HEPATIC DISORDERS

BIANCA

PRACTICAL RE-CAP

SIGNS & SYMPTOMS OF LIVER DISEASE
→ JAUNDICE
→ ASCITES
→ HEPATOMEGALLY/SPLENOMEGALLY
→ ENCEPHALOPATHY
→ BLEEDING TENDENCY

SPECIFIC DISEASES
→ HEPATITIS
→ LIVER FAILURE
→ CIRRHOSIS
→ PHT
→ SCHISTOSOMIASIS

The liver is evil and must be punished.
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The liver is evil and must be punished.
A. Practical Re-cap

History:
- Exposure to organic solvents (glue-sniffing)
- Use of pharmaceutical drugs (paracetamol, TB treatment)
- Use of traditional remedies (Senecio species)

Examination:
- Fullness of flanks → ascites
- Size of liver: should extend to 1-2cm below the costal margin in the mid-clavicular line
- Remember to comment on the border being: soft/firm / nodular / pulsating

B. Signs and Symptoms of Liver Disease
- Jaundice (1)
- Ascites and abdominal distension (2)
- Hepatomegally and/or hepatosplenomegally (3)
- Encephalopathy (4)
- Bleeding tendency

1. Jaundice
   60% term infants
   80% pre-term infants
   Children and adults with Total Serum Bilirubin $>35\mu mol/l$
   Neonates with TSB $>85\mu mol/l$

   a. Neonatal Jaundice

   | Physiological |
   | Non-Physiological (Persistent) |

   **Physiological**
   - Appears on Day 2 and declines after a week
   - Due to:
     - ↑ haemolysis (neonates RBC have shorter half life) therefore ↑ bili production
     - Liver immaturity therefore defective conjugation
     - ↓ excretion (β-glucoronidase deconjugates bili in the intestine to be re-up taken by placenta during fetal period, this enzyme is found in breast milk)
   - TSB upper limit: 200umol/l

   **Non-Physiological (Persistent)**
   - Onset before 24 hours or persistence beyond 10 days or a rise in TSB by 8.5umol/l/hour
   - Due to:
     - Infection
     - Mechanical Biliary Tract Obstruction
     - Genetic Defects
     - Metabolic Defects
     - Idiopathic Neonatal Hepatitis
   - Refer to pg 578 Table 28.1 for specifics

   Signs of underlying illness: vomiting, lethargy, poor feeding, excessive weight loss, apnoea, tachypnoea, temp instability

   → dark urine & pale stools
**Non-Physiological Persistent Neonatal Jaundice**

Present with dark urine, pale stools. On examination there is often: growth retardation, hepatosplenomegally and signs of a bleeding tendency.

**Approach:**

1. **Conjugated/Unconjugated?**
   - **Conjugated:** conjugated fraction is >20% of TSB
     Mainly neonatal hepatitis or obstructed biliary tree (together cause 90% of cholestatic jaundice in infants)
   - **Unconjugated:** conjugated fraction is <20% of TSB
     Mainly due to haemolysis or inadequate hepatic conjugation

2. If Conjugated differentiate between 2 main causes

<table>
<thead>
<tr>
<th>STOOL</th>
<th>NEONATAL HEPATITIS</th>
<th>OBSTRUCTED BILIARY TREE</th>
</tr>
</thead>
<tbody>
<tr>
<td></td>
<td>↑AST ↑ALT</td>
<td>↑AST ↑ALT ↑ALP</td>
</tr>
<tr>
<td></td>
<td>GGT:ALP &lt;2</td>
<td>GGT:AST &gt;2</td>
</tr>
<tr>
<td></td>
<td>Serology may be Positive</td>
<td>Serology Negative</td>
</tr>
<tr>
<td></td>
<td>Cultures may be Positive</td>
<td>Cultures Negative</td>
</tr>
<tr>
<td></td>
<td>Butyl IDA/Mebrofenin scan: no biliary obstruction</td>
<td>Butyl IDA/Mebrofenin scan: biliary obstruction</td>
</tr>
</tbody>
</table>

Find cause by doing the following tests:
FBC, CRP, TORCHes, TFT, LFT and urine dipstick

**MANAGEMENT**

- Treat according to cause, i.e:
  - syphilis, UTI
  - Neonatal hepatitis with giant-ell histology is confirmed on liver-biopsy
  - In most cases no definite cause will be determined and so management will be supportive

Supportive Mx:
- Phototherapy (risk of retinal damage, dehydration and lack of maternal bonding)
- *Exchange transfusions*
- *Phenobarbital*

- Early Surgery
- Explorative laparotomy
- Intra-operative cholangiogram
- Intra-hepatic Biliary Atresia→ inoperable
- Extra-hepatic Biliary Atresia→ porto-enterostomy (bowel drains directly from liver to bowel) Kasai operation

**Complications:**
Kernicterus which is characterized by a hight-pitched cry, lethargy, acidosis, hypotonia, and brain damage.
b. Acute Jaundice in Child/Adolescent

**Acute Jaundice**

**Appears Healthy + Jaundice**

- Limited investigations

**Unconjugated Hyperbilirubinaemia:**
  - Haemolysis / Gilbert’s Disease

**Conjugated Hyperbilirubinaemia:**
  - Hepatitis A / Hepatitis B
  - Diagnosis supported by ↑ Transaminases and + serology
  - If not: refer patient. Could be: non-A, non-B Hepatitis, gallstones, extra-hepatic obstruction, CF, Wilson's Disease, α1-antitrypsin deficiency

**Appears Ill + Jaundice**

- Toxic, Confused, Bleeding Tendency
  - Medical Emergency

- May be: Fulminant hepatic failure, Sepsis, Shock, Acute Cholangitis

- Refer to enable Dx to be made and Mx to be started immediately

**NB History:**
- Recent prodromal symptoms (fever, malaise, rash, arthritis)
- Signs of infectious mono-nucleosis (sore throat, lymphadenopathy, fever)
- Med Hx: jaundice / liver disease, drugs and toxins
- Fam Hx: Hepatitis A/B
- Surgical Hx: exposure to blood transfusions and blood products

**NB Examination**

**Hepatosplenomegally (relatively non-specific)**
- Hard: underlying cirrhosis therefore look for spider naevi, palmar erythema, clubbing and oedema
- Decrease in size of spleen: GI-haemorrhage in a patient with underlying PHT
- Massive: Budd-Chiari, Veno-Occlusive Disease, Right-Sided Heart Failure, Liver Neoplasm

2. Ascites and Abdominal distension

Collection of fluid in the peritoneal cavity

- Examination findings:
  - Abdominal distension
  - Shifting dullness
  - Fluid thrill

Approach depends on the type of fluid present and the presence of underlying pathology.
MECHANISM | AETIOLOGY | ASCITIC FLUID | DIAGNOSIS
--- | --- | --- | ---
Lymph Blockage | Congenital/Neoplastic | Lymph | Chylous fluid. Milky with fat globules and lymphocytes
↑Intra-vascular Hydrostatic Pressure | Causes of PHT | Transudate | Clear fluid with ↓ protein
↓Intra-vascular Oncotic Pressure | Hypo-proteinaemia in particular nephritic syndrome and cirrhosis | Transudate | Clear fluid with ↓ protein and ↓ serum albumin
↑Vascular Permeability and Inflammation | Causes of peritonitis (TB) | Exudate | Cloudy / Dark-Yellow Fluid with ↑ protein that clots. Polymorpho-nuclear leucocytes >25%

Management
Treat the cause.
Therapeutic paracentesis should be avoided because of the high risk of infection. Fluid tapped leads to re-accumulation which results in protein loss.
Loop-diuretics should not be used alone, rather in combination with spirinolactone.
In nephritic syndrome: furosemide + salt-free-albumin-infusion is effective.

3. Hepatomegaly and hepatosplenomegally
Clues from History and Examination
- Jaundice → INFLAMMATION
- TB / Generalised infection → RETICULO-ENDOTHELIAL (KNUPFER) CELL HYPERPLASIA
- CVS Symptoms → VENOUS CONGESTION
- Local Tenderness + Swelling + Acutely ill → LIVER ABSCESS
- Ascites + Absence of filling of jugular veins with pressure on liver → BUDD CHIARI
- Bleeding Tendency + Lymphadenopathy + Infection + Anaemia → INFLITRATIVE PROCESS
- AbN appearance + Neuro signs → STORAGE DISEASE
- Post-glycaemic episodes + metabolic acidosis → GLYCOGEN-STORAGE DISEASE
- Kwashiokor + Malnutrition → FATTY INFILTRATION

Page 582, Table 28.4 has a list of more causes and special investigations that should follow

4. Encephalopathy
An altered level of consciousness or abnormal behaviour
It can be staged as follows:

<table>
<thead>
<tr>
<th>STAGE</th>
<th>CLINICAL</th>
<th>SIGNS</th>
</tr>
</thead>
<tbody>
<tr>
<td>1</td>
<td>Subtle behaviour changes&lt;br&gt;Altered sleep pattern&lt;br&gt;Shortened attention span</td>
<td>Incoordination&lt;br&gt;Fine tremor</td>
</tr>
<tr>
<td>2</td>
<td>Personality changes&lt;br&gt;Drowsiness, disorientation&lt;br&gt;Agitation</td>
<td>Asterixis&lt;br&gt;Ataxia&lt;br&gt;Hypertonia</td>
</tr>
<tr>
<td>3</td>
<td>Temporal and spatial disorientation</td>
<td>Asterixis</td>
</tr>
<tr>
<td>Delerium, hallucinations</td>
<td>4</td>
<td></td>
</tr>
<tr>
<td>--------------------------</td>
<td>---</td>
<td></td>
</tr>
<tr>
<td>Mania</td>
<td>Comatose</td>
<td></td>
</tr>
<tr>
<td>Stupor, seizures</td>
<td>Semi/Unconscious</td>
<td></td>
</tr>
<tr>
<td>Rigidity</td>
<td>10</td>
<td></td>
</tr>
</tbody>
</table>

Asterixis: is a tremor of the wrist when dorsiflexed (flapping tremor)

C. Specific Diseases

1. Hepatitis

Inflammatory mononuclear cell infiltrate with varying degree of damage to hepatocytes, caused by infection, toxic substances and metabolic disorders. In children the majority of cases of hepatitis are caused from the virus: Hepatitis A / B

Causes of Hepatitis

- **Infection**
  - Viral: HAV, HBV, HCV, HDV, HEV, HGV, EBV; CMV, HSV, HIV
  - Bacterial: Leptospirosis, Pyogenic Bacteria
  - Parasitic: Toxoplasma Gondii, Entamoeba Histolytica, Schistosomiasis

- **Toxins and Drugs**
  - Toxins: Senecio alkaloids: Aflatoxins, Amanita Mushrooms
  - Drugs: Antimicrobials, Anti-TB, Anti-convulsants, Analgesic and Anti-inflammatory, Anaesthetic agents (halothane) For specific drugs look at page 583 Table 28.5

- **Physical Agents**
  - Burns, Irradiation, Hypothermia

- **Metabolic Errors**
  - Wilson’s disease

<table>
<thead>
<tr>
<th>HEPATITIS A</th>
<th>HEPATITIS B</th>
</tr>
</thead>
<tbody>
<tr>
<td>- Picorna virus</td>
<td>- Picorna virus</td>
</tr>
<tr>
<td>- Highly infectious in prodromal period</td>
<td>- All products from these patients infectious</td>
</tr>
<tr>
<td>- Infection mild/asymptomatic</td>
<td>- Chronic carrier state</td>
</tr>
<tr>
<td>- Dx confirmed with IgM antibody to HAV</td>
<td>- Dx confirmed by appropriate serology</td>
</tr>
<tr>
<td><strong>EPIDEMIOLOGY</strong></td>
<td><strong>EPIDEMIOLOGY</strong></td>
</tr>
<tr>
<td>- Faecal-Oral Transmission</td>
<td>- Parenteral and Venereal Transmission</td>
</tr>
<tr>
<td>- Via contaminated water and food</td>
<td>- Via blood, serum, semen, tears, saliva and breastmilk</td>
</tr>
<tr>
<td>- Common among children with inadequate sewage disposal</td>
<td>- Peak incidence: 2-11 years, due to horizontal transmission</td>
</tr>
<tr>
<td>- Chronic carriers will only be improved with improved living conditions</td>
<td>- Chronic carriers will only be improved with improved living conditions</td>
</tr>
<tr>
<td><strong>VIROLOGY</strong></td>
<td><strong>VIROLOGY</strong></td>
</tr>
<tr>
<td>- RNA virus</td>
<td>- DNA virus</td>
</tr>
<tr>
<td>- Enterovirus genus</td>
<td>- Outer lipoprotein coat = HBsAg</td>
</tr>
<tr>
<td>- Once infected the individual</td>
<td>- Inner nucleoside core = HBCAg</td>
</tr>
</tbody>
</table>
**A**

- develops lifelong immunity (IgG antibodies)
- Incubation period: 2-4 weeks
- Multiplies in liver
- Shed via bile to stools
- Infectivity maximal at onset of Prodrome

**B**

- Additional antigen = HBeAg
- HBV specific DNA polymerase indicates acute infection and viral replication
- IgM and IgG antibodies to HBcAg appear first
- Anti-HBe and Anti-Hbc develop in those who have overcome infection and have permanent immunity
- Carrier: HBsAg, HBeAg and HBV DNA polymerase persist. Despite the presence of anti-HBcAg they fail to develop anti-HBe and anti-HBs
- ↑ anti-Hbc indicate ongoing liver damage
- HBV causes hepatocellular damage by either a cellular & humoral response OR a direct effect on hepatocytes

<table>
<thead>
<tr>
<th>CLINICAL FEATURES</th>
<th>INVESTIGATIONS</th>
</tr>
</thead>
<tbody>
<tr>
<td>Assymptomatic to Mild</td>
<td>↑ serum transaminases (a few)</td>
</tr>
<tr>
<td>Often no jaundice (anicteric), if present it resolves in 2 weeks</td>
<td></td>
</tr>
<tr>
<td>Prodrome: nausea, vomiting, diarrhoea, fever, RUQ pain</td>
<td></td>
</tr>
<tr>
<td>Tender hepatomegallyin 30% of patients</td>
<td></td>
</tr>
<tr>
<td>Dark urine and pale stools</td>
<td></td>
</tr>
<tr>
<td>50% of patients have anicteric subclinical infections</td>
<td></td>
</tr>
<tr>
<td>Acute hepatitis from HBV is clinically indistinguishable from HAV</td>
<td></td>
</tr>
<tr>
<td>Adult prodrome: articulargia, skin rash (not in children)</td>
<td></td>
</tr>
<tr>
<td>Prolonged course</td>
<td></td>
</tr>
<tr>
<td>Chronic hepatitis (non-specific complaints: malaise, lethargy) may follow an acute episode.</td>
<td></td>
</tr>
<tr>
<td>Glomerulonephritis may occur due to HBV-circulating immune complexes. Present with nephritic syndrome</td>
<td></td>
</tr>
<tr>
<td>Arthritis and pericarditis can also occur due to these immune complexes</td>
<td></td>
</tr>
<tr>
<td>Carriers are unique to HBV. Indicators of active infection exist without evidence of liver disease</td>
<td></td>
</tr>
</tbody>
</table>

Look at page 584 Table 28.2 for graph

Look at page 586 Table 28.4 for graph

Biochemical investigations
<table>
<thead>
<tr>
<th>MANAGEMENT</th>
<th>No specific treatment</th>
<th>No specific treatment</th>
</tr>
</thead>
<tbody>
<tr>
<td>Supportive Management</td>
<td>Health-Workers must take appropriate precautions</td>
<td>Supportive Management</td>
</tr>
<tr>
<td>Diet: adequate fluids and energy</td>
<td>General measures as for HAV</td>
<td>Diet: adequate fluids and energy</td>
</tr>
<tr>
<td>Cster NOT indicated</td>
<td>↑risk of horizontal transmission, thus isolation may be necessary</td>
<td>Cster NOT indicated</td>
</tr>
<tr>
<td>Paracetamol and salicylates are CONTRA-INDICATED</td>
<td>Steroids, Anti-virals and interferon use is controversial</td>
<td>Paracetamol and salicylates are CONTRA-INDICATED</td>
</tr>
</tbody>
</table>

<table>
<thead>
<tr>
<th>PREVENTION</th>
<th>Most cases asymptomatic and those which are symptomatic are most infectious in prodromal phase therefore impossible to isolate</th>
<th>Priority in all blood products and should be carefully screened</th>
</tr>
</thead>
<tbody>
<tr>
<td>Improving sewage disposal, ensuring safe drinking water and encourage personal hygiene (washing hands)</td>
<td>HBV+ patients should be identified</td>
<td>Improving sewage disposal, ensuring safe drinking water and encourage personal hygiene (washing hands)</td>
</tr>
<tr>
<td>Normal Ig contains sufficient AB to HAV to give passive immunity for 3 months</td>
<td>Pregnant patients to take preventative measures to avoid vertical transmission</td>
<td>Normal Ig contains sufficient AB to HAV to give passive immunity for 3 months</td>
</tr>
<tr>
<td>HAV vaccine available and effective. Given as 2 doses in children over 2 years that are at risk of the infection</td>
<td>Improvement of hygiene will reduce the number of carriers</td>
<td>HAV vaccine available and effective. Given as 2 doses in children over 2 years that are at risk of the infection</td>
</tr>
<tr>
<td>ACTIVE IMMUNISATION</td>
<td>ACTIVE IMMUNISATION</td>
<td>ACTIVE IMMUNISATION</td>
</tr>
<tr>
<td>Recommended for all children (part of EPI at 6, 10 and 14 weeks)</td>
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<td>Recommended for all children (part of EPI at 6, 10 and 14 weeks)</td>
</tr>
<tr>
<td>Babies born to mothers that have HBsAg should receive anti-HBV gammaglobulin at birth and the vaccine at 4, 8 and 16 weeks</td>
<td>Babies born to mothers that have HBsAg should receive anti-HBV gammaglobulin at birth and the vaccine at 4, 8 and 16 weeks</td>
<td>Babies born to mothers that have HBsAg should receive anti-HBV gammaglobulin at birth and the vaccine at 4, 8 and 16 weeks</td>
</tr>
<tr>
<td>PASSIVE IMMUNISATION</td>
<td>PASSIVE IMMUNISATION</td>
<td>PASSIVE IMMUNISATION</td>
</tr>
<tr>
<td>Hyperimmune globulin is avised after HBV exposure</td>
<td>Hyperimmune globulin is avised after HBV exposure</td>
<td>Hyperimmune globulin is avised after HBV exposure</td>
</tr>
</tbody>
</table>

<table>
<thead>
<tr>
<th>COMPLICATIONS</th>
<th>Rare</th>
<th>Fulminating Hepatitis with Massive Liver Necrosis (10-20 in 1000, 8090% mortality) Look for: deepening jaundice, persistent prodromal symptoms, ↓liver size, sleepiness, confusion,</th>
</tr>
</thead>
<tbody>
<tr>
<td>Fulminant hepatic failure (5 in 1000)</td>
<td>Aplastic anaemia (↑mortality)</td>
<td>Fulminant hepatic failure (5 in 1000)</td>
</tr>
<tr>
<td>Aplastic anaemia (↑mortality)</td>
<td>No carrier status</td>
<td>Aplastic anaemia (↑mortality)</td>
</tr>
<tr>
<td>No carrier status</td>
<td></td>
<td>No carrier status</td>
</tr>
</tbody>
</table>
abnormal behaviour, bleeding tendency that fails to correct with Vit K
- Aplastic Anaemia
- Chronic Hepatitis
Hepatitis>3months and unclear whether child is a carrier. If HBsAg is + for 6 months do a liver biopsy. Results may be: chronic persistent hepatitis, chronic active hepatitis or established cirrhosis
- Hepatocellular Carcinoma

Other viral hepatitis
- HDV: incomplete RNA virus. Requires HBV for infectivity and replication. No treatment available although HBV vaccine should limit spread.
- HGV: cause of giant-cell hepatitis. Poor prognosis

2. Liver Failure
Impairment of liver functions: synthesis, detoxification and excretion.
Aetiology:
- Infection: HAV, HBV, HCV, other viruses during neonatal period due to immature immune system.
- Metabolic Disorders:
- Toxic: herbal remedies, overdose, idiosyncratic
Clinically:
- Liver disease → jaundice, ascites, enlarged/shrinking liver, anorexia, vomiting. Raised transaminases. If NO jaundice in liver failure; think of Reyes Syndrome!
- Systemic Complications → signs of encephalopathy, coagulopathy, renal failure, cardio-resp failure, sepsis, hypoglycaemia and metabolic acidosis.
Management:
- Determine the cause
- Degree of hepatic necrosis
- Medical support for recovery of hepatic lesion / consider hepatic replacement
- Transplantation is limited because of high costs, poor availability and lack of expertise

3. Other Chronic Liver Disease
Prolonged clinical course of hepatitis (>3months) → Biopsy liver and refer
Cirrhosis is usually the consequence
**CHRONIC PERSISTENT HEPATITIS**
- Benign condition
- Persistently ↑ transaminases after acute viral hepatitis
- Dx confirmed by biopsy (periportal inflammatory cell infiltration with normal histology)
- No specific treatment
- HBsAg+ have a higher risk of cirrhosis

**CHRONIC ACTIVE HEPATITIS**
- Auto-immune disorder
- Same histological picture as HBV
- Clinically: symptoms of acute hepatitis to non-specific complaints. Most patients have jaundice, hepatosplenomegaly, spider angiomata, striae, acne, palmar erythema. Extra-hepatic manifestations may be arthiritis, haemolytic anaemia, thyroiditis, glomerulonephritis, enteropathy and IBS
- Investigations: ↑ bili, ↑ transaminases, ↓ albumin, prolonged pro-thrombin time. ↑ IgG, auto-antibodies positive. Liver biopsy (inflame cell infiltrate from portal tracts to liver parenchyma with piecemeal necrosis and bridging fibrosis)
- Management: Steroids, Azathioprine (control inflame process with least SE)

**3. Cirrhosis**
= Necrosis + Fibrosis
Normal liver architecture is destroyed and replaced by regenerative tissue surrounded by fibrosis. It is progressive and will result in liver failure. It is the end-stage of many conditions and once established it is difficult to find the cause.

**CAUSES OF NECROSIS**

<table>
<thead>
<tr>
<th>Biliary Cirrhosis</th>
<th>Post-Necrotic Cirrhosis</th>
<th>Genetic</th>
</tr>
</thead>
<tbody>
<tr>
<td>Biliary Atresia</td>
<td>Neonatal/Viral/Chronic Hepatitis</td>
<td>Wilson’s Disease</td>
</tr>
<tr>
<td>Choledocal Cyst</td>
<td>Constrictive Pericarditis</td>
<td>Galactosaemia</td>
</tr>
<tr>
<td>Cystic Fibrosis</td>
<td>CHF</td>
<td>α1-Anti-Trypsin</td>
</tr>
<tr>
<td>Bile Duct Stenosis/Obstruction</td>
<td>Budd Chiari Syndrome</td>
<td>Deficiency</td>
</tr>
<tr>
<td>Ascending Cholangitis</td>
<td>Veno-Occlusive Disease</td>
<td>Glycogen Storage Disease</td>
</tr>
</tbody>
</table>
Clinically:
- Signs of underlying cause: Chronic liver disease / PHT
- Despite significant cirrhosis there may be no clinical signs

Investigations:
- ↓ albumin
- ↑ transaminases
- ↑ gammaglobulins
- ↑ ALP and ↑ cholesterol in biliary sclerosis
- Biopsy to determine the degree of cirrhosis (provided that the clotting profile is normal)

Management:
- Underlying cause
- Supportive
- Maintenance of general nutrition
- Liver transplantation

4. PHT

**EXTRA HEPATIC CAUSES**

- **Pre-Sinusoidal**
  - Portal Vein Obstruction
  - Splenic Vein Obstruction

- **Intrahepatic**
  - Cirrhosis
  - Schistosomiasis
  - Congenital Hepatic Fibrosis
  - Acute / Chronic Hepatitis

- **Post-Sinusoidal**
  - Budd Chiari Syndrome
  - IVC obstruction
  - Pericarditis
  - Heart Failure

Clinically:
- GI Haemorrhage (oesoph varices)
- Asymptomatic Slenomegaly
- Ascites
- Abdominal Distension
- Encephalopathy

Investigations:
- FBC
- Clotting Profile
- Endoscopic Examination (oesoph varices)
- Liver Scan
- Liver Biopsy

Management:
- Conservative
- Ascites: ↓ Na intake and diuretics
- Splenectomy (rare)
- Oesophageal Varices: Endoscopic Sclerotherapy / Banding
- GI Haemorrhage: Resuscitation and Transfer
5. Schistosomiasis
Intermediate host of Schistosomiasis Mansoni is a fresh-water snail, cercariae released from the snail pierce the skin and cause an infection. Parasites migrate to the liver. Deposition of eggs leads to inflammation response with healing by fibrosis this may lead to portal vein obstruction and thus splenomegally.

**Clinically:**
- Hepatomegally
- PHT
- Splenomegally
- Ascites
- Oesoph Varices

**Investigations:**
- Normal LFT’s
- Ova in urine, stool and liver biopsy

**Management:**
- Praziquantel 40mg/kg stat

6. Veno-Occlusive Disease
Hepatic vein obstruction caused by ingestion of alkaloids (Senecio plant species). The lesion is initially in the central vein of the hepatic lobule and branches of the hepatic vein. Endothelial oedema is followed by fibroblastic proliferation and occlusion of the vessel. Venous congestion → necrosis → cirrhosis.

**Clinically:**
- Commonly < 6 years
- Sudden onset
- Hepatomegaly
- Severe ascites
- Absence of jaundice

**Management:**
- No specific treatment
- Control ascites
- Patients that survive the acute phase progress to chronicity with PHT
- Educate patient on herbal enemas and traditional medicines

7. Gallstones and Cholecystitis
Uncommon. Contributing conditions: haemolytic anemia, CF, hypercholesterolaemia, congenital abN of biliary tract, TPN, ileal resection and Crohns.

Stones are radiolucent on Xray in children, therefore US preferred

D. Liver Transplantation
Careful election of families and candidates is critical because follow-up and long-term immunosuppressants (steroids, azothiaprine, mycophenolate mofetil, cyclosporine) are required for success.

Team: transplant surgeons, transplant physicians, radiologists and ICU staff.
85-90% 1 year survival
70-80% 5 year survival

Cx: growth retardation, infection, cosmetic effects and malignancy
<table>
<thead>
<tr>
<th>INDICATIONS</th>
<th>CONTRA-INDICATIONS</th>
</tr>
</thead>
<tbody>
<tr>
<td>Acute Fulminant Liver Failure</td>
<td>Hepatitis B (HBeAg+)</td>
</tr>
<tr>
<td>Biliary Atresia</td>
<td>Hepatitis C</td>
</tr>
<tr>
<td>Chronic Hepatitis</td>
<td>HIV</td>
</tr>
<tr>
<td>Cirrhosis</td>
<td>TB</td>
</tr>
<tr>
<td>Metabolic conditions</td>
<td>Malignancy outside of liver</td>
</tr>
<tr>
<td></td>
<td>Irrev disease of other organs</td>
</tr>
<tr>
<td></td>
<td>Psychosocial Factors</td>
</tr>
</tbody>
</table>
Diarrhoea
GORD
Pyloric Stenosis
Wilson's Disease
Meckel's Diverticulum
Appendicitis
IBD
Hirschprung's Disease
Biliary Atresia

**Bristol Stool Chart**

<table>
<thead>
<tr>
<th>Type</th>
<th>Description</th>
</tr>
</thead>
<tbody>
<tr>
<td>1</td>
<td>Separate hard lumps, like nuts (hard to pass)</td>
</tr>
<tr>
<td>2</td>
<td>Sausage-shaped but lumpy</td>
</tr>
<tr>
<td>3</td>
<td>Like a sausage but with cracks on its surface</td>
</tr>
<tr>
<td>4</td>
<td>Like a sausage or snake, smooth and soft</td>
</tr>
<tr>
<td>5</td>
<td>Soft blobs with clear-cut edges (passed easily)</td>
</tr>
<tr>
<td>6</td>
<td>Fluffy pieces with ragged edges, a mushy stool</td>
</tr>
<tr>
<td>7</td>
<td>Watery, no solid pieces. Entirely Liquid</td>
</tr>
</tbody>
</table>
Approach to Diarrhoea

- **History**
  - Careful feeding history
  - Frequency; number; character (blood etc) in stool
  - Area of cholera
  - Usage of drugs or antibiotics

- **Examination**
  Look for:
  - Signs of dehydration (sunken eyes, lethargy, skin pinch, thirst)
  - Signs of malnutrition
  - Abdominal mass or distention

- **Assess dehydration state**
  - Either severe, some or no dehydration (IMCI classifications)

1. **Severe dehydration**
   Diagnosis: 2x signs
   - Lethargy or unconscious
   - Sunken eyes
   - Skin pinch slow return
   - Not able to drink
   Treatment:
   - Start IV fluids (Ringers Lactate) and Oral rehydration solution (ORS) if child can drink
   - Administer 100ml/kg bolus as:
     - If 12 months and greater 30ml/kg in 30mins and 70ml/kg over 2 1/2 hours.
     - If below 12 months 30ml/kg over 1 hour and 70ml/kg over 5 hours
   - Initially reassess child every 15-30 mins then extend to hourly if child improving.

2. **Some Dehydration**
   Diagnosis: 2x signs
   - Restlessness/irritability
   - Sunken eyes
   - Thirsty and drinks eagerly
   - Skin pinch goes back slow
   Treatment:
   - ORS given in first 4 hours according to age or weight: <6kg = 300ml 6-10kg = 600ml >10-12kg = 800ml 12-19kg = 1200ml
   - After 4 hours reassess child and classify dehydration. If still dehydrated repeat treatment for another 4 hours
   - Begin refeeding or breastfeeding.

3. **No dehydration**
   Diagnosis: Does not have any signs as above
   Treatment: Treat child as outpatient, Counsel mom on home treatment ie. Extra fluid, Zn supplements, continue feeds and to follow up or to return if worsens.
Most common causes of diarrhea are:
1) Bacteria – Shigella, Salmonella, E. coli, Campylobacter
2) Parasites – Entamoeba histolytica, Giardia lamblia and Cryptosporidium
3) Viruses – Rotavirus, Echovirus etc

- Persistent diarrhea
  - Lasting for 14 days or more
  - Imperative to find a cause via stool sampling
  - If blood in stool or stool sample discloses presence of give oral antibiotics as per shigella, amoebiasis and giardia (Metronidazole 7.5mg/kg 3xday for 5days)

GORD

- Features
  - Risk factors: Preterm infants, Cerebral palsy, Congential oesophageal problems
  - Most often is mild with no treatment required but in a minority symptoms can be severe and complications such as failure to thrive, oesophagitis and aspiration pneumonia can occur. Coughing, bronchospasms and exacerbations of chronic lung diseases can occur.
  - Main symptom is recurrent regurgitation of vomiting
  - Oesophagitis can present as irritability, pain after feeding, blood in vomitus or even iron deficiency anemia.
- Diagnosis:
  - Normally clinical diagnosis
  - Barium studies and endoscopy
- Treatment:
  - Prokinetic drugs can be used but main line is H2 antagonists like Cimetidine

Pyloric Stenosis ⇒ Projectile Vomiting
- Hypertrophy of smooth muscle surrounding the pylorus
- More common in boys and is due to multifactorial inheritance patterns
- Causes a metabolic alkalosis
  - Features
  - Presents with persistent projectile vomiting which is non bilious in content
  - Normally starts to present at 2-6 weeks
  - Infant will be hungry and will want to feed even after vomiting
  - Weight loss, dehydration, and constipation occurs after a few days
- Diagnosis
  - Can be clinically diagnosed via palpation of pylorus during test feeding and peristaltic waves may be present.
  - Electrolyte disturbances will be present ie. Hypochloremic metabolic alkalosis
  - Ultrasound can show and confirm the diagnosis
- Treatment
Definitive treatment is surgery to divide the hypertrophic musculature. (Ramstedt’s procedure)

**Wilsons Disease**
- Due to autosomal recessive defect in copper metabolism with slow accumulation of copper with deposition in tissues.
  - Presentation:
    - **Liver**: cirrhosis, chronic active hepatitis, acute hepatitis, fulminating liver failure, there is low risk of HCC
    - **Eyes**: Kayser-Fleischer rings (copper in Descemet’s membranes) - more common in patients with
    - **CNS**: basal ganglia (wing flapping tremor, Parkinsonism), cerebellum (dysarthria, dysphagia, incoordination, ataxia), cerebrum (psychosis, affective disorder)
    - **Kidneys**: Fanconi’s syndrome (proximal tubule transport defects) and stones
    - **Blood**: intravascular hemolysis - may be initial presentation
    - **Joints**: arthritis, bone demineralization, calcifications
  - Diagnosis
    - suspect if increase liver function test (LFT’s) with clinical manifestations
    - Requires 2 of the following 3:
      1) Reduced total serum copper (ceruloplasmin)
      2) High liver copper on biopsy
      3) Kayser-Fleischer rings
  - Treatment
    - Chelators (penicillamine, trientine) – increased urinary excretion of copper
    - Zinc acetate – decreased absorption of copper in diet and enterohepatic circulation
    - Liver transplant in severe cases

**Meckel’s Diverticulum**
- Is a remnant of fetal vitello-intestinal duct and occurs in 2% of infants.
- is normally 2 inches longs 2 feet away from ileocecal valve (rule of 2)
  - Presentation
  - Majority are asymptomatic but can have peptic ulceration leading to rectal bleeding.
  - Treatment
    - Treatment is surgical removal of the meckel’s diverticulum

**Appendicitis**
  - Features
    - Central abdominal pain over umbilicus that slowly becomes localized in right inguinal fossa as the condition progresses
    - Anorexia with associated nausea and vomiting
- Signs of: Fever, Tachycardia, right inguinal fossa tenderness and guarding
  - Diagnosis
  - Usually on clinical grounds but if uncertain can use FBC and abdominal sonar
  - Treatment
  - Surgical removal of appendix: Depending on severity either done right away if condition worsening or patient can be stabilized and an elective surgery can be done 6 weeks later ie in case of phlegmon.

**Inflammatory Bowel Disease**

1) **Crohn's Disease**
- Affects any portion of the GIT from mouth to anus but distal ileum and colon are most commonly affected
- Cause is unknown but there is a genetic predisposition
  - The affected intestine is thickened

2) **Ulcerative Colitis**
- Chronic, Recurrent inflammatory disease involving the colon, normally starting in the rectum and extending proximally
  - The affected intestine is not normally thickened as disease is restricted to the mucosa

- **Features of IBD**
  - Cramp like lower abdominal pain
  - Bloody diarrhea
  - Weight loss and failure to thrive

- **Treatment**
  - Crohn's is usually managed with dietary measures and with systemic steroids for active relapses. Immunosuppressive therapies are also part but are a second line treatment strategy ie anti-TNF antibodies
  - Ulcerative colitis is treated with aminosalicylates and steroids are reserved for active disease.

**Hirschprung's disease**
- This is due to a genetic disorder where the bowel is not innervated, there is no ganglion cells in the myenteric and submucosal plexuses for variable segments of the bowel normally extending from the anus to the colon.
- The aganglionic portion of the bowel is often contracted and narrow.
- It is more common in males
  - **Features**
    - Delayed passage of meconium
    - Intestinal obstruction with abdominal distention
    - Severe constipation since birth
  - **Diagnosis**
    - Unprepared barium enema may show a narrowed area or a transition area between normal and aganglionic bowel.
Gold standard is Biopsy of rectum to show absence of ganglionic cells on histology
• Treatment
Resection of the sections of bowel involved

**Biliary Atresia**
- Rare but treatable cause of obstruction to biliary flow and thus neonatal jaundice
  • Features
    - Jaundice from second day of life with predominantly conjugated hyperbilirubinemia
      with dark urine and pale stools
    - Malabsorption and bleeding tendencies (due to Vit K malabsorption)
    - Failure to thrive
      • Diagnosis
    - Abdominal or liver ultrasound can be used to confirm diagnosis
  • Treatment
Surgical reanastomosis (Kasai procedure) carried out before 6 weeks of life
EXAMINATION DISORDERS OF THE:
→ EAR
→ NOSE
→ PARA-NASAL SINUSES
→ THROAT
→ MOUTH

sore throat
Examination of the ENT system
Disorders of the:
- Ear
- Nose
- Para-nasal sinuses
- Throat
- Mouth
Disorders of the ENT System

Examination of the ENT System

**EAR**
- Look at the pinna, noting its shape, size, deformity.
- Look behind the ears for any scars and see if there is a hearing aid. Removal of the hearing aid is mandatory.
- Gently pull the pinna and ask if it is painful.
- Look at the size of the meatus and if there is any discharge.
- Use the otoscope. Gently pull the pinna upwards and backwards to straighten the ear canal. Introduce the ear piece and inspect the skin for any abnormalities. Look at the amount and consistency of the wax.
- Visualize the tympanic membrane: You should be able to see a cone of light as the concave surface of the membrane reflects the light forwards. This is known as the light reflex. It should be on the same side as the ear that you are examining (i.e.) right ear: light reflex on right side. The membrane is a pearly grey colour and is translucent. Notice any perforations, discharges or bulges.

**Examination technique:**

![The normal eardrum.](image)

- Testing the hearing using the whispered voice test, the Weber's test and the Rinne's test. This may be difficult in children. Referral to an audiologist may be necessary.

**NOSE**
- Look at the external surface and appearance of the nose. Note any skin disease or deformities.
- Deviation of the nose is best appreciated by standing behind the patient and looking down the nose from above while asking the patient to look upwards.
- Note any peri-orbital swelling or conjunctivitis.
- Have a good light source. A head mirror or head-light allows both hands free to manipulate instruments.
- To see the anterior nares clearly, press on the tip of the nose to elevate it. You will see the nasal vestibule (with hair), the anterior end of the septum and the anterior end of the inferior turbinates.
- Look at the septum in the midline. Look for any deviations, bleeding points, clots, crusting or perforations.
- On the lateral walls, inspect the anterior end of the inferior turbinates. Look at the colour and size of the turbinates. In rhinitis, the mucosa is red and swollen. A pale grey moist swelling may be a polyp.
- Look for any nasal discharge. Note the colour, consistency, amount and smell.

![Nasal polyp](image)

**Palpation**
- Feel the nasal bones gently to distinguish bony from cartilaginous deformity.
- Feel for any facial swelling and note tenderness (Feel over the frontal, maxillary and ethmoidal sinuses).
- Block each nostril in turn and ask the patient to breathe in. Assess nasal obstruction.
A more detailed examination of the nasal cavity extending back to the nasopharynx can be performed using a fine-bore endoscopy. This is not routinely done in children.

**THROAT**

Have a good light source. A head mirror or head-light allows both hands free to manipulate instruments.

**Inspection**

- Observe the child’s demeanor and face for obvious abnormalities. These include skin abnormalities, scars, lumps, deformities, asymmetry or signs of trauma.
- Look at the lips. Then ask the child to open his/her mouth. Inspect the buccal mucosa, gums and teeth. Note the child’s dental hygiene and look for any gingivitis.
- Evert the child’s lips. Look for areas of discoloration, inflammation, ulceration or nodules.
- Look at the hard palate, uvula and tongue. Are there any deformities?
- Use a tongue depressor to inspect the child’s throat.
- Look at the nasopharynx. Notice the colour. Red = pharyngitis. Look at the tonsils. Are they inflamed? Do they have pus or tonsiloliths? Is there any post nasal drip? Are there any abnormalities such as membranes or swollen masses?
- Examine the neck by standing behind the child. Palpate the posterior triangle and the midline structures. Then palpate the structures in the anterior triangle.

**Midline structures:**

- Submental triangle
- Hyoid bone (C3)
- Thyroid membrane
- Thyroid cartilage (C4)
- Cricothyroid membrane
- Cricoid cartilage (C6)
- Isthmus of the thyroid gland and tracheal rings
- Infrathyroid (strap) muscles
  - sternothyroid, sternothyroid, thyrohyoid, omohyoid
- Anterior jugular vein
- Suprasternal notch (T2)

**Anterior triangle borders:**

Lateral: anterior border of SCM
Medial: midline structures
Superior: inferior border of the mandible
Apex: jugular notch

**Posterior triangle borders:**

Anterior: posterior border of SCM
Posterior: anterior border of trapezius
Base: middle third of the clavicle
Apex: superior nuchal line

**Abnormalities:**

- Most common cause of a lump in the neck is an enlarged lymph node and the source may be infection in the head, neck, chest or abdomen.
- Tenderness of the node indicates inflammation. Swollen jugulo-digartic lymph nodes indicate pharyngitis associated conditions.
- Mobile nodules are not ominous. Immobile, fixed nodes are.
- Never aspirate a pulsating mass.

**Tips:**

- Allow mother to co-operate during this examination.
- Allow the child to sit on her lap.
- Ask her to hold the child’s head (forehead) and ask her to turn the child’s neck, while you examine the child’s ear.
- Leave the examination of the ENT system (especially the ear) for the end.
- Use the IMCI guidelines during the consultation as this guide will help you to focus on specific aspects so that important points of the exam are not missed.
- Tongue depressors are not always necessary as they can open their mouths wide enough for a good view.
- Children’s sinuses develop at different stages. Neonates already have ethmoidal and maxillary sinuses. The sphenoid sinus develops at the age of 3 and the frontal sinus only develops at the age of 12/13 years.
Common clinical problems that occur in Paediatrics:

Section One: Disorders of the EAR

Otalgia: Pain in or around the ear.

<table>
<thead>
<tr>
<th>EAR</th>
<th>NOSE</th>
<th>THROAT</th>
</tr>
</thead>
<tbody>
<tr>
<td>Otalgia (earache)</td>
<td>Blocked nose</td>
<td>Painful throat</td>
</tr>
<tr>
<td>Otorrhoea (discharge of the ear)</td>
<td>Nasal discharge</td>
<td>Dysphagia (painful swallowing)</td>
</tr>
<tr>
<td>Hearing impairment</td>
<td>Epistaxis (bleeding from the nose)</td>
<td>Noisy breathing and stridor</td>
</tr>
</tbody>
</table>

Causes of Ear-ache:

**Table 30.1**

<table>
<thead>
<tr>
<th>Diagnosis</th>
<th>Clinical sign</th>
<th>Treatment</th>
</tr>
</thead>
<tbody>
<tr>
<td>Perichondritis</td>
<td>Inflammation of the pinna</td>
<td>Antibiotics</td>
</tr>
<tr>
<td>Furunculosis</td>
<td>Very tender swelling at the entrance of the ear canal</td>
<td>Antibiotics and antibiotics + incision + drainage + flucloxacillin 12-25 mg/kg/dose q.i.d 5 days</td>
</tr>
<tr>
<td>Swimmer's ear</td>
<td>White deposits in the auditory canal</td>
<td>Antibiotics</td>
</tr>
<tr>
<td>Otitis Media</td>
<td>Inflammation of the tympanic membrane and drum</td>
<td>Antibiotics and antibiotics + incision + drainage + flucloxacillin 12-25 mg/kg/dose q.i.d 5 days</td>
</tr>
<tr>
<td>Constipated ear</td>
<td>Inflammation of the tympanic membrane</td>
<td>Antibiotics and antibiotics + incision + drainage + flucloxacillin 12-25 mg/kg/dose q.i.d 5 days</td>
</tr>
</tbody>
</table>

**Perichondritis**
- Inflammation of the cartilage of the auricle
- Two types: infective (caused by *P. aeruginosa*) and autoimmune
- Clinical presentation: acute pain + swelling of the auricle
- Rx: IV Piperacillin (30mg/kg/day) + Amikacin (15mg/kg/day) + Metronidazole (20mg/kg/day) for 48 hours.

- If swelling subsides, continue Rx for 72h. If not, stop antibiotics and treat the relapsing polychondritis with Prednisone 10mg/kg/day for 10 days.
- Complications if not treated: cartilage necrosis + deformity

**Furunculosis**
- A boil (or Furuncle) is a deep infective folliculitis (infection of the hair follicle)
- It is almost always caused by infection by *Staph aureus*.
- Clinical presentation: painful swollen area on the skin caused by an accumulation of pus and dead tissue. Pyrexia may occur.
- Commonly found inferiorly at the entrance of the external ear canal
- Rx: incision + drainage + antibiotics if cellulitis around the ear: flucloxacillin 12-25 mg/kg/dose q.i.d 5 days.

**Otitis Externa**
- Inflammation of the external ear that can be either diffuse or furuncular
- Diffuse: due to a mixed infection involving Staphs, Streps, Proteus spp, *P. aeruginosa* and E.coli
- Infection due to - mixed infections
  - allergic dermatitis (caused by soaps or shampoo)
  - swimming pool chemicals
  - trauma caused by scratching (ear buds!)
- Furuncular: mentioned above

**Treatment:**
- General measures: exclude any chronic underlying otitis media
  - Thorough cleansing of the ear
  - Keep ear clean and dry afterwards
  - Do not leave pieces of cotton wool in the ear
  - Do not instill anything into the ear unless prescribed.
- Drug treatment: Diffuse
  - Does not usually require anti-biotics.
  - Make a wick where possible, using ribbon gauze with 2% acetic acid on alcohol, instilled in ear every 6 hours for 5 days. Instill 3-4 drops after cleaning and drying the ear.
  - Refer if there is no response to the treatment.
Keratosis obturans

- Mass of desquamated squamous epithelium filling the ear canal.
- Mass has a pearly white surface with underlying wax. Tympanic membrane is intact.
- Clinical presentation: pain and mild hearing loss.
- Dx: manual removal under microscopic vision using a Jobson Horne probe + cup forceps + suction. This is done under local or general anesthesia. It is NOT removed by ear syringing. After removal, apply 1cm thick ribbon gauze with Kenacomb ointment to external canal for 48 hours. Thereafter, apply Locacorten-Vioform daily for 5 days.

Haematoma of the auricle

- Follows blunt trauma. Bleeding between perichondrium and cartilage.
- Complications if not treated: The perichondrium gets pushed away from the cartilage and the cartilage becomes necrosed. Auricle then becomes deformed. ('Cauliflower ear' occurs)
- Rx: incision + drainage + placement of a corrugated drain + pressure dressing (with wet cotton wool) for 48 hours. Allows cartilage and perichondrium to be pressed back together.

Foreign body (FB)

- COMMON in children.
- Beads, stones, cotton bud tips, peas, beans, tissue paper.
- Rx: removal. Technique depends on the position of the object. If in the:
  - outer cartilage portion of the ear: Use forceps or a Jobson Horne probe. Forceps: tissue paper and tips of cotton buds only.
  - JH probe: everything else. Pass the loop end of the probe medial to the FB through the gap between the BF and the ear canal and the lever it outwards.
  - deep bony portion: Syringe the ear. If unsuccessful, refer to the ENT specialist.

NB: Organic foreign bodies (beans, peas) are considered an emergency because they swell in the ear canal. Manual removal with the JH probe + microscope under general anesthesia is the best method of management.

Acute Otitis Media

- Infection of the middle ear caused by an URTI. Infective organisms get into the middle ear via the Eustachian tube. The organisms cause inflammation and swelling which causes the Eustachian tube to become blocked. Organisms the remain within the middle ear and multiply. Typically there is pain and a build up of pus within the middle ear. This causes the eardrum to bulge. The ear drum may burst if the infection is not properly treated.
- Common causative organisms: H. influenzae, Strep pneumoniae or Staphylococcus.
- There are FOUR stages of Acute Otitis Media

<table>
<thead>
<tr>
<th>Stage</th>
<th>Clinical presentation</th>
</tr>
</thead>
<tbody>
<tr>
<td>1 Stage of tubal occlusion</td>
<td>Blocked ear and autophonia (echoing of one's own voice). Retracted ear drum. Absent light reflex of ear drum. Middle ear effusion: air bubbles seen in the middle ear.</td>
</tr>
<tr>
<td>2 Stage of presuppuration</td>
<td>Infected and increased middle ear effusion. Bulging and inflamed tympanic membrane. Throbbing pain in ear, fever and vomiting.</td>
</tr>
<tr>
<td>3 Stage of suppuration</td>
<td>Rupture of tympanic membrane due to an increase in pressure within the middle ear. Perforated tympanic membrane with pus in the ear canal. Pain subsides after eardrum has popped.</td>
</tr>
<tr>
<td>4 Stage of resolution</td>
<td>Perforation heals in 80% of cases (within one week).</td>
</tr>
</tbody>
</table>

Rx: Amoxicillin 40mg/kg/day t.d.s for five days + Paracetamol. Augmentin/Cefuroxime in resistant cases.

Chronic Otitis Media

- Persistent otorrhoea and ear drum perforation
- Two types: Non-cholesteatomatous and cholesteatomatous
Non-cholesteatomatous type

Rx: dry the ear to allow for spontaneous healing of the ear drum. This treatment is divided into 4 steps:

Step 1:
Amoxicillin or Augmentin
Aural toilet twice daily with cotton wool sticks (not the commercial ones)
Ear drops: 0.5% phenol drops after aural toilet
Duration: ONE MONTH
No response: move onto step 2.

Step 2:
Remove predisposing factors: tonsillectomy/adenoidectomy.
Continue with aural toilet and ear drops
No response: move onto step 3.

Step 3:
IV-antibiotics - Ampicillin 50mg/kg/day + Metronidazole 20mg/kg/day for 48 hours. If otorrhoea subsides, continue this treatment for 72 hours. If not, to MC&S and treat with the appropriate antibiotic.
No response: move onto step 4. 90% of cases will resolve if steps 1-3 are used.

Step 4:
Surgery. Mastoidectomy.

- Cholesteatomatous type:
  - In normal circumstances, squamous epithelium is only present in the outer layer of the ear drum and in the skin of the outer ear canal. A cholesteatoma forms when this epithelium enters the middle ear. Blockage of the Eustachian tube causes a negative pressure to accumulate within the middle ear. This causes the epithelium on the ear drum to move into the middle ear and to accumulate.
  - Cholesteatoma formation is serious because it favors bacteria proliferation and it releases proteolytic enzymes that destroy the bone around the middle ear.
  - These 2 factors result in a spread of infection within and outside the temporal bone, resulting in postauricular subperiosteal abscess formation, facial palsy, labyrinthitis or intracranial lesions.
  - Dx: white cheese like material commonly found in the attic + polyp in the ear canal can be seen
  - Rx: Excision via a modified radical mastoidectomy

Complications of OTITIS MEDIA: Extra and Intra cranial

<table>
<thead>
<tr>
<th>Table 30.3: Complications of Otitis Media</th>
</tr>
</thead>
<tbody>
<tr>
<td><strong>Complications</strong></td>
</tr>
<tr>
<td><strong>Intra cranial complications</strong></td>
</tr>
<tr>
<td><strong>Postauricular abscess</strong></td>
</tr>
<tr>
<td><strong>Facial palsy</strong></td>
</tr>
<tr>
<td><strong>Labyrinthitis</strong></td>
</tr>
<tr>
<td><strong>Hemidural abscess</strong></td>
</tr>
<tr>
<td><strong>Petrous apicitis</strong></td>
</tr>
</tbody>
</table>

| **Intracranial complications** | **Presentation** |
| **Subdural empyema** | **Silent or sudden death and premonia** |
| **Subarachnoid haemorrhage** | **Headache, delirium, hemiparesis** |
| **Cerebritis** | **Dizziness and unstable gait** |
| **Cortical sinus thromboses** | **Headache, paresthesia, and rigors, pain over the anterior border of the sternomastoid muscle** |
| **Meningitis** | **Severe headache, paresthesia, and neck stiffness** |
| **Epicranial abscess** | **Headache, nausea, and vomiting** |

Treatment of the complications:

**Extra cranial**: Ampicillin 150 mg/kg/day + Metronidazole 20 mg/kg/day. Mastoidectomy ASAP, preferably within 12 hours of presentation.

**Intra cranial**: IV antibiotics + mastoidectomy + surgical drainage within 12 hours of presentation.

Intra cranial complications must be suspected when patients present with SEVERE HEADACHES, NUCHAL RIGIDITY OR NEUROLOGICAL FALL OUT. Painless discharge.

**TB Mastoiditis**:

- Found in children <10 years
- Clinical presentation: painless and profuse otorrhoea, multiple tympanic membrane perforations, pale granulation tissue, lower motor neuron facial palsy, disproportionate hearing loss, bone necrosis and suquestrum formation. Pulmonary TB with pre-auricular lymphadenopathy.
- Dx: Biopsy - histology of granulation tissue from middle ear or mastoid cavity.
- Rx: TB for 6 months. Surgery is to obtain tissue sample and to remove the abscess / sequestra.
Otorrhoea: discharge of the ear

Common causes:

<table>
<thead>
<tr>
<th>Diagnosis</th>
<th>Type</th>
<th>Rx</th>
<th>Refer when?</th>
</tr>
</thead>
<tbody>
<tr>
<td>Chronic Otitis media</td>
<td>Muco-purulent or purulent</td>
<td>Refer to notes above</td>
<td>Complications or those unresponsive to Rx</td>
</tr>
<tr>
<td>CSF otorrhoea</td>
<td>Clear: (+) glucose on dipstix</td>
<td>ENT specialist referral</td>
<td>Refer ALL patients</td>
</tr>
</tbody>
</table>

Hearing Impairment:

Normal hearing is needed for the development of speech and language. Mild hearing impairment causes a delay in speech development and distorts the quality of speech. In severe sensor-neuronal hearing loss, speech will not develop.

There is a limited time period for the development of speech: the 1st 2 years of life. If the child is not speaking by the age of 5, it is unlikely that the child will develop intelligible speech thereafter.

Hearing loss must be detected as soon as possible, preferably before 12 months. High risk babies must be screened.

High Risk babies:
- family history of deafness
- intra-uterine rubella infection
- low birth weight
- congenital deformities
- neonatal jaundice
- meningitis
- low AFGAR score

Summary:

Table 30.4 Hearing impairment

<table>
<thead>
<tr>
<th>Type</th>
<th>Presentation</th>
<th>Diagnosis</th>
<th>Treatment</th>
<th>Referral criteria</th>
</tr>
</thead>
<tbody>
<tr>
<td>Mild conductive HL</td>
<td>Past performance at school and home, turns to volume or radio or TV</td>
<td>Conductive HL</td>
<td>Syringe</td>
<td>Hearing loss persist for many months or patient or child not responsive to Rx</td>
</tr>
<tr>
<td>CSOM</td>
<td></td>
<td>CSOM</td>
<td>Perforated tympanic membrane (PTM)</td>
<td>Refer to specialist</td>
</tr>
<tr>
<td>SOM</td>
<td></td>
<td>SOM</td>
<td>Secretory Otitis media</td>
<td>Refer to ENT specialist</td>
</tr>
<tr>
<td>CSOM</td>
<td></td>
<td>SOM</td>
<td>Antibiotics and anti-microbial</td>
<td>Treat as failure</td>
</tr>
</tbody>
</table>

Table 30.5 Hearing impairment in infancy and childhood

<table>
<thead>
<tr>
<th>Type</th>
<th>Presentation</th>
<th>Diagnosis</th>
<th>Treatment</th>
<th>Referral criteria</th>
</tr>
</thead>
<tbody>
<tr>
<td>Mild conductive HL</td>
<td>Past performance at school and home, turns to volume or radio or TV</td>
<td>Conductive HL</td>
<td>Syringe</td>
<td>Hearing loss persist for many months or patient or child not responsive to Rx</td>
</tr>
<tr>
<td>CSOM</td>
<td></td>
<td>CSOM</td>
<td>Perforated tympanic membrane (PTM)</td>
<td>Refer to specialist</td>
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<tr>
<td>CSOM</td>
<td></td>
<td>SOM</td>
<td>Antibiotics and anti-microbial</td>
<td>Treat as failure</td>
</tr>
</tbody>
</table>

Approach to hearing loss:

- Som = secretory Otitis media
- CSOM = chronic secretory Otitis media
Section Two: Disorders of the NOSE

Choanal Atresia

- Most common congenital malformation of the nose
- The narrowing is due to the persistence of the bucco-nasal membrane
- Atresia can be membranous, cartilaginous or bony. It may be unilateral but it is commonly bilateral.
- Clinical presentation – blue babies (due to asphyxia and cyanosis) that become pink when they cry. This is because they are unable to breathe through their noses. When they cry, air is drawn into their lungs.
- Dx: unable to pass a nasogastric tube through the nose (posterior choanae are closed. Conformed using contrast radiography or CT scan.
- Rx: place an oral air way and secure it with tape. Then, surgical correction ASAP. (To establish oral feeding)
- Unilateral atresia presents later with a blocked nose and a discharge. It is surgically corrected thereafter.

Bi-lateral choanal atresia:

Acute viral rhinitis

- Very common.
- Infection of the upper airway due to a viral infection conveyed by contact with air-borne droplets
- Complications if not treated: secondary bacterial infection (spreads via the lymphatic system) -> Otitis media, sinusitis, bronchitis or pneumonia.
- Rx: oral Allergex (anti-histamine) + multivitamins + nasal decongestant. ONLY ass antibiotics (amoxicillin emulsion) if there are signs of a bacterial infection
- Signs of a bacterial infection: yellow-green nasal discharge + post nasal drip + coughing + fever + noisy breathing.

Chronic recurrent viral rhinitis

- Common in children who attend daycare centers/ crèches. This causes recurrent contamination and re-infection (chronic nature)
- Causes: rhinovirus or adenovirus
- Complications if not treated: secondary bacterial infection (spreads via the lymphatic system) -> Otitis media, sinusitis, bronchitis or pneumonia.
- Rx: oral Allergex (anti-histamine) + multivitamins + nasal decongestant. ONLY ass antibiotics (amoxicillin emulsion) if there are signs of a bacterial infection

Mouth breathing and snoring

- Due to enlarged ADENOIDs
- Dx: history of restlessness during sleep, snoring, mouth breathing, sleep apnoea. Confirmed using a lateral X-ray.
- Complications if not treated: HYPOXIA -> pulmonary hypertension -> right ventricular hypertrophy -> cardiac failure.
- Rx: Adenoidectomy ASAP

Foreign Body (FB)

- TYPICAL presentation: offensive, unilateral, purulent nasal discharge.
- Examples of FBs: sponge, tissue paper, beads, stones, crayons
- Complication if not treated: dislodged in posterior space and aspirated into lung -> Airway obstruction or lung infection
- Rx: Sponge/ tissue paper -> remove using forceps
- Rest -> use a Jobson Horne probe to remove
Nasal Trauma

• Clinical Presentation: History of trauma + swollen nose with epistaxis
• Rx: Septum Haematoma (septum bulges and completely occludes nasal canal) -> Incise muco-perichondrium on the fluctuant side under local anaesthesia + drain the Haematoma + pack the nose with 2.5cm thick ribbon gauze with BIPP (bismuth, iodoform paraffin paste). Oral Erythromycin 10-15mg/kg/dose q.i.d for 5 days + refer to the ENT to repair the cosmetic deformity.
• Complication if not treated correctly: cartilage necrosis and nasal collapse (saddle nose deformity)

Clinical picture: septum hematoma

Furunculosis

• A boil (or Furuncle) is a deep infective folliculitis (infection of the hair follicle)
• Develops on the vestibule (hair bearing region within the nose)
• Clinical presentation: painful swelling of the tip of the nose. Tip of nose is red and inflamed
• Rx: puncture (hypodermic needle) + oral Erythromycin 10-15mg/kg/dose q.i.d for 5 days
• Prevent recurrence: apply antiseptic cream to the vestibule daily for one month.

Epistaxis: the bleeding nose

• Common on children. Isolated nosebleeds do not need medical attention. Recurrent bleeds do.
• Bleeding is from Little's area (caudal end of nasal septum where the arteries of the nose anastomose)
• Minimal blood loss = <10ml. Severe blood loss is uncommon in children.
• Approach = blood tests + first aid treatment + definitive treatment.

Blood tests: to rule out haemolytic disorders (thrombocytopenia or leukaemia). (i.e.) FBC + Platelets + Pro-thrombin index + Partial Thromboplastin time

First aid treatment: Explain to parents. Sit the child upright and bend his/her head forward. Apply pressure to Little's area (by compressing the nasal ala) until the bleeding stops. Apply ICE to the forehead or back of the neck. This causes a reflex vasoconstriction.

Definitive treatment: Cauterization of the blood vessels in Little's area.

Summary:

<table>
<thead>
<tr>
<th>Symptoms and signs</th>
<th>Diagnosis</th>
<th>Treatment</th>
<th>Referral criteria</th>
</tr>
</thead>
<tbody>
<tr>
<td>Bleeding + pressure through the nose at birth</td>
<td>Congenital aplasia</td>
<td>Have an ear, nose, and throat specialist examine the nose and refer to ENT as indicated.</td>
<td></td>
</tr>
<tr>
<td>Settings and nasal blockage with fever and upper respiratory tract symptoms</td>
<td>Acute suppurative sinusitis</td>
<td>Antibiotics and pain relief + refer to ENT if symptoms persist.</td>
<td></td>
</tr>
<tr>
<td>Infected nasal blockage worsening, with marked nasal congestion in font and back of neck</td>
<td>Acute otitis media</td>
<td>Maxilla or paranasal sinuses + refer to ENT.</td>
<td></td>
</tr>
<tr>
<td>Bleeding from the nose</td>
<td>Cystic sinus</td>
<td>Exclude haemorrhagic disorders; refer to ENT and maxillofacial surgeon.</td>
<td></td>
</tr>
<tr>
<td>Persistent  bloody discharge with mucous</td>
<td>Acute rhinorrhoea</td>
<td>Antihistamines + refer to ENT.</td>
<td></td>
</tr>
<tr>
<td>Persistent nasal discharge with mucus</td>
<td>Chronic rhinitis</td>
<td>Nasal polyps or hay fever + refer to ENT.</td>
<td></td>
</tr>
<tr>
<td>Persistent nasal discharge with mucus</td>
<td>Chronic sinusitis</td>
<td>Referral to ENT specialist.</td>
<td></td>
</tr>
</tbody>
</table>

Table 3.8 Nasal and sinus disorders
The Para-nasal Sinuses

Acute Sinusitis

- Ethmoidal and maxillary sinusitis are common
- Precipitated by viral rhinitis
- Other causes: swimming with a URTI
- Causative organisms: Pneumococ, Strep, Staphs and H. influenzae
- Clinical presentation: headache + pain over affected sinus = per orbital cellitis + nasal obstruction + percussion tenderness over sinus + purulent nasal discharge.
- Dx: clinical + confirmed on a CT-scan of the para-nasal sinuses
- Rx: abscess drainage + antibiotics OR antral lavage + oral amoxicillin

Complications of sinusitis: Orbital or Intra-cranial

Orbital:
- per orbital oedema
- lid abscess
- chemosis
- proptosis
- ophthalmoplegia
- blindness

Intra- cranial
- extra-dural abscess
- sub-dural abscess
- meningitis
- brain abscess
- cavernous sinus thrombosis

Chronic Sinusitis

- Non specific, purulent sinusitis that may follow single or repeated acute sinusitis or it may be associated with allergic rhinitis.
- Clinical features: purulent post nasal drip + chronic nasal obstruction + headache (frontal and peri-orbital) + anosmia (decreased smell)
- Dx: coronal CT identifies the exact area.
- Rx: CONSERVATIVE – Amoxicillin + Metronidazole + Anti-histamine for one month. Rule out allergies and swollen adenoids. Endoscopic sinus surgery is only considered after conservative treatment.

Tonsillitis

- Tonsils = aggregates of lymphoid tissue. 3 types: adenoids (found in the nasopharynx), the lingual tonsils (found at the base of the tongue) and the palatine tonsils (found in the oropharynx)
- They initially become inflamed by a virus and then get secondarily infected by bacteria. This is referred to as tonsillitis (bacterial inflammation of the tonsils – commonly the palatine tonsils)
- The most causative organism is Streptococcus pyogenes

Acute Tonsillitis:

Three stages of inflammation

1. Parenchymatous stage: the tonsils are uniformly inflamed + hyperemic
2. Follicular stage: the tonsils are uniformly inflamed but also have yellow exudates in the crypts
3. Membranous stage: the exudates coalesce to form a membrane over the tonsils

- Clinical presentation: painful throat + fever + pain on swallowing
- On examination: red inflamed tonsils with pustules + large tender jugulo-digastric lymph nodes.
-Rx:
  Benzathine penicillin, IM, STAT
  < 15kg = 30 000 units
  15-30kg = 60 000 units
  > 30kg = 1.2 million units
  If injection is refused: use Phenoxymethylpenicillin, oral, b.d for 10 days.
  > 11-35 kg = 250mg dose
  > 35 kg = 500mg dose
  > 55 kg = 500mg dose
  If allergic to penicillin, give Erthromycin, oral, 10-15mg/kg/dose 6 hourly.
Pain relief: Paracetamol, oral, 15mg/kg/dose, 6 hourly
Conservative Rx: homemade salt mouthwash, gargle for one minute, b.d. Advise on adequate hydration + avoid irritants (e.g.) vapo-rubs inserted into nostrils

- Complications of tonsillitis. Chronic tonsillitis, peri-tonsillar abscess or a para-pharyngeal abscess.

Chronic
- Follows follicular tonsillitis
- The micro abscess that formed becomes walled off by fibrous tissue
- This produces a chronic sore throat
- Rx: Tonsillectomy

Indications for a tonsillectomy:
- recurrent tonsillitis
- chronic tonsillitis
- tonsillolith**
- peri-tonsillar abscess
- chronic suppurative otitis media (blocked Eustachian tube)
- airway obstruction due to enlarged tonsils
- tonsilar tumour

** Tonsillolith: secretions from the tonsilar crypts which solidify. Looks like rice grain. Has an offensive odour (common cause of halitosis)

- Complications of a tonsillectomy:
  - Hemorrhage – primary within 24 hours and secondary within 10 days. Secondary is due to a secondary infection. Can be life threatening. Treat ASAP
  - Altered speech – settles after 2 weeks
  - Death – 1: 20,000

Peri-tonsillar abscess (Quinsy)
- Infection spreads from the tonsil to the peri-tonsillar space.
- Clinical presentation: Pain on swallowing + trismus + drooling
- On examination: bulging of the soft palate above the swollen tonsil
- Rx: depends on age of child

Older: incision + drainage under local anesthesia. Add procaine penicillin injections daily for 3 days and oral amoxicillin for 5 days.
Younger: admit + give IV penicillin G. Fluid balance is importance.

Para-pharyngeal abscess
- Management the same as a peri-tonsillar abscess

Retro-pharyngeal abscess
- Common in children under the age of 2 years
- Due to inflammation and breakdown of the retropharyngeal lymph nodes leading to unilateral swelling of the posterior pharyngeal wall
- Infection spreads to the adenoids
- Clinical presentation: fever, torticollis, drooling, swelling of the posterior wall of the oropharynx just lateral to the midline.
- Dx: clinical and lateral X-ray. Typical picture = enlarged pre-vertebral soft tissue shadow, greater than twice the width of the vertebral body.
- Rx: IV antibiotics + incision + drainage under general anesthesia.
**Aphthous ulcer**
- Rare
- Pinhead sized growth with a sloughing base and a erythromatous margin.
- Clinically: painful and recur
- Rx: oral hygiene (salt water mouth wash) + paracetamol (for pain) + Kenalog in a Orabase ( topical steroid)

**Recurrent parotitis**
- Parotid gland pain and swelling.
- Cause unknown (ruled out mumps)
- Unilateral but alternates sides
- Sialectasia is often present
- Rx: Paracetamol + lemon juice (sialogogue)
- Sialography performed if sialectasia is suspected.
- If no improvement: antibiotics
- Still no improvement: parotidectomy

**Tumours of the parotid gland**
- Mesothelial tumours (hemangiomas and lymphangiomas)
- Hard/persistent tumours can be glandular or epithelial origin and are in 50% of cases malignant
- Rx: refer

**Rannula**
- Retention cyst of the sublingual salivary gland
- Thin walled blue cyst found on the floor of the mouth on one side of the frenulum.

**Stridor**
- noisy respiration
- Due to an incomplete airway obstruction
- It can be inspiratory, expiratory or both
- Neonatal stridor is due to laryngomalacia, Sub-glottic stenosis, Vocal fold palsy, Laryngeal webs, Bifid epiglottis, Laryngeal cysts, Tumours, Micrognathia and Glossopteris.

**Summary**

<table>
<thead>
<tr>
<th>Conditions and disease</th>
<th>Diagnosis</th>
<th>Treatment</th>
<th>Referral criteria</th>
</tr>
</thead>
<tbody>
<tr>
<td>Acute salivary infection</td>
<td>Antibiotics</td>
<td>Antibiotics</td>
<td>Patients with complications</td>
</tr>
<tr>
<td>Sialectasia</td>
<td>Antibiotics</td>
<td>Antibiotics</td>
<td>Refer to radiologist</td>
</tr>
<tr>
<td>Parotiditis</td>
<td>Antibiotics</td>
<td>Antibiotics</td>
<td>Refer to radiologist</td>
</tr>
<tr>
<td>Sialectasia</td>
<td>Antibiotics</td>
<td>Antibiotics</td>
<td>Refer to radiologist</td>
</tr>
<tr>
<td>Neoplasms</td>
<td>Surgery</td>
<td>Surgery</td>
<td>Surgery</td>
</tr>
<tr>
<td>Neoplasms</td>
<td>Referral</td>
<td>Referral</td>
<td>Referral</td>
</tr>
<tr>
<td>Neoplasms</td>
<td>Referral</td>
<td>Referral</td>
<td>Referral</td>
</tr>
</tbody>
</table>
NEUROLOGICAL EVALUATION
DISORDERS OF THE SPINAL CORD
CNS INFECTIONS
CP
SEIZURES
THE FLOPPY INFANT SYNDROME
STROKE
ABNORMAL MOVEMENTS IN CHILDREN
HEADACHE
HYDROCEPHALUS
NEUROLOGICAL EVALUATION
DISORDERS OF THE SPINAL CORD
BRAIN TUMOURS
INFECTIONS OF THE CNS
CP
SEIZURES
THE FLOPPY INFANT SYNDROME
STROKE
ABNORMAL MOVEMENTS IN CHILDREN
HEADACHE
HYDROCEPHALUS
Neurological and muscular disorders

The neurological examination of a child differs according to various age groups.

1. The chronological age should be corrected for prematurity when a developmental assessment is done. E.g., 3 months should be subtracted from a premature baby's age when he was born 3 months too early.

NEUROLOGICAL EVALUATION OF A CHILD

1. HISTORY

2. CLINICAL EXAMINATION ACCORDING TO VARIOUS AGE GROUPS

<table>
<thead>
<tr>
<th>Age</th>
<th>Gross Motor</th>
<th>Fine Motor</th>
<th>Speech and language</th>
<th>Adaptive and social skills (psychosocial)</th>
</tr>
</thead>
<tbody>
<tr>
<td>Birth</td>
<td>Brain stem reflexes</td>
<td>Continuous movements</td>
<td>Very frequently &amp; well</td>
<td>Learning to focus</td>
</tr>
<tr>
<td>2 weeks</td>
<td>1 month</td>
<td>Follows with eyes to moving</td>
<td>Very frequently &amp; well</td>
<td>Learning to focus</td>
</tr>
<tr>
<td>6 weeks</td>
<td>prone-lie-in, social smile infrequently</td>
<td>Infrequent</td>
<td>Infrequent</td>
<td>Infrequent</td>
</tr>
<tr>
<td>8 weeks</td>
<td>prone-lie-in, social smile infrequently</td>
<td>Infrequent</td>
<td>Infrequent</td>
<td>Infrequent</td>
</tr>
</tbody>
</table>

1. History

1. Main Complaint
   - Reason for referral
   - Chronological exposition
   - Related complaints and symptoms

2. Family History
   - *Mother, father, grandparents, siblings

3. Pregnancy History
   - Age of mother
   - Antenatal risk factors
   - Prenatal
   - Abortion
   - Illnesses during pregnancy
   - Drugs taken

4. Birth History
   - Gestational age, type of delivery
   - APGAR scores, infant weight, height, skull circumference

5. Neonatal Period
   - Condition after delivery
   - Admission to neonatal unit
   - Course
   - Duration of hospitalization

6. Medical History
   - Nature of onset, duration
   - Complications
   - Medications
   - *Previous surgery
   - *Previous trauma

7. Family History

8. Behaviour

9. Progress in school

10. Developmental history
    - *Milestones

The table above outlines the neurological examination of a child according to various age groups, focusing on gross and fine motor skills, speech and language development, and adaptive and social skills. This evaluation helps in understanding the child's development and identifying any delays or concerns.
### 2. Examination

#### Objectives of Neurological examination

1. **General**
2. **Higher function**
3. **Head and face**
4. **Cranial nerves**
5. **Neck and back**
6. **Signs of raised intracranial pressure**
7. **Motor system**
8. **Sensory system**
9. **Basal ganglia**
10. **Cerebellar function**
11. **Autonomic function**
12. **Markers**
13. **Developmental assessment**

#### Examination

- **Joints**
  - **Inspection:** Gait, contour, swellings or effusions, skin, position, joint stability
  - **Palpation:** Tenderness, warmth, effusions, crepitations

- **Skeleton**
  - **Inspection:** Gait, posture, symmetry, body ratios, spinal column
  - **Palpation:** Tenderness, Rickety, rickets, cranial bones, broad wrists

#### Clinical Examination

1. **General:**
   - Level of consciousness
   - Weight, Height, Skull circumference
   - Dysmorphic features, Posture, Movement
2. **Higher function**
   - General behavior: Normal/Hyperactive/quiet/meatness
   - Mood: Appropriate/abnormal/flat
   - Thought content: Fears/Hallucinations
   - Intellectual ability: Multi/Language/Communication
   - Sensation: Consciousness/Attention span
   - Speech: Fluency
   - Dysphonia/Dysarthria/Dysphasia

#### Head Shapes

- **Plagiocephaly:** Head looks like a parallelogram
- **Scaphocephaly:** Head is long in AP diameter
- **Turricephaly:** Head is tall
- **Brachycephaly:** Back of head is wide

#### Modified Glasgow Coma Scale for Children

<table>
<thead>
<tr>
<th>Feature</th>
<th>Description</th>
</tr>
</thead>
<tbody>
<tr>
<td><strong>Opening of eyes</strong></td>
<td>(4) Spontaneous, (3) To verbal commands, (2) To pain stimulus, (1) No response</td>
</tr>
<tr>
<td><strong>Motor response</strong></td>
<td>(6) Oriented surroundings, (5) Withdrawal on pain stimulus, (4) Decorticate posture, (3) Decerebrate posture, (2) No response</td>
</tr>
<tr>
<td><strong>Best verbal response</strong></td>
<td>(6) Oriented, (5) Oriented-crying, (4) Unoriented, (3) Continuous crying, (2) Incoherent sounds, (1) No response</td>
</tr>
</tbody>
</table>

#### Clinical Features

- **Skull growth**
  - **Macrocephaly:** Skull circumference more than 2 standard deviations above mean for age and sex class
  - **Microcephaly:** Skull circumference less than 2 standard deviations below mean for age and sex class

---

*Note: The table and diagram are not fully transcribed due to the limitations of text extraction. Further reading and interpretation are recommended.*
4. Cranial nerves

**Visual acuity definition**

Distance from that point to the Snellen Chart.

Mesures:
- 20/200
- 20/2000
- 20/2000
- 20/1000
- 20/2000

A normal pre-natal condition is.

---

**Cranial nerves**

**Visual acuity definition**

Distance from that point to the Snellen Chart.

Mesures:
- 20/2000
- 20/2000
- 20/2000
- 20/1000
- 20/2000

A normal pre-natal condition is.

---

**Palpation:**
- Sutures, Frontanelles (closes # 6-18 months)
- Cranial tabs

**Auscultation:**
- For breaths

**Percussion:**
- MacEwen cracked pot sign

---

**Neck and back**

5. Neck and back

- Signs of meningial irritation:
  - Neck stiffness, Kernig, Brudzinski
  - Spina Bifida
  - Kyphosis
  - Lordosis
  - Scoliosis
  - Tenderness

---

6. Signs of raised intracranial pressure

1. Early morning headache, throbbing
2. Vomiting
3. Diplopia (VI)
4. Bulging frontanelle
5. Distasis of sutures
6. Setting sun sign

---

7. Motor system

**Distribution of power:**

- Upper limbs:
  - Proximal: Shoulder falls through
  - Can’t walk on hands like a wheelbarrow and can’t press against wall
- Distal: Can’t pull himself up when held by his hands

- Lower limbs:
  - Proximal: Power
  - Can’t climb stairs
  - Distal: Can’t stand on his toes

---

**Motor milestones**

- Can’t walk
- Broad or narrow-based
- Toe or flat walking
- Eversion of feet
- Limping, ataxia, waddling
- Hemiplegia
- Slapping = distal limb weakness

---

**Muscle bulk**

- Hypertrophy
Muscle tone
Power
Pseudohypertrophy
Atrophy
Hypertonia, hypotonia
Distribution of fall out
Range of movement of joints
Grading
Proprioception (4-5y)
Stereognosis (1mm on the tongue, 3mm on the finger tips, 8-12mm on the palm of the hand)
Cerebellar system
Fasciculations
Myotonia
Palpate
Tenderness (myositis, texture (doughy in polymyositis))
DEEP:
Grading
0: No movement
1: Flicker of movement
2: Active movement, Gravity excluded
3: Active movement against gravity
4: Movement against resistance
5: Normal
Choreo
dystonia
Dystonic movements of the face and limbs
9. Basal ganglion
10. Cerebellar function
WARNICHD
11. Autonomic system
Sphincter control
Blood pressure
Ability to sweat
Horner's syndrome
12. Markers
Dysmorphic features
Neurocutaneous markers (Hypo or hyperpigmentation)
Abnormal smells
Grass motor
Fine motor
Language
Psychological development
13. Developmental assessment
Disorders of the Spinal Cord
Always consider 4 pathways when examining a patient for a spinal cord lesion:
1. Descending corticospinal tracts (Pyramidal tracts Motor)
2. Descending spinothalamic tracts (Pain and temperature)
3. Descending spinocerebellar tracts (Coordination)
4. Posterior columns (Proprioception, vibration and pressure)

Spinal cord compression syndrome
Clinically presents with pyramidal tract (UMN signs) initially. As compression progresses, sensory loss occurs and then autonomic impairments. Blood supply to spinal tract: 1. One Anterior spinal artery supplying most of tracts 2. Two Posterior arteries supplying the posterior columns

ANTERIOR SPINAL ARTERY SYNDROME: Most compression lesions initially compress the anterior spinal artery
leading to ischaemia of the watershed areas of the spinal cord (Corticospinal tracts). That is why motor symptoms occur first with compressive spinal cord lesions.

Most common cause:
1. TB spondylitis (Anterior erosion of the adjacent vertebrae with anterior collapse and compression of the anterior spinal artery)
2. Spinal tumours
3. TB Abscess
4. Blurred granuloma
5. Trauma

TRANSVERSE MYELITIS: Characterised by a SUDDEN onset of PROGRESSIVE WEAKNESS of lower limbs, loss of BLADDER and BOWEL, shiver control and SENSORY loss. Often preceded by a RESPIRATORY tract infection. Initially FLACID weakness but gradually UMN signs with spasticity, Babinski reflex and increased deep tendon reflexes develops. Posterior column function spared. On biochemistry: Children have a pleocytosis and raised proteins in CSF.

SPINAL DYSPHASIA (NEURAL TUBE DEFECTS)
Caused by failure of primitive neural tube closure.
1. Spina Biliosa Occulta: Dorsal spine fails to close. Underlying meninges and neural tissue intact with no neurological deficit. Clinical clues includes a hump or a dimple on the skin over low lumbar-sacral spine
2. Meningocele: Dorsal spine is bony and the meninges of the cord extends as a cystic swelling over the defect. Underlying neural tissue of the cord usually not involved
3. Meningomyelocele: Neural tube is exposed and damaged with resultant neurological defects at the site of the lesion and at levels below. UMN signs at the level of the lesion (Flaccid paralysis, sensory loss, Dysesthesia of bladder and bowel) control. Refer for surgical management in the first 48 hours.

BRAIN TUMOURS
Second most common tumour in children after childhood leukemias. Usually presents with cerebellar ataxia, pyramidal tract signs and cranial nerve palsy as most of the tumours are found in the posterior fossa (Brainstem and cerebellum)

CLASSIFICATION OF BRAIN TUMOURS acc. to WHO’s classification according to location of the tumour:

SUPRATENTORIAL (More common in infants, esp. 1.)
- Cerebral hemispheres and meninges:
  - Astrocytoma *
  - Ependymoma
  - Meningioma
  - Malignant gliomas
  - Midline Structures:
    - Pituitary adenoma
    - Craniopharyngioma
    - Cystic cyst
- Meningoencephalitis
- Germ cell tumour
- Astrocytoma
- Optic nerve glioma

INFRAVENTRIAL (more common in childhood, esp. *)
- Cerebellum and 4th Ventricle
- Astrocytoma *
- Medulloblastoma
- Ependymoma
- Brainstem
- Astrocyma

RISK FACTORS ASSOCIATED WITH DEVELOPMENT OF BRAIN TUMOURS
- Genetic: Neurofibromatosis
- Listeriosis, Polio
- Von Hippel-Lindau disease
- Basal cell naevus syndrome
- Immunosuppression
- Renal transplant suppression
- Ataxia telangiectasia
- Environmental: Aromatic hydrocarbons
- Nitrates
- Trichloroethylene
- System hydrazines
- Background radiation
- Maternal consumption of alcohol

Clinical features:
Features of raised Intracranial pressure:
1. Headache and vomiting (especially morning)
2. Visual disturbances in supratentorial tumours
3. Ataxia and CN palsies common in posterior fossa tumours
4. Relative bradykardia and hypertension (Cushing’s reflex)
5. Change in level of affect
6. Supratentorial tumours may cause shift of cerebral tissue and herniation
7. Infratentorial tumours commonly presents with hydrocephalus due to obstruction of the 4th ventricle.

Diagnostic evaluation:
- Skull X-ray (Signs of Raised ICP and calcification)
- CT scan with contrast
- MRI for brainstem tumours
- Cerebral angiography is useful for vascular tumours or vascular malformations.

Management:
use Dexamethasone to control ICP
Infections of the CNS

BACTERIAL

Acute bacterial meningitis

<table>
<thead>
<tr>
<th>Age related meningitis</th>
<th>Cause</th>
</tr>
</thead>
<tbody>
<tr>
<td>Neonates (0-29 days)</td>
<td>1. E. coli</td>
</tr>
<tr>
<td></td>
<td>2. Group B Haemolytic streptococci</td>
</tr>
<tr>
<td></td>
<td>3. Listeria Monocytogenes</td>
</tr>
<tr>
<td></td>
<td>4. Micromonospora</td>
</tr>
<tr>
<td></td>
<td>5. Entacoccus</td>
</tr>
<tr>
<td>Infants (1 month - 2 years)</td>
<td>1. Group B Haemolytic streptococci</td>
</tr>
<tr>
<td></td>
<td>2. H. Influenza type B</td>
</tr>
<tr>
<td></td>
<td>3. S. Pneumoniae -</td>
</tr>
<tr>
<td></td>
<td>4. N. Meningitidis -</td>
</tr>
<tr>
<td></td>
<td>5. salmonella gravis</td>
</tr>
<tr>
<td>Childhood and adolescence</td>
<td>1. S. Pneumoniae</td>
</tr>
<tr>
<td></td>
<td>2. N. Meningitidis</td>
</tr>
<tr>
<td></td>
<td>3. H. Influenzae type B</td>
</tr>
</tbody>
</table>

Clinical features:

1. Common in all age groups:
   - Fever, headache, vomiting
   - Signs of raised ICP (neck stiffness, focal neurology)
   - Encephalopathy showing cortical involvement

2. Neonatal period: ([Age specified])
   - Inability
   - Leathery
   - Failure to thrive
   - Hypothermia
   - Poor feeding
   - Respiratory distress
   - Convulsions
   - Septicaemia

3. Older babies and children:

   - S. Pneumoniae: Gram pos. diplococci
   - Should be suspected in patients with meningitis associated with a feature of the skull, paranasal sinuses or frontal bones leading to CSF leak and otitis media. High risk in patients with sickle cell anaemia.

   - Characterized by a petechial or purpuric rash. Organism can be found in the rash and can be cultured after a scrape of the skin lesion.
   - Endotoxin of the organism can lead to shock, bilateral adrenal haemorrhages and DIC.

   - H. influenzae
When to perform a CT scan?

1. Prolonged or persistent coma
2. Persistent focal neurological signs
3. Respite after initial response
4. Hydrocephalus
5. Decreasing level of consciousness
6. Subdural effusion suspicion

In patients with suspected Meningitis, look for a history of fevers and do appropriate Mantoux (PPD) skin testing

Complications of Meningitis

1. Vasculitis
2. Brain oedema
3. SIADH
4. Convulsions
5. Subdural effusions
6. Brain abscess
7. CN palsies
8. Fever
9. Hydrocephalus
10. Deafness and blindness
11. Recurrent meningitis
12. Learning problems

Management:

Supportive management:

1. Fluid balance
2. Fluid restriction
3. Electrolytes
4. S-glycose
5. Blood pressure
6. Oxygen saturation
7. ICP

Management of bacterial meningitis in infants and children:

Initial Rx:

1. Antibiotics
2. Fluid balance
3. Corticosteroids
4. Phenobarbitone
5. Anticonvulsants
6. Oxygen therapy
7. ICU support
When organism identified:

- **Group B Streptococcus**
  - Parenteral or Amoxicillin alone or combined with Ampicillin
  - Duration: 14 days

- **Gram Neg Bacilli**
  - Parenteral alone or combined with Ampicillin
  - Duration: 14 days

Duration of therapy:

- Listeria and group B streptococci: 14 days
- Gram negative bacilli: 31 days

Prophylaxis:

- Children who are close contacts of patients with meningitis caused by **N. Meningitidis** will need prophylaxis.

Causes of acute bacterial meningitis:

1. Partially treated meningitis
2. **Streptococcus pneumoniae**
3. Hemolytic streptococci
4. Viral meningitis
5. Leukemia
6. Uncommon infections: Syphilis, Mycoplasma, Leptospirosis

Viral infections:

Some viruses have a predilection for specific areas in the brain:
- **Polio** - anterior horn cells
- **Vaccinia** - Cerebellum
- **HBV** - Temporal lobe
- **Rabies** - Cerebral

Encephalitis:

If results from invasion of the CNS by the virus, the inflammatory response may also be an indirect effect of the interaction between the host and the virus (post-infectious meningencephalitis or post-encephalitic encephalitis).

Common features include fever and disturbances in the areas of consciousness. Neurological manifestations are more common than with viral meningitis, it may affect any part of the CNS including:

- Meningoencephalitis
- Myeloneuroencephalitis
- Transverse myelitis
- Encephalitis

Causes include:

- HSV type 1 and 2
- **Mumps**
- **Varicella**
- Enteroviruses
- **Rabies**

Treatment includes Supportive care and Anti-viral.

HSV encephalitis:

Characterized by a neurotropism of specific herpes virus, which has a predilection for temporal, orbital areas and brainstem.

Prophylaxis:

- Children who are close contacts of patients with meningitis caused by **N. Meningitidis** will need prophylaxis:
  1. **Ceftriaxone** 250mg im (adults), 125mg im staf (children)
  2. **Ciprofloxacin** 500mg bd (adults only)

Causes of septic meningitis:

1. Partially treated meningitis
2. **Streptococcus pneumoniae**
3. **Staphylococcal**
4. Viral meningitis
5. Leukemia
6. Uncommon infections: Syphilis, Mycoplasma, Leptospirosis
Chronic encephalitis

1. HIV encephalopathy
   Clinical picture:
   1. Acute encephalopathy
   2. Slow neurodegeneration
   3. Loss of intellectual capacities
   4. Ataxia
   5. Myoclonus
   6. Seizures

2. SSPE (Subacute sclerosing panencephalitis)
   As a result of persistent (HTLV-1) infection usually 1-3 years after initial infection. Symptoms usually begin with a slow onset, with personality changes and intellectual deterioration. It then progresses to pyramidal and extrapyramidal signs in the form of dystonia and spasms. The disease presents in a few months to a progressive dementia often leading to death.

3. Progressive rubella panencephalitis

The disease manifests with progressive neurodegeneration, dementia, incontinence and it is a fatal condition.

4. HLV-1:
   It causes a progressive psychosis, panencephalitis and is mainly seen in adults.

5. Poliomyelitis

Parasitic Infections

1. Malaria:
   Symptoms are a complication of Plasmodium falciparum and manifest with intermittent fever, headache, dyspnea, agitation, delirium, vomiting and seizures.

2. Neurocysticercosis:
   Causes: Infection of brain by the tapeworm, Taeniasis solium. The human becomes the intermediate host. These bitches in the large intestine and penetrate the intestinal mucosa and lodge in various parts of the body, including the brain. Most commonly found in the ventricular part of the brain, particularly the gray matter. The cysts cause an inflammatory response in the brain, resulting in edema. As a result, seizures can occur 2-3 years after infection. When it occurs in the ventricular part, it may cause a hydrocephalus.

   Symptoms and signs:
   - Focal seizures are common but may also present with headache, focal neurological signs, and a raised ICP. A cyst can cause a ventricular and lead to obstructive hydrocephalus.

   Investigations:
   1. Eosinophilia
   2. CSF shows mild pleocytosis, elevated proteins, and albuminocytosis, which can be identified by an ELISA test.
   3. The diagnosis is confirmed with a CT scan revealing a solitary cyst or multiple cysts within the cerebral hemispheres, as well as patches of hypodensity with surrounding edema and single or multiple calcified lesions. This produces the cysts in various stages of development, the cysts are often thin-walled and surrounded by a layer of macrophages.

   Treatment:
   Albendazole and praziquantel for 8 days. Praziquantel 30 mg/kg for 10 days is the treatment of choice. Steroids and corticosteroids may be helpful.

   The disease presents with progressive neurodegeneration, dementia, incontinence and it is a fatal condition.
Fungal infections of the CNS

Cryptococcosis: MAE 10

The fungus found in soil and often in pigeon droppings. Primary infection occurs via the respiratory system, spreading via blood to the rest of the body, including the brain.

The organism causes a granulomatous arachnoiditis resembling cryptococcal meningoencephalitis, similar to that found in patients with AIDS. Headaches and meningitis are the main symptoms experienced. Other neurological signs that develop include:

- CN palsies
- Raised ICP with papilloedema
- Impairment of consciousness
- Signs of hypertension of hydrocephalus

Cryptococcosis may present as:

- Malaise
- Headache
- Disorientation

Diagnosis: Culture of the organism in the CSF will confirm the diagnosis and demonstration of antigen in the CSF by latex agglutination is reliable and sensitive. CSF may show raised protein, leukocytes, and elevated pressure. Intrathecal cryptococcal antibodies are absent in AIDS patients, but may be present in non-AIDS patients.

Treatment: Amphotericin B and flucytosine are used in combination. Amphotericin B may be given intravenously and flucytosine orally. A total of 150 mg/kg is given daily in divided doses.

Relapses are common and can still occur 2–3 years after completion of treatment.

Cerebral Palsy

Definition: CP is a result of an insult to the developing brain that produces a disorder of movement and posture that is permanent but not unchangeable.

Causes of CP

Before pregnancy

1. History of factors
2. Congenital malformations
3. Smoking
4. Genetic
5. Multiple gestation

During pregnancy

1. Low SES
2. Congenital malformations
3. Abnormal fetal presentations
4. Birth asphyxia
5. Anoxic brain injury
6. Fetal death
7. Infection
8. Hypertension
9. Stroke
10. Prematurity

Prenatal

1. Congenital malformations
2. Infections
3. Drug abuse
4. Maternal smoking
5. Maternal alcohol consumption
6. Maternal temperature
7. Obstetric complications
8. Maternal illness

Perinatal

1. Prematurity
2. Infection
3. Birth asphyxia
4. Hypoxia
5. Birth trauma
6. Metabolic disorder
7. Intracranial hemorrhage
8. Neonatal stroke

Postnatal

1. Head injury
2. Meningitis
3. Metabolic disorder
4. Spinal cord injury
5. Cord compression
6. Traumatic brain injury
Early signs of CP:
1. High pitch cry
2. Tonic bite
3. Spastic movement
4. Asymmetrical movement
5. Spontaneous clonus
6. Muscle developmental delays (MBK)
7. Abnormal movements
8. Thumbs adducted and fingers extended
9. Hand held closed at 4 months
10. Early visual and hearing impairment
11. Feeding difficulties
12. Abnormal protective reflexes; poor development in two arm/pedestal support at 18 months
13. Gait delay at 5 months
14. Functional hypotonia
15. Increased extensor tone in ventral suspension
16. Kyphoscoliosis in ventral suspension

Clinical presentation:
1. Usually have UMI signs
2. Deteriorate motor milestones
3. Increase tone
4. Contractures
5. Muscle mass and hypotonia
6. Pathological reflexes including a grasp response
7. Seizures may be hypotonic and with abnormal posture.

CLASSIFICATION

<table>
<thead>
<tr>
<th>According to Topography</th>
<th>According to Functionality</th>
</tr>
</thead>
<tbody>
<tr>
<td>1. Hemiplegic (One side of body)</td>
<td>1. Spastic</td>
</tr>
<tr>
<td>2. Diplegic (Legs only)</td>
<td>MILD - INDEPENDENT</td>
</tr>
<tr>
<td>3. Quadriplegic (All four limbs)</td>
<td>Moderate - Partial care</td>
</tr>
<tr>
<td>4. Monoplegic (One limb)</td>
<td>and may need orthotics</td>
</tr>
<tr>
<td>5. Triplegic (Limb)</td>
<td>Severe - Total and</td>
</tr>
<tr>
<td></td>
<td>Comprehensive care</td>
</tr>
<tr>
<td></td>
<td>needed.</td>
</tr>
</tbody>
</table>

NB: Component of CP classification:
1. Motor abnormalities
   a) Nature and typology of motor disorder
   b) Functional motor abilities (ESP, Ommotor and Speech)
2. Associated impairments (e.g. epilepsy)
3. Anatomical and radiological findings
   a) Anatomical distribution
   b) Radiological findings
4. Causation and timing

Comorbidities:
- Mental retardation
- Learning difficulties
- Speech problems
- Oral hypersensitivity and drooling
- Feeding problems
- Constipation
- Reflux
- Visual and hearing impairment
- Orthopaedic problems (fixed contractures of hemiplegia)
- Behavioural problems
- Epilepsy

Management:
- AMI (PREP FUNCTION, OPTIMIZE DEVELOPMENT AND PREVENT COMPLICATIONS!!!)
- The treatment involves a multidisciplinary team including OT / Physio / Speech / Dietician / OMM / Paediatrician and an Orthopedic surgeon.
- Feeding and nutrition should be re-evaluated regularly as well as contractures.
- Epilepsy if present should be controlled. Orthotics might be needed.
- Specific treatment modalities include:
  1. Spasticity: Baclofen, BTD, Botulinum
  2. Surgical: Tendon release
  3. Feeding: Gastronomy

Seizures in Childhood

- Focal, Partial Seizures: State impairment of consciousness if applicable
- Generalized seizures:
  - Tonic clonic
  - Absence
  - Tonic
  - Clonic
  - Myoclonic
  - Atonic

Self-limiting or continuous
Criteria for a febrile fit: (RF)

1. Age: 6 months to 3 years
2. Extracranial cause of a fever
3. Medical history of febrile convulsions
4. Temp >38.5
5. Generalized convulsions duration not >30min
6. No neurological deficit pre or post convulsion
7. Normal EEG after one week

1/3 may have at least one recurrence. 2% risk of developing epilepsy.

Infantile Spasms: West's syndrome
Characterized by triad of:
1. Flexor or extensor spasms
2. Hypsarrhythmia on EEG
3. Mental retardation

Management: BZD, Valproate and Steroids.

Lennox-Gastaut syndrome (LGS)
Characterized by triad of:
1. Stare, jerk and fall seizures
2. Slow spike wave discharges on EEG
3. Mental Retardation

Stare, jerk and fall seizures are of several types usually:
- Brief tonic
- Atonic drop attack (more than 4 seconds)
- Myoclonic attacks
- Dysphasic absence
- Status

Temporal lobe epilepsy (TLE)
This syndrome comprises seizures arising from the temporal lobes and their connections. Thus, the seizure manifests in the form of olfactory, gustatory, psychosensory (visual and auditory hallucination) and autonomic symptoms. Memory and emotional distortions may also be involved. Sensations of extreme depression, pleasure, fears, anger, tempest tantrums or laughter may occur as may visceral manifestations such as hollow feeling in stomach, feeling of illness nausea, abdominal pain, palpitations, elevation, sweating.

The prophylactic management of a febrile seizure is only indicated if there are one or more of the following: first convulsion at a temperature ≥38.5°C or previous history of febrile seizures but not of non-febrile seizures.

Management of febrile seizures:
1. Identify the cause (LPE)
2. CT or MRI not warranted
3. Routine labs except for glucose
4. Long term use of antiepileptics drug (PEE) not necessary
5. Phenobarbital, sodium valproate
6. Rectal chlormethiazide
7. Antipyretic

Associated Symptoms
- Seizures occurring in children 3 months-3-5 years in association with fever but without the evidence of an intracranial infection. Boys > girls.

Causes of Epilepsy:
1. Idiopathic
2. Congenital: Trauma, HIE, Congenital brain anomalies
3. Trauma
4. Infection
5. Vascular
6. Neoplastic
7. No neurological deficit pre or post convulsion
8. Extracranial cause of a fever
9. Family history of febrile convulsions
10. Normal EEG after one week

Seizures usually due to a structural abnormality of the CNS
- Lesional: At one, structural abnormality
- Metabolic: Hypoglycemia, hypocalcemia, hypomagnesemia, hypoparathyroidism, hypothyroidism, hypoglycemia
- Vascular: Periventricular abnormalities
- Infections: Infections meningitis
- Trauma: Brain injuries
- Antecedent: Recognizable

Emergency and early childhood (2 months-3 years)
- Usually initial seizures, intaniltile spasms.
- LENNOX GASTAUT syndrome: myoclonic seizures, Status epilepticus and paroxysmal discharges on EEG

Childhood to early adolescence (usually typical absence seizures, TLE, benign partial seizures with Rolandic or centromedial spikes)

Nine years to adulthood, focal epilepsy is associated with brain injury. Primary generalised epilepsy is uncommon.

Leptin and leptin receptors play role in the pathogenesis of obesity and related diseases, including metabolic disorders and neuronal disorders. Leptin is a hormone that regulates appetite and metabolism. Its levels are upregulated in obesity and downregulated in anorexia nervosa. Leptin receptors are also expressed in the brain, suggesting a role in neural function. Dysregulation of leptin and its receptors may contribute to obesity, diabetes, and neurological disorders.
Treatment of neonatal seizures:
1. Optimize ventilation, cardiac output, BP, glucose, electrolytes and pH
2. Treat the underlying disease
3. IV fluids
4. Vent the chest
5. Treat seizures with phenobarbitone or Lorazepam or Phenobarbitone

Treatment of epilepsy:
1. Drug treatment should be started
2. Keep on profile
3. Maximum side effects
4. Monitor therapy, use one drug to minimum before adding a 2nd drug
5. Changes provided should be gradual
6. Continue with treatment for at least 2 years after last attack and keep away since decided to stop
7. High initial dose may cause side effects
8. Complex, learning disabilities, the list of provoking status
9. Always calculate the dose according to the weight

Anti-epileptic drugs in children

<table>
<thead>
<tr>
<th>Drug</th>
<th>Dose (mg/kg/day)</th>
<th>Half life</th>
</tr>
</thead>
<tbody>
<tr>
<td>Generalized and local</td>
<td></td>
<td></td>
</tr>
<tr>
<td>Phenobarbitone</td>
<td>5-10</td>
<td>9h</td>
</tr>
<tr>
<td>Phenytoin (Epanutin)</td>
<td>4-8</td>
<td>14</td>
</tr>
<tr>
<td>Carbamazepine (Tegretol)</td>
<td>10-20</td>
<td>12</td>
</tr>
<tr>
<td>Valproate (Eptin)</td>
<td>20-40</td>
<td>8</td>
</tr>
<tr>
<td>Lamotrigine</td>
<td>1.5</td>
<td>24</td>
</tr>
<tr>
<td>Sodium</td>
<td>40-60</td>
<td></td>
</tr>
<tr>
<td>Partial</td>
<td></td>
<td></td>
</tr>
<tr>
<td>Valproate</td>
<td>20-60</td>
<td>40</td>
</tr>
<tr>
<td>Ethosuximide</td>
<td>20-60</td>
<td>10-20</td>
</tr>
<tr>
<td>R - Carbamazepine</td>
<td>10-20</td>
<td>40</td>
</tr>
<tr>
<td>Focal seizures</td>
<td></td>
<td></td>
</tr>
<tr>
<td>Phenobarbitone</td>
<td>5-10</td>
<td></td>
</tr>
<tr>
<td>Valproate</td>
<td>20-40</td>
<td></td>
</tr>
<tr>
<td>Myoclonic seizures</td>
<td></td>
<td></td>
</tr>
<tr>
<td>Valproate</td>
<td>20-40</td>
<td></td>
</tr>
<tr>
<td>Clonazepam (Kovale)</td>
<td>0.015-0.05</td>
<td></td>
</tr>
<tr>
<td>Clonazepam (Rabion)</td>
<td>0.5</td>
<td></td>
</tr>
</tbody>
</table>

| Nitrazepam (Magadan)  | 0.2-1            |

STATUS EPILEPTICUS
Medical ER!!!

DEFINITION: Epileptic seizure with episodes of continuous dysrythmia lasting > 30 min
with no recovery of consciousness between seizures. Thus, seizures that is prolonged
and repeated
The most severe form of status epilepsy that is associated with severe brain damage is
generalized tonic clonic status.

Classification of status epilepticus

<table>
<thead>
<tr>
<th>Generalized</th>
<th>Partial</th>
</tr>
</thead>
<tbody>
<tr>
<td>Tonic-clonic</td>
<td>Simple</td>
</tr>
<tr>
<td>Myoclonic</td>
<td>Complex</td>
</tr>
</tbody>
</table>

Management of Status epilepsy: (EXCL. neonates)

1. Abort the seizure
   STEP 1: Lorazepam 0.1-0.5mg/kg IV or
           Clonazepam 0.015-0.05mg/kg IV at 0.5-1.5mg/kg rectally
           Look out for respiratory depression
           The dose of valium may be repeated!
   STEP 2: Phenytoin 20mg/kg slowly IV (max rate of infusion is 1mg/kg/min) OR
           10mg/kg IV infusion then 3mg/kg in 6 hours and then 3mg/kg in 6 hours again.
           Repeat dose only once at 3mg/kg
           An alternative is Valproate 20mg/kg IV in children > 2 years
   STEP 3: If seizures haven't stopped in 15min arrange for an ICU bed and initiate
           Thiopentone IV stat
           1-3mg/kg/hour
Alternative is midazolam loading dose of 0.2mg/kg with a maintenance dose of 0.1mg/kg/hour. OR Propofol loading dose 1mg/kg with a maintenance dose of 0.5mg/kg/hour.

2. ABC of resuscitation
3. Mannitol for cerebral oedema
4. Correct metabolic and biochemical abnormalities:
   - Drop in BP
   - Impaired brain perfusion
   - Increased liver enzymes
   - Clotting defects
   - Hyperkalemia
   - Hypoglycaemia
   - Inappropriate ADH release
   - Renal failure
5. Treat hypervolemic
6. Treat the cause of the seizure. DO a drug/toxic screen, CT/MRI or LP to find a cause!

[Diagram of LMN disorder]

The floppy infant syndrome / Hypotonia and Lower motor neuron disorders

[Diagram of infantile spinal muscular atrophy (SMA)]

Infantile spinal muscular atrophy (SMA) — Sweating paralyses

Common hereditary disorder affecting proximal symmetrical muscle atrophy associated with progressive degeneration of the anterior horn cells of the spinal cord and bilateral lower motor neurons (5th-12th CNs).

Clinical features:
1. Severe weakness of various degrees
2. Face more affected than the arms
3. Facial weakness seen in SMA of cases
4. Fasciculations of the tongue and small muscles of the hands
5. Tone reflexes are reduced
6. Marked hypotonia

Clinical classification of SMA

<table>
<thead>
<tr>
<th>Clinical classification of SMA</th>
<th>Course</th>
<th>Course</th>
<th>Age of death</th>
</tr>
</thead>
<tbody>
<tr>
<td>1. Severe Werdnig Hoffmann</td>
<td>First 6 months</td>
<td>Never stands</td>
<td>&lt;= 2 years</td>
</tr>
<tr>
<td>2. Intermediate Werdnig Hoffmann</td>
<td>&lt; 16 months</td>
<td>Never stands</td>
<td>&lt;= 2 years</td>
</tr>
<tr>
<td>3. Mild Kugyelberg Weiland</td>
<td>18 months</td>
<td>Stand alone</td>
<td>Adult</td>
</tr>
</tbody>
</table>

TYPE 1 SMA: Present in first days of the infant shows generalized hypotonia with marked weakness and head down trunk and poor head control. Legs more involved than the arms. Bulbar weakness present with difficulties in swallowing and sucking.

The chart shows:
- Difficult swallowing
- Weakness
- Leans during
Onset: Infantile Poliomyelitis:
- Presents in infants and young children.
- Muscles involved:
  - Spinal: Lower limbs (weakness, paralysis)
  - Cranial: Palsy affecting the muscles of the face and limbs
- Systemic signs: Fever, vomiting, rash
- Therefore, clinical picture is that of a systemic illness extending to involve the CNS.

Guillain-Barré syndrome:
- Onset: Several weeks after a viral infection (e.g., respiratory, gastroenteritis, etc.).
- Clinical presentation: Flaccid paralysis, areflexia, sensory loss.
- Systemic signs: Fever, malaise, headache, rash.
- Diagnosis:
  1. CSF: Increased protein, increased IgM, decreased glucose.
- Management:
  1. Supportive care (hospitalization).
  2. Plasma exchange or IV immunoglobulin.
- Prognosis: Depends on severity and duration of illness.

Peripheral Nerves (peripheral neuropathies are uncommon in children):
- Possible causes:
  1. Hereditary motor and sensory neuropathy
  2. Congenital hypomyelinating neuropathy
  3. Axonal neuropathy
  4. Metabolic neuropathies
- Diagnosis:
  1. Clinical presentation
  2. Electrophysiological studies
  3. Imaging studies (MRI, EMG)
- Treatment:
  1. Symptomatic management
  2. Nutritional supplements
  3. Physiotherapy

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  1. CSF: Increased protein, increased IgM, decreased glucose.
- Management:
  1. Supportive care (hospitalization).
  2. Plasma exchange or IV immunoglobulin.
- Prognosis: Depends on severity and duration of illness.
**Diphtheria**

**Neuromuscular junction**

**Acquired Myasthenia Gravis (MG)**

- Characteristic weakness and decremental fatigue after repetitive or sustained activity, and improvement after rest.
- Weakness affects muscles of the eye, face, and extraocular muscles.
- Also affects facial muscles, including the face, neck, and extremities.
- May be present in the legs.
- Diagnosis is made clinically by fatiguing certain muscle groups, measuring the power before and after fatigue. This can be confirmed by performing repetitive stimulation of the nerve.
- Anti-AChR antibodies are detected in 80-90% of cases.
- There is evidence of associated disorders such as thyroid disease, diabetes, and sarcoidosis.
- Treatment includes immunosuppressive drugs such as pyridostigmine (mestinon), 150 mg (dosage and frequency are adjusted to the patient). The treatment is symptomatic in nature.
- Immunosuppressive and immunomodulatory therapy can be used to treat myasthenia gravis.

**Muscle disorders**

**Muscular dystrophies**

These are defined as a group of genetically determined disorders with progressive degeneration of skeletal muscle and no structural abnormalities in the central or peripheral nervous system.

1. **Duchenne's muscular dystrophy**
   - Most common.
   - X-linked disorder characterized by progressive weakness of predominantly proximal muscles due to a deficiency in the neuromuscular junction.
   - One of the middle-sized muscles where the child starts walking but may have difficulty walking at any age.
   - Signs include:
     - Abnormal gait
     - Frequent falls
     - Difficulties in climbing stairs
     - Weakness in shoulder muscles
     - Progressive weakness initially

2. **Becker's muscular dystrophy**
   - Similar to Duchenne's but milder. It has the same location on the Xp21 chromosome.
   - Onset of symptoms occurs later, with a more benign course.

3. **Congenital muscular dystrophy**
   - This consists of a group of muscular dystrophies affecting infants with muscle weakness at birth or within a few months of life.
**Myotonic syndromes:**

**Myotonic dystrophy**

This clinical form occurs in children and adults with myotonia dystrophy of the 
(Eisenhardt, 1995). It typically presents as a delayed component of the leg 
atrophy. Associated features include cardiac involvement, cataract formation, 
growth abnormalities, postural instability and intellectual impairment.

The inheritance pattern is autosomal dominant with the gene located on 19q13.

The neonatal form has its onset promptly and the fetus may present with

Acute onset of symptoms and a rapid progression to death with rapid loss of

Muscle strength.

Myotonia congenital

**Myotonia congenital**

Characterized by delayed relaxation of muscles after voluntary movement.

<table>
<thead>
<tr>
<th>THUNDER syndrome</th>
<th>MYTH syndrome</th>
</tr>
</thead>
<tbody>
<tr>
<td>Autosomal dominant</td>
<td>Autosomal recessive</td>
</tr>
<tr>
<td>Myotonia and muscle hypertrophy</td>
<td>Myotonia and muscle hypertrophy</td>
</tr>
<tr>
<td>More benign course</td>
<td>More benign course</td>
</tr>
<tr>
<td>Myotonia occurs when the child has difficulty in rising from their seat</td>
<td></td>
</tr>
<tr>
<td>Weakness is usually more severe in the upper body</td>
<td></td>
</tr>
<tr>
<td>Progress is more slow and more symptom is usually noted after a period of rest</td>
<td></td>
</tr>
<tr>
<td>Pain is less</td>
<td></td>
</tr>
<tr>
<td>Cold fatigue and muscle weakness may exaggerate the myotonia</td>
<td></td>
</tr>
</tbody>
</table>

**Paramyotonia congenital (Eulenberg's disease)**

Myotonia that is brought on by cold. It is mild and affects the face, eyelids and hands. Responds equally to warming.

**Chondrodystrophic myotonia (Schwartz-Jampel syndrome)**

Consists of a myotonia, chondrodystrophy and short stature.

Children have a distinct appearance of blepharoptosis, narrow palpebral fissures, macroglossia, and flattened faces. The muscles are hypertrophied and stiff with limitation of joint movements.

---

**Stroke in Children**

Most strokes in children are due to cerebrovascular abnormalities. The stroke can be acute or chronic progressive.

**Classification:**

1. Embolism of cerebral vessels
2. Thrombosis – Arterial or venous
3. Cerebral haemorrhage

**Embolic:**

Sudden loss of neurological function with an acute hematoma. This rare in children.

**Cause:**

1. Atherosclerosis
2. Vascular anomalies
3. Air / fat / septic emboli
4. Cerebral congenital heart disease
5. Rheumatic heart disease

**Arterial thrombosis**

Faster to develop. It is rare in children.

The cause is usually due to a vascular abnormality:

1. Infections
2. Diphtheria
3. Haemoglobinopathies

**Venous thrombosis**

More common in children. Usually present with seizures, an altered mental state, raised ICP and focal neurological signs.

It is associated with:
1. Bacterial meningitis
2. Cerebral sinus thrombosis
3. Severe dehydration (thrombosis of superior sagittal sinus and superficial cortical veins)
4. Hypocalcaemia

**Cerebral haemorrhage**

Cause includes:
1. AV Malformations – Intracranial haemorrhage
2. Cerebral aneurysms
3. Haematological disorders
4. Trauma
<table>
<thead>
<tr>
<th>Causes of Stroke in Children Include:</th>
</tr>
</thead>
<tbody>
<tr>
<td><strong>Heart diseases</strong></td>
</tr>
<tr>
<td>Congenital</td>
</tr>
<tr>
<td>Acquired</td>
</tr>
<tr>
<td>Intravascular:</td>
</tr>
<tr>
<td>1. Hemodynamical disorders</td>
</tr>
<tr>
<td>2. Sickle cell disease</td>
</tr>
<tr>
<td>3. Infections:</td>
</tr>
<tr>
<td>4. Arterial border zone - associated with resuscitation and hypotension</td>
</tr>
<tr>
<td>5. Multi artery infarction - congenital heart disease, DIC and polycythemia</td>
</tr>
<tr>
<td>6. Single artery infarction - injury to carotid artery during delivery</td>
</tr>
<tr>
<td>7. Maternal use of cocaine can cause cerebral infarction</td>
</tr>
<tr>
<td>8. Idiopathic</td>
</tr>
<tr>
<td>Vascular:</td>
</tr>
<tr>
<td>9. Moyamoya</td>
</tr>
<tr>
<td>10. Anemia</td>
</tr>
<tr>
<td>11. Infections:</td>
</tr>
<tr>
<td>12. Neurofibromatosis</td>
</tr>
<tr>
<td>13. Migraine</td>
</tr>
<tr>
<td>14. Idiopathic</td>
</tr>
<tr>
<td>Trauma:</td>
</tr>
<tr>
<td>15. Alternating hemiplegia in childhood</td>
</tr>
<tr>
<td>16. Idiopathic</td>
</tr>
<tr>
<td>Other:</td>
</tr>
<tr>
<td>17. Alternating hemiplegia in childhood</td>
</tr>
<tr>
<td>18. Idiopathic</td>
</tr>
</tbody>
</table>

**Causes of neonatal infarction:**
1. Arterial border zone - associated with resuscitation and hypotension
2. Multi artery infarction - congenital heart disease, DIC and polycythemia
3. Single artery infarction - injury to carotid artery during delivery
4. Maternal use of cocaine can cause cerebral infarction
5. Idiopathic

**Management:**
1. **Thorough History:**
   - Trauma
   - Drug ingestion
   - Developmental status
   - MR
   - Regression
   - Seizures
   - Family history
2. **Examination:**
   - Skin birthmarks, Abnormal pigmentation, Nodules, Papules, Signs of trauma

**Abnormal Movements in Children**

**Abnormal movements:**

**SLOW - Dystonias and Athetosis**

**FAST - Stereotyped (involuntary)**

**Rhythmic = Tardor**

**Not Rhythmic = Tics and myoclonus**

- Non stereotyped
- Choreo

**TICS**

- Most common movement disorder in children
- MOTOR: eye blinking, shoulder shrug
- VOCAL: squeaking, cough, sniffing
- SENSORY: sensation, non comfortable in skin
- Can be suppressed
- Letsens in sleep
IRANSIENT CHRONIC TOURETTE'S

<table>
<thead>
<tr>
<th>TRANIENT</th>
<th>CHRONIC</th>
<th>TOURETTE'S</th>
</tr>
</thead>
<tbody>
<tr>
<td>Single / multiple motor</td>
<td>Single / Multiple motor OR Aural/Oral</td>
<td>Multiple motor AND single / multiple vocal</td>
</tr>
<tr>
<td>6-12 weeks &lt; 1 year</td>
<td>&lt; 1 year but not free for 3 months</td>
<td>&lt; 1 year and not free &lt; 3 months</td>
</tr>
</tbody>
</table>

Tourette Syndrome:
- Compromise of motor and vocalic
- Present from early years
- Occurring in most patients daily nearly everyday
- Causing significant impairment or marked distress
- Onset <18 years
- Not due to drug or illness

![Diagnostic criteria:](image)

PANDAS:
(Paediatric Autoimmune Neuropsychiatric Disorder associated with Streptococcal Infection)
- Associated with Tics and Dystonia post a GABHS infection
- Emotional and behavioural changes are common - OCD, Anxiety, personality changes.
- Causes by auto antibodies to basal ganglia
- Diagnostic criteria:
  1. Prepubertal
  2. Tics or OCD
  3. Sudden onset / fluctuating course
  4. Ass with GABHS inf.
  5. Neurological abnormalities
- Associated disorders:
  - OCD
  - ADHD (90% of Tourette's syndrome precedes for by 2 years)
  - Sleep disorders
  - Learning problems
  - Behavioural problems
  - Mood disorders
- There is currently a lot of data pointing to inflammation of cerebellum — probably autoimmunity dominant.
- Treatment: Pharmacological treatment is only indicated if the tics become incapacitating but is rarely needed. Treatment includes haloperidol. But the management of co-morbid disorders probably more important.

CHOREA: [IN Voluntary, A RYTHMICAL, ASYMMETRICAL SUDDEN BRIEF > PROXIMAL]
There are about 150 causes described, to mention a few:

1. Infections: Rheumatic fever, Sydenham Chorea, Herpes Encephalitis, HIV
2. Systemic disease: SLE
3. Metabolic: Wilson's disease, Celiac sprue
4. Vasculitis: Cerebral artery lesions
5. Intoxication: CO, Methanol alcohol
6. Primary genetic: Benign hereditary, Huntington's chorea

Sydenham's Chorea
- Major feature of Rheumatic fever
- More common in older age group and girls
- Often sore throat
- Febrile illness
- Headache, difficulty writing, stiffness and then over weeks/years the chorea becomes evident
- Persistent for weeks to months
- Usually good outcome
- Clinical manifestations include: MUSCULAR SYNDROME, let the child grasp index finger of examiner.

Let child extend arms above head with palm upwards will find it difficult to maintain this posture and chorea will be aggravated.

Dystonia: Co-contraction, Abnormal Posture
Another description of dystonia is involuntary sustained or intermittent muscle contractions causing twisting of repetitive movements, abnormal postures or both. Can affect any part of the body and it can be painful. It disappears with sleep. It is assumed to be due to an imbalance of neurotransmitters.

Classification:

1. Primary
   - Idiopathic torsion dystonia
   - Inherited idiopathic dystonia of infants
   - Juvenile parkinsonism

2. Secondary
   - Metabolic disease
   - Drug induced
   - Stroke, traumatic, tumors, neurodegenerative

3. Or by Age: In children there are 2 types:
   - Primary torsion dystonia
   - Dopa-responsive dystonia

Dystonia is genetic in origin - mostly autosomal dominant with variable penetrance. It usually begins in the feet and then spreads to become generalized. It is severely disabling but cognition is spared.

Dyskinetic CP:
2 videos exist:

1. Chorea-athetoid
2. Dystonic

Dystonia is usually severe type in term babies and previous heritabilities. There is also usually a history of insult and trauma. The incidence of MRS 30%.

Sterotypes:
Voluntary, often rhythmic movements, e.g. head banging, head rolling, thumb sucking. Self stimulating behaviors can also included here.
It can occur in otherwise normal children. Some patterns are more prevalent in children with Mental retardation and behavioral disorders like autism - e.g. self mutilation, brushing, hand washing.

Myoclonus (Simple, Sudden, Single):
1. Physiological: Sleep myoclonus, Startle response (Aware)
2. Non-epileptic: Benign neonatal sleep myoclonus, benign myoclonus of early infancy
3. Epileptic: Myoclonic epilepsy, component of epilepsy syndromes with different seizure types.

1. Headache

Headaches tend to become more frequent with increasing age. Features associated with an organic brain lesion, then WARNING SIGNS:
- Severe headache of recent onset
- Chronic or progressive headache
- Localized pain
- Wakes the child at night
- Exacerbated by straining, valvula or change in position
- Associated with neurological symptoms and signs
- Change in headache pattern

Classification of headache according to the temporal pattern:

<table>
<thead>
<tr>
<th>TEMPORAL PATTERN</th>
<th>CAUSE</th>
<th>HEADACHE TYPE</th>
</tr>
</thead>
<tbody>
<tr>
<td>Acute</td>
<td>1. Infections and fever</td>
<td>Sometimes associated with neurolologic signs and symptoms</td>
</tr>
<tr>
<td></td>
<td>2. Toxins</td>
<td></td>
</tr>
<tr>
<td></td>
<td>3. Post convulsions</td>
<td></td>
</tr>
<tr>
<td></td>
<td>4. Anemia - Hypotension</td>
<td></td>
</tr>
<tr>
<td></td>
<td>5. Electrolyte</td>
<td></td>
</tr>
</tbody>
</table>
### Trigger/precipitating factors in migraine:
1. Anxiety
2. Fatigue/Lack of sleep
3. Stress - Headache occurs after the stress during relaxation
4. Head trauma - especially in males
5. Exercise - during or after
6. Menstruation - at menarche or associated with PMS.
7. Diet - Tyramine containing foods (releases serotonin) such as ice-cream, red wine, cheeses. We hope that the child is not taking any red

### Nature of a migraine headache:
- They are throbbing
- Unilateral (can be bilateral and alternating in sides)
- Relieved after sleep
- An aura is usually present
- Abdominal pain might be the only preceding symptom in children
- Nausea and vomiting usually present as well as a
- Family history

### 3 types of migraines:

<table>
<thead>
<tr>
<th>COMMON (Without aura)</th>
<th>CLASSIC (With aura)</th>
<th>COMPLICATED</th>
</tr>
</thead>
<tbody>
<tr>
<td>Usually no well-defined aura except autonomic; e.g. neuropathy, nausea, vomiting, abdominal pain, facial pain, and hair loss under the eyes.</td>
<td>Aura preceded by problems, e.g.:</td>
<td>Associated with neurologic deficit due to prolonged ischemia caused by vascular constriction.</td>
</tr>
<tr>
<td>Sensation - paresthesia of both hands/palmar erythema</td>
<td>Visual - light flashes, blurred vision, scotoma, transient blindness, or hemianopia</td>
<td>The deficit precedes the headache but may also follow it and can last for up to several days.</td>
</tr>
<tr>
<td>1/2 a week</td>
<td>Auditory, and olfactory</td>
<td></td>
</tr>
<tr>
<td>Lasts 2-6 hours</td>
<td></td>
<td></td>
</tr>
<tr>
<td>Relieved by sleep</td>
<td></td>
<td></td>
</tr>
</tbody>
</table>

### Migraine

Delineates: Episodic, periodic paroxysmal attacks of vascular constriction and vasodilation of cerebral blood vessels.

Migraine is the most common form of headache in children in 5% of patients it occurs 10-20 y of age and in 20% of y of age.

In childhood boys right, but after puberty, girls right.
Presumed symptom:

1. Tension headache

Involves bihemiparietal and/or frontal pain. Treatment includes relaxation, anti-inflammatory, and analgesics.

Cluster headache:

Characterized by episodes of unilateral, severe pain lasting 45-180 minutes. Trigger is usually associated with consumption of alcohol or food that contains monosodium glutamate. Treatment includes ergotamines, triptans, and intranasal or intravenous sumatriptan.

Migraine:

Characterized by throbbing pain, often with nausea and sensitivities to light, sound, and touch. Treatment includes prophylactic medications, such as beta-blockers and NSAIDs, and acute medications, such as triptans and ergotamine.

Tension headache:

Characterized by generalized or localized pain in the forehead, scalp, neck, or upper back. Treatment includes relaxation techniques, such as biofeedback and massage, and over-the-counter pain medications.

Hydrocephalus:

Definition: Abnormal accumulation of CSF in the ventricles of the brain, leading to increased intracranial pressure and dilation of the brain. Treatment includes placement of a shunt to drain excess CSF and alleviate pressure.

Management of headaches:

1. Obtain a detailed history
2. Perform a thorough physical and neurological examination
3. Consider the use of diagnostic studies
4. Initiate treatment
5. Identify triggers and environmental factors
6. Consider the presence of ocular symptoms
7. Monitor for complications

CSF is produced in the choroid plexus of the lateral ventricles and is drained into the subarachnoid space through the interventricular foramen of Monro and the cerebral aqueduct. It then flows through the subarachnoid space to the spinal cord and eventually exits the body via the arachnoid granulations in the dura mater.
In 12% travels through spinal pathways, eventually it is absorbed through the subarachnoid cavities into the sagittal sinus.

Path of formation: 0.3-0.4 mm/in and continuous even when the intraventricular pressure is increased.

Total CSF volume in newborn is 50 mL increasing with age to adult volume of 150 mL.

Normal intraventricular pressure is 50-120 mm H2O in child.

**Classification of Hydrocephalus:**

<table>
<thead>
<tr>
<th>Pathway</th>
<th>Cause</th>
</tr>
</thead>
<tbody>
<tr>
<td>Intraventricular Block</td>
<td>Tumour / Cyst</td>
</tr>
<tr>
<td>3rd Ventricle</td>
<td>Tumour / Cyst</td>
</tr>
<tr>
<td>Sylvian Aqueduct</td>
<td>Tumour, inflammation, haemorrhage, congenital aqueduct stenosis.</td>
</tr>
<tr>
<td>4th Ventricle</td>
<td>Post. Fossa tumour / cysts (Dandy Walker), neural tube defects.</td>
</tr>
</tbody>
</table>

**Signs and Symptoms of Hydrocephalus:**

1. Large head / progressively enlarging head circumference (crossing sutures)
2. Pseudotumoural headache
3. Headache
4. Spasticity
5. Distortion of scalp veins
6. Sunsetting sign or loss of upward gaze
7. Neck retraction or neck stiffness
8. Perinatal failure signs

**Management of Hydrocephalus:**

1. Do CT scan or MRI
2. Measure the CSF pressure and measure the intraventricular pressure.
3. Lumboperitoneal or ventriculoperitoneal shunt.
4. Medical reduction of CSF production: Diuretics
5. Surgical diversion of CSF flow by means of SHUNTING: Ventriculoperitoneal (VP) shunt / Lumbo-peritoneal shunt.
6. Follow up to monitor for complications of shunt procedures.
ORAL & DENTAL DISORDERS
Oral and Dental Disorders

Systemic diseases may often be reflected by abnormalities in the mouth.

**Mouth (Oral Cavity)**

- Gingiva (gums)
- Hard palate
- Uvula
- Papillae of tongue
- Premolar
- Canine
- Lips
- Soft palate
- Palatine tonsil
- Molars
- Tongue

**NORMAL TOOTH**

- Enamel: Constitutes the bulk of the tooth. Yellow/Opaque in colour. Considered to be calcified pulp.
- Dentine: Covers the Dentine at the root of the tooth where it is held in the alveolar bone.
- Crown: Part of tooth projecting above the gingiva and it is covered with ENAMEL (Enamel is translucent in colour). The thinner the enamel, the more yellow the teeth appear.

Dental pain is felt when the dentine becomes exposed to noxious stimuli. Fluid then moves into the innervated pulp. A number of factors initiate this, e.g. sugar, temp. changes.

**Teething:**
Process whereby teeth cut through gum.

<table>
<thead>
<tr>
<th>6th months of age</th>
<th>1st tooth erupts</th>
<th>Lower central incisor</th>
</tr>
</thead>
<tbody>
<tr>
<td>3 years</td>
<td>all milk teeth/primary teeth present.</td>
<td></td>
</tr>
<tr>
<td>6 years</td>
<td>the permanent molars erupt distal to the primary molars, not replacing them.</td>
<td></td>
</tr>
<tr>
<td>6-12 years</td>
<td>period of mixed dentition</td>
<td>primary dentition exfoliates and is replaced by permanent teeth.</td>
</tr>
<tr>
<td></td>
<td>3rd molars/wisdom teeth (32) are the last of permanent dentition to erupt.</td>
<td></td>
</tr>
</tbody>
</table>

The whole process of teething is accompanied with general (Fever, GI disturbances) and local (Swelling and gingival irritation).

**Treatment:** symptomatic with smooth biting toy and topical medication.

**Problems with erupting teeth**

<table>
<thead>
<tr>
<th>Problem</th>
<th>Pathophysiology and symptoms</th>
<th>Treatment</th>
</tr>
</thead>
<tbody>
<tr>
<td>Eruption cyst</td>
<td>Follicular sac surrounding the developing tooth fills with fluid or blood when traumatized. Therefore, gum over erupting tooth forms a blue, purple swelling. Fluctuating, painless.</td>
<td>No treatment. Eruption will follow normal course.</td>
</tr>
<tr>
<td>Premature eruption</td>
<td>Tooth/teeth present at or soon after birth. Usually hyper mobile. High ASPIRATION risk</td>
<td>Remove</td>
</tr>
<tr>
<td>Premature loss of primary teeth</td>
<td>The closer to actual eruption time of the successor the primary tooth is lost, the speedier its eruption.</td>
<td>Remove only retained primary incisors.</td>
</tr>
<tr>
<td>Abnormal number</td>
<td>Supernumerary (more teeth in dental arch) Most common is a mesiodens tooth lying in ant, palatal midline. Indicates possible underlying</td>
<td>Extraction</td>
</tr>
<tr>
<td>Abnormality</td>
<td>Description</td>
<td></td>
</tr>
<tr>
<td>-----------------------------</td>
<td>---------------------------------------------------------------------------------------------------------------------------------------------</td>
<td></td>
</tr>
<tr>
<td>Hypodontia</td>
<td>Fewer than normal teeth. Common with ectodermal dysplasia. Down's and cleft palate.</td>
<td></td>
</tr>
<tr>
<td>Abnormal shape</td>
<td>Microdontia. Macroodontia. Fusion of 2 developing teeth. Germination/twinning results from an incomplete division of a tooth bud. Syphilis commonly affects developing teeth e.g. causing HUTCHINSON'S incisors (Screw driver shaped) or Mulberry molars.</td>
<td></td>
</tr>
</tbody>
</table>
| Abnormal structure          | Hereditary or environmental.  
Amelogenesis imperfecta: Groups of hereditary enamel defects. Enamel hypoplasia: Due to infection or nutritional deficiencies. This results in mottling or white spots in the enamel.  
Dentinogenesis imperfecta: affects primary and permanent dentition. Teeth vary in colour, grey to brownish blue. Associated with osteogenesis imperfecta. |
| Abnormal colour             | Congenital: Porphyria. Obstructive Jaundice. Haemolytic anaemias stain teeth with blood borne products which are taken up during tooth formation.  
Acquired: Tetracyclines stains teeth yellow-brown when taken by mother during pregnancy or the first three months after birth. Permanent teeth affected when taken from 3 months up to 8 YEARS of age.  
Systemic fluoride in excessive amounts, tea, coffee, tobacco, iron.  
This brownish yellow staining can be removed with |
Dental caries and toothache (primarily a disease of children)

Pathophysiology: Follows the action of plaque bacteria on dietary carbohydrates (sugar). Sugar is rapidly fermented to acid which is then held in plaque. Destruction spreads to dentine. Undermining of the softened enamel occurs. Cavity forms and tooth is open for bacterial invasion. When cavity forms, process is irreversible.

The longer and more frequent the sugar is in contact with the plaque, the longer the acid will be in contact with the tooth surface and the more the enamel will be demineralized by the acid. Caries usually starts in grooves on biting surfaces of the tooth or in between teeth, often hidden away. Demineralization leads to white spot on the tooth, this is still reversible with vigorous oral hygiene as long as there is no break in the enamel. A fracture in the enamel is visible as an obvious cavity in the tooth called an active carious lesion. The active carious lesion is soft in consistency and has a yellow-tan to brown colour. The active lesion can now follow one of 2 routes:

1. The lesion can become inactive (dark brown to black in colour with a hard consistency), now called an arrested...
caries is painless requiring no further management.

2. The lesion can also become one of two types of rapidly advancing caries: ECC (Early childhood caries) or rampant caries.

ECC: Caries occurring in primary teeth. Children aged between birth and 6 years of age. The cause is usually prolonged bottle feeding usually associated with sucrose containing liquid (sweetened milk, sugared water, fruit juices and carbonated or non carbonated beverages) in the bottle or by the use of sweetened dummies.

Rampant caries: Also seen in teenagers. Very destructive pattern. Widespread and rapid attack at a number of sites not normally susceptible to decay. (Areas between lower incisors, outer surfaces of upper incisors and inner surfaces of upper teeth.). Patients can present with dental abscesses.

Symptoms and signs: Pain. Pain is felt when carious process reached sensitive dentine. Stimuli initiating the pain will be hot, cold or sweet drinks and food stuff. Pain usually throbbing in nature and tooth is painful to bite on.

Bacteria soon invades the pulp through the dentine causing death and necrosis of the surrounding tissues with pus accumulating under and around the roots of the tooth. This causes severe pain and swelling of the gum (a boil or dento-alveolar abscess) which often ruptures spontaneously in children. Persistent infection causes damage to the underlying tooth below the damaged primary tooth.

Complications: Abscess/boils, asphyxiatation and cavernous sinus infection (When pus accumulates without the formation of a tract or sinus to the outside now it spreads to the cheek and via tissue planes to the cavernous sinus.)
Treatment:
(1) Local Anaesthesia and cleaning out of the cavity.
(2) Temporary filling made from Zinc oxide powder and oil of cloves or Glass ionomer Cement. (3) Analgesia.
(4) Dental referral for permanent restorations.

An tooth with an abscess should be extracted unless root canal repair is available. If non of the above is available suppress the abscess with either antibiotics alone or with a combination of antibiotics and incision and drainage. The antibiotic of choice is Penicillin or Erythromycin.

Prevention:
Reduction and elimination of dietary sugar and increasing the tooth's resistance to acid with incorporation of fluoride into the developing enamel. (The safest way of giving fluoride to the entire population is via drinking water) Where the water content of fluoride is less than 0.3 parts per million (ppm) dietary supplementation is advocated in the form of drops or tablets.

<table>
<thead>
<tr>
<th>AGE</th>
<th>DAILY DOSE</th>
</tr>
</thead>
<tbody>
<tr>
<td>0-2 y</td>
<td>0.25mg (0.55mg sodium fluoride)</td>
</tr>
<tr>
<td>2-4 y</td>
<td>0.5mg (1.1 mg sodium fluoride)</td>
</tr>
<tr>
<td>4-12 y</td>
<td>1.0mg (2.2 mg sodium fluoride)</td>
</tr>
</tbody>
</table>

The most cost-effective agent is fluoride toothpaste and fluoride mouthwashes. Weekly fluoride rinses can be organized for school children. They should not have anything to eat or drink 30 min before and after rinsing. If commercial mouthwash not available make your own; mix 2g sodium fluoride powder in 1l of water. Dilute this 10 x for a daily mouth rinse. Fluoride is beneficial on smooth surfaces of the teeth. Pit and
Fissure sealants are effective in the occlusal or grooved areas of the teeth.

**Gingival and periodontal conditions**

Prevention of gingivitis and periodontitis is mainly by tooth brushing and the use of dental floss.

<table>
<thead>
<tr>
<th>Condition</th>
<th>Features</th>
<th>Treatment</th>
</tr>
</thead>
<tbody>
<tr>
<td>Chronic inflammatory gingivitis</td>
<td>Inflammation to the gingiva due to plaque accumulation. The signs are redness, swelling and bleeding. Periodontal ligament and bone are not involved. Usually painless.</td>
<td>Reversible with removal of plaque.</td>
</tr>
</tbody>
</table>
| ANUG: Acute necrotizing ulcerative gingivitis | Destructive gingivitis caused by fusiform bacilli and spirochaetes. (Borrelia vincentti and fusobacterium). Gingival marginal ulceration, bleeding and extreme pain. Lesions covered by a pseudomembrane or slough. Contributing factors include poor oral hygiene, existing chronic marginal gingivitis, malnutrition and systemic illness (HIV). | Reassurance  
Corrective nutrition  
Oral hygiene instruction and gentle ultrasonic cleansing  
Metronidazole or Penicillin  
Chlorhexidine mouthwash bd. (0.2% solution). |
| Cancrum oris  
(noma)       | Gangrenous form of ANUG usually seen in malnourished children often with an underlying history of a viral infection such as measles. |                                                     |
<p>| ANUG: Acute herpetic gingivostomatitis | Vesicles, ulcers, redness of the gingiva and oral mucosa, pain, fever | Tetracycline mouth rinses.                          |</p>
<table>
<thead>
<tr>
<th>Condition</th>
<th>Description</th>
<th>Management/Prevention</th>
</tr>
</thead>
<tbody>
<tr>
<td>Acute leukaemia</td>
<td>Gingival hyperplasia, ulceration, bleeding, red/purple colour.</td>
<td>Specialist referral</td>
</tr>
<tr>
<td>Hormonal or puberty gingivitis</td>
<td>Gingival swelling, redness, bleeding. Hormonal changes associated with puberty enhances the tissue response to plaque. Steroid hormones causes the gingival tissue to be more sensitive to microbial challenges.</td>
<td>Oral hygiene</td>
</tr>
<tr>
<td>Periodontitis</td>
<td>Chronic gingivitis progresses into chronic periodontitis. Inflammation of the gum, bone and supporting structures of the teeth. Gingivitis, pockets, plaques, calculus, loose teeth, bone loss.</td>
<td>Oral hygiene root planing, scaling</td>
</tr>
<tr>
<td>Prepubertal periodontitis</td>
<td>Primary dentition affected. Associated with severe congenital or acquired defects involving immunodeficiency such as Chediak-Higashi syndrome, chronic and cyclic neutropenia and HIV. Deep pockets form, bone loss, few or all teeth affected, systemic defects present.</td>
<td>Broad spectrum Antibiotics Oral hygiene Extraction of loose teeth, scaling and root planing.</td>
</tr>
<tr>
<td>Juvenile periodontitis</td>
<td>Familial condition. Hereditary. 2 clinical forms: (a) Localized, involves only 1st molar and incisor teeth. (b) Generalized, many or all teeth affected. Usually patients have a relative depression of systemic and local polymorphonuclear leucocyte function. Usually associated with Actinobacillus infection. Gingiva appears normal. Limited plaques and calculus deposits, deep pockets, bone loss. Usually adolescents.</td>
<td>Oral hygiene Scaling and prophylaxis, antibiotics and sometimes surgery to eliminate deep pockets.</td>
</tr>
<tr>
<td>Periodontal Abscess</td>
<td>Develops as a complication of gingival or periodontal pockets. Problem precipitated by entrapment of foreign objects such as popcorn, toothbrush bristles followed by</td>
<td>Drainage and debridement followed by saline or antiseptic irrigation of the pocket.</td>
</tr>
</tbody>
</table>
**Oral and mucosal lesions**

<table>
<thead>
<tr>
<th>Condition</th>
<th>Features</th>
<th>Treatment</th>
</tr>
</thead>
<tbody>
<tr>
<td>Recurrent aphthous ulceration</td>
<td>Oral ulceration first appearing between the ages 10-19 years. Usually caused by trauma, emotional stress, hormonal factors and bacteria. Minor ulcerations: Shallow, round or oval shaped, grey-yellow colour in centre with red border. Forms crops of 1-4, 2-4mm in size, rarely over 1 cm. Common on labial and buccal mucosa. Heals in 1-2 weeks. Major ulcerations: Larger often &gt;1 cm, takes 6 weeks/&gt; to heal. Seen on lips, soft palate and pillars of the fauces or oropharynx.</td>
<td>Topical anaesthetic or a steroid in the form of a protective paste or use of an antiseptic. Or Chlorhexidine/analgesic mouthwash. Recurrent Mostly actually self limiting.</td>
</tr>
<tr>
<td>Candidosis</td>
<td>Soft white gelatinous plaques, eroded ulcerated mucosa, redness, candidal invasion.</td>
<td>Correct systemic disorder, proper nutrition: antifungal drugs, mycostatin suspension.</td>
</tr>
<tr>
<td>Geographic tongue</td>
<td>White annular lesions on the tongue with atrophic centres. Found on dorsum of the tongue. (Appears like a geographic map). Asymptomatic.</td>
<td>None, regress over a few years. Steroids topically for</td>
</tr>
</tbody>
</table>
Tuberculosis occurs as early as 2 years of age. Ulcer – irregular, undermined edges, covered with grey/yellow slough. Granulating soft red bleeding gingiva.

HIV infection Ulceration of the gingiva, hairy leukoplakia, candidosis. Refer to specialist: Oral hygiene, scaling and prophylaxis.

<table>
<thead>
<tr>
<th></th>
<th>Prevention of dental disease</th>
</tr>
</thead>
<tbody>
<tr>
<td></td>
<td>DIET and DIRT aka Dietary and Plaque control. Plaque contains the micro-organisms responsible for acid production that initiates dental caries. This is achieved with tooth brushing, flossing and fluoride administration. Dietary control of refined sugars is NB for preventing caries formation. Babies should not be given bottles with sugar, sweetened drinks or sweetened dummies. Babies should be encouraged to drink from a cup from the age of 1 year. Sucrose is the most cariogenic followed by glucose and fructose.</td>
</tr>
</tbody>
</table>
Introduction to dermatology
Eczema
Bacterial diseases involving the skin
Viral infections
Fungal infections
Infestations
Immunologically mediated skin disorders
Genetic disorders
Miscellaneous skin disorders
**Dermatological Disorders**

When examining a child for a rash, the whole skin surface should be inspected in good light. The differential diagnosis depends on the primary lesions, their distribution, secondary effects and any associated features.

Primary lesions include:
- Erythema, a red discolouration
- Macule, a flat skin discolouration
- Papule, a small lump or nodule
- Vesicle, a small, clear blister
- Pustule, a yellowish blister containing pus
- Scaling, rough and flaking skin

Secondary effects include:
- Scratch marks
- Thickening of the skin (lichenification)
- Infection
- Pigment changes

The mouth, throat, ears and perineum must also be examined. Bacterial infections, eczema and scabies are the most common causes of rashes in children.

**Eczema (Dermatitis)**

- Most common skin disease
- Def: an inflammatory skin disease affecting mainly the epidermis, resulting in the formation of an intra-epidermal vesicle in the acute stage.
- Clinical signs in eczema:
  - Acute: oedema, erythema, vesicles, weeping
  - Subacute: papules, scaling
  - Chronic: papules, lichenification, scaling
  - 2° changes: excoriations, pigment changes, 2° infection

**Causes of eczema:**
- Dryness of the skin
- Infection
- Atopic eczema: there is often a history of hay fever, asthma and urticaria in the family. A papular and scaling eruption usually starts on the cheeks and extensor parts of the limbs, but may become generalised. Itch is usually severe and excoriations and secondary infection are common. It commonly occurs at the age of 3 months and recurs in a fluctuating manner. In older children the eczema tends to become localised in the flexures of the elbows and knees or around these and other joints.
- Seborrhoeic eczema: often familial and consists of yellowish scales on the scalp, so-called cradle cap. The flexural areas are commonly affected. The lesions often become moist and secondarily infected. It seldom persists beyond the 1st year of life.
- Contact dermatitis
- Napkin dermatitis (nappy rash): due to soap powders, rinses, or infrequent changing of soiled nappies. The skin may be red, glazed and shiny, or have an eczematous appearance. In severe cases, erosions, ulcers and nodular lesions may occur.

**Treatment:**
- Relieve dryness of skin by using an ointment such as ung. Emulsificans BP
- Treat secondary infection with a combined antibiotic-steroid cream or systemic antibiotics if necessary
- Moist, weeping areas should be treated with lotions or wet dressings
- Topical corticosteroids
- In the case of nappy rash, stop the use of nappy creams and disposable diapers. Change nappies frequently.
**Bacterial Diseases involving the Skin**

**Staph and Strep infections**
Staph infections result in the formation of pus and tend to be localised. Strep infections tend to spread diffusely with very little suppuration.

**Impetigo**
- Caused by staph and strep infections
- Round, confluent, superficial blisters which rupture early and form crusts
- Treatment: topical antibiotics and antiseptics, together with systemic antibiotics in severe or widespread infections

**Skin Eruptions Secondary to Strep Tonsillitis**
- Desquamation of the skin, esp. palms and soles is thought to be due to a previous asymptomatic infection which produces an erythrogenic toxin
- Fine rash, consisting of very small, diffuse, superficial papules together with a fever, may be due to strep tonsillitis
- Treatment: oral antibiotics

**DIC**
- Most commonly caused by gram-negative septicaemia but can also be caused by staph and strep infections
- Angulated, purpuric macules which may change to large blisters and skin infarcts

**Syphilis**

**Congenital**
- Widespread, scaling, maculopapular eruptions, mucous patches in the mouth, and moist condylomas in the perineum

**Venereal**
- Primary chancre with enlarged, rubbery regional lymph nodes suggest sexual abuse

**Tuberculosis**
- Skin lesions caused by direct infection with mycobacterium tuberculosis or hypersensitivity reactions called tuberculides.
- Confirmed with biopsy and a tuberculin test

**Tuberculous chancre**
- Extremely rare
- Nodule which ulcerates, together with enlargement of regional lymph glands

**Lupus vulgaris**
- Most common
- Lesions may be single or multiple and occur anywhere. May present as soft, flat or watery plaques or raised spongy nodules

**Tuberculides**
- Multiple, disseminated lesions resulting from an underlying tuberculous focus
- Due to hypersensitivity reactions to M. Tuberculosis
- 3 clinical types: papulonecrotic tuberculides, lichen scrofulosorum, nodular tuberculides

**Viral Infections**

**Herpes simplex**
- In infants, primary infection usually results in gingive-stomatitis; and fever blisters with clusters of vesicles grouped together on a red base in older children
- Should be considered in any vesicular rash of unknown cause
- Treatment: drying, antiseptic applications. Antibiotics only if 2° infection. Severe and generalised infections should be treated with oral or IV acyclovir

**Herpes zoster**
- Grouped vesicles arranged along the cutaneous distribution of a spinal nerve
- Heal within 2-3 weeks

**Hand, foot and mouth disease**
- Due to coxsackie virus type A
- Round/oval vesicles commonly on mouth, palms and soles
- Self-limiting

**Molluscum contagiosum**
- Caused by pox virus
- Numerous, umbilicated, dome-shaped papules containing a cheesy material
- Treatment: liquid nitrogen/ tretinoin/ benzyl benzoate emulsion

**Warts**
- Caused by HPV
- Warts may be single or multiple with a papillomatous appearance. Plane warts are small and flat and occur on the face and limbs. Condylomata acuminate occur on the genitalia and have a moist, vegetating, papillomatous surface. Sexual abuse should be considered
- Treatment: benzyl benzoate emulsion; podophylin

**Fungal infections**

**Candidiasis**
- Small, outlying pustules or macules with a peripheral scale surrounding red, glazed or moist skin
- Caused by candida overgrowth after systemic antibiotics, usually presenting with thrush in the mouth and GI tract
- Treatment: oral nystatin, as well as topical applications

**Ringworm**
- Commonly caused by T. violaceum and M. canus
- Scalp ringworm: patchy or diffuse areas of scaling and hair loss
- Body ringworm: ring-shaped lesions with active, raised, scaly margins which spread outwards
- Treatment: topical, anti-fungal preparations (imidazoles)

**Sporotrichosis**
- Chronic, purulent, ulcerative or granulomatous skin lesions
- Treatment: potassium iodide

**Infestations**

**Scabies**
- Caused by direct skin-to-skin contact with an infected person
- Small vesicles and short, superficial burrows often seen between fingers; small superficial papules on the trunk and limbs. Larger nodular lesions are sometimes found in the axillae, groins and genitalia. Lesions heal to form small white spots with a dark rim.
- Treatment: application of a scabicide over the entire skin surface from the neck to toes

**Sandworm**
- Lesions are itchy, superficial, large winding burrow. Secondary infection is common
- Caused by larvae of the cat and dog hookworm
- Treatment: albendazole

**Myiasis**
- Red, painful nodules
- Caused by larvae from the fly C. Anthropophag
- Treatment: Vaseline

**Lice**
- Treatment: shampoo with gamma benzene hexachloride

**Immunologically mediated skin disorders**

**Urticaria**
- Itchy, erythematous, papules and plaques which arise within seconds and disappear within hours. Lesions can be flat, red papules or large, confluent plaques or rings.
- Due to oedema, resulting from increased permeability of blood vessels following the release of histamine. When deeper vessels are involved, diffuse, ill-defined swellings result called angio-oedema and commonly involve the eyelids or lips.
- Common causes are infections, parasites, drugs and foods
- Treatment: oral antihistamines/ systemic antibiotics/ systemic corticosteroids

**Erythema multiforme**
- Small papules or vesicles which spread outward to form one or more concentric rings with dark centres (target/iris lesions). The rash is symmetrical and occurs mainly on the face and extensor surfaces of the limbs.
- Caused by drugs, bacterial and viral infections
- Treatment: systemic antibiotics corticosteroids for severe cases

**Erythema nodosum**
- Painful, deep nodules on the lower legs; usually bilateral. Overlying skin may be red and oedematous.
- Commonly caused by strep infection

**Genetic disorders**

**Ichthyosis**
- A group of inherited disorders of the epidermis characterised by dryness and scaliness
- Treatment: ung. Emulsificans BP

**Epidermolysis bullosa**
- Blisters under the epidermis
- Commonly seen in black neonates

**Miscellaneous Skin Disorders**

**Erythema toxicum**
- Small pustules on an erythematous base; mainly on the trunk and proximal parts of the limbs
- Cause is unknown and no treatment is required

**Pityriasis rosea**
- Usually confined to trunk and proximal parts of limbs but may be widespread involving the face and scalp.
- Small papules which enlarge to form oval macules covered with fine, superficial scales. On the back and chest they are arranged in lines which follow the direction of the ribs
- Treatment: bland cream with or without corticosteroid

**Psoriasis**
- Chronic, recurrent skin disease
- Raised, well-circumscribed plaques with shiny scales; become white and opaque if scratched. Lesions are usually large and round and occur over the knees, elbows and scalp.
- The cause is unknown.
- Treatment: corticosteroid ointment/ coal-tar containing ointments
Lichen planus
- Unknown cause
- Purplish, wee-demarcated polygonal flat-topped papules with a shiny scaly surface
- Lesions are itchy and sometimes spread along scratch marks (Koebner phenomenon)
- In the mouth it appears as milky-white lines or spots; the nail bed may also be involved
- Treatment: CS creams and occlusive dressings

Vitiligo
- Well-circumscribed symmetrical white patches with hypopigmentation
- Associated with auto-immune conditions
- Treatment: CS creams

Acne
- Seldom a problem before puberty

Alopecia areata
- May be auto-immune in origin
- Round, well-circumscribed areas of hair loss; most commonly in scalp
- Hair usually regrows within a few months but sometimes the patched recur and spread
- Treatment: CS creams/ tretinoin
Orthopaedic Disorders

Disturbance of Gait
Painful joints
Pyogenic arthritis
Non-pyogenic arthritis
Joint and spinal TB
Acute haemarthrosis
Perthe's disease
Osteomyelitis
Blount's disease
Congenital talipes equinovarus
Developmental dysplasia of the hip
**Orthopaedic disorders**

Patients with orthopaedic problems usually present with the following complaint:
- Abnormal gait or limp
- Swelling of a limb and/or pain
- Weakness of a limb
- Stiffness of a limb
- Spinal pain or tenderness
- Limb or spinal deformities

**Disturbance of Gait**

This may be due to ataxia or to a limp. Ataxia is of neurological origin and the patient has an uncoordinated, awkward, unbalanced wide based gait.

Limp – This may be due to:

- Shortening of a limb – caused by
- A true discrepancy in length (measured from ASIS to the medial malleolus)
- Adduction deformity of the hip causing a pelvic tilt

**Painful joint**

Muscle weakness – e.g. weak tibialis anterior will cause a drop foot deformity with a high stepping gait.

Joint stiffness (ankylosis)
- Intra-articular due to joint disease – whole range of movement impaired.
- Extra-articular – Due to contractures from muscle imbalance as seen in poliomyelitis.
  Only affects some movements.

NB – sudden onset of a limp due to pain can be the first sign of joint infection.

**Painful joints**

Commonly caused by: Trauma, Pyogenic arthritis, non-pyogenic arthritis, TB, arthritis, Juvenile rheumatoid arthritis, Perthes’ disease, Haemophilia (least common)

**Pyogenic arthritis**

The joint is invaded by a penetrating wound, blood spread or from an adjacent osteitis.

Pathology: The disease goes through 3 stages:
- Pyo – arthrosis. The joint becomes filled with pus
- Suppurative synovitis. The synovium and capsule become thickened and inflamed
- Suppurative arthritis. The inflammation spread across the articular cartilage, destroying it.

Later with healing fibrous or bony ankylosis may occur.

<table>
<thead>
<tr>
<th>Age</th>
<th>Organism</th>
<th>Antibiotic choice</th>
</tr>
</thead>
<tbody>
<tr>
<td>0 – 6 months</td>
<td>S. aureus, E. coli</td>
<td>Ciprofloxacin + Tobramycin</td>
</tr>
<tr>
<td>6 – 36 months</td>
<td>S. aureus, H. influenza</td>
<td>Ciprofloxacin + Ampicillin</td>
</tr>
<tr>
<td>&gt; 36 months</td>
<td>S. aureus, Staphylococci</td>
<td>Ciprofloxacin + Piperacillin</td>
</tr>
<tr>
<td>adult</td>
<td>S. aureus, N. gonorrhoeae</td>
<td>Ciprofloxacin, Ceftriaxone</td>
</tr>
</tbody>
</table>

TB can also affect the joints. Other – Salmonella, S. Schenkl, B. burgdorferi.

**Clinical picture:**

- Severe fever, tachycardia
- Severely painful, swollen, warm joint. Commonly affects the hip in children.
- Decreased ROM often completely absent due to pain and spasm (pseudoparalysis).
- In neonates septic arthritis may be painless. The emphasis is on sepsicaemia. Infection is suspected but it can be anywhere. Keep IV sites and the umbilical cord in mind.

**Investigations**

- WCC, CRP and ESR are usually raised
- Blood culture may indicate the causative organism – usually S. Aureus.
- X-ray. Early x-rays may show no abnormalities – there may be some increase in joint space and soft tissue swelling. After + 10 days osteoporosis, erosion and joint space narrowing can be seen.
- Joint aspiration and immediate mc+s can confirm the diagnosis.

**Treatment**

Treatment is started without further delay. Don’t wait for x-ray confirmation.
- IV antibiotics should be started. If gram positive organisms – Ciprofloxacin. If in doubt a 3rd generation cephalosporin will cover both gram negative and positive. IV administration is given for at least a week (up to 3) followed by oral administration.
- Treatment must be given for at least 6 weeks.
- Blood transfusion and other I.V. fluids may be needed.
- The affected limb should be splinted so that it does not become permanently stiffened.
- Drainage under anaesthesia or in a superficial joint it can be achieved by needle aspiration and irrigation.

**Complications**

- Early – Septic dislocation or AVN of the femoral head
- Late –Physeal and epiphyseal destruction, Ankylosis – (least common)

**Non-pyogenic arthritis**

In children a number of infections can cause this: Rubella, measles, pneumonia, typhoid or brucellosis.

There is pain and effusion of one or more joints

Usually responds to analgesia and aspiration.

**Joint and spinal TB**

Pathology:

It usually spreads through the blood to the synovium or nearby metaphysis, but less often it is transmitted directly from the bone. From here it spreads across the articular surface, destroying it and invading the subchondral bone. There is a chronic inflammatory reaction which leads to granuloma formation and cession. Spreading into soft tissue leads to a subacute abscess (cold abscess). The disease can still be cured medically if the disease is in the synovial stage.

Once bony erisions and destruction sets in, the granulation tissue converts to fibrous tissue and traps bacilli within. The bacteria may remain quiescent for years. Healing is by fibrosis resulting in a fibrous ankylosis. Injury could cause a flare up.

The knee, hip, elbow and wrist are most commonly affected.
The synovium of a tendon sheath can also become infected. Spinal Tb usually begins in the anterior part of the vertebral body. After progressive bone destruction it spreads to an adjacent vertebral body. The 2 collapse forward causing a sharp angulation of gibbus—usually in the lower thoracic or lumbar spine.

Clinical features
- Slight pain
- Swelling, stiffness and deformity. Swelling is due to synovial thickening, not effusion.
- Decreased ROM
- Muscle wasting
- Necrotic material may form a cold abscess. This may break through the skin causing a chronic discharging sinus.
- In spinal TB pain may be deceptively slight and the patient may have no signs until there is a visible abscess or gibbus. Occasionally a patient presents with weakness or loss of sensation in the lower limbs. If treatment is delayed this may progress to paraplegia (Pott's paraplegia).

Investigations
- Raised ESR
- Tuberculin test is positive
- X-ray – Erosion on both sides of the joint and joint space narrowing. Cystic lesions may occur in the bone. Look for paravertebral abscesses in spinal TB.
- Synovial biopsy or examine synovial fluid.

Treatment
- General – Bed rest, high-protein diet and anti-TB drugs for 6 months or more. TB drugs are given until the ESR normalises and x-ray shows evidence of healing.
- The joint must be splinted and rested in the position of function
- Surgery may be needed to drain a cold abscess or to to arthrodese a damaged joint.

Acute haemarthrosis
This can either be post-traumatic or due to a haemophilic bleed. The history is helpful and joint aspiration will resolve any doubt. Treatment is to stop the bleeding by appropriate factor replacement, compression bandages and a back splint.

Recurrent intra-articular bleeding leads to synovitis and later to joint surface destruction.

Perthes disease
This condition of a painful hip joint is due to AVN of the femoral head. Uncommon in black children.

Osteomyelitis
This needs early and intensive therapy. It may be caused by:
- Blood spread organisms – most commonly S. Aureus
- Direct spread – occurs from a wound. – staph, strep, e.coli, H. influenza or proteus.
- Patients with sickle cell anaemia are prone to develop salmonella bone infections.

Pathology
In the haematogenous type, infection begins in the metaphysis. Pus forms, the intraosseous pressure rises causing intense pain and obstructing blood flow. By the 2nd day the pus is in the medulla and forces its way along the Volkmann canals to the surface where it lifts the periosteum and forms a subperiosteal abscess. It then spreads along the shaft or bursts into the soft tissues. In infants it often extends into the epiphysis and then into the joint. If the pressure is not relieved necrosis will set in because of the decreased blood supply and thrombosis due to vascular stasis. This dead bone forms a sequestrum and the elevated periosteum forms a new layer of bone called the involucrum.

Clinical features:
- Pain, malaise and fever. In neglected cases toxaemia. There may be a history of a skin lesion, injury or sore throat.
- Warm, tender and swollen
- Decreased ROM
- In neonates – failure to thrive, irritable. Pseudoparalysis. Associated with septic arthritis.

Investigations
- Increased WCC and positive blood culture
- X-rays. The first 10 days shows no abnormalities. After +10 days -- rarefaction of the metaphysis and new bone formation. If treatment is delayed bone might have a ragged appearance.

With healing there sclerosis and thickening of the cortex. Sequestra can be seen.
- Radio isotope scan

Treatment
See septic arthritis
Remember adequate analgesia.
If pyrexia and local tenderness persists for longer than 24hr after treatment with antibiotics drainage will be needed.

Chronic osteitis is the result of inadequate treatment of acute osteomyelitis. The changes are incurable and permanent. The disease is liable to acute exacerbations with pain swelling and pyrexia. In these cases antibiotics may subdue the infection but an abscess must be drained and the sequestra removed.

Blount's disease
Is one of the causes of genu varum and is seen mainly in black children.

Pathology
The posterior medial part of the tibial metaphysis doesn't develop normally and with growth causes the bone to angulate medially.

Clinical features
Children are often overweight and start to walk early
Medial angulation of one or both legs (bowing of the legs). Otherwise the child is normal.
X-rays show abnormal flattening of the medial half of the epiphysis.
Treatment
Corrective osteotomy – not done before the age of 4.

Congenital talipes equinovarus
1/1000 births, 50% bilateral, m>f, severity F>r
3 parts to deformity
Talipes – talus is inverted and internally rotated
Equines – ankle is planter flexed
Varus – heel and forefoot are in varus

Causes:
• May be idiopathic, neurognic or syndrome-associated
• Examine the back for dysraphism
• Examine the hips for DDH
• Examine the knees for deformity

Clinical features:
• The deformity is usually obvious at birth. The foot is both turned inward the
  that the sole faces posteromedially.
• In a normal baby the the foot can be dorsiflexed and everted until the toes almost touch
  the front of the leg. In club-foot there is considerable resistance and sometimes the
  deformity is fixed.
• Always look for DDH and spina bifida.

Types:
Extrinsic: It corrects easily with passive manipulation and probably due to intrauterine position.
Intrinsic: More difficult to correct. It has a worse prognosis.

Treatment:
• Manipulation and adhesive strapping. Parents should be taught.
• Application of plaster-of-paris cast
• Splintage
• Surgery – mostly for relapses – lizarov fixator.
• Relapses may be prevented by using a Dennis Browne boot

Developmental dysplasia of the hip
There is partial or complete displacement of the femoral head from the acetabulum. It is due to
ligamentous laxity and abnormal sloping of the acetabular roof.

Predisposing factors – family history, breech presentation, postnatal factors

Diagnosis
• Diagnosis is clinical
• Limited abduction of the flexed hip (<50-60 degrees)
• Ortolanis test
• Barlows test
• Galeazzi sign

Treatment
• Tendelenburg test and gait useful when children > 2 years.
• X-ray and ultra sound are helpful
Disorders
Metabolic
Metabolic Disorders

- Acid-Base
- Blood sugar disturbances
- Electrolyte disturbances
- Metabolic bone diseases
  - Vit D
  - Rickets
  - Osteoporosis
  - Osteosclerosis

Metabolic disorders
Metabolic disorders:

- Acute acquired metabolic disturbances
- Inherited metabolic disorders
- Disorders of acid-base regulation
- Disorders in glucose metabolism
- Electrolyte disturbances

Disorders of acid-base regulation:

**Structured Approach to Diagnosis of Disturbances of Acid-Base Balance**

**First: Initial Clinical Assessment**
A clinical assessment based on history and clinical examination is essential.

**Second: Acid-Base Diagnoses**
Perform a systematic evaluation of the blood gas and other results and make an acid-base diagnosis.

**Finally: Clinical Diagnosis**
Synthesize the information to make an overall clinical diagnosis.

**How to evaluate the blood gas:**
1. Assess deviation of pH from normal
   - Normal pH = 7.4 ± 0.02
   - Increased pH = alkalaemia, decreased pH = acidemia
   - Normal pH = acid-base disorder or mixed acid-base disorder with opposing disorders
   - Note: except for chronic respiratory alkalosis, simple acid-base disorders do not fully compensate
2. **Pattern:** check pattern of pCO2 (if deranged = respiratory disturbance) and HCO3- (if deranged = metabolic disturbance).
3. **Compensation:** assess appropriateness of compensation

**Simple Acid-Base Disorders**

<table>
<thead>
<tr>
<th>Disorder</th>
<th>pH</th>
<th>Clinical examples</th>
</tr>
</thead>
<tbody>
<tr>
<td>Respiratory acidosis</td>
<td>&lt; 7.35</td>
<td>Laryngeal oedema, bronchospasm, asthma, brainstem herniation, sedatives, muscle paralysis</td>
</tr>
<tr>
<td>(CO2 retention by lungs)</td>
<td></td>
<td></td>
</tr>
<tr>
<td>Metabolic acidosis</td>
<td>&lt; 7.35</td>
<td>Shock, hypoxia, DKA, starvation, glycogen storage diseases, Salicylate poisoning, NH4Cl administration</td>
</tr>
<tr>
<td>(Lactic acidosis)</td>
<td>7.35</td>
<td>Mapa syrup urin disease</td>
</tr>
<tr>
<td>(Keto-acidosis)</td>
<td>7.35</td>
<td>Salicylate poisoning, NH4Cl administration, Renal failure, RTA, renal immaturity</td>
</tr>
<tr>
<td>(Organic acidemia)</td>
<td></td>
<td>Acute diarrhea, Acetazolamide treatment</td>
</tr>
<tr>
<td>(Exogenous acids)</td>
<td></td>
<td></td>
</tr>
<tr>
<td>(Reduced H+ excretion)</td>
<td></td>
<td></td>
</tr>
<tr>
<td>(Gastro-intestinal HCO3 losses)</td>
<td></td>
<td>Acute diarrhea, Acetazolamide treatment</td>
</tr>
<tr>
<td>(Renal HCO3 losses)</td>
<td></td>
<td></td>
</tr>
<tr>
<td>Respiratory alkalosis</td>
<td>&gt; 7.45</td>
<td>Salicylates, Emotional, brain stem involvement</td>
</tr>
<tr>
<td>(Hyperventilation – CO2 exhalation)</td>
<td></td>
<td></td>
</tr>
<tr>
<td>Metabolic alkalosis</td>
<td>&gt; 7.45</td>
<td>Vomiting eg. pyloric stenosis, gastric drainage</td>
</tr>
<tr>
<td>(H+ losses from gut)</td>
<td></td>
<td>Purosemede</td>
</tr>
<tr>
<td>(H+ losses from kidneys)</td>
<td></td>
<td>Hypochloremia, hypokalaemia, hyperaldosteronism</td>
</tr>
<tr>
<td>(Excess HCO3 retention from kidneys)</td>
<td></td>
<td>Bicarbonate, citrate or lactate administration</td>
</tr>
<tr>
<td>(Exogenous alkali)</td>
<td></td>
<td></td>
</tr>
</tbody>
</table>

**Some Add to Interpretation of Acid Base Disorders**

<table>
<thead>
<tr>
<th>Clue</th>
<th>Significance</th>
</tr>
</thead>
<tbody>
<tr>
<td>High anion gap</td>
<td>Always strongly suggests a metabolic acidosis</td>
</tr>
<tr>
<td>Hyperglycaemia</td>
<td>If ketones also present in urine -&gt; DKA</td>
</tr>
<tr>
<td>Hyperkalaemia and/or hypochloremia</td>
<td>If ketones absent -&gt; Hypoosmolar non-ketotic state</td>
</tr>
<tr>
<td>Hypochloremia</td>
<td>Suggests metabolic alkalosis</td>
</tr>
<tr>
<td>Elevated creatinine and urea</td>
<td>Common with normal anion gap acidosis</td>
</tr>
<tr>
<td>Elevated creatinine</td>
<td>Suggests azotemic acidosis or hypovolaemia (prerenal renal failure)</td>
</tr>
<tr>
<td>Elevated osmolar gap</td>
<td>Consider ketoacidosis: ketones interfere in the laboratory method (Jaffe reaction) used for creatinine measurement &amp; give a falsely elevated result.</td>
</tr>
<tr>
<td>Anion Gap (AG) = (Na+ + K+) – (Cl- + HCO3-), normal values = 7 – 14 mmol/L</td>
<td></td>
</tr>
</tbody>
</table>

1. Causes of low AG
   - Hypoalbuminemia
   - Myeloma
   - Laboratory error
2. Causes of high AG metabolic acidosis (MUDPILES)
- Renal failure
- Diabetic ketoacidosis
- Ethanol, methanol, ethylene glycol, salicylate
- Lactic acidosis

Blood sugar disturbances

Hypoglycaemia

Definition: A blood glucose level of less than 2.2 mmol/l
Pathology: Glucose utilization occurs DIRECT (RBC’s, brain, liver) or INDIRECT (under influence of insulin). Glucose is needed to facilitate normal respiration.
Causes:
- Decreased production
  - SUBSTRATE DEFICIENCY: Small for GA, PEM, chronic diarrhea, starvation
  - DEFECTIVE GLUCONEOGENESIS: Glycogen storage disease
  - DEFECTIVE GLUCOGENOLYSIS: Endocrine or enzyme deficiency
  - HEPATIC DAMAGE: Liver disease or failure, impella or salicylate poisoning, alcohol, Jamaican vomiting sickness
  - IDIOPATHIC

Increased utilization

HYPERINSULINISM: Infant of diabetic mother, Beckwith syndrome, iatrogenic, insulinoma, Nesidioblastoma

Clinical presentation:
- In neonates: non-specific – lethargy, poor feeding, convulsions, apnoea attacks, hypotonia. (Even asymptomatic hypoglycaemia can impair brain development in the neonate.)
- In infants:
  - An acute episode: hunger, weakness and a sympathetic effect – pallor, tachycardia, sweating, drowsiness, coma, convulsions.
  - A gradual drop in blood sugar: malnourishment with limited signs until neuroglycopenia (coma, convulsions).

Management:
- If symptomatic or confirmed; 1 g/kg or 2 ml/kg of a 10-20% glucose solution IV, if no line available give 10-20% glucose solution at 1 g/kg per NG tube.
- Do serial blood glucose checks to confirm appropriate rise.
- If recurrent, unexplained episodes, investigate for metabolic disorder.

Hyperglycaemia (discussed further in endocrine section under DM)

Causes:
- Insulin deficiency: Diabetes Mellitus
- Impaired uptake: Hypokalaemia, sick-cell deficiency
- Increased gluconeogenesis: steroid therapy, cushing’s.
- Increased glucogenolysis: stress mediated catecholamine release
- Iatrogenic

Management:
- Treat underlying cause.
- If metabolic acidosis: Give 8.4% NaHCO3 (contains 1 mmol HCO3 and 1 mmol Na+/ml), IV slowly, according to the formula:
  BASE DEFICIT x (0.3) x BODY MASS = MMOL HCO3 REQUIRED
  (rapid administration or >8 mmol/kg/day = hypernatraemia, hypokalaemia, hypercalcaemia, fluid overload.
- If metabolic alkalosis: Give 5% NH4CL (contains mmol/ml) according to the formula:
  BASE DEFICIT x (0.3) x BODY MASS = MMOL NH4CL REQUIRED
## Electrolyte Disturbances:

**Hypokalaemia**
- Causes: (1) Treat if $K^+ > 6.5$ or symptomatic or ECG-changes: 2) Eliminate all $K^+$ intake 3) Correct metabolic acidosis with NaHCO₃ 4) Dilute $5mg$ of $24ml$ Salbutamol in $0.9%$ Saliue and Nebulise over 20 mins 5) Inject $10\%$ Ca-gluconate, $1ml/kg$ IVI slowly 6) Give $1g/kg$ IVI glucose over 1 hour and add 1 unit of Insulin for every $3g$ of glucose 7) Give Sodium Polystyrene Sulphonate $1g/kg$ PO or PR 8) Give Furosemide $1mg/kg$ IVI 9) Last resort consider peritoneal dialys 10) Treat if $K^+ < 2.5$ or symptomatic or ECG-changes 2) Give $3 - 6mEq/KCl/kg/day PO$ 3) If severe losses eg Bartter's, give up to $10mEq/kg/day$, but never exceed an IV dose of $40mEq/KL (1mI 15%KCl = 2mmol Kcl)$

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**Hyperammonaemia:**
- Causes: liver failure, liver immaturity with excessive protein intake, inherited disorders of metabolism  
  Clinical presentation: hepatic encephalopathy (vomiting, confusion, coma, seizures)  
  Management: restrict dietary proteins, give $250mg/kg/day$ IV sodium Benzoate
Hyperphosphataemia

Pathology: A raised phosphate level leads to a decreased serum calcium level which causes soft tissue calcifications and secondary hyperparathyroidism (PTH).

<table>
<thead>
<tr>
<th>Causes</th>
<th>Management</th>
</tr>
</thead>
<tbody>
<tr>
<td>Increased intake</td>
<td>1) Decrease dietary intake</td>
</tr>
<tr>
<td>Decreased renal excretion</td>
<td>2) If caused by increased renal excretion:</td>
</tr>
<tr>
<td></td>
<td>give AIOH or Mg-trisilicate</td>
</tr>
</tbody>
</table>

Causes of hypophosphataemia:
- Increased intake
- Decreased renal excretion
- Cow's milk feeds
- Enemas containing phosphate

Clinical presentation:
- Soft tissue calcification
- Low s-Ca²⁺
- High s-PTH

Management:
1) Decrease dietary intake
2) If caused by increased renal excretion: give AIOH or Mg-trisilicate

Hypercalcaemia

Pathology: Symptoms occur with a decrease in the IONIZED fraction of total serum-Ca²⁺ (accounts for half of s-Ca²⁺ measured if the albumin is normal). This results in neuromuscular, cardiac, eye and skin changes.

Causes: Lack of intake: neonatal hypocalcaemia

Clinical presentation: Neonatal: non-specific: tremors, cyanotic attacks, metabolic disturbances

Management: 1) Reverse underlying cause
2) Give 2ml/kg 10%Ca-glucorate IV slowly while monitoring heart rate to avoid a bradycardia
3) Oral Ca²⁺ supplements

Hypomagnesaemia

Pathology: Mg is an essential co-factor for many enzyme systems and is also required for the normal release of PTH.

Causes:
- Decreased intake: PEM
- Increased losses: chronic diarrhoea, diuretic therapy

Management: 250 – 500mg MgSO₄ IM or IV for 3-5 days.

Inherited Metabolic Disorders

Definition: Genetically determined deficiency of enzymes or co-factors that can be classified into 3 groups:

Group 1:
- Problem with the synthesis or catabolism of large molecules
- Presenting periodically with accumulation of toxic compounds proximal to the metabolic block
- Eg. disorders of gluconeogenesis or mitochondrial disorders

Group 2:
- Inborn errors of intermediate metabolism,
- Presenting with accumulation of toxic compounds proximal to the metabolic block
- Eg. amino-acidopathies, sugar intolerance, PKU

Group 3:
- Disorder of deficient energy production
- With accumulation of toxic products and lack of energy
- Eg. disorders of gluconeogenesis or mitochondrial disorders

Clinical presentation:
- Vomiting and acidosis after feeding initiation (amino acid (M) or carbohydrate (CHO) metabolic disorder)
- Hepatosplenomegaly (metabolites accumulate in the liver)
- Neurologic syndrome: acute and chronic encephalopathy, mental retardation (MR), megalencephaly
- (Mucopolysaccharide disorders)
- Severe acidosis (aminoaciduria)
- Hypertension (renal cycle and organic acid disorders)
- Growth retardation
- Seizures
- Hypoglycaemia
- Family history of early infant death

Examination – clues:
- Odour: burnt sugar, sweaty feet, musty, ammonia-like
- Skin: hypohyperpigmentation, rash, ichthyosis, xanthomas
- Hair: alopecia, hirsutism, abnormal architecture, fair colouring
- Eyes: cornea (clouding, crystals), lens (cataracts, dislocation), retina (macular cherry red spot, pigment retinopathy, optic atrophy)
Screening of a newborn

<table>
<thead>
<tr>
<th>BLOOD</th>
<th>URINE</th>
</tr>
</thead>
<tbody>
<tr>
<td>Glucose</td>
<td>Colour</td>
</tr>
<tr>
<td>Electrolytes (FBC, UCE)</td>
<td>Smell</td>
</tr>
<tr>
<td>ABG – acid-base</td>
<td>Reducing substances</td>
</tr>
<tr>
<td>Ca, P, Mg</td>
<td>Ketotest</td>
</tr>
<tr>
<td>Phenistick</td>
<td>2,4-DNP-hydralazine</td>
</tr>
</tbody>
</table>

Preliminary Management Plan in a suspected metabolic disorder:
1. Total protein restriction
2. Maintain high urine output
3. Intake of 150ml/kg/day with bicarb for alkaline urine
4. Give dextrose
5. Give Multivitamins
6. Give Sodium benzoate for hyperammonaemia

PHENYLKETONURIA (PKU)

1 in 12,000
deficiency of phenylalanine hydroxylase prevents conversion of phenylalanine to tyrosine and subsequent build up toxic metabolites phenylactic acid and phenyllactic acid
symptoms seen later in infancy and during childhood
mothers who have PKU may have infants with congenital anomalies

Presentation
mental retardation, neurological symptoms (hypertonic, tremors, behaviour disorders), skin hypopigmentation

Treatment
PKU screened at birth
dietary restriction starting at one month of age

Suspect Metabolic Bone Disease IF:
Bone deformities: due to path fractures (all MBD's) or growth plate disturbances (Rickets)
Short stature: in long standing MBD's
Hypotonia and proximal muscle weakness: due to Vit D deficiency (used to diff between Vit D deficient and hypophosphataemic rickets)
Delayed developmental milestones esp. gross motor: due to hypotonia and muscle weakness
Convulsions and other stigmata of hypocalcaemia: early stages of Vit D deficiency in young infants or in severe rickets
Acidoses: soft ribs and thoracic cage abnormalities
Recurrent lower resp tract infections: due to soft pliable ribs and ineffective ventilation (rickets and osteogenesis imperfecta)

Vitamin D metabolism

Sources of Vit D
D3 (cholecalciferol) from 7-dehydrocholesterol in skin
Under the influence of UV-light
D2 (ergocalciferol) dietary source
From irradiated plant material

Stored in muscle or fat and then transported to the liver
Hydroxylated to 25-hydroxy-cholecalciferol in the liver (inactive metabolite)

Transported to the kidney under the influence of PTH or hypophosphataemia and hydroxylated to 1-25-dihydroxy-cholecalciferol (active metabolite) by the enzyme 1-α-hydroxylase

1-25-dihydroxy-cholecalciferol (CALCITRIOL) plays central role in Ca²⁺ homeostasis by working synergistically with PTH

In the KIDNEY: increase Ca²⁺ reabsorption, decrease PO₄ and HCO₃ reabsorption
In the BONE: increased Ca²⁺ release and osteoclastic reabsorption
In the GUT: increased Ca²⁺ and PO₄ absorption

And one more thing about Ca²⁺ that is probably useful:
Remember that s-Ca²⁺ levels vary according to the s-albumin levels, thus s-Ca²⁺ measurements should be CORRECTED:
If s-alb<40 = [Ca²⁺] + 0.02 x (40 – [alb]) = corrected Ca²⁺ in mmol/l
If s-alb>45 = [Ca²⁺] - 0.02 x ([alb] – 45) = corrected Ca²⁺ in mmol/l

Causes of Vit D deficiency:
- Lack of sunlight exposure due to environment, cultural traditions or weather
- Increased skin pigmentation
- Vegetarian diets
- Prolonged breast or cow milk feeding without adding Vit D
- Infants delivered to Vit D-deficient mothers (inactive metabolite crosses placenta)

Rickets
DEFINITION: a MBD where there is a failure in or delay in mineralisation of uncalcified osteoid, resulting in delayed mineralisation of the growth plate (only occurs in children whose epiphysis are not yet fused). The ratio of uncalcified osteoid to calcified bone is increased.

CLASSIFICATION: CALCIUM-deficiency rickets vs PHOSPHORUS-deficiency rickets

Ca²⁺-deficiency rickets
1) ABNORMALITIES in VIT D METABOLISM
2) DIETARY CA²⁺ DEFICIENCY

<table>
<thead>
<tr>
<th>CAUSES</th>
<th>Nutritional Vit D deficiency</th>
<th>Impaired Vit D absorption</th>
<th>Impaired hydroxylisation to 25-OH-Vit D</th>
<th>Increased Vit D metabolism</th>
<th>Decreased synthesis of 1-25-(OH)₂-Vit D</th>
<th>End-organ resistance to 1-25-(OH)₂-Vit D</th>
<th>Dietary deficiency of Ca²⁺</th>
</tr>
</thead>
<tbody>
<tr>
<td>PATHOLOGY</td>
<td>See above: causes of Vit D-deficiency</td>
<td>Vit D and Ca²⁺ is absorbed with mechanism similar to dietary fats – any cause of steatorrhoea = malabsorption * Coeliac disease * Biliary atresia * Liver immaturity (prem)</td>
<td>* Severe liver disease * Liver failure * Liver immaturity</td>
<td>* Anti-convulsants esp. Phenobarbital increases hepatic hydroxylation and excretion of Vit D</td>
<td>* Renal failure = renal osteodystrophy * Vit D-dependency rickets: rare, Autosomal recessive condition, absent gene coding for 1-α-hydroxylase</td>
<td>* Rare syndrome of peripheral resistance to Vit D (Vit D – dependent rickets Type 2)</td>
<td>* Diet lacking milk and other dairy products</td>
</tr>
</tbody>
</table>

<table>
<thead>
<tr>
<th>CLINICAL</th>
<th>See below: Signs of Vit D-deficiency rickets</th>
<th>See below</th>
<th>See below</th>
<th>See below</th>
<th>See below</th>
<th>See below</th>
<th>See below</th>
</tr>
</thead>
</table>

<p>| RADIOGRAPHY | See below | See below | See below | See below | See below | See below | See below |</p>
<table>
<thead>
<tr>
<th>BIO-CHEM</th>
<th>1) HypoCa2+ 2) HyperPTH 3) HypoPO4- 4) Raised ALP 5) Decreased 25-OH-VitD 6) Decreased urinary Ca2+ (&lt; 2mg/kg/24hrs)</th>
<th>Same as for VitD-deficiency</th>
<th>Same as for VitD-deficiency</th>
<th>Same as for VitD-deficiency BUT raised ALP is a poor indicator of bone loss as it could also be due to effect of Phenobarbital on the liver.</th>
<th>Raised calcium levels</th>
<th>1) HypoCa2+ 2) Normal PO4- 3) Raised ALP 4) Normal 25-OH-VitD</th>
</tr>
</thead>
<tbody>
<tr>
<td>MANAGE-MENT</td>
<td>1) Prevent: Adequate sunlight OR Fortified formula OR Supplement breast milk with 400 IU/day from birth 2) Treat: 1000 - 5000 IU/day for 3 weeks to 6 months. Add Ca2+ in early stages 3) If pt convulsing or having apneic attacks give 10% Ca-gluconate, 1-2ml/kg IV slowly (monitor pulse)</td>
<td>1) Prevent: Adequate sunlight OR Fortified formula OR Supplement breast milk with 400 IU/day from birth 2) Treat: 1000 - 5000 IU/day for 3 weeks to 6 months. Add Ca2+ in early stages</td>
<td>1) Treat: 1000 - 5000 IU/day for 3 weeks to 6 months.</td>
<td>* Renal failure: 1) Raised urea and creatinine 2) Hyper PO4- * Vit D dependency: 1) normal 25-OH-vitD 2) Very low calcium</td>
<td></td>
<td>1) HypoCa2+ 2) Normal PO4- 3) Raised ALP 4) Normal 25-OH-VitD</td>
</tr>
</tbody>
</table>

**Signs of Ca2+ deficient rickets:**

**Signs of hypoCa2+:**
- Acute: jitteriness, seizures, tetany, carpopedal spasm (flexion and adduction at the wrist and MP joints and ankles), laryngeal spasm (laryngismus stridulus = tremolous, crowing, inspiratory noise due to tetany of the vocal cords), parastesias, Chovstek’s sign (tapping of facial nerve anterior to ear = twitching of ipsilateral facial muscles), Trousseau’s sign (BP - cuff on arm inflated to just above systolic for 3 minutes = carpal spasm), arrhythmias.
- Chronic: dystrophic features: lenticular cataracts, dry and scaling skin, coarse hair, brittle nails, enamel hypoplasia of teeth.

**Signs of bone deformities:** cranialstes (ability to depress the skull bones in front of and behind the ears), delayed fontanellae closure, hot bun appearance of skull (due to frontal and parietal bossing), asymmetrical skull (infant lies mostly on one side of head), enlarged metaphysis of wrists and ankles, long bone deformity (bowing or knock knees), rickety rosy (enlarged costochondral junctions), Harrison’s sulcus (at rib margin) and Violin case upper ribs (due to diaphragm’s and intercostal muscles’ effect on pliable ribs), recurrent lower resp tract infections, acidosis

**Radiographic changes of Ca2+ deficient rickets:**
- Delay in epiphyseal development
- Widening of the growth plate (> 1 mm in distance)
- Splaying and cupping of the metaphysis
- Irregular and fraying metaphyseal ends
- Osteopenic bone with thin cortices
- Subperiosteal erosions (due to secondary PTH)

**The complicated pathogenesis of renal osteodystrophy:**

1) Decreased hydroxylation to produce 1,25-dihydroxy-Vit D 2) Decreased excretion of phosphate with secondary hypoca2+ and hyperPTH 3) Chronic acidosis aggravates bone loss

**Phosphorus-deficiency Rickets**

**Distinguished from Ca2+-deficiency by:**
- History: diet, sunlight exposure, climate
- Radiological changes:
  - No g's of secondary hyperPTH
  - No osteopenia
- Biochemistry:
  - normocalcaemia
  - normoPTH
  - normal levels of urinary Ca2+

**Notes:**
- **Phosphorus** is always elevated in chronic renal failure.
- **Calcium** is elevated in chronic renal failure.
- **Hyperparathyroidism** should not be treated unless **renal failure** is also present.
- **Vitamin D** is effective for both hyperparathyroidism and renal osteodystrophy.
- **Calcitriol** is effective for both hyperparathyroidism and renal osteodystrophy.
- **Calcium** should be given to patients with chronic renal failure who have osteoporosis.
- **Calcium** should be given to patients with chronic renal failure who have osteopenia.
- **Calcium** should be given to patients with chronic renal failure who have osteomalacia.
- **Calcium** should be given to patients with chronic renal failure who have osteodystrophy.
**Causes of phosphorus deficiency rickets:**

1. **Inadequate intake or absorption**
   - Inadequate intake: intestinal phosphorus absorption is decreased.
   - Increased renal excretion: due to increased tubular reabsorption.

2. **Fanconi's syndrome**
   - X-linked or sporadic.
   - Excess renal phosphate losses in the absence of hyperPTH.
   - No amino-aciduria or glycosuria.
   - No muscle weakness.
   - TREAT (until late adolescence when epiphyses are fused):
     1. Joulies solution of 1.5 – 3g/24hrs phosphorus in 5 divided doses PO.
     2. Mx secondary hyperPTH with:
        -铝ca2+ and hyperPTH.

**Osteoporosis**

- Clinically: Will present with pathological fractures rather than bone deformities.
  (Remember that 50% of boys and 30% of girls, without bone abnormalities, will fracture a long bone during childhood).
- Biochemically: normal serum: calcium, phosphorus and ALP.
- Radiologically: Picture of osteopenia – decreased bone density (decreased radiolucency with thin cortices and trabeculae not thickest at stress points (does not adhere to Wolff's law). Total width of bone narrowed.
- Dxa (dxa scan) = energy x-ray absorptiometry measuring a decrease in bone mineral density is the gold standard but not widely available.

**Causes of osteoporosis:**

<table>
<thead>
<tr>
<th>Decreased bone matrix formation</th>
<th>Increased bone resorption</th>
</tr>
</thead>
<tbody>
<tr>
<td>Osteogenesis Imperfecta</td>
<td>Immobilization</td>
</tr>
<tr>
<td>Steroid Induced</td>
<td>Juvenile osteoporosis</td>
</tr>
<tr>
<td>PEM</td>
<td>Marrow hyperplasia eg Thalassaemia major</td>
</tr>
<tr>
<td>Vitamin C deficiency</td>
<td>HyperPTH</td>
</tr>
<tr>
<td>Copper deficiency</td>
<td></td>
</tr>
</tbody>
</table>

**Osteogenesis Imperfecta (OI):**

**DEFINITION:** Inherited (mostly autosomal dominant), MBD, characterized by an abnormality in collagen synthesis.

**CLASSIFICATION:** Divided into 7 subgroups (5-7 very rare)

<table>
<thead>
<tr>
<th>OI CLASS:</th>
<th>SIGN:</th>
<th>SEVERITY</th>
<th>INHERITANCE</th>
<th>BLUE SCLERA</th>
<th>DENTINOGENESIS IMPERFECTA*</th>
<th>PATH</th>
<th>FRACTURES</th>
<th>STATURE</th>
<th>OTHER</th>
</tr>
</thead>
<tbody>
<tr>
<td></td>
<td></td>
<td>mild, commonest</td>
<td>AD</td>
<td>present</td>
<td>in 25% fractures</td>
<td>decreases after puberty</td>
<td>normal or mildly affected</td>
<td>hydrocephalus, joint hyperlaxity, hypermetabolism (increased sweating, heart rate and temperature)</td>
<td></td>
</tr>
<tr>
<td>1</td>
<td></td>
<td>lethal</td>
<td>AD</td>
<td>present</td>
<td>birth = severe deformity</td>
<td>neonatal = death</td>
<td>shortened with triangular faces</td>
<td></td>
<td></td>
</tr>
<tr>
<td>2</td>
<td></td>
<td>severe, deforming</td>
<td>AD</td>
<td>absent</td>
<td>in 80% fractures</td>
<td>frequent</td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>3</td>
<td></td>
<td>moderately severe (more than 1, less than 3)</td>
<td>AD</td>
<td></td>
<td>in 60% fractures</td>
<td>occurs</td>
<td></td>
<td></td>
<td></td>
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<tr>
<td>4</td>
<td></td>
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</tr>
</tbody>
</table>

*teeth defects due to lack of Dentin (contains collagen), teeth translucent with frequent carries.

**RADIOGRAPHIC** mild to severe signs of osteoporosis – in severe cases the skull can show multiple wormian bones with playasia (soft cranial bones cause post. fossa to buldge upwards to foramen magnum – syn. basilar invagination).

**MANAGEMENT = MULTI-DISCIPLINARY:**

1. Paediatrician: Bisphosphonates increase bone mass and counselling.
2) Orthopedic: fractures prevented and treated promptly. Osteotomies and bone realignment surgery where necessary
3) Physio: mobilize to prevent further osteoporosis

Steroid induced: CUSHING'S SYNDROME (chronic glucocorticoid excess)
CAUSES: ACTH-dependant vs ACTH-independent:
- Ant. Pituitary adenoma (Cushing’s disease) - iatrogenic (eg treatment for Duchenne’s)
- Ectopic tumour (SCLC, bronchial ca) - Primary adrenal tumour
PATH: Primary = failure of matrix formation due to osteoblast suppression
Secondary = decreased Ca2+ absorption from GIT + immobilization from primary illness
TREATMENT: Large doses of Vit D and Ca2+ supplement from onset of steroid therapy.

Vitamin C deficiency: SCURVY
CAUSE: decreased intake in infants fed almost exclusively on cow’s milk (presents during second half of first year)
CLINICALLY: irritability, loss of appetite, tenderness of long bones progressing to refusal to stand and later refusal to move (lie with limbs in flexed position) with movements of the limbs eliciting acute pain. Also may have swelling and ecchymosis around joints.
RADIOLOGY:
- Ground glass appearance of bone shaft due to osteopenia
- Cortices thin but well defined
- Areas of rarefaction beneath otherwise well demarcated bone ends
- Areas of healing sub-periosteal haematomas (Caffey’s sign)
TREATMENT: 250mg Vit C PO daily

Copper deficiency:
Caused by prolonged IV alimentation and GIT disorders like chronic diarrhea
Present with anaemia, neutropenia and severe osteoporosis.

Juvenile Osteoporosis:
Aetiology of unknown origin
Presents like osteogenesis imperfect in the absence of a family history
Symptoms often subside at puberty
Treatment with bisphophonates appears to be useful

HyperPTH
CAUSES:
- Primary: rare, parathyroid adenoma or Ca = increased PTH production
- Secondary: Increased PTH due to decreased Ca2+ = renal failure of Vit D deficiency
- Tertiary: parathyroid glands become hyperplastic after prolonged secondary hyperPTH

CLINICAL:
BONES – hypotonia, bone pain
STONES – kidney stones associated with haematuria, frequency, renal colic
MOANS – depression, irritability, drowsiness
ABDOMINAL GROANS – decreased appetite, nausea, constipation

**Osteosclerosis**

<table>
<thead>
<tr>
<th>CAUSES</th>
<th>Osteoporosis</th>
<th>Diaphyseal dysplasia</th>
</tr>
</thead>
<tbody>
<tr>
<td>AETIOLOGY</td>
<td>Autosomal Dominant</td>
<td>Recessive</td>
</tr>
<tr>
<td>PATHOLOGY</td>
<td>Failure of osteoclast production OR osteoclast function: failure of bone remodeling with accumulation of bone</td>
<td></td>
</tr>
<tr>
<td>CLINICAL</td>
<td>- anemia, thrombocytopenia, neutropenia, increased infection susceptibility due to bone marrow suppression - deafness and blindness due to nerve compression - hydrocephalus - dental sepsis with jaw osteomyelitis</td>
<td></td>
</tr>
<tr>
<td>- milder symptoms becoming apparent in adolescence and adulthood - anaemia uncommon - nerve compression symptoms common</td>
<td></td>
<td></td>
</tr>
<tr>
<td>TREATMENT</td>
<td>Lethal if not treated with bone marrow transplants</td>
<td></td>
</tr>
</tbody>
</table>

**Diaphyseal dysplasia**

- Hyperostosis of the skull
- Bone pain
- Muscle weakness

**RADIOGRAPHY:**
- Localized areas of subperiosteal resorption esp along the metacarpals and phalanges
- Severe demineralization of the clavicles
- Dense line along teeth (lamina dura) disappears due to resorption
- Metaphysis become ragged and frayed

**TREATMENT:**
- If primary remove parathyroid glands, secondary treat cause
Nutritional Disorders

ANEMIA - PEM PTX

Assessment
PEM
Vitamins, minerals & trace elements
Nutritional Disorders

Assessment
PEM
Vitamins, minerals & trace elements
NUTRITIONAL DISORDERS

Predisposing factors to undernutrition:

- Prem babies/low birth weight babies
- Babies who are not breastfed in poor socio-econ circumstances
- Twins
- Diarrhoea / pneumonia / HIV / TB / other infections / Ca / chronic disease
- Mother with: poor social circumstances, poor health, many children or is incompetent
- Father is: unemployed, alcoholic
- Inadequate child-minded with working mother
- Home with out piped, clean water

Assessment of nutritional status:

Dietary history: obtained from care giver with sensitivity and tactfully

Clinical assessment:

Generalised malnutrition symptoms: irritability, apathy, crying, failure to thrive

1. Anthropometry
   - weight for age: in normally grown infants this reflects mainly an energy deficit
   - weight for height: indicates recent weight loss or wasting
2. Skin changes
3. Skin appendages
   - hair
   - fingers and toe nails
4. Eyes
5. Mouth and tongue
6. Subcutaneous oedema

Pathophysiology of PEM: due to profound changes in body composition

1. Total Body Water (TBW): Increases with disappearance of fat stores and muscle bulk. An increased TBW is associated with and increased extracellular fluid volume and therefore oedema. On recovery this ECF volume is taken up by cells and lost by diuresis causing the initial loss in body weight on recovery.
2. Potassium: Wasting of lean tissue and loss in diarrhoeal stools = severe hypokalaemia with secondary sodium retention and oedema. Also causes hypotonia, decreased insulin secretion and disturbed renal function.
3. Other minerals: Mg²⁺, Ca²⁺, PO₄⁻, and Fe²⁺
5. Total body protein: Non-collagen protein affected, collagen protein affected to very limited extent.
6. Plasma protein: Total albumin decreased by 50%, mostly the extra-vascular pool is affected, this decreased colloid osmotic pressure increases the oedema.
7. Body Fat: Drops to about 5% in marasmus (normal 19%).
8. Endocrine changes:
   - Pituitary: Increased human growth hormone and TSH.
   - Thyroid: Intense metabolism and growth adequately stimulated with recovery, thus no deficiencies during PEM
   - Adrenals: Increased plasma cortisol levels due to hypoalbuminaemia with greater free fraction. Presents with impaired glucose tolerance, moon facies and oedema

Causes of HYPOALBUNINAEMIA:

<table>
<thead>
<tr>
<th>Mechanism</th>
<th>Condition</th>
</tr>
</thead>
<tbody>
<tr>
<td>Insufficient protein intake or absorption</td>
<td>PEM</td>
</tr>
<tr>
<td>Reduced protein synthesis</td>
<td>Chronic liver disease or liver failure</td>
</tr>
<tr>
<td>Increased protein losses</td>
<td>Renal (nephritic syndrome), GUT (protein losing enteropathy), Skin and other surfaces (burns, bullous, exudates)</td>
</tr>
<tr>
<td>Catabolic states</td>
<td>Sepsis (acute phase response)</td>
</tr>
</tbody>
</table>

PROTEIN ENERGY MALNUTRITION:

CAUSES:

1. Insufficient intake:
   - 1.1. Insufficient total amount of protein (Quantity)
   - 1.2. Correct amount of insufficient quality protein
   - 1.3. Insufficient total amount of energy

Protein requirements:

<table>
<thead>
<tr>
<th>Protein source</th>
<th>Milk/egg</th>
<th>Mixed veg</th>
<th>Single veg</th>
</tr>
</thead>
<tbody>
<tr>
<td>Chemical score</td>
<td>1.19</td>
<td>1.70</td>
<td>2.08</td>
</tr>
<tr>
<td>g/kg/day (WHO)</td>
<td>100</td>
<td>70</td>
<td>60</td>
</tr>
</tbody>
</table>

Energy requirement:

<table>
<thead>
<tr>
<th>Components of requirement</th>
<th>kJ/kg/day</th>
</tr>
</thead>
<tbody>
<tr>
<td>Maintenance (1.5)BMR</td>
<td>330</td>
</tr>
<tr>
<td>Growth</td>
<td>20</td>
</tr>
<tr>
<td>Physical activity</td>
<td>80</td>
</tr>
<tr>
<td>TOTAL</td>
<td>430</td>
</tr>
</tbody>
</table>

THE RELATIONSHIP between QUALITY and QUANTITY of protein:

At low levels of intake, nitrogen uptake in tissue is not as much from a vegetable mixture as a mixture containing milk (as animal protein). At higher levels of intake the nitrogen retention of the vegetables is equal to that of the milk. Thus, with poor protein intake, animal proteins should be included in the diet.
appearance due to absence of and 'flaking enamel paint' a reversible Parkinson-like tremor. Distinguished from pellagra by its appearance depigmentation were

CLINICAL Presentation of PEM:

1) Growth faltering (nutritional dwarfing):
   - The first effect of PEM
   - Slowing or cessation of linear growth
   - Slowing or cessation of weight gain or weight loss
   - Decreased MUAC
   - Normal/decreased skin fold thickness

   NOTE: Plot the height for age and weight for height, but also consider weight for age, otherwise PEM can be missed due to relatively normal body proportions.

2) Kwashiorkor
   Epidemiology:
   - Weaning from breast milk or bottle (between nine months and 2 years)
   - Diet devoid of milk or high protein food
   - Diet often consists of carbohydrates, cereals and vegetables

   Presentation:
   1. Anthropometry: according to Welcome and Waterlow
   2. Skin changes: desquamation, pigmented, dermatosis (pellagra in type). With 'crazy paving'—dry, scaling pigmentation and 'flaking enamel paint'—depigmentation were skin flaked off—signs. Distinguished from pellagra by its appearance on sun exposed and unexposed areas, especially the buttocks and perineum in children
   3. Skin appendages
      = hair: brittle, discolored to red or grey, sparse, easily pulled out
      = fingers and toe nails: thinned and brittle
   4. Eyes: xerophthalmia if associated Vit A deficiency
   5. Mouth and tongue: angular stomatitis (fissuring at the mouth corners) with atrophic tongue papillae (atrophic glossitis).
   6. Subcutaneous oedema: Starts on dorsum of the feet or on the lower tibia. Can be slight or generalized depending on hydration state. Ascites rare and distinguishing feature if differential of cardiac, renal or hepatic oedema
   7. Subcutaneous tissue and fat: child can be deceptively chubby due to high carbohydrate diet and oedema
   8. Bones: growth failure and muscle wasting leads to inability to run, walk, sit or hold up head
   9. Organomegaly: A large, fatty liver due to triglyceride deposition
   10. Mental state: apathy and irritability common as well as 'kwashi shakes/Khan's syndrome'—a reversible Parkinson-like tremor
   11. Other: abdominal distension (pot belly) with small bowel dilation, diarrhea (due to atrophic bowel)

6. Kwashiorkor

Weaning from breast milk or bottle (between nine months and 2 years) Diet devoid of milk or high protein food Occurs mostly during the first year of life Can be due to early weaning, poor feeds, chronic diarrhea, malabsorption syndromes or infections (AIDS/TB)

Presentation:
1. Anthropometry: according to Welcome and Waterlow
2. Skin changes
3. Skin appendages
4. Eyes
5. Mouth and tongue
6. Subcutaneous oedema
7. Subcutaneous tissue and fat: shrunken, wizened, stark appearance due to absence of subcutaneous fat.
8. Bones: Slimming with voluntary muscles weak and atrophic
9. Organomegaly: Not a feature
10. Mental state: apathy, irritability
11. Other: slight abdominal distension, diarrhea, vomiting

Other than the five mechanisms mentioned above, a decreased cardiac output and lowered glomerular filtration rate also contributes to the pathophysiology of oedema in kwashiorkor.

CLASSIFICATION of PEM:

<table>
<thead>
<tr>
<th>WELLCOME CLASSIFICATION of PEM</th>
<th>Oedema absent</th>
</tr>
</thead>
<tbody>
<tr>
<td>Weight (% of standard)</td>
<td>Oedema present</td>
</tr>
<tr>
<td>&gt;80-8000</td>
<td>Kwashiorkor</td>
</tr>
<tr>
<td>&lt;80</td>
<td>Underweight</td>
</tr>
<tr>
<td>&lt;60</td>
<td>Marasmus</td>
</tr>
</tbody>
</table>

WATERLOW CLASSIFICATION

<table>
<thead>
<tr>
<th>Category</th>
<th>Height-for-age (%)</th>
<th>Weight-for-height (%)</th>
</tr>
</thead>
<tbody>
<tr>
<td>Normal</td>
<td>&gt;80% standard</td>
<td>&gt;80% standard</td>
</tr>
<tr>
<td>Stunted</td>
<td>&lt;80% standard</td>
<td>&gt;80% standard</td>
</tr>
<tr>
<td>Wasted</td>
<td>&gt;80% standard</td>
<td>&lt;80% standard</td>
</tr>
<tr>
<td>Wasted and stunted</td>
<td>&lt;80% standard</td>
<td>&lt;80% standard</td>
</tr>
</tbody>
</table>

Poor prognostic factors in kwashiorkor: (VERY IMPORTANT)

Severe infection
Hypoglycaemia
Jaundice
Collapse due to dehydration

3) Marasmus:

Epidemiology:

- Diet fails to meet energy requirement and often also protein requirements
- Occurs mostly during the first year of life
- Can be due to early weaning, poor feeds, chronic diarrhea, malabsorption syndromes or infections (AIDS/TB)

Presentation:
1. Anthropometry: according to Welcome and Waterlow
2. Skin changes
3. Skin appendages
4. Eyes
5. Mouth and tongue
6. Subcutaneous oedema
7. Subcutaneous tissue and fat: shrunken, wizened, stark appearance due to absence of subcutaneous fat.
8. Bones: Stunting with voluntary muscles weak and atrophic
9. Organomegaly: Not a feature
10. Mental state: apathy, irritability
11. Other: slight abdominal distension, diarrhea, vomiting
4) Marasmic Kwashiorkor:
Intermediate form of PEM with marasmus as well as clinical dermatosis and or oedema
characteristic of kwashiorkor.
“Kwashiorkor over night”: cases of marasmus where oedema and skin lesions become more
apparent when hydration improves.

TREATMENT of PEM:

<table>
<thead>
<tr>
<th>OUTPATIENT vs INPATIENT therapy:</th>
<th>Hospital Care</th>
</tr>
</thead>
<tbody>
<tr>
<td>Uncomplicated malnutrition</td>
<td>Severe or complicated malnutrition:</td>
</tr>
<tr>
<td>Weight for height &gt;70% of expected</td>
<td>Marasmus or kwashiorkor + one:</td>
</tr>
<tr>
<td>Anorexia</td>
<td>Dehydration</td>
</tr>
<tr>
<td>Clinically well</td>
<td>Lethargy</td>
</tr>
<tr>
<td>Eating well</td>
<td>Fever&gt;85</td>
</tr>
<tr>
<td>No lethargy</td>
<td>Pneumonia</td>
</tr>
<tr>
<td></td>
<td>Severe anaemia</td>
</tr>
<tr>
<td></td>
<td>Diarrhoea</td>
</tr>
</tbody>
</table>

OUTPATIENT Rx of mild to moderate PEM:
With early identification and management:
- Check and treat for infections, especially intestinal parasites
- Assess social and economic circumstances
- Advise and assist where possible:
  1) If animal protein too expensive, substitute with beans, peas, lentils and nuts
  2) Milk = 1 gram protein/30 ml milk, daily requirements of 1-2g protein/kg sufficient.
  3) If available distribute dried milk powder or RTUF (ready-to-use-therapeutic-foods)
- Road to health chart used to monitor efficacy of treatment: weight gain best parameter.

HOSPITAL TREATMENT of severe PEM:
Immediate Mix:
- Resusc and stabilize: If shocked = Ringer’s Lactate IV at 15 ml/kg for first hour and then 30ml/kg over next hour, then switch to ORS. If satisfactory peripheral circulation: Oral
  Rehydration Solution at 10ml/kg/hour.
- Metabolic: Give K+ 3-4mmol/kg/day, Mg2+4mmol/kg/day, Zinc acetate 2mg/kg/day, Cu2+ 0.3mg/kg/day.
- Hypoglycemia: Do routine screens. If <3mmol/l give 10% dextrose water, 50ml PO and
  follow with first feed ASAP.
- Hyperthermia: Check and warm child adequately
- Infection: Give anti-biotics immediately after blood culture if: critically ill child OR open
  skin lesions OR swab infection.

Diet and supplementation after resusc:
- Small, frequent milk feeds given 2-3hrly around the clock to supply a volume of
  130ml/kg/day

* ORS if necessary
* Gradually increase milk feeds to 150ml/kg/day (too rapid increase = gastric distention and
  vomiting)
* WHO recommends milk formula F-75: 75kCal/100 ml, 0.9g/100ml protein, aiming for an
  energy intake of 100kCal/kg/day
* Cereal and other foods introduced after 3 or 4 days

Vitamin Supplementation
- Vit A, Vit K, folic acid and iron

Course of the disease:
- Mortality highest within first three days due to bad prognostic factors.
- Weight gain for first three days and then weight reduction due to massive diuresis (loss of
  oedema)
- From there a steady weight gain of 10mg/kg/day should be achieved

Follow up:
- At monthly intervals to ensure growth is parallel to percentiles

Long term effects:
With corrected social circumstances and adequate food intake, growth retardation and cognitive
performance can be corrected

Vitamins, minerals and trace elements
The daily requirement for vitamins does not change much through the age ranges. Vitamins and
minerals are present in most mixed diets but deficiencies can occur. In SA vitamin A, D and iron
deficiencies are the most common.

Daily Requirements:

<table>
<thead>
<tr>
<th>Vitamin A (ug)</th>
<th>Vitamin C (mg)</th>
<th>Vitamin D (IU)</th>
<th>Calcium (mg)</th>
<th>Iron (mg)</th>
</tr>
</thead>
<tbody>
<tr>
<td>300-750</td>
<td>40</td>
<td>400</td>
<td>500</td>
<td>5-10</td>
</tr>
<tr>
<td>200-500 IU</td>
<td></td>
<td></td>
<td></td>
<td>12-24 (female adolescents)</td>
</tr>
</tbody>
</table>

Mineral deficiencies

<table>
<thead>
<tr>
<th>Mineral</th>
<th>Cause</th>
<th>Consequence</th>
</tr>
</thead>
</table>
| Iron    | Diet / loss (i.e. blood) | Anaemia
|         | Immunodeficiency | GIT dysfunction
|         | Impaired thermoregulation | Impaired cognition
|         | Weakness | |
| NA, K, Mg | Vomiting/ diarrhoea | Electrolyte abnormalities |
| Calcium | Decreased milk intake | Rickets/ metabolic bone diseases |

These deficiencies must be replaced with supplements.
Trace element deficiencies

<table>
<thead>
<tr>
<th>Trace element</th>
<th>Cause</th>
<th>Consequence</th>
</tr>
</thead>
<tbody>
<tr>
<td>Fluoride</td>
<td>Decrease levels in drinking water</td>
<td>Dental caries</td>
</tr>
<tr>
<td>Iodine</td>
<td>Dietary deficiency</td>
<td>Goiter and hypothyroidism</td>
</tr>
<tr>
<td>Zinc (difficult to measure)</td>
<td>Dietary deficiency</td>
<td>Rashes and growth stunting</td>
</tr>
</tbody>
</table>

As with mineral deficiencies, trace elements should be supplemented if necessary.

**Vitamin A (Retinol)**

**Sources:**
This is a fat soluble vitamin and is found in milk/butter, egg yolk and fish oils etc. Its provitamin is beta carotene (transformed in gut to Retinol) and is found in carrots and green leaves.

**Function:**
- Essential for vision in dim light and for integrity of tissues (especially the epithelial tissue of the skin and the eye).

**Causes of deficiency:**
- infants = decreased milk intake
- older children = decreased fruit and veg intake
- often occurs with PEM
- infection decreases vitamin A levels

**Signs of deficiency:**
- infants = xerophthalmia and keratomalacia
- older children = night blindness
- silver-grey blind's spot

**Confirmation of diagnosis:**
- serum retinol

**Management:**
- 30mg of retinol daily for 3 days (1/2 PO, ½ IM)

**Toxicity:**
- seen in overdose of vitamin supplements or eating too much fish liver
- presents with restlessness headache and vomiting
- may have yellow staining of skin
- think roacutan SE's, dry skin, cracked lips, arthralgia, headache and weakness

**Vitamin E**

**Sources:**
- in many foods so deficiency is unlikely

**Function:**
- Anti-oxidant

**Causes of deficiency:**
- biliary atresia or other cholestatic liver disease

**Signs of deficiency:**
- haemolytic anaemia in prem infants
- peripheral neuropathy
- ataxia

**Confirmation of diagnosis:**
- history and examination

**Management:**
- supplementation

**Vitamin B complex**

- individual B vitamin deficiencies are rare
- most deficiencies are associated with kwashiorkor or pellagra

**Sources:**
- proteins (not maize) i.e. milk, meat, cheese, eggs, fish, beans, peas and lentils.
- pyridoxine B6 = in most foods
- B12 = animal proteins

**Function:**
- important co-factors for many metabolic processes

**Causes of deficiency:**
- decreased breast milk levels (diet) or alcoholism in adults
- high maize diet
- B12 = true vegans (rare in children)/ malabsorption

**Signs of deficiency:**
- B1 deficiency = beri beri
- beri beri = acute cardiac failure with pseudomeningeal signs and drowsiness
- nicotinic acid deficiency = pellagra (just about only in Africa)/ children over 5y
- pellagra = dermatitis (erythema on sun-exposed areas), diarrhea and dementia (rare in children), commonly with kwashiorkor
- B12 deficiency = megaloblastic anaemia, smooth red tongue, irritability and failure to thrive

**Confirmation of diagnosis:**
- beri beri = decreased erythrocyte traksetolase activity
- pellagra = clinical and history and decreased N-methyl nicotinamide in the urine

**Management:**
- in beri beri = 50-100mg thiamine IV/IM stat, oral maintenance of 5-10mg daily for several days
- in pellagra = prevention by maize enrichment programs
- nicotinamide or nicotinic acid 100mg orally every 4hrs but it is important to change the diet
- B12 deficiency = supplementation

**Vitamin C (ascorbic acid)**

**Sources:**
- fresh vegetables and fruit
- breast milk/ fruit juice/ supplements in infancy

**Function:**
- facilitates the absorption of iron
- releases free folic acids from ingested food
- involved in collagen synthesis

**Causes of deficiency:**
- unsupplemented processed milks as main source of food
Signs of deficiency:
- scurvy (at 4-6m if fed on unsupplemented processed milks), irritability with pain on movement resulting in frog like posturing
- defective function of intercellular ground substance
- poor wound healing, rupture of capillaries and haemorrhage (defective collagen)
- haemorrhage into skin or muscle in response to minor trauma.
- anaemia
- X-rays show signs of periosteal elevation due to haemorrhage (later calcified), ground glass appearance of the shaft and ringing of the epiphysis

Confirmation of diagnosis:
- clinical and history
- X-ray changes

Management:
- immediate treatment is necessary to prevent further bleeding
- luckily the condition is completely reversible
- 250mg ascorbic acid 4 times a day
- prevention = 40 mg ascorbic acid daily/ fruit juice to infants on bottle feeds

Vitamin K
Sources:
- fat soluble vitamin occurring in plants
- also produced by bacteria in the gut

Function:
- co-factor for the synthesis of prothrombin in the liver

Causes of deficiency:
- sterile gut of the neonate
- breast and cows milk = low vitamin K levels
- after newborn period = biliary obstruction, liver disease, fistulae, PEM, intestinal malabsorption syndromes and long term antibiotic treatment

Signs of deficiency:
- haemorrhagic disease of the newborn (2nd-5th day of life)

Confirmation of diagnosis:
- clinical and history
- clotting factors

Management:
- 1mg vitamin K IM at birth
- for older children = vitamin K by mouth or injection

Obesity

DEFINITION: Weight for age and weight for length excessive (sex-specific), due to an excess fat intake.

DIAGNOSIS: BMI (weight in kg/height in meters²), at or above the 90th percentile on the sex-specific BMI-chart OR on normal growth charts: in weight: 10-20 or more percentiles above percentile of height for age (example: child on 60th centile height-for-age but 90th centile weight for age). BMI of >19 at 5 years, >20 at 10 years at >25 at 18 years is generally pathological

PATHOGENESIS: Due to a chronic, positive energy balance = energy intake exceeds energy expenditure.

- Low birth weight due to fetal undernutrition associated with metabolic X-syndrome later in life
- 7 known genes are associated, but interactions with environmental factors more important, even with genetic predisposition
- In infancy artificial opposed to breast feeding predisposes to obesity
- Older children: eating habits (refined carbohydrates) and lack of exercise

COMPLICATIONS:
INFANCY AN PRE-SCHOOL:
- Repeated respiratory infections, hypoventilation and airway disease. Pickwick syndrome (carbon dioxide retention) can occur.
- Difficult to assess dehydration
- Impaired glucose tolerance

SCHOOL AGE:
- Psychological: rejection, poor self image – predisposes to further over-eating or anorexia nervosa
- Orthopaedic problems:
  - Slipped Upper Femoral Epiphysis
  - Blount’s: medial half of tibial epiphysis, fuses with progressive bow-leg deformity (diagnose with metaphyseal-diaphyseal angle ≥11 degrees
  - Flat feet: loss of medial foot arch (pes planus)

LONG TERM: Development of metabolic X syndrome (hypertension, DM type 2, hypercholesterolaemia, hyperlipidaemia), associated with an increased risk for coronary artery disease

DIFFERENTIAL:
- Large infant – weight and length correlates
- Endocrine disorders: hypothyroidism, Cushing’s, craniopharyngioma (hypothalamic problem)
- Congenital disorder: Down’s, Prader Willi syndrome

TREATMENT:
- Involve a dietician
- In infants aim to keep weight static rather than to loose weight: allow child to grow into his weight. Cut feeds down to requirements
- In adolescence: sensible dieting, exercise, psychological support