    - Pallor and icterus in the eyes
    - Central cyanosis, dental caries in the mouth
    - Intravenous cannula
    - Clubbing and peripheral stigmata of infective endocarditis (Osler's nodes, Janeway lesions) in the hands
    - Look for features of Marfan's syndrome
    - Ankle/ pedal oedema

Examination of the cardiovascular system proper

Examination of the pulses

- Pulse – Examine the rate, rhythm, character of the pulse. Do not forget to look for the peripheral pulses (especially femoral and dorsalis pedis). Look for radio-femoral delay and radio-radial delay.
- JVP – Is not that important in paediatrics

Examination of the precordium

- Inspection – look for chest deformities, surgical scars, and visible pulsations in the precordium.
- Palpation
    - Palpate for the location of the apex beat- show that you are counting the ribs
    - Get an idea of the character of the apex beat
    - Palpate for thrills
    - Left parasternal heave – Indicates right ventricular hypertrophy
    - Epigastric pulsations
    - Palpable heart sounds, especially P2 – indicates pulmonary hypertension
- Auscultation – Listen to the 1st and 2nd heart sounds and any murmurs

Examine the liver for hepatomegaly (indicates right heart failure) and listen to the lung bases for bilateral end- inspiratory fine crepitations (indicates left heart failure).

Tips and tricks to make a diagnosis (Read and remember these)

General examination

- Age of the child – In pediatrics there are 2 types of cases given at the examination. These are congenital heart disease and patients with rheumatic heart disease. If you are given an older child rheumatic heart disease is more likely and lesions such as PDA are more unlikely.
- Look for the features of Down's syndrome. This is commonly given with underlying cardiovascular lesions.
- Clubbing and cyanosis – This finding narrows down the differential diagnosis considerably. Of the large range of cyanotic congenital cardiac disease TOF is the one given 99% of the time at exams.
- IV cannula – A child given for CVS examination with an IV cannula in situ is most likely a case of infective endocarditis.

Pulse examination

- The pulse is extremely valuable in making a diagnosis. Bounding peripheral pulses especially the dorsalis pedis and posterior tibial pulses indicate a diagnosis of PDA. Remember that PDA is difficult to diagnose based on the murmur alone and the pulses become very important.
- Always remember to feel the femoral pulses. A difficult to feel femoral and lower limb pulses may indicate coarctation of the aorta. (Not commonly given at cases)
- If the pulses are difficult to feel this indicates aortic stenosis (Rare).

Palpation

- Location of the apex beat is important. A displaced apex beat indicates volume overload (MR, AR) or another lesion with heart failure.
- Character of the apex is not that useful in pediatrics.
- A parasternal heave indicates RVH.
- If a prominent thrill is felt the diagnosis is most likely to be VSD.

Auscultation

- Remember that all murmurs in pediatrics are systolic. If the murmur is best heard below the line connecting the nipples it is pansystolic and murmurs best heard above this line are usually ejection systolic.
- Try to have a tentative diagnosis before auscultation.
- If you cannot hear a prominent murmur ASD is a possibility. Therefore listen carefully for the wide fixed splitting of S2

Diagnosis

- The diagnosis should be stated in full. For example
  "This child has an acyanotic congenital heart disease probably a ventricular septal defect with L to R shunt not complicated by heart failure or pulmonary hypertension"

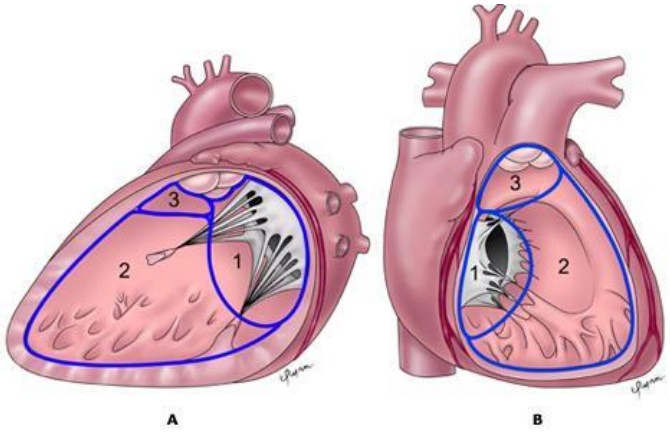
	VSD	ASD	PDA	MR	AS	TOF
Inspection	Child may be small for age and dyspnoeic if in heart failure		Child may be small for age and dyspnoeic if in heart failure			Central and peripheral cyanosis Child may be small for age
Pulse examination			Bounding peripheral pulses Lower limb pulses are extremely prominent Look especially for dorsalis pedis and posterior tibial pulse		The pulses are usually difficult to palpate. Slow rising pulses	
Palpation	May have a prominent displaced apex Systolic thrill in the lower left sternal edge Left parasternal heave Loud P2 if in pulmonary hypertension	Apex is usually normal Left parasternal heave Loud P2 if in pulmonary hypertension	Left parasternal heave Loud P2 if in pulmonary hypertension	Displaced hyperdynamic apex beat, thrusting in character May have an apical systolic thrill	Non displaced hyperdynamic apex which is heaving in nature	Left parasternal heave
Auscultation	Harsh pan systolic murmur in the lower left sternal edge Also may have a mid diastolic murmur at the apex in moderate and large lesions May have a loud P2 if in pulmonary hypertension	Ejection systolic murmur in the pulmonary area Wide fixed splitting of S2 May have a loud P2 if in pulmonary hypertension (**PS** is a similar murmur but no splitting)	Continuous machinery murmur in the left infraclavicular area	Pan systolic murmur best heard at the apex which radiates to the axilla	Ejection systolic murmur in the aortic area which radiates to the neck	Ejection systolic murmur best heard at the mid-upper left sternal edge Soft P2

Ventricular Septal Defect (VSD)

Classification and anatomical basis of the defect

- The interventricular septum is formed by 3 basic anatomical components. These are the
  1. Septum of the atrioventricular canal
  2. Muscular septum
  3. Part of the conotruncal septum (This divides the right and left ventricular outflow tracts)

- **Classification of VSD**
  Membranous – Most common and accounts for 75% of all VSD. Lesions in the component number 3 with extension into number 2
  Subpulmonic – Occur in the outlet (component 3)
  AV canal defects
  Muscular type – Occur in the muscular part of the interventricular septum



- **Clinical classification of VSD**
  Small
  Moderate
  Large
- Clinical differentiation of these types may be asked during the discussion. This is based on the history and examination

	Small VSD	Moderate and large VSD
History	Child is usually asymptomatic	Usually develop features of heart failure such as dyspnoea, poor feeding and inadequate weight gain
Examination	Murmur is shorter and louder Evidence of pulmonary hypertension is absent No evidence of heart failure	Murmur is longer and softer Other murmurs – Mid diastolic rumble may be present Evidence of pulmonary hypertension will be present There may be clinical features suggestive of heart failure

Investigations to be carried out

- **ECG**
  Is normal in patients with small VSD's
  In moderate to large VSD's may show left atrial and ventricular enlargement
  If there is significant pulmonary hypertension the ECG will show evidence of Right ventricular hypertrophy
- **CXR**
  May be normal in small VSD
  In moderate to large VSD it will show increased pulmonary vascular markings and cardiomegaly
- **Echocardiography**
  This is the confirmatory investigation

Possible complications

- Heart failure
- Pulmonary hypertension
- Reversal of the shunt and Eisenmenger syndrome
- Infective endocarditis

Management options

- **Medical management of heart failure**
  Dietary management and growth monitoring
  Diuretics – Frusemide, spiranolactone
  ACE inhibitors
- **Management of pulmonary hypertension**
  This is done with pulmonary vasodilators
- **Surgical management**
  Close follow up is necessary. Most defects with spontaneously close within the 1st few years. But early surgery is indicated in the following
  Severe symptoms with failure to thrive
  Pulmonary hypertension (early)
  Associated valvular lesions especially aortic regurgitation
  Significant left to right shunting (Qp:Qs > 2:1)

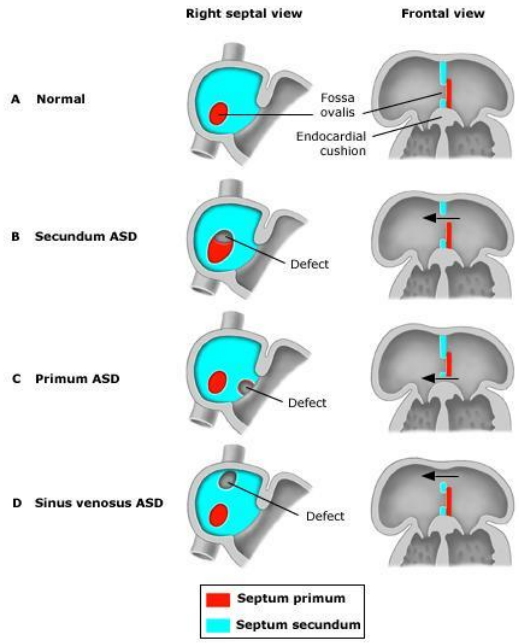
Complications of surgery

- Heart block
- Residual VSD

Atrial septal defect (ASD)

Classification and anatomical basis of the defect

- There are 3 basic types of ASD. There are
  Ostium secondum ASD
  Ostium primum ASD
  Sinus venosus ASD



Associations

- Holt- Oram syndrome (ASD with abnormalities of the radius and thumbs)

Investigations

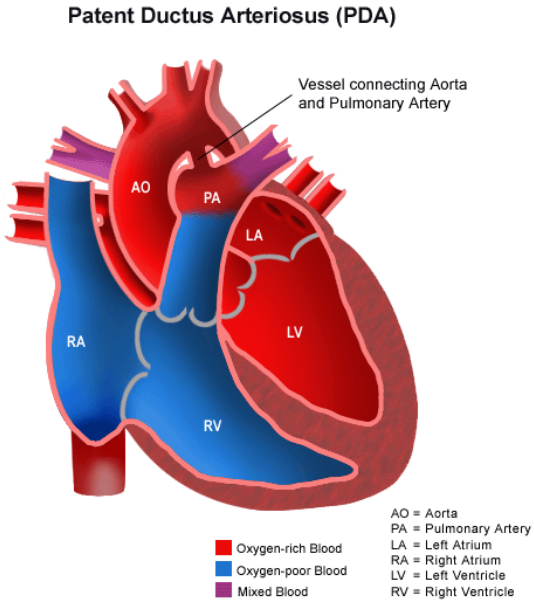
- **ECG**
  This may show 1st degree heart block, right axis deviation, increased PR interval
  Ostium primum defect may have associated RBBB
- **CXR**
  The typical ostium secondum ASD will show evidence of increased pulmonary vascular markings
- **Echocardiogram**
  This offers the definitive diagnosis

Management options

- Ostium secondum ASD's are usually followed up and closed electively before school entry at the age of 5-6 years
- However symptomatic ASD's or ASD's with a significant L to R shunt should be closed before this age
- The options for surgical closure are open surgery and transcatheter device closure. The latter is preferred as it is less invasive. But transcatheter closure may not be possible in large ASD's and complicated ASD's
- Ostium primum defects should be surgically repaired

Patent ductus arteriosus (PDA)

- The ductus arteriosus is a fetal vascular connection between the main pulmonary artery and the aorta
- Failure of ductal constriction at birth causes a PDA
- Is classified according to the ratio between the pulmonary and systemic flow



Associations

- Congenital rubella syndrome
- Maternal warfarin therapy

Other causes of continuous murmurs

- Venous hum
- Systemic arteriovenous shunts
- Coronary artery fistulas
- Ruptured sinus of valsalva

Investigations

- **Echocardiogram**
  This is the investigation of choice. This can make the diagnosis and also assess the severity by estimation of the pulmonary to systemic flow ratio by Doppler echocardiography
- **ECG**
  (May show biventricular hypertrophy)
- **CXR**
  Shows a prominent main pulmonary artery, enlargement of the heart and prominent pulmonary vasculature

Complications

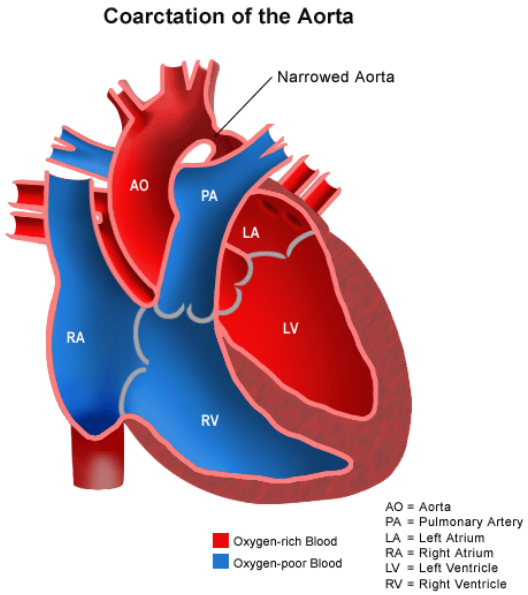
- Heart failure
- Pulmonary hypertension
- Eisenmenger's syndrome
- Infective endocarditis

Management

- The main mode of management is closure of the PDA
- Closure of the PDA is indicated in patients who are symptomatic, have evidence of early (reversible) pulmonary hypertension and significant L to R shunts. Some advocate closure of asymptomatic PDA but this is controversial

- Closure of the defect is achieved in several ways. These are,
- Pharmacological management is used for PDA closure in the preterm. The drugs used are inhibitors of PG synthesis – Indomethacin and ibuprofen
- Surgical management – Coil, device or open surgery used in older children

Coarctation of the aorta



- This is a rarely given case at the exam

Associations

- Turner syndrome
- Berry aneurysms

Investigations

- **Echocardiogram**
- **CXR**
  Cardiomegaly in infants
  Notching of the ribs in older children
- **ECG**

Management options

- **Surgical repair with end to end anastomosis**
- In neonates with a duct dependent circulation keep the duct open – PG infusions

Aortic stenosis (AS)

- This is a rarely given case at the exam

Investigations

- Definitive diagnosis is by echocardiography
- Other investigations are usually of limited importance
- Cardiac catheterization is carried out to look for the severity of the lesion

Management options

- Valvotomy – balloon or surgical
- The indication for intervention depends on the symptoms and pressure gradient across the aortic valve

Pulmonary stenosis

- This is a rarely given case at the exam
- **Investigations –** Definitive diagnosis is by echocardiography

Management options

- Valvotomy
- Indication for intervention depends on the pressure gradient across the pulmonary valve

Mitral regurgitation (MR)

- Mitral regurgitation in children may be due to a primary defect in the valve mechanism or secondary to another cardiac disease
- **Primary**
  Congenital
  Mitral valve prolapse
  Acute rheumatic carditis
  Infective endocarditis
  Secondary
  Left ventricular dysfunction
  Cardiomyopathy
- The discussion may then shift to one on acute rheumatic fever or infective endocarditis

Acute rheumatic fever

- Diagnosis of acute rheumatic fever is based on the Duckett- Jones criteria
- This has 3 components
  Evidence of preceding Group A streptococcal infection
  Major criteria
  Carditis
  Polyarthritis
  Chorea
  Erythema marginatum
  Sub cutaneous nodules
  Minor criteria
  Arthralgia
  Fever

Elevated acute phase reactants (ESR, CRP)
Increased PR interval

- A diagnosis is made if there is evidence of a preceding streptococcal infection with 2 major criteria or 1 major and 2 minor criteria
- Preceding streptococcal infection is investigated with ASOT

Management

- Consists of 4 basic principles. These are
- Anti inflammatory therapy – Aspirin is the drug of choice
- Antibiotic therapy – IM benzathine penicillin once or oral penicillin for 21 days
- Management of heart failure – This can be achieved using medical management. Surgical intervention may be needed in a minority of cases that do not respond to the treatment
- Prophylaxis (secondary prevention) – This is achieved with monthly IM benzathine penicillin injections usually up to the age of 18 years

Infective endocarditis

- Usually presents with pyrexia of unknown origin
- Diagnosis is by the Duke's criteria

Investigations

- Blood culture – 3 blood culture samples should be taken from 3 different sites within 1 hour
- Echocardiogram

Principles of management

- Start empirical antibiotic therapy after collection of the blood cultures. Then change to a suitable antibiotic
- Monitor for the response and the complications
- Complications of infective endocarditis
  Cardiac effects – Valvular dysfunction and heart failure, perivalvular abscess formation
  Septic embolization
  Immune complex mediated damage – Glomerulonephritis
  Complications due to the treatment

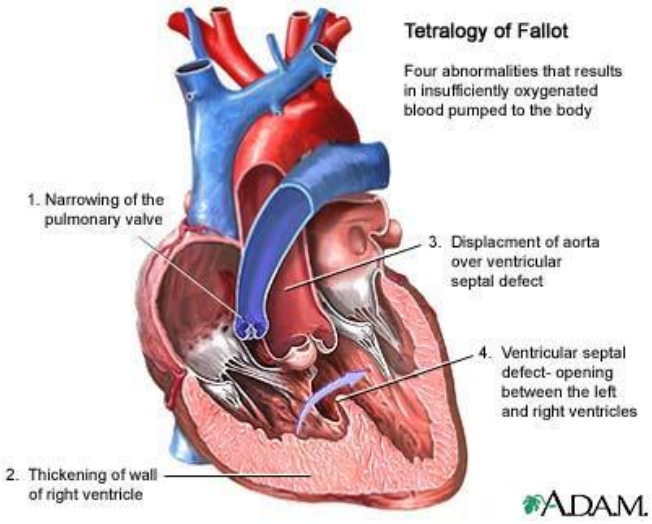
Cyanotic heart disease and Tetralogy of Fallot

Cyanotic congenital heart disease

- Tetralogy of Fallot
- Transposition of great vessels
- Tricuspid atresia
- Truncus arteriosus
- Total anomalous pulmonary venous drainage
- Ebstien's anomaly
- Pulmonary atresia
- Univentricular heart

Tetralogy of Fallot

- Consists of 4 anatomical abnormalities.
  These are
  Pulmonary infundibular stenosis
  Right ventricular hypertrophy
  VSD
  Overriding aorta



Associations

- Down syndrome
- Di George anomaly
- Velocardiofacial syndrome

Investigations

- **ECG**
  Shows features of right ventricular hypertrophy
- **CXR**
  Shows the classical boot shaped heart with an upturned apex and a concave main pulmonary artery segment
- **Echocardiogram**
  This is used for the definitive diagnosis and also the assessment of other abnormalities which may affect the surgical management
- **Cardiac catheterization**
  This should also be performed in preparation for surgery

Complications

- Hypercyanotic spells
- Infective endocarditis
- Cerebral abscess

Management options

- The final definitive option for management of TOF is total correction. This can be performed within the first year of life
- However if the child is extremely symptomatic a palliative shunt can also be considered earlier in life with a plan for total correction later. This is termed a Blalock – Tassing shunt and involves an anastomosis of the subclavian artery to the pulmonary artery to improve the pulmonary blood flow
- **Management of a Hypercyanotic spell**
  Child in the knee chest position
  100% Oxygen
  IV fluid bolus

  IV morphine
  IV beta blockers – Propranolol
  Emergency surgery for shunt creation

	TGV	TAPVD
Anatomical defect	Aorta arising from the right ventricle and pulmonary artery arising from the left ventricle Survival due to mixing of blood via PDA, VSD, ASD or PFO	All pulmonary veins drain into the right atrium instead of the left atrium Mixture of blood PDA, ASD
Clinical features	Cyanosis within a few hours of birth Clinical signs are minimal	Cyanosis
Investigations	CXR shows the “egg on a side” appearance	CXR shows typical snowman appearance in supracardiac lesions
Management	Emergency neonatal PG infusion to keep the ductus arteriosus patent Atrial balloon septostomy (Rashkind) **Corrective surgery** Arterial switch operation (Should be ideally done before 4 weeks)	Emergency neonatal PG infusion Corrective surgery

Respiratory system

Examination scheme

This is one of the easiest short cases in pediatrics. Be extremely observant. Sometimes auscultation is not required to make a diagnosis.

- Ask the age of the child
- Look at the child and get a general impression – Is the child ill looking or well looking? Is the child adequately grown for his or her age?
- Note any dysmorphic features
- Is the child dyspnoeic?
- Note if the child is on supplementary oxygen
- General examination – Look at the child from head to toe.
    - Warm to touch/not
    - Central and peripheral cyanosis
    - Cervical lymphadenopathy
    - Intravenous cannula
    - BCG scar
    - Mantoux scar
    - Ankle edema

Examination of the respiratory system proper

Inspection

- Start with child lying on the bed
- Go to the foot end of the bed
- Look for chest deformities, signs of hyperinflation (barrel shaped chest), Harrison's sulcus
- Count the respiratory rate
- Look for features of respiratory distress – Tachypnoea, cyanosis, nasal flaring, subcostal and intercostal recessions, use of accessory muscles of respiration.
- Listen for any audible sounds – Stridor, wheeze
- Do not forget to look at the back for pleural aspiration scars and the axilla for scars due to IC tube insertion

REMBEMBER – The respiratory system should be examined from both back and front. But most of the signs are best seen on examination of the back. Therefore try to start from the back especially in an older child.

Palpation

- Feel for the position of the trachea (Do not do this in very small children as it is very distressing)
- Chest expansion – Use a tape if the child is very small
- Apex beat – Important to detect mediastinal shift
- Tactile vocal fremitus – In older children

REMEMBER- The next step in a routine examination would be percussion. But percussion is distressing to the child and he/she may start crying. If this happens auscultation will be a nightmare. Therefore in paediatrics always leave percussion to the end.

Auscultation

- Listen to the air entry in all zones of the lung. Compare the sides as you go on
- Listen to the type of breathing and determine whether it is vesicular breathing or bronchial breathing
- Listen for any added sounds – ronchi, crepitations
- Examine vocal resonance in a older child

Finish with examination of the liver and spleen

	Pleural effusion	Consolidation	Asthma	Bronchiectasis
General examination		Child may be febrile		Clubbing
Inspection	May have IC tube insertion scar Pleural aspiration/ biopsy scar Decreased chest wall movement on the affected side	Decreased chest wall movement in the affected zones	Features of hyperinflation Harrison's sulcus	
Palpation	Mediastinal shift away from the affected side (only in large effusions) Decreased chest movements on the affected side Decreased tactile vocal fremitus	No mediastinal shift Decreased chest movements in the affected zone Increased tactile vocal fremitus		
Percussion	Stony dull on percussion	Dull to percussion		
Auscultation	Decreased breath sounds Decreased vocal resonance	Decreased breath sounds Bronchial breathing Increased vocal resonance	Ronchi +/- crepitations	Coarse crepts especially in the lower zones

Discussion
Pleural effusions in children

Causes

Exudative effusion	Transudative effusion	Other
Infections Pneumonia Tuberculosis **Neoplastic** Lymphoma **Connective tissue disease** SLE	Nephrotic syndrome Cardiac failure Liver failure Dengue	Chylothorax Hemothorax

Investigation of a patient with a pleural effusion

- **CXR**
- **Pleural tap**
  Several aspects of the pleural fluid is examined
  The appearance
  Pale yellow clear fluid indicates a transudate while a grossly purulent fluid indicates an empyema
  Milky fluid indicates a chylothorax
  Blood stained pleural fluid indicates a hemothorax or TB

  Exudate vs. transudate
  Exudates will have a protein content of > 3g/dL
  Light's criteria is a more complete assessment which is based on the protein and LDH in serum and pleural fluid

  Further analysis of exudative effusions
  Pleural fluid glucose – Very low levels are seen in parapneumonic effusions, TB and malignancy
  LDH - >1000 in empyema
  pH < 7.3 - parapneumonic effusions, TB and malignancy

  Microbiological assessment
  Gram stain and culture
  Acid fast bacilli
  Adenosine deaminase
- **Further investigations with FBC, mantoux etc.**

Management

- A pleural effusion usually resolves with treatment of the underlying disorder
- Some cases will require IC tube drainage
  Effusions that are enlarging or compromising respiratory function in a febrile unwell child require drainage. Other risk predictors indicating the need for chest tube placement include frank pus on thoracentesis, a positive pleural fluid gram stain and culture finding, pleural fluid pH level of less than 7, a glucose concentration of less than 40 mg/dL, or an LDH level of more than 1000 IU.
- Resistant cases require surgical drainage of the effusion

Pneumonia

- Is an inflammation of the parenchyma of the lung
- Classified as
  Community acquired
  Hospital acquired
  Pneumonia in the immunocompromised
- Basics of investigations and management may be asked for in short case
- **Investigations**
  FBC – May show a neutrophil leukocytosis in a bacterial infection
  CXR – May show features of consolidation and effusion or patchy infiltrates in a patient with bronchopneumonia
  Sputum for gram stain and culture
  Blood culture

- **Management**
  If there is a clinical suspicion of pneumonia empirical antibiotics should be started. However these should be given after blood culture is collected

  The antibiotic can be changed after the ABST to a suitable antibiotic
  Initially IV antibiotics are given which may be changed to oral antibiotics once the fever and respiratory signs are resolving. (Duration of therapy is usually 5-7 days)

Bronchiectasis

Possible causes

- As a sequale of whooping cough and measles
- Kartagener's syndrome
- Cystic fibrosis
- Immune deficiency

Investigations

- **CXR –** Shows ring shadows and tramline shadows
- Investigation of choice is an HRCT
- Further investigations can be performed to find a possible cause

Management

- Postural drainage
- Antibiotics may be used in infective episodes and as prophylactic therapy for recurrent exacerbations
- Bronchodilators may be used if there is significant airflow limitation

Abdomen

Examination scheme

General examination

- Ask the age of the child
- First get a general impression of the child. Is the child ill looking or well looking? Is the child adequately grown for the age
- Look for dysmorphic features
- Continue with the general examination from head to toe

  Warm to touch/not
  Eyes - Look for pallor, Icterus, evidence of nutritional deficiency (vitamin A), cataract
  Mouth – Oral pigmentation, angular stomatitis, glossitis, look at the teeth and examine the palate for palatal petichiae
  Examine all groups of lymph nodes for lymphadenopathy
  Hands – Look for clubbing, leuconychia, koilonychia and palmar erythema, widened wrist joint (rickets)
  Chest – Look for spider naevi
  Lower limbs – Any deformities, ankle edema (Look for sacral edema in infants)
  Skin – Examine for rashes, scratch marks and any petichiae or echymotic patches

- At this point reflect on your findings on the general examination. The most important findings are
- Pallor
- Icterus
- Lymphadenopathy
- Stigmata of chronic liver disease – Clubbing, leuconychia, palmar erythema, spider naevi (These are not commonly seen in children)
- Nutritional deficiency
- Ankle/sacral edema

Examination of the abdomen

- **Inspection**
  Look for distension of the abdomen and any visible lumps
  Visible hernia (umbilical hernia)

Surgical scars including biopsy scars (these are commonly missed)
Dilated veins on the abdomen

- **Palpation and percussion**
  Always keep your eye on the child's face and palpate all 9 quadrants in an orderly fashion
  Examine for palpable organs – Liver, spleen and renal masses
  Do not forget to measure any palpable organ with a measuring tape
  Confirm with percussion
- **Percuss for free fluid**- First look for flank dullness and shifting dullness. Then look for horseshoe dullness
- **Auscultation**
  Auscultate over the liver for a hepatic bruit, listen for a renal bruit and a splenic rub

General points on the presentation and discussion

- The usual cases which will be given for the exam usually involve palpable organs. Therefore one should know how to present a liver, spleen and renal mass
- See the table below for the method of presentation

Liver	Spleen	Renal mass
Right hypochondrial mass Cannot get above Moves with respiration Dull to percussion over the mass and continues with the liver dullness	Left hypochondrial mass Cannot get above Moves diagonally with respiration Notch felt/not Dull to percussion over the mass and the dullness is continuous with the splenic dullness	Mass in the loin Can get above (If the mass is very large you may not be able to get above it) Moves with respiration Ballottable Dull to percussion over the mass but with the presence of resonant bands
Mention the distance from the costal margin to the lower border of the liver in the mid clavicular line	Mention the distance from the costal margin to the tip of the spleen	Measure the size of the lump from upper to lower pole
Mention the site of the upper border of the liver (mid clavicular line State the span of the liver		
Describe the lower border of the liver, the surface and the consistency	Describe the consistency of the spleen	Describe the consistency
Mention if there is a hepatic bruit/not	Mention if there is a splenic rub/not	

- The next step is to know the differential diagnosis for hepatomegaly, splenomegaly, hepatosplenomegaly and renal mass
- Study the following table for the differential diagnosis

	Hepatomegaly	Splenomegaly	Hepatosplenomegaly
Infective	**Viral** Viral hepatitis CMV Rubella Dengue EBV **Bacterial** Leptospirosis **Protozoal** Toxoplasmosis Amoebiasis	**Viral** EBV **Bacterial** Typhoid Infective endocarditis **Protozoal** Malaria Visceral leishmaniasis	**Viral** EBV CMV **Bacterial** Typhoid **Protozoal** Toxoplasmosis
Hematological		Hemolytic anaemias Congenital and acquired	Hemolytic anaemias Congenital and acquired
Neoplastic	Hepatoblastoma Hematological malignancy	Leukaemia, lymphoma	Leukaemia, lymphoma
Storage disease	**Carbohydrate** Glycogen storage disease Mucopolysaccharidosis **Fat** Gaucher's Niemann-Pick **Amino acid** Tyrosinaemia	 **Fat** Gaucher's	**Carbohydrate** Glycogen storage disease **Fat** Gaucher's disease
Autoimmune	Autoimmune hepatitis	SLE, JIA	
Congestive	Cardiac failure Budd chiari syndrome	Portal hypertension Especially with cirrhosis of the liver And prehepatic PH	Chronic liver disease with portal hypertension
Metabolic	Wilson's disease Alpha 1 antitrypsin deficiency		

Hepatomegaly

- After presenting the case you will be asked for a differential diagnosis of hepatomegaly. The table given above will give you a good guide
- Then use the findings on the general examination to narrow down your differential diagnosis
- Given below are the possible clinical scenarios

Clinical scenario	Differential diagnosis
Febrile child	Infective hepatitis
Lymphadenopathy +/- fever	Infective hepatitis Hematological malignancy
Pallor	Hematological malignancy Chronic infection
Icterus	Chronic liver disease (Wilson's) Infective hepatitis
Normal general examination	Chronic liver disease Storage disease (May have dysmorphic features, developmental delay)

Investigations

- This will be guided by the differential diagnosis but there are some general investigations that should be done
- Full blood count and blood picture
- Ultrasound scan of the abdomen

Discussion can continue on glycogen storage disease, infective hepatitis or chronic liver disease

Glycogen storage disease

- There are several types of glycogen storage disease. Type one is the most important
- Presents with hypoglycaemia and early morning seizures
- Investigations
  Fasting blood sugar – Low
  Serum lactate, uric acid, triglycerides are elevated
- Other types are associated cardiomyopathy and skeletal myopathy
- Management is with avoidance of fasting
- Corn starch is added to the diet

Infective hepatitis

Viral hepatitis

- Is caused mainly by hepatitis viruses A-E but can also be caused by other viruses such as CMV and EBV
- The clinical course is usually described in 4 phases. These are the incubation, prodrome, icteric phase and recovery phase
- Investigations in the initial phase
  FBC – Decreased WCC with a relative lymphocytosis
  Liver function tests – Elevated AST with normal serum bilirubin
- Specific investigations
  Include those for the diagnosis (See table below)
- Management is usually supportive

	Microbiology	Complications	Investigations	Prevention
Hepatitis A	RNA virus Transmission is via the faeco-oral route	Acute fulminant hepatitis	**Prodrome** HAV in stool (by electron microscopy or RNA detection. This is not commonly used **IgM HAV** Positive at the onset of symptoms. Persists for about 4-6 months	Personal hygiene Vaccination
Hepatitis B	DNA virus Transmission is by the intravenous route, sexual contact Vertical transmission can also occur	Chronic infection Asymptomatic carrier Cirrhosis Hepatocellular carcinoma Acute fulminant hepatitis is rare	**HBsAg and positive IgM anti-HBc** Indicates acute hep B infection **HBeAg** Indicates severe infection	Vaccination

			HBsAg persisting for more than 6months with positive IgG anti-HBc and negative IgM anti-HBc Indicates chronic infection	
Hepatitis C	RNA virus Transmission is via the intravenous route and by sexual contact	Higher risk of developing chronic liver disease, cirrhosis and hepatocellular carcinoma	**Anti HCV**	Vaccine?
Hepatitis D	Incomplete RNA particle Can only replicate in the presence of hepatitis B infection This can be superinfection (infection in a person already having hep B) or coinfection	Acute fulminant hepatitis more common in coinfection Chronic liver disease more common in superinfection	**Co infection** IgM anti HDV and IgM anti HBc **Superinfection** IgM anti HDV and IgG anti HBc	Vaccine?
Hepatitis E	RNA virus Similar to Hep A	Fulminant hepatitis	HEV RNA in stools	No effective prophylaxis

Chronic liver disease

- Suspected chronic liver disease in children should be investigated according to the cause

Cause	Investigations
Wilson' s disease	Ophthalmology referral to examine for the presence of K-F rings Serum ceruloplasmin, 24h urinary copper excretion
Hep B/C	See above
α1 antitrypsin deficiency	Enzyme levels
Hemochromatosis	Serum ferretin and serum iron

Congenital hepatic fibrosis	Liver biopsy
Biliary cirrhosis	Unconjugated hyperbilirubinaemia, elevated alkaline phosphatase

Splenomegaly

- As in a case of hepatomegaly use the findings of the general examination to narrow down the diagnosis

Clinical scenario	Differential diagnosis
Pallor +/- Icterus	Hemolytic anaemia Cirrhosis with portal hypertension
Lymphadenopathy	Infection (IMN) Hematological malignancy
Normal general examination	Storage disease Cirrhosis with portal hypertension

- Massive splenomegaly is also encountered at the exam. The differential diagnosis is unique
- This is when the lower pole of the enlarged spleen crosses the midline. Causes are

  Thalassemia major
  Lymphoma
  Chronic leukaemia
  Gaucher's disease
  Chronic malaria
  Visceral leishmaniasis

  (Langerhan cell histiocytosis
  HIV
  Autoimmune lymphoproliferative disease)

Investigations in a patient with splenomegaly

- **FBC and blood picture** is a key investigation
  Look for evidence of infection – neutrophil leukocytosis
  Anaemia – hemolytic anaemia
  Pancytopenia – could be due to hypersplenism or bone marrow suppression due to a malignancy

Blood picture will show leukaemic blast cells, specific cells in certain subtypes of lymphomas and specific cells in certain hemolytic anaemias

- **USS of the abdomen**
  This will help visualize the enlarged spleen. Also look for intra abdominal lymphadenopathy in lymphomas, ascites in chronic liver disease
  Combining with the Doppler should be done to assess for portal hypertension

Discussion will continue with hemolytic anaemia or portal hypertension

Hemolytic anaemia

- Basic investigation of a suspected hemolytic anaemia starts with the FBC and blood picture. The blood picture has a specific appearance according to the type of hemolytic anaemia
- **Other investigations**
  Reticulocyte count - Increased reticulocytes
  Unconjugated hyperbilirubinaemia
  Hemoglobinuria and hemosiderinuria in intravascular hemolysis
  Bone marrow – erythroid hyperplasia
- After confirmation of a hemolytic anaemia investigations should focus on finding the cause or underlying pathology
- Classification of hemolytic anaemia is into congenital and acquired

Congenital	Acquired
Membrane defects Hereditary spherocytosis Hereditary elliptocytosis	**Immune** **Autoimmune** Warm Cold **Alloimmune** Transfusion reactions Hemolytic disease of the newborn
Metabolic defects G6PD deficiency PK deficiency	**Red cell fragmentation syndromes** Prosthetic valves HUS, TTP, DIC
Disorders of hemoglobin Thalassemia Sickle cell anaemia	**Systemic disease** **Infection** **Toxins**

Congenital hemolytic anaemias

Disease	Investigations	Management
Hereditary spherocytosis (AD)	Blood film – Spherocytes Osmotic fragility test	Definitive management is splenectomy which is usually

		planned when the child is 5-6 years of age In the meantime the management is based on symptoms with severe anaemia managed by blood transfusion
G6PD deficiency **(X linked)**	Hb normal between attacks Blood film – Bite cells, blister cells, Heinz bodies Features of intravascular hemolysis G6PD levels in the RBC	Is precipitated by antioxidant drugs and substances Avoiding these is the most important aspect of the management A crisis can be treated with blood transfusions as necessary
Thalassemia **β – Thalassemia major**	Blood film - microcytic hypochromic anaemia Target cells, nucleated red blood cells Reticulocyte count may be low Serum iron studies Bone marrow – Erythroid hyperplasia Serum hemoglobin electrophoresis- Absent HbA with increased HbF and HbA2	Management is with recurrent blood transfusions Consider splenectomy **Monitor for the complications of iron overload** Cardiomyopathy Liver disease Endocrine organ dysfunction – growth, hypothyroidism, diabetes **Iron chelation therapy** Subcutaneous desferrioxamine Can cause auditory and ophthalmological side effect **Counseling and parent education** **Counseling of the child**
Sickle cell anaemia	Blood film - Sickle cells, Howell-Jolly bodies Sickling test Hb electrophoresis - HbS	Rare in SL Avoid precipitants Management of crisis with analgesia, adequate fluid and transfusion

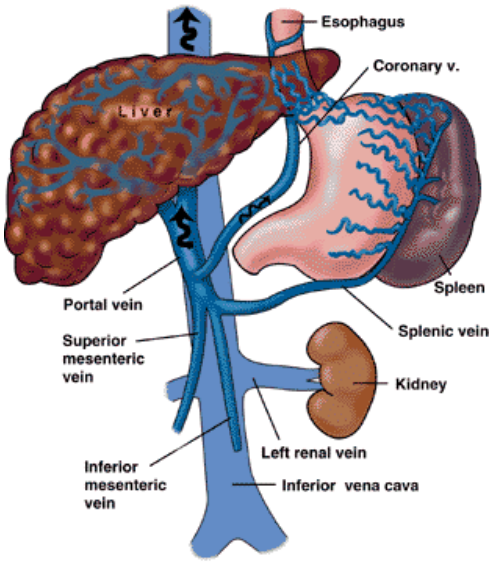
Acquired hemolytic anaemia

	Warm AIHA	Cold AIHA
Type of antibody	IgG	IgM
Causes	Idiopathic Autoimmune – SLE Lymphoma CLL Drugs – Methyldopa	Idiopathic Infections – EBV, CMV Mycoplasma
Investigations	Those of hemolytic anaemia Spherocytes on the blood film	Those of hemolytic anaemia Less spherocytes Cold agglutination test

Management	Treat cause Blood transfusion of necessary Steroids Immunosuppressive drugs – Azathioprine, cyclophosphamide IVIg Splenectomy is also a final option of there is poor response to the medical management	Treat cause Keep warm

Portal hypertension

Classification is based on the anatomy



Prehepatic	Hepatic	Posthepatic
Small liver, big spleen	Small/large/normal liver Big spleen	Big liver, big spleen
Portal/splenic vein thrombosis Neonatal sepsis Dehydration Hypercoagulability **Increased portal flow** AV fistula	**Hepatocellular** Chronic liver disease (Cirrhosis) **BIliary tract disease**	**Hepatic vein thrombosis** (Budd -Chiari syndrome) Polycythaemia Leukaemia Coagulopathy **Right heart failure** **Constrictive pericarditis**

Investigations

- **USS of the abdomen with doppler –** Can outline the anatomy of the portal system, and study the flow and estimate the pressures within the portal system
- **Upper GI endoscopy –** To look for evidence of gastroesophageal varices

Management

- These patients can present with an acute episode of hematemesis due to bleeding from the esophageal varices
- If this is the case manage the emergency
- Initial assessment of the patient, especially for features of hemodynamic instability
- If features of hemodynamic instability are present insert 2 wide bore cannulae and collect blood for investigations – FBC, grouping and DT, PT/INR
- IV Proton pump inhibitors – Omeprazole
- Consider giving other drugs – IV vasopressin, IV octreotide
- Correct the coagulopathy with FFP, administer vitamin K
- Emergency endoscopy if possible for banding, ligation

Follow up

- Elective endoscopy
- Drugs to reduce the portal pressure – oral propranolol (titrate dose based on the resting heart rate)

Hepatosplenomegaly

- This is the commonest case given at the exam
- The general examination is extremely useful in making a diagnosis

Clinical scenario	Differential diagnosis
Pallor + Icterus	Hemolytic anaemia Chronic liver disease
Lymphadenopathy	Hematological malignancy IMN Typhoid
Normal general examination	Storage disease Chronic liver disease Transfused hemolytic anaemia

Investigations

- Do the initial investigations with FBC, blood picture and USS of the abdomen
- After this initial discussion there may be further questions asked on individual diseases

Hematological malignancy
Leukaemias

- Are the most common malignancy in childhood
- Classified as acute and chronic leukaemias.
- ALL is the commonest and accounts for about 75% of all leukaemias. It is also about 5 times more common than AML
- Chronic leukaemias are rare in childhood
- CML is rarely encountered

	ALL	AML
Clinical presentation	Features of bone marrow failure Organ infiltration	Features of bone marrow failure Organ infiltration Can have other features DIC, gum hypertrophy
Associations	Trisomy 21 Translocation (9:22)	
Investigations	FBC – Pancytopenia Blood picture – Blasts Bone marrow - >30% blast cells Special stains and classification tests	Similar Positive for myeloperoxidase stain

Management	General supportive therapy Chemotherapy **Induction of remission** Prednisilone **Consolidation of remission** Intensive multi agent chemotherapy CNS prophylaxis **Intensification** **Maintenance chemotherapy**	General supportive therapy **Induction of remission** **Consolidation**

Lymphomas

	Hodgkin's lymphoma	Non Hodgkin's lymphoma
Clinical	Lymphadenopathy usually begins from 1 group of peripheral lymph nodes and spreads contiguously to the others Can have mediastinal involvement Extra nodal spread rare Leukaemic phase rare Constitutional symptoms common	Has a more unpredictable and haphazard spread Involves oropharyngeal lymph nodes Extra nodal spread common Leukaemic phase more common Constitutional symptoms rare
Investigations	Lymph node biopsy shows Reed – Sternberg cells	No RS cells
Management	**Early stage disease** Radiotherapy **Advanced disease** Chemotherapy +/- radiotherapy	Multi agent chemotherapy

Renal mass

- The differential diagnosis for a renal mass is as follows

Intrarenal	Extrarenal
Solid Wilm's tumor Renal vein thrombosis Horseshoe kidney	Adrenal mass Neuroblastoma
Cystic Hydronephrosis Single cyst Polycystic kidney	

Hydronephrosis

- Is an aseptic dilation of the whole or part of the kidney due to incomplete or intermittent obstruction to the outflow of urine
- A diagnosis of hydronephrosis first needs confirmation by ultrasound scan. The subsequent investigations are focused on finding the cause
- The ultrasound scan can also assess the severity of the hydronephrosis by the diameter of the renal pelvis
- The following features will help to find the level of obstruction
  No megaureter = PUJ obstruction
  Megaureter = VUJ obstruction
  Large bladder = bladder outlet obstruction (PUV)

Wilm's tumor

- Is one of the commonest abdominal tumors in children
- Associated with other abnormalities of the GU tract, aniridia and hemihypertrophy
- Investigations are for imaging of the tumor and staging
- Definitive management is by nephrectomy

PCKD

- There are 2 types of polycystic kidney disease. These are the autosomal dominant and autosomal recessive forms

AR PCKD	AD PCKD
Presents with renal failure in childhood	Presents with renal failure in adulthood
Associated with liver cysts and pulmonary hypoplasia Antenatal diagnosis possible	Liver cysts and berry aneurysms

Nervous system

Examination scheme

The examination of the nervous system covers several components. These are

- General examination relevant to the nervous system
- Examination of the cranial nerves
- Examination of the motor system including the gait

The scheme of examination should be adapted according to the age of the child

Examination of the nervous system in an infant/toddler

General examination

Observe the child carefully. This is usually the key to picking up the physical signs. Therefore be extremely observant

Look at

- Alertness of the child and interactions with the environment
- Dysmorphic features
- Posturing of the child – hypotonic posture, spasticity
- Movements – Is the child moving all the limbs? Look for asymmetry. Look for abnormal movements such as tics, chorea, athetoid movements and tremor
- Examine the skin for evidence of neurocutaneous manifestations – tuberous sclerosis, neurofibromatosis
- Look carefully for pressure sores
- Look if the child is wearing pampers – This is unusual in a child >3years and may indicate urinary/faecal incontinence

Examine the head

- Shape
- Sutures
- Fontanelles
- Measure the OFC and palpate for a VP shunt

Cranial nerve examination

- This is mostly through observation
- Eyes – For eye to eye contact and fixation, look for squint and any other abnormalities in the eyes, examine the pupils
- Face (VII) – Look for symmetry of the smile

- Mouth – Look at the tongue (XII)

Examine the back – This is frequently missed at the exam. Look for scars and evidence of neural tube defects

Proceed to examination of the motor system of the limbs

- Examine the following
- Inspection – For wasting, fasciculations, scars
- Tone
- Power – This is mostly done through careful observation
- Reflexes
- Coordination
- Examine the gait if possible

Examination of the nervous system in an older child

General examination

Look at

- Alertness of the child and interactions with the environment
- Ask to measure the OFC
- Dysmorphic features
- Posturing of the child – hypotonic posture, spasticity
- Movements – Is the child moving all the limbs? Look for asymmetry. Look for abnormal movements such as tics, chorea, athetoid movements and tremor
- Examine the skin for evidence of neurocutaneous manifestations – tuberous sclerosis, neurofibromatosis

Cranial nerve examination

- **Cranial nerve I**
  Not usually tested. Ask the child if he/she has any problems in smelling food

Examination of the eyes

- **Cranial nerve II**
  Check the visual acuity with a pocket Snellen chart. If this cannot be read gradually work down to counting fingers, detecting hand movements and distinguishing light from dark
  Check the direct and consensual light reflex
  Visual fields – This is very difficult to examine in children. The confrontation method (similar to adults) can be done in older children
- **Cranial nerves III, IV and VI**
  Examine the ocular movements

Examination of the face

- **Cranial nerve V**
  Ask the child to clench the teeth
  Sensory is usually not examined
- **Cranial nerve VII**
  Examine the muscles of facial expression

Examination of the mouth

- **Cranial nerve IX and X**
  Ask the patient to say ‘ah’ and examine the symmetry of the soft palate
- **Cranial nerve XII**

Examine the tongue while it is inside the mouth for wasting and fasciculations. Then ask the child to protrude the tongue and look for any obvious deviation

Examination of the neck

- **Cranial nerve XI**
  Examine the sternocleidomastoid and trapezius muscle

<u>Examination of the upper limbs</u>

- **Inspection –** Expose completely and look for wasting, fasciculations, deformities and surgical scars
- **Tone**
- **Power**
  Examine the power in the key muscle groups
  Shoulder abduction – **C5, C6**
  Shoulder adduction – **C7, C8**
  Elbow flexion – **C5, C6**
  Elbow extension – **C7, C8**
  Wrist flexion and extension – **C7, C8**
  Small muscles of the hand – **T1**
- **Reflexes**
  Biceps **– C5, C6**
  Triceps – **C7, C8**
  Supinator- **C6, C7**
- **Coordination**
  Check for intentional tremor, past pointing and positive finger nose test and dysdiadochokinesia
- **Sensory**
  Usually not examined

<u>Examination of the lower limbs</u>

- **Examine the gait**
  Just observe the gait
  Examine for tandem walking
  Examine for Gower's sign
- **Inspection –** As for the upper limbs. A commonly missed point is a tendon release scar. These are on the posterior aspect of the lower limbs
- **Tone**
- **Power**
  Examine the power in the key muscle groups
  Hip flexion – **L1, L2**
  Hip extension- **L5, S1**

Hip abduction – **L2, L3**
Hip adduction- **L3, L4**
Knee flexion – **L5, S1**
Knee extension – **L3, L4**
Foot dorsiflexion – **L4, L5**
Foot plantarflexion – **L5, S1**
Inversion – **L4, L5**
Eversion – **L5, S1**

- **Reflexes**
  Knee – **L3, L4**
  Ankle – **L5, S1**
  Examine the plantars – in children above 2 years of age
- **Coordination**
- **Sensory**

Discussion

Basic neurology

- The initial topic of discussion in any case in neurology will focus on 2 basic questions. These are
- What is the site of the lesion?
- What is the pathology?

Examination of the limbs

- After completing examination of the lower limbs the findings should be categorized as UMN or LMN

Upper motor neuron lesion	Lower motor neuron lesion
Increased tone	Decreased tone
Increased reflexes	Diminished or absent reflexes
Ankle and patellar clonus may be present	
Extensor plantar response (In children above 2 years)	Plantars may be flexor or equivocal

LMN lesion

- If the lesion is a lower motor neuron lesion further analyze your findings to localize the site of the lesion

Site of the lesion	Pattern of neurological signs
Muscle	Bilateral and symmetrical weakness Proximal>Distal weakness Reflexes – Knee jerk is lost while the ankle jerk may be preserved Waddling gait and Gower sign positive No sensory impairment
NMJ (Not given at the exam)	Fatigable weakness
Peripheral nerve	**Polyneuropathy** Bilateral and symmetrical weakness Distal>Proximal weakness Sensory may or may not be impaired. If impaired will be in a glove and stocking distribution **Mononeuropathy** (Rare in paediatrics)

	Motor and sensory pattern related to the supply of the nerve **Multifocal neuropathy** (Rarely given as cases) Patchy involvement of peripheral nerves
Root	Will have motor and sensory loss in a root distribution
Anterior horn cell	Bilateral and symmetrical weakness Proximal>distal Prominent wasting and fasciculations No sensory impairment
Spinal cord lesions	Spinal cord lesions may present as LMN lesions Associated bladder and bowel incontinence Sensory level Evidence of neural tube defects
Cerebral cortex	This is also a possibility in paediatrics and has special features Axial hypotonia (truncal hypotonia) with preserved or exaggerated reflexes is characteristic
Systemic disease	Are important causes in infantile hypotonia Syndromes Hypothyroidism

A special case given at the exam is LMN lesion in infants. There is no change in the approach

Further discussion may focus on muscle disease, peripheral neuropathy or anterior horn cell disease

Muscle disease

- Muscle disease is of 2 types. These are
  Myopathy – Can be congenital, metabolic, mitochondrial
  Muscular dystrophy
- Of these 2 muscular dystrophy is the most important

Duchenne muscular dystrophy

Genetics

- Is an X-linked recessive disease
- The primary defect is a mutation in the dystrophin gene on chromosome 21

Presentation and complications

- Presents with progressive weakness initially presenting from the age of 3 years. Will be completely wheelchair bound by the age of 12
- Associated cardiomyopathy and intellectual impairment

- Respiratory muscle weakness leads to aspiration and recurrent pulmonary infections

Investigations

- The initial investigation is CK. This is usually 10-20 times the upper limit of normal
- **ECG**
  To assess for cardiomyopathy and arrhythmias
- **EMG –** myopathic changes
- **Muscle biopsy**
  This shows significant muscle degeneration and replacement by fat and connective tissue

Management

- This is usually supportive
- Prednisilone has been used in the management

Peripheral neuropathy

- Peripheral neuropathy in children is classified into 2 broad categories. These are congenital neuropathies and acquired neuropathies. The following table lists some of the causes

Congenital	Acquired
Hereditary motor and sensory neuropathy (HMSN)	**Infection** Leprosy Diphtheria **Inflammatory** Guillain- Barre syndrome CIDP Vasculitis and connective tissue disease **Metabolic and endocrine** DM Vitamin deficiency – B1, B6, B12, E **Organ failure** Chronic renal failure **Drugs** **Toxins** Arsenic Lead Organophosphates **Malignancy**

- Investigation of a suspected neuropathy should be started with a nerve conduction study. Then specific investigations should be performed to find the possible cause
- The NCS identifies 2 major categories of peripheral neuropathy
  Demyelinating – Reduced nerve conduction velocity
  Axonal – Reduction in the amplitude of the action potential with relative preservation of the conduction velocity

Guillain – Barre syndrome

- Is a post infectious demyelinating disease
- Diagnosis is on clinical suspicion. Presents as an ascending paralysis which may follow a respiratory tract infection or an episode of diarrhea

Management

- The most important aspect of the management is close monitoring of the patient. The following are the most important
- **Progression of the neurological symptoms and signs**
- **Respiratory function**
  This is done with the single breath count and cough effort at the bedside. A more accurate assessment can be made by a respirometer
- **Autonomic function**
  A life threatening complication is autonomic dysregulation. Therefore monitor the pulse rate and blood pressure
- If there is deterioration in the respiratory function ICU care is necessary
- IV IG or plasmapharesis is used as the definitive management
- The child should be given limb and chest physiotherapy and DVT prophylaxis until recovery
- Proper nursing care is essential

Investigations

- Confirmatory investigations
- LP – shows cytoprotein dissociation with elevated proteins and normal white cell count
- Nerve conduction study

Spinal muscular atrophy

- Are a group of autosomal recessive disorders characterized by progressive lower motor neuron type weakness
- 3 important types

	SMA1	SMA2	SMA3
Clinical	Present before 6 months of age Hypotonia Bulbar muscle weakness Respiratory paralysis No extraocular muscle involvement	Presents between 6-18 months Typically presents with motor delay	After 18 months Slower progression when compared to the other types Bulbar dysfunction occurs late

UMN lesions

- If the lesion is an upper motor neuron lesion the next step is to classify the lesion into the following clinical categories
- **Spastic quadriplegia**
- **Spastic paraplegia**
- **Hemiplegia**

Spastic quadriplegia

- If the child has spastic quadriplegia the lesion can be at 3 possible sites. These are
  Cerebral cortex – Will have associated intellectual impairment, developmental delay (cerebral palsy)
  Brainstem – associated cranial nerve lesions
  Cervical spinal cord – is extremely rare. Will have associated sphincter dysfunction

Spastic paraplegia

- The lesion can be
  Cerebral cortex – Cerebral palsy
  Spinal cord. Between T1 and L1

Hemiplegia

- The lesion can be
  In the contralateral cerebral hemisphere – associated abnormalities in speech and other higher functions
  Brainstem – associated cranial nerve palsies

Causes of hemiplegia in children

- Hemiplegic cerebral palsy
- Stroke
- Space occupying lesion

If you find signs of an UMN type of lesion the diagnosis is almost always cerebral palsy. Therefore try to examine or at least offer to examine the following

- Developmental assessment
- Primitive reflexes
- Rheumatological assessment for contractures

Discussion on cerebral palsy will focus only on the basics in a short case

Cerebral palsy

- Is a disorder of posture and movement due to a non progressive lesion in the motor pathways of the developing brain
- The aetiology can be classified as prenatal, perinatal or postnatal

Prenatal	Perinatal	Postnatal
Cerebral dysgenesis and malformations Congenital infections (TORCH)	Prematurity (PVL) Perinatal asphyxia – HIE	Cerebral ischaemia IVH Hydrocephalus Trauma Neonatal encephalitis Hyperbilirubinaemia

- Classification of CP is based on the neurological findings
- Spastic
  Spastic quadriplegic
  Spastic diplegic
  Hemiplegic
- Ataxic hypotonic
- Dyskinetic

Associated disorders

- Impaired vision
- Impaired hearing
- Epilepsy
- Learning disability
- Psychiatric disorders
- Urinary problems
- Growth failure

Investigations

- Cerebral palsy is a clinical diagnosis
- Neuro imaging is usually not performed in these children unless there is an uncertainty of the clinical diagnosis
- EEG should be performed in children who have associated seizures

Management

- Management of CP should be done as a multidisciplinary team activity and should involve the paediatrician, rheumatologist, physiotherapist, occupational therapist, speech therapist and social worker
- The ultimate objectives of management are to prevent or treat medical problems and make the child as independent as possible in activities of daily living
- The rheumatological management includes management of spasticity. There are several options
  Botulinum toxin
  Oral antispasticity drugs
  Orthopaedic interventions
  Splints and orthosis
  Tendon release surgery

Cranial nerve lesions and cerebellar lesions

These are very rarely given at exams. A few important cases are discussed below.

Ptosis

Site of the lesion	Clinical condition	Features
Muscle	Ocular myopathy	Bilateral and symmetrical ptosis
Neuromuscular junction	Myasthenia gravis	Fatigability
Occulomotor (CN III) palsy	Midbrain tumor/stroke	Associated hemiplegia
	Posterior communicating artery aneurysm	Isolated III CN palsy
	Cavernous sinus thrombosis	III, IV and VI cranial nerve palsy
	Superior orbital fissure lesion	III, IV and VI cranial nerve palsy
	Other DM, HT, Vasculitis	III nerve palsy with intact pupils
Horner's syndrome	Congenital Neuroblastoma Post cardiac surgery Klumpke's paralysis Brainstem tumor	Partial ptosis, enophthlamos, meiosis and anhydrosis

Squint

Type of squint	Features	Causes
Non paralytic squint	Deviation unchanged in all directions of gaze. Convergent or divergent	Refractive error Eye disease- optic atrophy, retinal disease
Paralytic squint	Deviation changes in all directions of gaze	**IIIrd nerve palsy –** (See above) **VIth nerve palsy** Pontine lesion (associated VII CN palsy and hemiplegia) Increased intracranial pressure

Facial nerve palsy

- If a facial nerve palsy is given for a case the first step is to find out whether the lesion is an upper motor neuron lesion or a lower motor neuron lesion
- The whole face will be affected in a LMN facial nerve palsy while only the lower half of the face will be affected in an UMN lesion

UMN	LMN
The lesion should be above the facial nerve nucleus which is located in the pons	**Pontine lesion** Associated 6th nerve palsy and hemiplegia **Lesion in the CP angle** Associated 8th and 5th nerve palsy **Lesion in the facial canal** Loss of taste in the anterior 2/3 of the tongue Hyperacousis **Lesion at the stylomastoid foramen** Isolated 7th nerve palsy **Lesion of the geniculate ganglion (Ramsay – Hunt syndrome)** Vesicles at the external ear

Ataxia

Causes of ataxia in children

- Congenital anomalies – Agenesis of the cerebellar vermis, Dandy – Walker malformation
- Cerebral palsy
- Cerebellar tumors - Medulloblastoma
- Infections
  During infections – coxsackie, echo, EBV
  Postinfectious – varicella
- Degenerative conditions – Friedrich's ataxia, ataxia telangectasia, Batten's disease
- Drugs and toxins – Phenytoin
- Seizures

Developmental assessment

- This is a very common short case at the exam and is easy to score from as it is extremely straight forward.
- Most of the developmental assessment is done through careful observation. The child given is usually an infant

General examination

- This is similar to the general examination in a neurology short case
- Look for
- Alertness of the child and interactions with the environment
- Ask to measure the OFC
- Dysmorphic features
- Posturing of the child – hypotonic posture, spasticity
- Movements – Is the child moving all the limbs? Look for asymmetry. Look for abnormal movements such as tics, chorea, athetoid movements and tremor
- Examine the skin for evidence of neurocutaneous manifestations – tuberous sclerosis, neurofibromatosis

Proceed to examine the main categories of the developmental assessment

- **Social and behavioural**
  Observe the interaction between the mother and the child and with you. This is a good estimate
- **Speech**
  Again observe the interactions between mother and child and assess the speech (can be done at any point in the examination
- **Vision and fine motor**
  Show the child an attractive toy and check for fixing and following. Hold the toy in your hand and see if the child reaches out to grasp the object
  Look for hand preference.
  See what the child does with the toy – transferring, playing

  Examine fine motor development – Do this by showing the child a small colored pebble. (Make sure the child does not swallow this). Look for pincer grasp

  If the child has well developed pincer grasp give some crayons and a book. Ask the child to draw
  Check for the ability to copy certain figures – circle, cross, triangle

  The draw a man test may also be utilized for this purpose
- **Gross motor**
  Perform the 180 degree technique to examine gross motor development

Start with the child in the supine position
Pull the child to sit and look for any obvious head lag
See if the child can sit (with support and then without support)
Pull the child to stand and see if he can stand (with support and then without support)
Hold the child in ventral suspension
Place the child on the bed in the prone position
Remember if the child is older direct examination of the gait is sufficient

- **Hearing**
  Offer to do the distraction test

Age	Gross motor	Fine motor and vision	Hearing and speech	Social and behavioural
6 weeks	Symmetrical limb movements Adopts fencing posture in the supine position	Fixes and follows to 90 degrees Grasp reflex	Cries/coos Startles to noise	Smiles
3 months	Moves limbs vigorously No head lag Lifts chest up from the prone position	Fixes and follows 180 degrees Plays with own hands	Turns to sound	Laughs
6 months	Sits without support Lifts chest on extended arms Rolls from supine to prone Downward parachute	Palmar grasp Transferring of objects	Says vowels and syllables	Laughs and screams
9 months	Pulls to standing Stands with support Rolls from prone to supine Forward parachute (7months)	Reaches for objects Points with index finger Early pincer grasp	Distraction test Says ‘mama’, ‘dada’ non specifically	Stranger anxiety Plays peek a boo Understands ‘no’ and ‘bye bye’
12 months	Walks if held and may take a few steps unsupported	Well developed pincer grasp	Knows name Understands simple commands Says a few words	Drinks from cup and uses spoon Finger feeds Waves ‘bye bye’
15 months	Broad based gait	Tower of 2 bricks To and fro scribble	2-6 words Communicates wishes and obeys commands	Uses cup and spoon
18 months	Steady purposeful walk Runs, squats Pushes/ pulls	Circular scribble Hand preference Points to pictures in book Turns pages of book	6-20 words	Points to named body parts Feeds independently Symbolic play
2 years	Kicks ball	Tower of 6 bricks	2-3 word sentences Proper grammar	Begins toilet training
3 years	Walks on tip toe	Tower of 9 bricks Copies circle	Gives first and last name Recognizes colours	Washes hands and brushes teeth
4 years	Hops	Builds steps of bricks Copies cross Draws a man	Counts to 10 or more	Able to undress
5 years	Skips Runs on toes Catches ball	Copies triangle	Asks how and when questions	Able to put on clothes and do large buttons

General short cases

Technique of examination

Head

- Large head
- Small head

Eyes

- Pallor
- Icterus

Cyanosis

Clubbing

Lymphadenopathy

Dysmorphic features and syndromes

- Down syndrome
- Turner syndrome
- Other

Short stature

Skin

- Description of a skin lesion
- Neonatal skin lesions
- Rashes associated with systemic disease
- Infection
  Impetigo
  Scabies
  Molluscum contagiosum
  Fungal
- Atopic dermatitis
- Sebhorrhoeic dermatitis
- Vasculitic rashes - HSP

Deformities

- Bow legs
- Knock knees

Neurological

- Abnormal posturing

Large head

- After picking up the large head from the initial observation continue with the examination as follows
- Look for the alertness of the child, activity, movements. Is the child adequately grown for the age?
- Look for dysmorphic features, evidence of neurocutaneous syndromes

Head

- Measure the head circumference in the proper manner
- Palpate the sutures and fontanelles
- Look for a VP shunt. If VP shunt is present track the tube down to the abdomen and inspect the abdominal scar. Check for the functioning of the VP shunt

Eyes

- Observe eye movements and pupils. Look for a squint (VI CN palsy)
- Look for nystagmus
- Sun setting sign
- Offer to examine the fundus with an ophthalmoscope

Examine the back for evidence of neural tube defects or scars suggesting surgical repair of these

- Meningomyelocoele is commonly associated with hydrocephalus

Examine the lower limbs (motor system)

Offer to examine the development of the child

Discussion

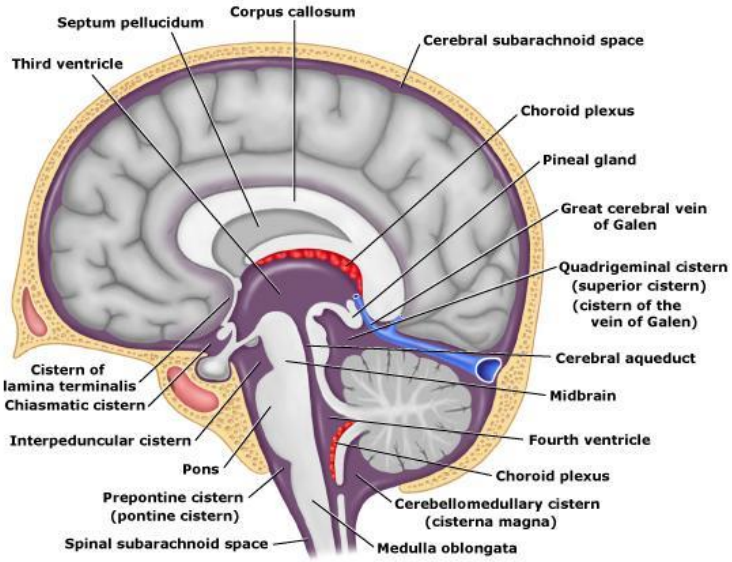
The following table gives the differential diagnosis for a child with a large head

Causes	Examples
Large bones	Achondroplasia Bone expansion – rickets, osteogenesis imperfecta Bone marrow expansion – chronic hemolytic anaemias (thalassemia)
Large brain	Normal variant Familial Soto's syndrome Metabolic – storage diseases

	(mucopolysaccharidosis)
Large ventricles	**Hydrocephalus** **Non communicating** Aqueduct stenosis Posterior fossa tumors Arnold – Chiari malformation **Communicating** Meningitis SAH
Large bleed	
Other	**Chronic cerebral edema** **Chronic subdural effusion**

Hydrocephalus

- This is the most important cause of large head in children
- Neuroimaging is used for confirmation of the diagnosis
- USS is the investigations of choice as it can be performed through the open fontanelles
- MRI or CT scan can be used in older children
- LP is contraindicated if the child has evidence of a space occupying lesion



Management

- This is with the placement of a VP shunt
- The proximal end of the catheter is placed into one lateral ventricle. This is connected through a one way valve to a reservoir
- The distal end is introduced into the peritoneal cavity
- Complications
  Blockage of the shunt
  Infection of the shunt
  Slit ventricle syndrome – occurs due to overdrainage of the CSF
  Abdominal complications – peritonitis
- Seizures are commonly associated with hydrocephalus

Small head

Causes of small head

- Normal variation
- Familial
- Genetic
- Secondary microcephaly
  Perinatal – congenital infection (look for other signs) and birth asphyxia (examine for developmental delay and features of cerebral palsy)
  Fetal alcohol syndrome
  Syndromes associated with learning difficulty
  Craniosynostosis

Pallor

- If you find pallor in the general examination complete the general examination and look for more clues
- Icterus
- Lymphadenopathy
- Then go on to examination of the abdomen – the most common finding will be hepato splenomegaly
- See discussion of hepatosplenomegaly under abdominal short cases

Icterus

Use the following table to assess further

	Further examination	Causes	Further assessment
Pre hepatic	Look for pallor Examine the abdomen for hepatosplenomegaly	Hemolytic anaemia	Elevated reticulocyte count Unconjugated hyperbilirubinaemia
Hepatic	Look for features of decompensated liver disease Look for stigmata of chronic liver disease	Acute liver disease (viral hepatitis, drugs) Chronic liver disease	Mixed hyperbilirubinaemia Transaminase rise more pronounced than the rise of alkaline phosphatase in acute hepatitis but may not be that pronounced in chronic liver disease
Post hepatic (cholestatic)	Remember that the jaundice will be extremely deep	**Intrahepatic** Due to chronic liver disease **Extrahepatic** Disorders of the biliary tree Gallstones Pancreatic structural anomalies	Conjugated hyperbilirubinaemia Alkaline phosphatase rise more pronounced than rise in transaminases

Clubbing

Causes of clubbing in children

Cardiovascular	Respiratory	Gastrointestinal	Other
Cyanotic congenital heart disease Infective endocarditis Atrial myxoma	Cystic fibrosis Bronchiectasis Lung abscess Empyema	Chron's Ulcerative colitis GI lymphoma Cirrhosis	Graves disease Familial

Lymphadenopathy

Given below are some important causes of generalized lymphadenopathy

Cause	Examples
Infections	
Bacterial	Brucellosis, typhoid fever, syphilis
Viral	CMV, EBV, HIV, measles, rubella
Protozoal	Toxoplasmosis
Malignancy	Lymphoma Leukaemia
Immune	SLE, still's disease

A child with dysmorphic features

- Remember that a syndromic diagnosis may not always be asked. Describe the dysmorphic features the child has

Head

- Compare the proportions of the head and the rest of the body
- Measure the OFC and look for macrocephaly and microcephaly
- Look for any abnormalities in the skull – scaphocepaly, brachycephaly, plagiocephaly

Eyes

- Look for hyper or hypotelorism, microphthalmos, buphthalmos
- Epicanthic folds, palpebral fissures
- Look at the orbital ridges – Are they shallow or prominent?
- Look at the cornea for clouding, blue sclera and abnormalities of the iris

Ears

- Look for low set ears
- Examine the shape and size of the auricle

Face

- Look for facial symmetry, maxillary/mandibular hypoplasia
- Mouth – Look for cleft lip/palate, high arched palate, macroglossia
- Examine the teeth
- Look for cyanosis (associated cardiac lesion)

Neck

- Webbing of the neck

Hands

- Simian crease, brachydactyly, clinodactyly, polydactyly, syndactyly, arachnodactyly, broad thumbs, absent thumbs

Feet

- Look for a wide sandal gap
- Rocker bottom feet

Skin

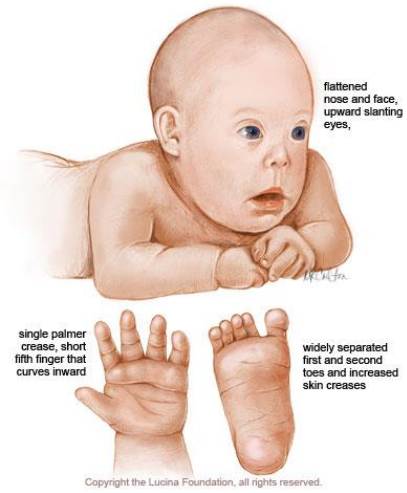
Examine the genitalia for any abnormalities

Discussion
Down syndrome

- Look at a patient and review the features of Down syndrome

Genetic basis

- Is due to trisomy 21
- Trisomy 21 can occur due to the following genetic defects
- Non disjunction at meiosis (13 -15,21 or 22)
- Robertsonian translocation
- Mosaicism

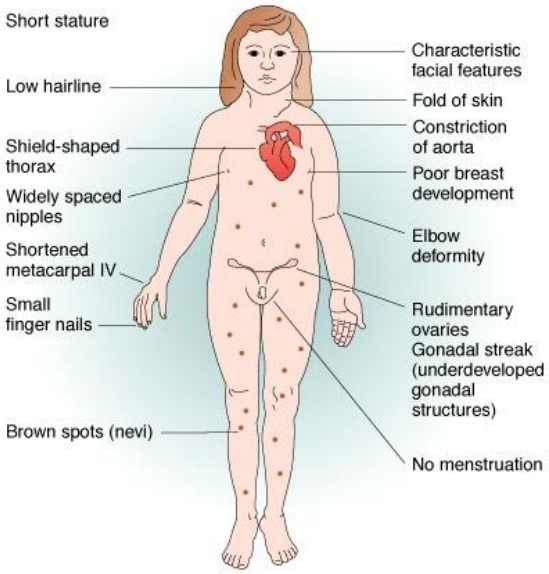


Other associated defects

System	Abnormalities
Cardiovascular	**AVSD, VSD, ASD, PDA, TOF** (in order of incidence)
GI	**Duodenal atresia, Hirschprung's disease, imperforate anus, esophageal atresia**
Neurological	**Developmental delay, intellectual impairment**
Special senses	**Refractive errors, hearing abnormalities**

Hematological and immunological	**Leukaemia, immunological disorders**
Growth	**Short stature, obesity**

Turner syndrome



Short stature

Causes of short stature

- Normal variation
- Familial
- **Inadequate nutrition**
- **Malabsorption**
- **Emotional deprivation**
- **Syndromes – Look for associated dysmorphic features**
  Down syndrome
  Turner syndrome
  Noonan syndrome
  Silver- Russel syndrome
- **Skeletal dysplasia**
  Osteogenesis imperfecta
  Rickets
- **Endocrine**
  GH deficiency
  Pituitary defect - craniopharyngioma
  Hypothyroidism
  Cushing syndrome or exogenous steroids
- **Chronic diseases**
- **Disproportionate short stature**

Investigations in a child with short stature will be guided based on the history and examination

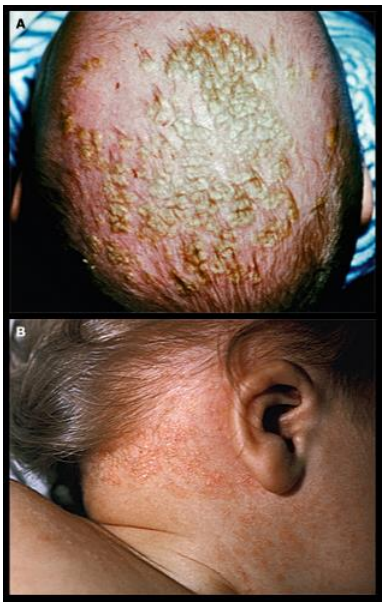
- X ray of the wrist and hands for the bone age – There will be some delay in constitutional delay of growth and puberty and marked delay for GH deficiency, hypothyroidism and other endocrine anomalies
- **FBC, LFT –** Features of nutritional deficiency
- **SE, blood urea, serum creatinine –** CRF
- **TSH**
- Karyotyping
- GH levels, IGF -1, GH stimulation testing
- MRI - Craniopharyngioma

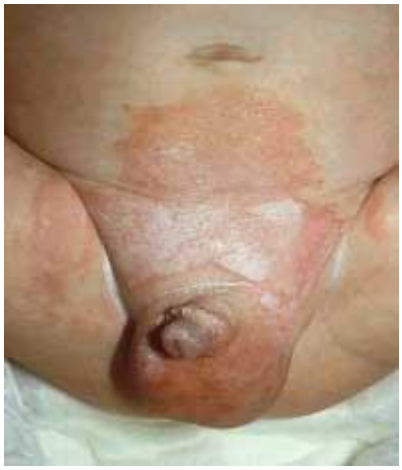
Skin short cases

Description of a skin lesion

Lesion	Description
Macule	Flat disc with alteration in colour or texture
Papule	Circumscribed palpable elevation <0.5 cm in diameter
Nodule	Solid mass in the skin >0.5cm in diameter
Plaque	Elevated area >2cm in diameter
Scale	A flake of stratum corneum
Wheal	A transient area of dermal edema
Vesicle	Visible fluid accumulation <0.5cm in diameter
Bulla	Visible fluid accumulation >0.5cm in diameter
Ulcer	Loss of dermis and epidermis

Nappy rash

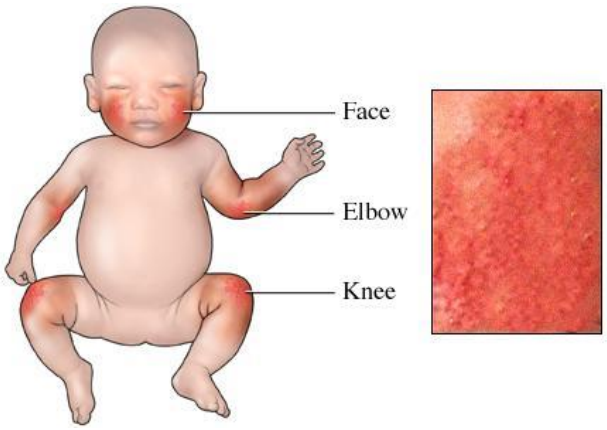


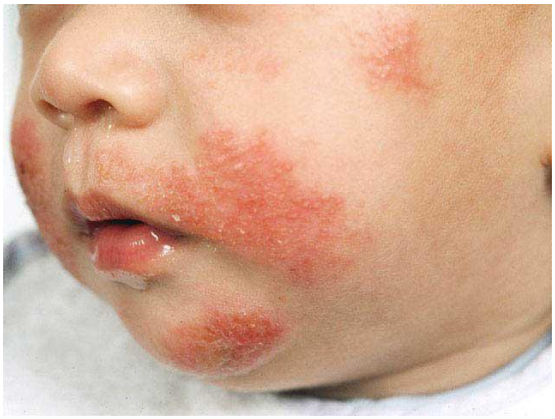


The main causes of nappy rash are

- Irritant dermatitis
  Is due to the irritant effects of urine and faeces. The flexural creases are spared. Treat with regular nappy change, emollient creams and in severe cases mild topical corticosteroids
- Candida infection
  No sparing of the flexural creases. Satellite lesions may be seen
  Treatment is with topical antifungal agent or combined antifungal and steroid preparation
- Seborrhoeic dermatitis
  Red moist rash with fine yellow scales. Starts in the scalp – known as cradle cap. Then affects the flexural creases and the nappy area
  Treatment
  Sulphur and salicylic acid containing ointment to the scalp
  Other lesions with combined steroid and antifungal ointments and emollients

Atopic eczema





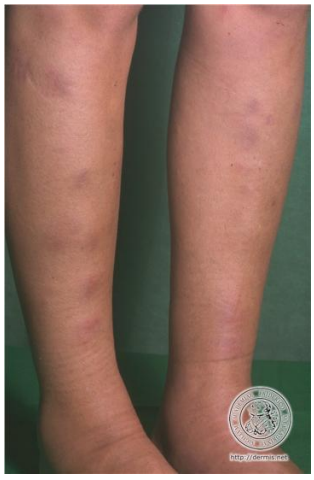
Management

- Avoid irritants and precipitants
- Emollients
- Topical corticosteroids
- Occlusive bandages – These are used especially when itching is a major problem. May be impregnated with zinc paste
- Antibiotics for infected eczema
- Anti histamines for suppression of itching
- Dietary management if associated food allergies

Rashes associated with systemic disease

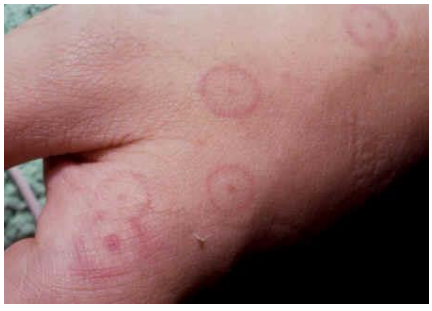
Erythema nodosum

Causes



Infections	Bacterial – streptococcal, Mycoplasma, TB, salmonella Viral – EBV, hepatitis B Fungal
Inflammatory bowel disease	UC, chron's disease
Autoimmune	Sarcoidosis, SLE
Drugs	Sulphonamides

Erythema multiforme



Causes

- HSV infection
- Mycoplasma infection
- Drug reaction

Henoch – Schonlein purpura

- Is a type of vasculitis affecting small vessels
- There are 4 classical features. These are
  Rash – purpuric rash typically involving the buttock and extensor surfaces of the lower limbs
  Joint pain – Mainly affects weight bearing joints
  Abdominal pain – Intestinal colic, intususception
  Hematuria – This is due to glomerulonephritis

Management

- Supportive

Infections of the skin

Disease	Clinical features	Investigations	Management
Impetigo *(Staphylococcus aureus)*	Honey coloured crusting around the mouth and nose		Topical antibiotics Systemic antibiotics if widespread
SSSS	Brightly erythematous skin, fissuring and crusting around the eyes, nose and lips May be systemically unwell	Skin and blood culture Hematological investigations	Manage the systemic disease Emollients Systemic antibiotics
Dermatophyte infections	Annular, scaling, erythematous lesion with an active, raised border and central clearing	Skin scrapings, nail clippings and air for microscopy	Antifungals
Molluscum contagiosum	Papules with central umbilication		Spontaneous resolution
Scabies	Burrows, papules and vesicles Occur in finger webs, palms, soles and axillae		Permethrin Benzyl benzoate Malathion

Petechial rash

- These are pinpoint, erythematous, non blanching lesions
- Are caused by abnormalities in the haemostatic pathway

Disruption in vascular integrity	Disorders of coagulation
Infection Dengue **Trauma** **Vasculitis** HSP	**Platelet disorders** **Thrombocytopenia** As a component of pancytopenia (i.e. bone marrow infiltration) Inherited thrombocytopenia ITP TTP DIC Drug induced thrombocytopenia **Platelet functional disorders** **Clotting factor deficiency** Von Willebrand disease Hemophilia Liver disease Vitamin K deficiency

ITP

Investigations

- **FBC**
  Confirm the thrombocytopenia and exclude a pancytopenia
- **Blood picture**
  This is done to exclude any abnormalities of the white cells which could indicate a leukaemia causing pancytopenia
- Further assessment may be done by a bone marrow biopsy – in ITP there may be a large number of megakaryocytes

Initial management

- Education
- Pharmacological management
  Steroids
  IVIG
- Consider platelet transfusion if there is life threatening bleeding
- Chronic ITP

Defined as persistent thrombocytopenia beyond 6 months of presentation

- **Pharmacological management**
- Steroids
  Immunosuppressant drugs
- Splenectomy may be considered as a last option in selected cases

Bow legs

- **Rickets is the most likely diagnosis**
- **Other likely causes**
  Physiological
  Trauma
  Blount's disease

Look for the other features of rickets

Head

- Frontal bossing
- Wide open fontanelles
- Craniotabes
- Examine the teeth

Chest

- Rachitic rosary – enlargement of the costochondral junctions along the anterolateral aspect of the chest
- Harrison's sulcus

Upper limbs

- Widening of the wrists

Lower limbs

- Widening of the ankles
- Bow legs
- (Or) knock knees
- Hypotonia

Discussion

Important investigations which should be performed

- **X – Ray B/L wrists**
  Widening of the epiphyseal plate
  Cupping, fraying and splaying of the distal radial and ulnar metaphysis
- Serum alkaline phosphatase – will be elevated

Causes

Hypocalaemic rickets	Hypophosphatemic rickets
Disorders related to vitamin D Dietary deficiency of vitamin D Malabsorption of vitamin D Liver disease Chronic renal failure Vitamin D receptor defects **Deficiency of calcium**	**Renal tubular disease**
Low serum calcium **Low serum phosphate** **(Can be increased in rickets associated with chronic renal failure)** **Increased PTH**	**Normal serum calcium** **Low serum phosphate** **Normal PTH**

Management of vitamin D deficient rickets

The most widely used treatment for vitamin D deficiency consists of vitamin D2 (ergocalciferol). The following dosing scheme is commonly recommended for treatment of vitamin D deficient rickets

- 1000 Int. Units daily – age less than 1 month
- 1000 to 5000 Int. Units daily – age 1-12months
- 5000 Int. Units daily for children one year and older

Treatment is continued at the above doses until radiographic evidence of healing is seen; then the dose of vitamin D is reduced to 400 Int. Units daily thereafter.

Neonatal examination

This is also a common case given at the exam. **The most important aspect is to wash your hands before examination.**

General appearance of the patient

Ask the mother to undress the patient completely and look at the following

- Look for gross deformities or dysmorphism
- Assess the respiratory effort
- Look at the position and movements of the child
- Assess the colour of the child – look for pallor, cyanosis and Icterus

Measurements of the child – offer to plot in a centile chart

Head

- Look for any abnormalities of the skull
- Palpate the fontanelles and sutures
- Look for caput succedaneum and cephalhematoma

Face

- Look for symmetry and facial palsies

Eyes

- Look at the spacing
- Symmetry
- Palpebral fissures
- Eye movements
- Look at the sclera, conjunctiva, cornea
- Examine the pupils
- Ask to examine for the red reflex with the ophthalmoscope

Examine the ears and nose

Look at the mouth and teeth

Look at the neck for any lumps

Examine the upper limbs

Examine the chest and auscultate the lungs

Examine the CVS

- Examine the pulse
- Examine the apex
- Auscultate for murmurs

Examine the abdomen

- Look for abdominal distension
- Scaphoid abdomen – associated with diaphragmatic hernia
- Look for visible lumps, defects in the anterior abdominal wall
- Palpate for masses in the abdomen
- Examine the umbilical cord

Examine the external genitalia

Do Ortolani's test and Barlow's test for dislocation of the hip

Do a quick neurological examination of the limbs

Look at the back and examine the anus

Look for the primitive reflexes