Paediatrics Revision Guide 2023

Disclaimer: This resource has been written by a medical student with the aim of aiding revision. It is not an exhaustive list of content which could be examined and has no affiliation with the Medical School therefore the use of this for learning is at your own discretion.

Advice

- This guide contains a lot of the main conditions which would might encounter on the wards/learn about but is not an exhaustive list for exams so use this in combination with other materials
- Guidelines and treatment pathways may have changed since the time of writing so these should always be double checked
- This has been double checked for mistakes however there may still be errors present
- Once you've gone through the material, I would recommend using questions to consolidate and apply your learning

Feedback Form

Please fill in the following feedback form regarding the revision guide as we will use these comments to continually improve the resource

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Peer Teaching Society

Cardiology

VENTRICULAR SEPTAL DEFECT

- Most common form of heart defect
- Causes a left to right shunt
- Moderate: Enlarged atria and ventricles can lead to pulmonary HTN and congestive heart failure
- Severe: Severe pulmonary HTN and early onset heart failure

Risk Factors
- Premature birth
- Certain genetic conditions such as Down's syndrome, Edward's, Patau
- Family history of congenital heart defects

Clinical Presentation
- Often can by symptomless
- **Pansystolic murmur at the lower left sternal border**
- Poor feeding
- Tachypnoea
- Dyspnoea
- Failure to thrive

Diagnosis
- **Echo**
- ECG
- X- ray which may show cardiomegaly

Treatment
- Diuretics to relieve pulmonary congestion
- ACE inhibitors to reduce systemic pressure
- Surgical repair

Complications
- Eisenmengers
- **Endocarditis**
- Heart failure

ATRIAL SEPTAL DEFECT

- Defect in the septum between the 2 atria causing a **left to right shunt** due to the higher pressure in the left atria

Aetiology
- Maternal smoking in 1st trimester
- Family History of CHD

- **Maternal diabetes**
- Maternal rubella

Clinical Presentation
- Tachypnoea
- Poor weight gain
- Recurrent chest infections
- **Soft, systolic ejection murmur heard in 2nd intercostal space**
- Wide, fixed split S2 sound

Investigations
- ECG
- **Echo**

Management
- If small, can be managed conservatively and will close within 12 months of birth
- **Surgical closure**, usually if ASD >1cm

Complications
- Stroke from DVT
- Atrial fibrillation
- Pulmonary HTN
- Eisenmenger's syndrome

TETRALOGY OF FALLOT

Most common **cyanotic** congenital heart disease
- Overriding aorta
- Large VSD
- Pulmonary stenosis
- Right ventricular hypertrophy

Epidemiology
- More common in males
- Rubella
- Increased age of the mother (>40)

Pathophysiology
- Decreased right ventricular outflow
- Dilated and displaced aorta
- Mild: asymptomatic but as heart grows, develops cyanosis aged 1-3 year
- Moderate: Cyanosis and respiratory distress in the first few months of life
- Extreme: Often detected on antenatal scan, present with cyanosis in first few hours of life

Clinical Presentation
- Irritability
- Cyanosis

- Clubbing
- Poor feeding
- Poor weight gain
- **Ejection systolic murmur in pulmonary region (caused by pulmonary stenosis)**
- **Tet spells**

Investigations
- CXR: **boot shaped heart**
- MRI/Cardiac catheter
- **Echo**

Treatment
- **Prostaglandin infusion PGE1 to maintain ductus arteriosus**
- Beta blockers
- Morphine to reduce respiratory drive
- **Surgical: repair under bypass 3 months - 4 years but needs ICU post op**

Complications
- Pulmonary regurgitation
- **Lifelong follow up**

TRANSPOSITION OF THE GREAT ARTERIES

Aorta rises from right ventricle and pulmonary artery from left ventricle

Epidemiology
- More common in males
- Mum>40
- Rubella
- Maternal diabetes
- Alcohol consumption

Pathophysiology

Deoxygenated blood delivered systemically

Mixing needs to be possible to sustain life: patent foramen ovale, VSD or patent ductus arteriosus must be present alongside

Clinical Presentation
- **Cyanosis in the first 24 hours of life**
- Right ventricular heave
- Loud S2 heart sound
- Systolic murmur if VSD present
- Sometimes PD/VSD can make symptoms however within a few weeks they will develop respiratory distress, poor feeding etc

Diagnosis

Low SATS

Echo

CXR: **'egg on a string'** due to narrowed mediastinum and cardiomegaly
Metabolic acidosis

Treatment
PGE1 infusion to ensure PDA and mixing of blood
Surgical correction before 4 weeks

PATENT DUCTUS ARTERIOSUS

- Persistent connection between the aorta and pulmonary artery
- **Normal in utero** but usually closes within first 10-15 minutes of life
- **Left to right shunt**

Risk Factors
- Female
- Prematurity

Clinical Presentation
- Respiratory distress
- Apnoea
- Tachypnoea
- Tachycardia
- **Continuous machinery murmur** at the left sternal edge

Diagnosis
- **Echo**
- ECG/CXR

Management
- **Cardiac catheterisation** to close around 1 years old or sooner in more severe cases
- Premature infants: **Indomethacin or Ibuprofen** inhibits prostaglandin and stimulates closure

Respiratory

CROUP

- Acute laryngotracheobronchitis

Epidemiology
- Children: 6 months - 3 years old
- Peak incidence is at 3 years old
- **Common in Autumn and Spring**
- More common in boys

Pathophysiology

- Mucosal inflammation anywhere between the nose and the trachea
- Causative organisms: **Parainfluenza virus** mainly, Adenovirus, Rhinovirus, Enterovirus

Clinical Features
- Mild: Occasional **barking cough** with no audible stridor, no recession, child happy to eat and drink as normal
- Moderate: Frequent **barking cough with audible stridor at rest**, suprasternal recession, child not agitated
- Severe: **Frequent barking cough, prominent stridor** (high pitched breathing indicating an upper airway obstruction), marked sternal recession, agitated and distressed child potentially with tachycardia

Examination and History
- 1-4 days history of non-specific rhinorrhea (thin, nasal discharge), fever and **barking cough**
- Worse at night
- Stridor
- Decreased bilateral air entry
- Tachypnoea
- Costal recession

Respiratory Failure Red Flags
- Drowsiness
- Lethargy
- Cyanosis
- Tachycardia
- Laboured breathing

Diagnosis
- FBC, CRP U+E
- CXR to exclude foreign body

Treatment
- Symptoms can last 48 hours - 1 week
- Paracetamol/Ibuprofen for fever/sore throat
- Admission if moderate/severe and consider if dehydrated
- **Single dose dexamethasone 0.15mg/kg** or prednisolone
- Nebulised adrenaline for relief of severe symptoms
- Oxygen if required
- Monitor for needed ENT intervention if suspected airway blockage

Complications
- **Otitis Media**
- Dehydration due to reduced fluid intake
- Superinfection: pneumonia

BRONCHIOLITIS

- Viral infection of the bronchioles

Epidemiology
- Affects under 2's
- Very common
- Peaks in the winter and spring months
- Most commonly caused by **RSV (Respiratory Syncytial Virus)**

Risk Factors
- Breastfeeding for < 2 months
- Smoke exposure
- Older siblings who attend nursery/school
- Chronic lung disease of prematurity

Clinical Features
- Symptom onset in 2-5 days
- Low grade fever
- Rhinorrhea and nasal congestion
- Cough
- Reduced feeding
- Signs of Respiratory distress: nasal flaring, tracheal tug, head bobbing, grunting, sub/intercostal recessions
- Inspiratory crackles

Investigations
Nasopharyngeal aspirate for RSV culture
FBC, Urine, Blood gas if severely unwell
CXR - not used usually but shows hyperinflation, air trapping and flattened diaphragm

Management
Supportive management from home
Palvizumab vaccine against bronchiolitis should be considered in
- Children <9 months with chronic lung disease of prematurity
- Children < 2 years with severe immunodeficiency require long term ventilation

Urgent Hospital Admission
- Apnoea
- Resp Rate > 70
- Central cyanosis
- SpO2 < 92%

Non-Urgent Admission
- Resp Rate > 60
- Clinical dehydration

Inpatient Management
- Oxygen to bring SpO2 up
- Fluids

- CPAP if in respiratory failure
- Suctioning of secretions
- Ribavirin for severe cases
- No evidence for bronchodilatory, antibiotics or steroids in bronchiolitis

PNEUMONIA

- Infection of the lower respiratory tract and lung parenchyma which leads to consolidation.

Epidemiology
- Highest incidence in infants
- Viral cause more common in young infants
- Bacterial more common in older children
- Viral disease more common in the winter

Aetiology
Neonates: Group B Strep, E coli, Klebsiella, Staph Aureus
Infants: Strep pneumoniae, Chlamydia
School age: Strep pneumoniae, Staph Aureus, group A Step, Mycoplasma pneumoniae

Clinical Presentation
- Usually precede an upper respiratory tract infection
- Fever
- SOB
- Lethargy
- Signs of respiratory distress
- Auscultation signs: dullness to percuss, crackles, decreased breath sounds, bronchial breathing
- Wheeze and hyperinflation more typical of viral infection

Investigations
- Mainly clinical
- CXR - fluid in the lunds (associated with Staph)

Treatment
- Management at home with analgesia
- If admitted: Oxygen therapy and IV fluids
- Abx
    1. Neonates: Broad spec IV Abx
    2. Infants: Amoxicillin/Co-Amoxiclav
    3. Over 5s: Amoxicillin/Erythromycin

Complications
- Risk of parapneumonic collapse and empyema if so follow up at 4-6 weeks with a fluid sample

WHOOPING COUGH

- Bacterial URTI (bronchitis)
- **Bordetella Pertussis** - Gram -ve bacillus
- Can last up to 6-8 weeks without treatment

Epidemiology
- Much less incidence now due to vaccination programme
- Vaccinations: 2,3,4 months, booster at 3 years 4 months
- Impacts infants more dramatically

Clinical Presentation
- Catarrhal Phase: Lasts 1-2 weeks: coryzal symptoms
- Paroxysmal Phase: Occurs week 3-6: characteristic **'inspiratory whoop'**
- Cough worse at night
- Spasmodic coughing episodes - can lead to vomiting
- Low grade fever
- Sore throat
- Convalescent phase - downgrade of cough, may last up to 3 months

Diagnosis
- **Nasal-pharyngeal swab with pertussis**
- FBC
- Antibody test

Treatment
- **Macrolide e.g. Clarithromycin**
- Prophylactic Abx given to close contacts who are in higher risk health groups
- Isolation for 21 days after symptom onset or 5 days after antibiotics

Complications
- Seizures
- Pneumonia
- Bronchiectasis
- Encephalopathy
- Otitis Media

ASTHMA
- **Reversible paroxysmal constriction of the airways** with inflammatory exudate and followed by airway remodelling
- Most chronic condition of children

Aetiology
- **Genetic**
- Prematurity
- Low birth weight
- Parental smoking
- Viral bronchiolitis in early life
- Cold air

- Allergen exposure e.g. dust

Clinical Presentation
- **Episodic wheeze** which is infrequent/frequent and persistent most days and nights
- Dry cough often worse at night
- SOB
- Wheeze
- Reduced peak flow

Investigations
- FEV1 significantly reduced
- FVC normal
- FEV1:FVC may be <70% if poorly controlled
- Reversible spirometry is highly suggestive of asthma
- ENO levels of nitric oxide correlate to inflammation
- Baseline chest x ray

Management
Step 1: SABA PRN - Salbutamol
Step 2: ICS Preventer therapy - Beclomethasone
Step 3: LTRA Montelukast
Step 4: Strop LTRA if hasn't helped and add LABA - Salmeterol
Step 5: Switch ICS/LABA for ICS MART: Formoterol and ICS
Step 6: Add a separate LABA
Step 7: High dose ICS (>400mcg), referral

Under 5's
Step 1: SABA PRN - Salbutamol
Step 2: SABA + 8 week trial of ICS if symptoms reoccur within 4 weeks, restart ICS
Step 3: Refer to specialist

VIRAL INDUCED WHEEZE

- Episodic Wheeze - a symptom of viral URTI and symptom free in between events
- Multiple trigger Wheeze - URTI and other factors trigger wheeze

Management
Symptomatic treatment
- SABA inhaler via spacer maximum of 4 hourly up to 10 puffs
- LTRA and ICS via spacer
- Multiple trigger wheeze: trial ICS or LTRA for 4-8 weeks

RESPIRATORY DISTRESS SYNDROME

- Affects premature neonates, before the lungs start producing adequate surfactant, common in below 32 week babies.

Pathophysiology

- Inadequate surfactant leads to high surface tension within alveoli leading to atelectasis (lung collapse) as it is more difficult for the alveoli and the lungs to expand leading to inadequate gaseous exchange and hypoxia, hypercapnia and respiratory distress.

Management

- Dexamethasone is given to mothers with suspected or confirmed preterm labour to increase production of surfactant and reduce the incidence and severity of respiratory distress syndrome in the baby
- Intubation and ventilation may be needed to fully assist breathing if the distress is severe
- Endotracheal surfactant, which is artificial surfactant delivered into the lungs via an endotracheal tube
- CPAP via a nasal mask to keep the lungs inflated during breathing
- Supplementary oxygen to maintain sats between 91 and 95%

Complications

Short Term

- Pneumothorax
- Infection
- Apnoea
- Intraventricular haemorrhage
- Pulmonary haemorrhage
- Necrotising Enterocolitis

Long Term

- Chronic lung disease of prematurity
- Retinopathy of prematurity
- Neurological, hearing and visual impairment

BRONCHOPULMONARY DYSPLASIA

- Infants who still require oxygen at a postmenstrual age of 36 weeks are described as having BPD.
- The lung damage comes from pressure and volume trauma of artificial ventilation, oxygen toxicity and infection with the CXR characteristically showing widespread areas of opacification and sometimes cystic changes, fibrosis and even lung collapse.
- These infants are mainly weaned onto CPAP followed by additional oxygen where needed and sometimes given corticosteroids to facilitate weaning from the ventilation however there is a risk of abnormal neurodevelopment with these.
- A few infants with severe disease can die from intercurrent infection or pulmonary HTN.

Gastrointestinal

PYLORIC STENOSIS

- Progressive hypertrophy of the pyloric sphincter causing gastric outlet obstruction

Epidemiology
- Much more common in boys
- Affects 1 in 500
- Family history
- First borns

Clinical Presentation
- Presents 4-6 weeks of age
- **Non bilious, forceful, projectile vomiting** after every feed approx 30 mins
- Will continue to feed despite vomiting
- Weight loss
- Dehydration
- Constipation
- Visible peristalsis
- Palpable **olive sized pyloric mass** felt

Investigations
- Test feed with NG tube and empty stomach to feel for visible peristalsis and olive shaped mass
- USS - Hypertrophy of the muscle
- Blood gas - **Hypochloremic, Hypokalemic Metabolic Alkalosis** (loss of hydrogen and chloride ions due to vomiting gastric contents)

Management
- Correct metabolic imbalances - NaCl
- Fluid bolus for hypovolemia
- NG tube and aspiration of the stomach
- **Ramstedt's Pyloromyotomy** - feeding can commence 6 hours after procedure

HIRSCHSPRUNGS DISEASE

- Nerve cells of the myenteric plexus are absent in the distal bowel and rectum, specifically the parasympathetic ganglionic cells resulting in lack of peristalsis.

Epidemiology
- 90% present in the neonatal period
- Average age of presentation: 2 days
- Males
- Down's syndrome

Pathophysiology

- Short segment is the most common type where the disease is confined to the rectosigmoid part of the colon
- Ganglion cells of the submucosal plexus aren't present
- Failure of peristalsis and bowel movements causing obstruction
- Can lead to bacterial build up and enterocolitis (inflammation) and sepsis

Clinical Presentation

1. **Failure to pass meconium** (within 48 hours of birth)
2. Abdominal distention
3. Bilious vomiting

- Palpable faecal mass in the left lower abdomen
- Empty rectal vault

Investigations
Rectal suction biopsy to test for ganglionic cells in anyone who has:

- Delayed passage of meconium
- Constipation in the first few weeks
- Chronic abdominal distention
- Positive family history
- Faltering growth

Contrast Enema - Shows short transition zone between proximal and distal colon and a small rectal diameter

Management
IV Abx
Bowel decompression
NG tube
Surgery is definitive treatment: Swenson, Soave, Dunhamel pull through surgery

MALROTATION AND VOLVULUS

- Twisting loop of bowel leading to intestinal obstruction
- Generally presents in the first month of life

Clinical Presentation

- Abdominal pain
- **Bilious vomiting**
- Caecum at the midline
- Reflux symptoms

Investigations

- Barium enema
- **Abdominal X-ray with contrast** to look for obstruction (dilated stomach and proximal small bowel - **double bubble sign**)

Treatment
Emergency surgical repair

INTUSSUSCEPTION

- One piece of the bowel telescopes inside another leading to ischaemia and bowel obstruction
- Most common in the distal ileum at the ileocecal junction

Epidemiology
- 3 months to 3 years
- Most commonly < 1
- **Most common cause of obstruction in neonates**

Risk factors
- CF
- Meckel's diverticulum
- HSP
- Rotavirus vaccine > 23 weeks

Clinical Presentation
- Colic abdominal pain
- Pallor
- **Sausage shaped mass palpable in the RUQ**
- **Redcurrant jelly stools**, often late in presentation
- Abdominal distention
- Shock
- Peritonitis: Guarding, rigidity,pyrexia

Investigations
USS: target shaped mass
Abdominal X Ray: Distended small bowel, absence of gas in the large bowel

Treatment
Medical emergency
IV fluids
Air Enema using USS to stretch the walls of the bowel and reduce the intussusception -> if this is unsuccessful then **surgery to repair manually**
If perforation and peritonitis, broad spec Abx e.g Gentamicin

NECROTISING ENTEROCOLITIS
- Acute inflammatory disease affecting preterm neonates leading to bowel necrosis and multi system organ failure

Epidemiology
- Low birth weight (1500g)
- Most common surgical emergency in neonates
- Commonly presents in the first 2 weeks of life
- Prematurity
- Abx therapy > 10 days
- Genetic

Clinical Presentation

- New feed intolerance
- **Vomiting (+ bile)**
- **Fresh blood in stools**
- Abdominal distention
- Reduced bowel sounds
- Palpable abdominal mass
- Visible intestinal loops
- Sepsis

Investigations
- Bloods: thrombocytopenia, neutropenia
- Cultures
- Blood gas: acidotic
- USS: Air in the portal system, ascites, perforation
- **X ray:**
    1. **Rigler's sign**: both sides of the bowel are visible due to gas in the peritoneal cavity
    2. Dilated bowel loops
    3. Distended bowel
    4. Thickened bowel wall
    5. Air outlining falciform ligament

Management
- Nil by mouth
- Bowel decompression by NG tube
- IV Cefotaxime
- Surgery to remove necrotic bowel

MECKEL'S DIVERTICULUM
- Congenital diverticulum of the small intestine containing ileal, gastric and pancreatic mucosa
- Occurs in 2% of the population
- Is 2cm from the ileocecal valve
- Supplied by the omphalomesenteric artery
- Risk of peptic ulceration

Clinical Presentation
- Abdominal pain
- **Rectal bleeding in children age 1-2 years**
- **Obstruction due to intussusception and volvulus**

Management
- **Removal if symptomatic (resection)**

BILIARY ATRESIA
- Obstruction of the biliary tree due to sclerosis of the bile duct, reducing bile flow

Epidemiology
- Females
- Neonatal cholestasis 2-8 weeks
- Associated with CMV
- Congenital malformations

Pathophysiology
- Type 1: Common duct is obliterated
- Type 2: Atresia of the cystic duct in the porta hepatis
- Type 3: Most common atresia of the right and left ducts at the level of the porta hepatis

Clinical Presentation
- **Jaundice post 2 weeks**
- Dark urine
- Pale stools
- Appetite disturbance
- Hepatosplenomegaly
- Abnormal growth
- Duodenal atresia - presents in the first 24 hours of life and more common in trisomy 21

Investigations
- Serum bilirubin: **conjugated bilirubin high**
- LFTs raised
- Alpha 1 antitrypsin to rule out deficiency
- Sweat test to rule out CF
- USS to look for structural abnormalities

Management
- Surgical dissection of the abnormalities: **Kasai procedure**
- Abx

Complications
- Cirrhosis and HCC
- Progressive liver disease

NEONATAL JAUNDICE
- Caused by hyperbilirubinemia

Pathophysiology
1. Physiological
- Breakdown of in-utero Hb
- Immature liver cannot break down high bilirubin concentrations
- Starts 2-3 days of life, peak at day 5, usually resolves by 14 days

2. Pathological Jaundice

- Onset less than 24 hours is pathological
- G6PD deficiency
- Spherocytosis

3. Prolonged Jaundice
    - Jaundice > 14 days in term infants and 21 days in preterm
    - Biliary atresia
    - Hypothyroidism
    - Breast milk jaundice resolves 1.5-4 months
    - UTI/Infection

Aetiology
- Prematurity
- Small for dates
- Previous sibling with neonatal jaundice

Clinical Presentation
- **Colour**
- Drowsiness
- Signs of infection

Investigations
- **TCB can be used over 35 weeks gestations** -> measures bilirubin in the skin
- Serum bilirubin if TCB cannot be used
- Total and conjugated bilirubin
- Coombs test
- Infection screen

Management
- Treatment threshold graphs for specific gestation
- **Phototherapy** will be used for those above threshold
- Those below the threshold should have levels rechecked within 24 hours
- Repeat bilirubin 4-6 hours after commencing phototherapy and 6-12 hourly once levels stabilise
- Stop phototherapy once over 50umol/L from the treatment line
- Recheck bilirubin 12-18 hours after stopping
- Exchange transfusion
    1. Exchange of blood with donated plasma to decrease level of circulating bilirubin
    2. Via umbilical artery or vein
    3. Indicated in rise of >8.5umol/L/hour
    4. Indicated in signs of Kernicterus

Complications
- **Kernicterus** - bilirubin induced encephalopathy and irreversible neurological damage: > 360umol/L

CONSTIPATION

- An extremely common condition affecting children characterised by decreased frequency, increased harness of the stool and painful defecation.
- Most cases of constipation are idiopathic however it is important to think about secondary causes.

'Red Flag' symptom/ signs	Diagnostic concern
Failure to pass meconium within 24 h of life	Hirschsprung disease
Failure to thrive/growth failure	Hypothyroidism, coeliac disease, other causes
Gross abdominal distension	Hirschsprung disease or other gastrointestinal dysmotility
Abnormal lower limb neurology or deformity, e.g. talipes or secondary urinary incontinence	Lumbosacral pathology
Sacral dimple above natal cleft, over the spine – naevus, hairy patch, central pit, or discoloured skin	Spina bifida occulta
Abnormal appearance/ position/patency of anus	Abnormal anorectal anatomy
Perianal bruising or multiple fissures	Sexual abuse
Perianal fistulae, abscesses or fissures	Perianal Crohn disease

Secondary causes

- Hirschprung's disease
- Cystic fibrosis
- Hypothyroidism
- Spinal cord lesions
- Sexual abuse
- Intestinal obstruction
- Cows milk intolerance

Clinical Presentation

- Less than 3 stools a week
- Hard stools that are difficult to pass
- Rabbit dropping stools
- Straining and painful passages of stools
- Abdominal pain
- Overflow soiling caused by faecal impaction
- Palpable hard stools in the abdomen

Management

- Idiopathic constipation can be diagnosed clinically, once red flags have been considered.
- Correction of any reversible contributors e.g. high fibre diet, good hydration
- Laxatives: **Movicol is first line**
- Disimpaction regimen may be needed with high dose of laxatives at first followed by half the disimpaction dose as maintenance
- Encouragement of visiting the toilet to reduce withholding.

ENT

EPIGLOTTITIS

- Caused by Haemophilus Influenzae Type B

Clinical Presentation

- Rapid onset
- High temperature
- Stridor

- Drooling and saliva
- Patient finds it easier to breathe leaning forward and extending neck **(tripoding position)**

Diagnosis
- Clinical
- CXR - **Thumb sign** and acute epiglottis swelling
- **Do not examine throat**

Management
- O2
- Nebulised adrenaline
- IV Abx - 3rd gen cephalosporins e.g. **Ceftriaxone**

OTITIS MEDIA
- Infection of the middle ear

Aetiology
- Viral
- Pneumococcus/Haemolytic Streptococcus/Hib

Clinical Presentation
- Ear pain
- Fever
- Bulging tympanic membrane
- Discharge

Secondary otitis media (glue ear)
- Child may have hearing loss
- Retracted eardrum
- > 3 months: referral for grommets and adenoidectomy

Management
- 5 days Amoxicillin/Erythromycin

Ophthalmology

SQUINT
- Also known as strabismus, this is misalignment of the eyes
- When the eyes are not aligned, the images on the retina will not match and the patient will experience double vision

Pathophysiology
- In childhood, as the eyes have not fully established their connections with the brain, the brain copes with this by reducing the signal from the less dominant eyes.
- This results in one dominant eye and one eye which will be ignored (lazy eye)

- When left untreated, this lazy eye becomes more and more disconnected from the brain and the problem worses - this is known as **amblyopia**

1. Concomitant squint - differences in the control of the extra ocular muscles
2. Esotropia - inward position squint -> affected eye deviated towards the nose
3. Exotropia - outward position quint -> affected eye deviated towards the ear
4. Hypertropia - upward moving affected eye
5. Hypotropia - downward moving affected eye

Aetiology
- Idiopathic
- Hydrocephalus
- Cerebral palsy
- Space occupying lesion e.g retinoblastoma
- Trauma

Investigations
- Eye movements and inspection
- Fundoscopy
- Visual acuity
- Hrischberg's test: shine a pen torch at the patient from 1 metre away, when they look at it, observe the reflection of the light source on their cornea - this should be central and symmetrical.
- Cover test: cover one eye and ask the patient to focus on an object in front of them. Move the cover to the opposite eye and watch the movement of the other eye and observe for any exo/esotropia.

Management
- Treatment must start before 8 years of age
- **Occlusive patch** can be used to cover the good eye and force the weak eye to develop
- Atropine drops can be used in the good eye causing blurry vision and again forcing the bad eye to develop.

PERIORBITAL CELLULITIS

- An eyelid and skin infection in front of the orbital septum where the inflammation and infection remains confined to the soft tissue layers superficial to the orbital septum.
- It is important to differentiate this from orbital cellulitis which is where the muscles of the orbit are affected and is usually due to bacterial sinusitis and is a life threatening condition.

Risk factors
- Boys
- Previous sinus infection
- Lack of Hib infection
- Recent eyelid injury

Clinical Presentation

- Swelling, redness and hot skin around the eyelids and the eye

Investigations

- Clinical examination
- **CT sinus and orbits with contrast** will help to differentiate between periorbital and orbital

Management

Empirical antibiotics either an inpatient or outpatient depending on severity e.g Cefotaxime/Clindamycin

Neurology

EPILEPSY

- An umbrella term for a tendency to have seizures which are transient episodes of abnormal electrical activity in the brain.

Types of Seizures

1. Generalised Tonic-Clonic Seizures
    - Loss of consciousness with tonic (rigidity) and clonic (rhythmic jerking) phase with possible tongue biting, incontinence, groaning and irregular breathing
    - After this there is a postictal period where the person is confused, drowsy and feels irritable/low.
    - Management is sodium valproate, **lamotrigine** or carbamazepine.
2. Focal Seizures
    - These begin in the temporal lobes and affect speech, memory and emotions.
    - They can present with hallucinations, memory flashbacks and deja vu.
    - Management is with **lamotrigine or levetiracetam**
3. Absence Seizures
    - The most common in children, the become blank and stare into space then abruptly return to normal
    - During this, they are unaware of their surroundings and will not respond and these generally last for around 10-20 seconds.
    - Most patients stop having these with age
    - Management is **ethosuximide**.
4. Myoclonic Seizures
    - Sudden brief muscle contractions where the person remains awake
    - These are often part of juvenile myoclonic epilepsy
    - Management includes **sodium valproate or levetiracetam**
5. Tonic/Atonic Seizures
    - Sudden tension/stiffness affecting the body
    - Management is with **sodium valproate or lamotrigine**

Investigations

- Full history
- EEG should be performed after the second simple tonic-clonic seizures
- MRI brain to help diagnose structural abnormalities
- Blood electrolytes, glucose, cultures and LP can also be considered.

Management of Acute Seizures

- Recovery position if possible
- Put something soft under the head to prevent injury
- Remove any obstacles that could lead to injury
- Make a note of time of start and end of seizures
- Call an ambulance if the seizures lasts longer than 5 minutes

Side Effects

- Sodium Valproate - Teratogenic, Liver damage, Hair loss, Tremors
- Carbamazepine - Agranulocytosis, Aplastic anaemia
- Ethosuximide - Night tremors, Rashes, N+V
- Lamotrigine - **DRESS syndrome**, Leukopenia

Status Epilepticus

- **Medical emergency** where a seizures lasts more than 5 minutes or 2 or more seizures without regaining consciousness
- Management includes securing airway, high concentration oxygen, assess cardiac and respiratory function and **IV lorazepam** which can be repeated after 10 minutes if it continues
- Medical options in the community include **buccal midazolam and rectal diazepam**.

FEBRILE CONVULSIONS

- Seizures which occur in children with a **high fever** between the ages of 6 months and 5 years
- Simple febrile convulsions are generalised tonic clonic and last less than 15 minutes and only occur once during a single febrile illness
- Complex febrile convulsions are focal seizures, last more than 15 minutes or occur multiple times during the same febrile seizures

Investigations

- Rule out other causes such as epilepsy, syncopal episode, trauma, space-occupying lesions and neurological infection.

Management

- Identify and manage source of infection
- **Control fever with simple analgesia**
- Parental education

- Prognosis is slightly higher for seizures/epilepsy in the future

Dermatology

ECZEMA

- A chronic atopic condition caused by defects in the normal continuity of the skin barrier, leading to gaps which allow irritants, microbes and allergens to enter, creating an immune response and leading to inflammation.
- Some patients will identify environmental triggers such as changes in temperature, certain dietary products, washing powders, cleaning products and emotional events/stress.

Clinical Presentation

- Usually in infancy
- **Dry, red, itchy skin** with sore patches over the flexor surfaces (elbows, knees) and face and neck
- Often **episodic with flares**

Management

- For maintenance, emollients e.g. E45, Diprobase should be used as often as possible, especially after washing and before bed which help create an artificial barrier over the skin
- Flare ups can be treated with thicker emollients such as Cetraben ointment or topical steroids such as Hydrocortisone and Betnovate (beclomethasone) which help keep moisture locked in overnight
- Other specialist treatments include topical tacrolimus, oral corticosteroids and methotrexate.

STEVENS-JOHNSON SYNDROME

- A disproportional immune response causing epidermal necrosis resulting in blistering and shedding of the top layer of the skin - less than 10% of body surface area affected

Aetiology

1. Medications
    - Anti epileptics
    - Antibiotics
    - Allopurinol
    - NSAIDs
2. Infections
    - Herpes simplex
    - Mycoplasma pneumonia
    - Cytomegalovirus

- HIV

Clinical Presentation

- Some cases will be mild whilst others will be severe and potentially fatal
- **Non-specific symptoms initially** with fever, cough, sore throat, sore mouth, sore eyes and itchy skin
- **Purple/red rash** which spreads across the skin and blisters, this then breaks away and leaves the raw tissue underneath
- Pain, blistering and shedding can also happen to the lips and mucous membranes
- Inflammation and ulceration of the eyes can also happen
- It can also affect the urinary tract, lungs and internal organs

Management

- **Medical emergency**: supportive care is essential
- Steroids, immunoglobulins and immunosuppressant medications can all be given with specialist guidance

Complications

- Secondary infection such as cellulitis, sepsis
- Permanent skin damage

URTICARIA

- Also known as hives, these are small itchy lumps which appear on the skin and may be associated with angioedema.

Pathophysiology

- Release of histamine and other pro-inflammatory chemicals by mast cells in the skin
- These may be part of an allergic reaction in acute urticaria or an autoimmune reaction in chronic idiopathic urticaria

Aetiology

- Allergies to food, medications or animals
- Contact with chemicals, latex or stinging nettles
- Medications
- Viral infections
- Insect bites

Chronic Urticaria

- An autoimmune condition where autoantibodies target mast cells and trigger them to release histamines and other chemicals

Management

- Antihistamines
- **Fexofenadine** is the antihistamine of choice for chronic urticaria
- Oral steroids may be given for flare ups
- Omalizumab which targets IgE

NAPPY RASH

- Contact dermatitis in the nappy area, normally caused by friction between the skin and nappy and contact with the urine and faeces.
- Most common between 9-12 months of age
- Breakdown of skin and the warm moist environment can lead to candida (fungus) or bacteria infection (staphylococcus/streptococcus)

Risk factors

- Delayed changing of nappies
- Irritant soap products and vigorous cleaning
- Diarrhoea
- Oral antibiotics predispose to candida
- Preterm infants

Clinical Presentation

- Sore, red, inflamed skin in the nappy areas
- **No rash on the creases of the groin**
- The rash may be itchy and the infant may be distressed
- Severe and long standing rash can lead to erosion and ulceration

Differentiating Candida vs Nappy Rash

- **Rash extending into the skin folds**
- **Large red macules**
- Well demarcated scaly border
- **Circular pattern to the rash spreading outwards**, similar to ringworm
- Satellite lesions - small, similar patches of rash near the main rash

Management

- Switching to highly absorbent nappies
- **Change the nappy and clean the skin** as soon as possible after wetting or soiling
- Use water or gentle alcohol free products
- Ensure the nappy area is dry before replacing the nappy
- **Maximise time not wearing a nappy**
- Infection requires **antifungal/antibiotic cream**

NON-BLANCHING RASHES

- Caused by bleeding under the skin
- Petechiae are small, non blanching, red spots on the skin caused by burst capillaries

- Purpura are larger, non-blanching, red-purple macules or papules caused by leaking of blood from vessels under the skin
- **Any child with non-blanching rash needs immediate investigation due to the risk of meningococcal sepsis.**

Differentials

- Meningococcal septicaemia: feverish, unwell child which requires immediate antibiotic management due to significant morbidity/mortality.
- HSP: purpuric rash on the legs and buttocks and may have associated abdominal or joint pain
- ITP: rash which develops over several days in an otherwise unwell child
- Leukaemias: gradual development of petechiae with other signs such as anaemia, lymphadenopathy and hepatosplenomegaly.
- HUS: Presents in a child with recent diarrhoea alongside oliguria and signs of anaemia.
- Mechanical: Strong coughing, vomiting or breath holding can product petechiae above the neck and most prominently around the eyes
- Traumatic: Tight pressure on the skin e.g NAI can lead to traumatic petechiae
- Viral illness can often cause rashes

Investigations

- FBC, U+e, CRP, ESR, Blood cultures, Meningococcal PCR, LP, Blood pressure and urine dipstick

ANAPHYLAXIS

- 85% of this is caused by a food allergy with an IgE mediated response causing significant respiratory/cardiovascular compromise.
- Other causes include drugs, insect stings, latex, exercise and idiopathic
- Most occurs in children under 5 due to food allergy however the most fatal is in adolescence.
- Acute management is early administration of adrenaline and long term management involves a detailed plan for allergy avoidance and the presence of adrenaline auto-injectors.

Infectious Diseases

KAWASAKI DISEASE

- Mucocutaneous, lymph node syndrome - a systemic medium-sized vessels vasculitis
- Typically affects young children under 5 years with no clear cause or trigger
- More common in boys, usually Japanese and Korean children

Clinical Presentation

- **Persistent high fever for more than 5 days**
- Child will be unwell and unhappy

- Widespread erythematous **maculopapular rash and desquamation on the palms and soles**
- **Strawberry tongue**
- Cracked lips
- **Cervical lymphadenopathy**
- Bilateral conjunctivitis

Investigations

- FBC to show anaemia, leukocytosis and thrombocytosis
- LFTs can show hypoalbuminemia
- Raised ESR
- Urinalysis can show raised WBC
- **Echo to rule out major complication: coronary artery aneurysms**

Disease courses

1. Acute phase - child will be unwell with fever, rash and lymphadenopathy - 1-2 weeks
2. Subacute phases - acute symptoms will settle but the arthralgia and risk of coronary artery aneurysms form - 2-4 weeks
3. Convalescent stage - remaining symptoms return back to normal and blood tests return to normal - 2-4 weeks

Management

- **High dose aspirin** to reduce the risk of thrombosis
- **IV immunoglobulins**
- **Public health should be informed**

MENINGITIS

- Inflammation of the meninges which are the lining of the brain and the spinal cord
- **Neisseria meningitidis** is a **gram negative diplococcus** bacteria which occur in pairs
- Meningococcal meningitis is when the bacteria is infecting the meninges and the CSF

Bacterial

- Most common cause is Neisseria Meningitidis and Streptococcus pneumoniae
- In **neonates**, the most common cause is **Group B Strep** which is contracted during birth from GBS bacteria that live in the mothers vagina

Clinical Presentation

- Fever, neck stiffness, vomiting, headache, photophobia, altered consciousness and seizures
- Neonates and babies can have non specific symptoms such as hypotonia, poor feeding, lethargy, hypothermia and a bulging fontanelle.

- Kernig's test: Lay patient on back and flex one hip and knee then straighten the knee whilst keeping the hip flexed, this will product pain/resistance in meningitis
- Brudzinski's test: Lay the patient on their back and lift their head and neck off the bed and flex chin to the chest, this will cause involuntary flexion of hips and knees.

All children under 1 month with a fever, 1-3 months with a fever and unwell and under 1 year with unexplained fever and other signs of serious illness should have an LP.

Management

- Community: **Stat injection of benzylpenicillin** and then transfer to hospital
- Hospital: blood culture and LP should be performed before antibiotics are started and a meningococcal PCR should be sent
- **Under 3 months: IV Cefotaxime plus IV Amoxicillin, above 3 months - IV Ceftriaxone**
- Dexamethasone is sometimes used
- **Public health should be informed**
- A single dose of ciprofloxacin should be given as post exposure prophylaxis.

Viral Meningitis

- Most common causes are HSV, enterovirus and VZV
- Sample of CSF should be sent for viral PCR
- Only supportive treatment is needed, sometimes aciclovir can be used

Cerebrospinal Fluid	Bacterial	Viral
Appearance	Cloudy	Clear
Protein	High	Mildly raised or normal
Glucose	Low	Normal
White Cell Count	High (neutrophils)	High (lymphocytes)
Culture	Bacteria	Negative

MEASLES

- Despite significant vaccination, this is still a major cause of morbidity and death worldwide.
- Initial exposure occurs through droplet spread and it is highly infectious during viral shedding

Clinical Presentation

- Fever
- Koplik spots (blue, white spots on the inside of the cheek)
- Conjunctivitis
- Coryza
- Cough
- Rash which spreads downwards from behind the ears to the whole of the body - maculopapular rash.

Management

- Supportive treatment depending on symptoms
- Avoid school for at least 5 days after initial development of rash
- **Notify Public Health**

Complications

- **Otitis media** (most common)
- Pneumonia
- Febrile convulsions
- Encephalitis/Subacute sclerosing panencephalitis

CHICKENPOX

- This is a common virus which is spread by respiratory droplets and is very infectious during viral shedding.

Clinical presentation

- Fever
- **Vesicular rash beginning on head and trunk which spreads to peripheries**
- Itching can lead to permanent scars/secondary infection

Management

- **Symptomatic treatment**
- Immunocompromised children/higher risk groups can be given aciclovir
- **Avoid school until lesions have crusted over**

Complications

- Bacterial superinfection
- Pneumonitis
- DIC

RUBELLA

- A generally mild disease in childhood which occurs in winter and spring
- Incubation period is 15-20 days and it is spread through respiratory contact.

Clinical Presentation

- Low grade fever
- Maculopapular rash on the face which then spreads across the whole body.

Management

- No treatment necessary
- Diagnosis should be confirmed serologically if there is any risk of exposure for non-immune pregnant women

Complications

- Arthritis
- Encephalitis
- Myocarditis

DIPHTHERIA

- Infection which causes local disease with membrane formation affecting the nose, pharynx or larynx or systemic disease with myocarditis and neurological manifestations.
- It has generally been eradicated in the UK.

SCALDED SKIN SYNDROME

Aetiology

- Caused by an exfoliative staphylococcal toxin which causes separation of the epidermal skin through the granular cell layers

Clinical Presentation

- Fever
- Malaise
- Purulent, crusting, localised infection around eyes/nose/mouth with widespread erythema and tenderness
- Areas of epidermis separate on gentle pressure (**Nikolsky sign**) leaving denuded areas of skin which then dry and heal without scarring.

Management

- IV anti-staph antibiotics such as **flucloxacillin**
- Analgesia
- Fluid balance

TUBERCULOSIS

- TB had a real decline however there is increased incidence in patients with HIV infection and the emergency of drug resistant strains
- It is spread through respiratory droplets.
- TB infection is latent TB and is more likely to progress to TB disease in infants and young children however children will generally not be infectious compared to adults.

Clinical Presentation

- Asymptomatic children will have minimal signs of infection as a local inflammatory reaction limits the progression of the disease however it remains latent and so a Mantoux test may become positive.

- If the local infection response fails, it can spread through the lymphatic system causing fever, anorexia and weight loss, cough and chest x-ray changes such as **hilar lymphadenopathy.**
- There may be enlargement of the peribronchial lymph nodes which can cause consolidation, bronchial obstruction and pleural effusions.
- There may also be other organ involvement including the gut, skin and superficial lymph nodes.
- A dormant stage occurs and this can be reactivated and present as post-primary TB where the infection can be local or spread across systems including bones, joints, kidneys and CNS (can lead to TB meningitis)

Investigations

- Given the difficulty in obtaining sputum samples, gastric washings on 3 consecutive mornings can be obtained to culture **acid-fast bacilli** through an NG tube.
- Mantoux tests can be performed however this can be positive due to past vaccination rather than infection.
- IGRA is a new blood test which is used to assess the response of T cells to antigens found in TB but not the BCG vaccine.

Management

- **Rifampicin, Isoniazid, Pyrazinamide, Ethambutol therapy initially and then reduced to just Rifampicin and Isoniazid after 2 months. This whole therapy normally lasts around 6 months.**
- After puberty, pyridoxine is given weekly to minimise the peripheral neuropathy side effects of isoniazid.
- **Contact tracing** is essential as children often pick up the infection from adults

HIV INFECTION

- Affecting over 2 million children a year, the main route of transmission is mother-children transmission during pregnancy, at delivery or through breast feeding.

Investigations

- In children over 18 months, **presence of antibodies is diagnostic**.
- In those under 18 months, a **HIV DNA PCR** is needed as antibodies may be present from merely exposure not active infection.

Clinical Presentation

- A proportion of HIV infected infants progress rapidly to symptomatic disease and AIDS in the first year of life however most children will remain **asymptomatic for several years.**
- Children with mild immunosuppression may present with lymphadenopathy, those with moderate may have recurrent bacterial infections, chronic diarrhoea and lymphocytic interstitial pneumonitis.

- Severe AIDS diagnosis can be indicated with pneumocystis jirovecii pneumonia, severe faltering growth and encephalopathy.

Treatment

- **Antiretroviral therapy** should be started in all infants and some older children depending on clinical status, HIV load and CD4 count.
- **Immunisations** are important due to the higher risk of other infections
- MDT approach with management for the family as well
- **Regular follow up** including weight, development and clinical signs of disease.

Transmission Reduction

- Mother who are positive should be on **antiretroviral drugs to reduce viral load at time of delivery**
- They should also **avoid breastfeeding and active management of labour** and delivery to prevent prolonged rupture of membranes.
- If breastfeeding cannot be avoided, antiretroviral drugs should be given to both mother and baby.

ENCEPHALITIS

- Inflammation of the brain which can be either infectious or non-infections (this is usually autoimmune)
- The most common cause is viral however bacterial and fungal are also possible.
- The most common type is **herpes simplex 1** from cold sores in children and **herpes simplex 2 in neonates from genital herpes** contracted during birth.
- Other causes include VZV associated with chickenpox, EPV, enterovirus, adenvirus and influenza.

Clinical Presentation

- Altered consciousness
- Altered cognition
- Unusual behaviour
- Acute onset of focal neurological symptoms
- Acute onset of focal seizures
- Fever

Investigations

- **LP to send CSF fluid for PCR testing**
- CT if an LP is contraindicated e.g. active seizures, hemodynamically unstable
- **MRI after the LP for visualisation**
- HIV testing is also recommended in all patients with encephalitis

Management

- **Aciclovir to treat Herpes and VZV, Ganciclovir to treat Cytomegalovirus**
- **Repeat LP** are needed to ensure successful treatment before antivirals are stopped

Complications

- Lasting fatigue
- Change in personality/mood/memory/cognition
- Headaches and chronic pain
- Sensory disturbance
- Seizures

SLAPPED CHEEK SYNDROME/FIFTH DISEASE

- **Parvovirus B19** causes erythema infectiosum/slapped cheek syndrome
- Outbreaks are common during the spring months and transmission is via respiratory secretions from infected patients/mother to foetus.
- It infects the erythroblastosis red cell precursors in the bone marrow

Clinical Presentation

- Asymptomatic infection
- Erythema infectiosum - the most common illness with **fever, headache and myalgia** followed by a **characteristic rash on the face which progresses to maculopapular rash on the trunk and limbs**
- Aplastic crisis: occurs in children with haemolytic anaemia where there is an increased rate of red cell turnover and immunodeficiency.
- Foetal disease where transmission can lead to foetal death due to severe anaemia.

Management

- Supportive therapy

IMPETIGO

- A superficial bacterial skin infection usually caused by the staphylococcus aureus bacteria
- A ‘golden crust’ is characteristic for this

Non-bullous

- This typically occurs around the nose or mouth and the exudate from the lesions dries to form a **golden crust**
- The children will have **no systemic symptoms and will not be unwell**
- This can be treated with **topical fusidic acid** or antiseptic cream if it is localised.
- For widespread, oral flucloxacillin is used
- Patients should avoid touching or scratching the lesions

Bullous

- This is always caused by **S.aureus** which produce toxins that break down the proteins that hold skin cells together causing 1-2 cm fluid filled vesicles to form on the skin which then burst and form the **golden crust.**

- This is much more common in neonates and children under 2 and patients will have **systemic symptoms such as fever and malaise**.
- Swabs of the vesicles can confirm diagnosis to help tailor antibiotic management, usually **flucloxacillin**.

Complications

- Sepsis
- Scarring
- Post strep glomerulonephritis
- Scarlet fever
- Staphylococcal scalded skin syndrome

TOXIC SHOCK SYNDROME

Aetiology

- Toxin producing **Staphylococcus aureus and group A streptococci**

Clinical Presentation

- Characterised by fever > 39
- Hypotension
- **Diffuse erythematous, macular rash**
- Causes organ dysfunction
    1. Mucositis: Conjunctivae, oral mucosa, genital mucosa
    2. Gastrointestinal: Vomiting/Diarrhoea
    3. Renal impairment
    4. Liver impairment
    5. Clotting abnormalities and thrombocytopenia

Management

- Intensive care support is needed to manage the shock and areas of infection should be debrided
- Antibiotics such as **ceftriaxone with clindamycin** are used to stop toxin production
- After 1-2 weeks, there is desquamation of the palms, soles, fingers and toes.
- There is a risk of recurrent skin and soft tissue infections

SCARLET FEVER

- An infectious disease caused by toxin producing strains of the bacterium **Streptococcus pyogenes.**
- It is highly contagious and is transmitted through infected saliva or mucus with aerosol transmission or direct contact.
- Common in those between 2-8 years of age

Risk factors

- Neonates

- Immunocompromised
- Concurrent chickenpox or influenza

Clinical Presentation

- Initial sore throat, fever, headache, fatigue and nausea/vomiting
- **Pinpoint, sandpaper like blanching rash** initially on the trunk then spreads to the rest of the body and flexures
- **Strawberry tongue**
- **Cervical lymphadenopathy**

Management

- Oral antibiotics such as **benzylpenicillin for 10 days**
- **Notify Public Health**

Renal

URINARY TRACT INFECTION

- Common infection which is important to investigate properly in children due to potential for structural abnormalities in the urinary tract and scarring of the kidneys if pyelonephritis develops which can lead to renal failure.
- Common organisms: E coli, Klebsiella, Proteus, Pseudomonas

Clinical Presentation

- Infants: Fever, vomiting, lethargy, poor feeding, jaundice, septicaemia, smelly urine and febrile convulsions
- Older Children: **Dysuria**, abdominal pain, fever, lethargy, vomiting/diarrhoea, haematuria, **smelly/cloudy urine**

Investigations

- A **clean catch urine sample** needs to be collected for dipstick which often can be very difficult for children.
- In the older child, a midstream urine sample can be used and cultured.
- **Ultrasound of urinary tract and kidneys**

Management

Antibiotics - IV for all those <3 months e.g Cefotaxime

Atypical UTI

- Seriously ill/Septicaemia
- Poor urine flow
- Abdominal mas
- Raised creatinine
- Failure to respond to Abx within 48 hours
- Infection with non E coli organism

All those with an atypical UTI should undergo ultrasound to look for abnormalities with potential **DMSA and MCUG scans** to look for scarring and vesicoureteric reflux.

Vesicoureteric reflux: Developmental abnormality where the ureters are displaced and enter directly into the bladder rather than at an angle causing reflux of urine into the renal pelvis and can cause scarring with UTI

UTI Prevention

- High fluid intake to produce a high urine output
- Regular voiding
- Ensuring complete bladder emptying
- Prevention/Treatment of constipation
- Prophylactic Abx can be considered

NOCTURNAL ENURESIS

- Known as 'bed wetting', this is a common problems of middle childhood
- There is a genetically determined delay in acquiring sphincter competence and emotional stress can cause secondary enuresis however underlying disorders should always be considered:
    1. UTI
    2. Faecal retention which is severe enough to reduce bladder volume and cause bladder dysfunction
    3. Polyuria from osmotic diuresis

Management

- Explanation to the child and the parent that this is common and beyond conscious control
- **Star charts**
- **Enuresis Alarm** which sounds when it becomes wet to awaken the child
- **Desmopressin** can be used to provide short term relief from bedwetting

NEPHROTIC SYNDROME

- **Proteinuria with 3+/4+ on urine dipstick or a urine protein:creatinine ratio of >200mmg/mol**
- **Hypoalbuminaemia <25g/l**
- **Oedema**

Aetiology

- Primary is generally an idiopathic cause: **Minimal change disease** is the most common cause in children
- Secondary can be caused by systemic diseases such as HSP, SLE

Clinical Presentation

- **Periorbital oedema**, often on awakening
- **Scrotal, vulval, leg and ankle oedema**

- Ascites
- **SOB** due to presence of pleural effusions and abdominal distension

Investigations

- **Urine dipstick**
- **Urine protein:creatinine ratio**
- Urine microscopy
- Urine cultures
- Bloods - FBC, U+E, albumin, bone profile

Management

Medical

1. Corticosteroid therapy:
- **Prednisolone** 60 mg/m_2 /day in a single morning dose (maximum 80mg/day) for 28 days.
- Then reduce dosage to 40mg/m_2 /alternate day (maximum 50mg/alternate day) given once daily, for 28 days and then stop without tapering.
2. Diuretics may be needed to control oedema whilst the steroids are taking effect: **furosemide 1-2mg/kg/day**
3. Diet with reduced salty food diet
4. **Pneumococcal immunisations** - 23 valent pneumococcal polysaccharide vaccine

Most children will have remissions and relapses however the relapses will generally become less frequent and may stop once they are in teenage years.

Steroid-resistant Nephrotic Syndrome

- Common in Asian boys, this is a potential complication of nephrotic syndrome and requires specialist paediatric nephrologist involvement.
- It can lead to hypovolaemia, thrombosis, infection and hypercholesterolaemia
- It may resolve or can cause relapses and can progress to renal failure
- Some patients may respond to cyclophosphamide, tacrolimus or rituximab
- Causes include focal segmental glomerulosclerosis (most common) and membranous nephropathy.

NEPHRITIC SYNDROME

Inflammation within the nephrons on the kidneys causing

- **Reduction in kidney function**
- **Haematuria**
- **Proteinuria**

Aetiology

1. Post Strep Glomerulonephritis
    - Occurs 1-3 weeks after a **B-haemolytic streptococcus infection** such as tonsillitis

- Immune complexes made up of streptococcal antigens, antibodies and complement proteins get lodged in the glomeruli and cause inflammation and AKI

2. IgA Nephropathy
    - This condition is related to HSP which is an IgA vasculitis
    - **IgA deposits in the nephrons of the kidneys** causing inflammation

Investigations

- Urine microscopy
- Protein and calcium excretion
- Kidney and urinary tract ultrasound
- Bloods including FBC, U+E, creatinine

Management

- Supportive therapy of the renal failure
- Diuretics and antihypertensive medications can be used for complications like HTN and oedema
- Immunosuppressant medications such as steroids

HYPOSPADIAS

- A condition affecting males where the urethral meatus (opening of the urethra) is abnormally displaced to the underside of the penis towards the scrotum.
- It is a congenital condition which affects babies from birth and is usually diagnosed on NIPE exam
- Management is surgery which is usually performed after 3-4 months of age and aims to correct the position of the meatus and straighten the penis.

ACUTE KIDNEY INJURY

Acute renal failure with oliguria (<0.5ml/kg/hour) is usually present

Aetiology

1. Prerenal (the most common cause in children): Hypovolaemia caused by infections such as gastroenteritis, burns, sepsis, haemorrhage and nephrotic syndrome
2. Renal: HUS, vasculitis, renal vein thrombosis, acute tubular necrosis, glomerulonephritis, pyelonephritis
3. Post renal: obstructions such as posterior urethral valves, blocker catheters

Management

1. Regular monitoring of circulation and fluid balance
2. Ultrasound scan to identify any obstruction of the urinary tract
3. Treatment depending upon the cause e.g. fluid replacement, assessment of the site of obstruction, renal biopsy.

4. Dialysis in severe cases

HAEMOLYTIC URAEMIC SYNDROME

- A triad of **acute renal failure, microangiopathic anaemia and thrombocytopenia**.
- Usually occurs secondary to GI infection, contact with farm animals or eating uncooked beef.

Pathophysiology

- Toxin enters the GI mucosa and localises to the endothelial cells in the kidney causing activation of the clotting cascade and consumption of platelets.
- Anaemia is caused by damage to RBC as they circulate

Clinical Presentation

- Reduced urine output
- Haematuria
- Abdominal pain
- Lethargy and irritability
- Confusion
- Oedema
- HTN

Management

- Early supportive therapy including dialysis usually gives a good prognosis
- Anti hypertensives if needed
- Careful maintenance of fluid balance
- Blood transfusions if needed

CHRONIC RENAL FAILURE

- **eGFR < 15ml/min**

Aetiology

- Structural malformations
- Glomerulonephritis
- Hereditary nephropathies
- Systemic diseases

Clinical Presentation

- Symptoms generally do not develop until renal function falls to less than ⅓ of normal and is often picked up on antenatal ultrasound
- Anorexia and lethargy
- Polydipsia and polyuria

- Faltering growth
- HTN
- Acute-on-chronic renal failure precipitated by infection/dehydration
- Incidental finding of proteinuria

Management

- Sufficient feeding with good protein intake to maintain growth -> this can supplemented with NG/gastrostomy feeding if necessary
- Phosphate restriction and activated vit D to prevent renal osteodystrophy
- Bicarbonate supplements to prevent acidosis
- EPO to prevent anaemia
- Growth hormone
- Dialysis and transplantation if necessary

Oncology

LEUKAEMIA

- Cancer of the stem cells in the bone marrow

1. ALL - most common in children - peaks ages 2-3 years
2. AML - next most common - peaks aged under 2 years
3. CML - very rare

Risk factors

- **Down's syndrome**
- Kleinfelters syndrome
- Radiation exposure during pregnancy
- Noonan syndrome

Clinical Presentation

- Persistent fatigue
- Unexplained fever
- Faltering growth
- Weight loss
- Night sweats
- Anaemia
- Petechiae and abnormal bruising
- Unexplained bleeding
- Abdominal pain
- Generalised lymphadenopathy
- Hepatosplenomegaly

Investigations

- NICE recommend any child with unexplained petechiae or hepatomegaly require specialist assessment

- FBC: if suspected, should be done within 48 hours: anaemia, leukopenia, thrombocytopenia
- Blood film which can show blast cells
- **Bone marrow biopsy** is the definitive investigation.

Management

- Chemotherapy
- Potential radiotherapy and bone marrow transplant

BRAIN TUMOURS

- These are almost always primary in children and are the leading cause of childhood cancer deaths in the UK.

1. Astrocytoma - varies from benign to highly malignant
2. Medulloblastoma - arises in the midline of the posterior fossa and may seed through the CNS via the CSF giving spinal metastases
3. Ependymoma - mostly in the posterior fossa where it behaves like medulloblastoma
4. Brainstem glioma
5. Craniopharyngioma - developmental tumour arising from an embryological remnant.

Clinical Presentation

- Often related to **raised ICP**: Headache worse on waking, coughing, straining or bending forward along with papilloedema
- Focal neurological signs may be detected
- Back pain, Peripheral weakness of arms/legs or Bladder/bowel dysfunction can also be present depending upon the level of the lesion.

Investigations

- **MRI scan**

Management

- **Surgery** is usually the first treatment and aimed at treating hydrocephalus
- Chemo and radio depends upon the tumour type and age of the patient.

NEUROBLASTOMA

- Tumour arising from the neural crest tissue in the adrenal medulla and sympathetic nervous system.
- It is a biologically unusual tumour that usually occurs before 5 years of age
- Generally, **prognosis is very good** with more than 80% of patients being cured

Clinical Presentation

- Abdominal mass
- Pallor
- Weight loss

- Hepatomegaly
- Bone pain
- Limp
- Cervical lymphadenopathy
- Periorbital bruising
- Skin nodules

Investigations

- Raised **urinary catecholamine levels**
- Biopsy
- **Bone marrow sampling**

Management

- **Surgery** can often be curative when there is no metastatic disease
- Metastatic disease will need chemotherapy, surgery and radiotherapy

WILMS TUMOUR

- Originating from embryonal renal tissue, it is the **most common renal tumour** of childhood which most often presents before 5 years of age

Clinical Presentation

- **Large abdominal mass**, often incidentally found in an otherwise well child
- Abdominal pain
- Anorexia
- Haematuria
- HTN

Investigations

- CT/MRI are often characteristic and show the intrinsic renal mass

Management

- Initial chemotherapy followed by delayed nephrectomy

BONE TUMOURS

- Ewing's sarcoma is seen more often in younger children but Osteogenic sarcoma is more common than Ewings.

Clinical Presentation

- Limbs are the most common site
- Persistent, localised bone pain is the most common symptom otherwise patients are generally well.

Investigations

- Plain X-ray is often first line and then followed by MRI and bone scan
- Bone x-ray shows destruction and variable periosteal new bone formation and with Ewings, there is often a substantial soft tissue mass **(onion skin appearance)**
- CT can be used to assess for lung metastasis

Management

- Combination chemotherapy is given before surgery
- In Ewings, radiotherapy is also used, especially if surgical resection is impossible.

RETINOBLASTOMA

- Malignant tumour of retinal cells, which accounts for around 5% of visual impairment in children and can be unilateral or bilateral.
- Bilateral tumours are hereditary
- Retinoblastoma susceptibility is on chromosome 13 with dominant inheritance
- Most cases present in the first 3 years of life

Clinical Presentation

- **White pupillary reflex replaces the red one**
- Squint

Investigations

- MRI and examination under anaesthetic

Management

- Aim is to cure but still preserve vision
- Treatment is based around ophthalmological findings
- Chemotherapy is used to shrink the tumour, particularly in bilateral disease and then followed by local laser treatment.
- Most patients are cured but many can be visually impaired but there is a significant risk of secondary malignancy

HEPATOBLASTOMA

- Primary malignancy liver tumour
- Usually presents with abdominal distension or with a mass, pain and jaundice is usually rare
- Elevated a-fetoprotein is detected in nearly all cases of hepatoblastoma.
- Management includes chemotherapy, surgery and in inoperable cases, liver transplantation is needed.
- Prognosis is generally good and children will be cured.

Psychiatry

ATTENTION DEFICIT HYPERACTIVITY DISORDER

- Altered levels of dopamine in the brain
- Structural and functional changes in the brain
- Genetic component

Aetiology
- More common in boys than girls (4:1) this is attributed to girls being able to cover up their symptoms until a later age
- 50% of children with ADHD also have another co-morbid such as ASD, dyslexia or depression/anxiety

Clinical Presentation:
1. Inattention
    - Does not complete instructions
    - Does not want to engage in intense tasks
    - Easily distracted
    - Difficulty organising tasks
    - Forgetful
    - Loses important things
2. Hyperactivity/Impulsivity
    - Cannot play quietly
    - Talks excessively
    - Does not wait their turn
    - Continuously on the go
    - Interrupt others
    - Answers questions prematurely

Diagnostic Criteria:
- Diagnosis< 16 years old = child must have at least 6 criteria from either category
- Diagnosis> 17 years old = child must have at least 5 criteria from either category
- Symptoms must have been present BEFORE 12 years old for > 6 months
- Symptoms must be present in more than one setting
- Must be evidence of impairment of child's function but also in line with child's developmental level

Investigations
- Conners questionnaire
- School observation
- Home visit and information from other relatives etc

Management
1. Non Medical
    - Care plans from teachers
    - CBT
    - Behavioural strategies
2. Medical
    - **Methylphenidate**- first line (S/E: **Cardiotoxic** so need baseline ECG before starting)
    - Lisdexamfetamine

- Dexamfetamine

AUTISM SPECTRUM DISORDER

- A neurodevelopmental disorder impacts social interaction, communication and behaviour.

Aetiology

- Genetic with multi gene involvement
- Structural changes within the brain

Clinical Presentation

1. Abnormal social interaction
    - Poor eye contact
    - Plays alone
    - Uninterested in social interaction
    - Difficulty forming close relationships
2. Impaired social communication
    - Failure to develop spoken language
    - Failure to initiate conversation
    - Abnormal rhythm, pitch and tone of speech
3. Repetitive ideas
    - Need for routine/rituals
    - Motor mannerisms: repetitive compulsive movements
    - Sensory issues: Only eat certain foods, do not like loud noises

Diagnosis

- Features from all 3 categories plus one of before aged 3
    1. Lack of social attachments
    2. Abnormal/delayed expression
    3. Abnormal symbolic play

Management

- Education care plans
- Applied behaviour analysis
- Family support/counselling
- MDT approach

Genetics

KLINEFELTER SYNDROME

- An additional X chromosome for men making them 47 XXY

Clinical Presentation

- **Taller height**
- Wide hips

- **Gynaecomastia**
- Weaker muscles
- **Small testicles**
- Reduced libido
- Shyness
- **Infertility**
- Subtle learning difficulties

Management

- Testosterone injections
- Advanced IVF techniques can potentially allow for IVF
- Breast reduction surgery
- MDT input to improve speech, language, strengthen muscles and provide education support.

Complications

- There is an increased risk of breast cancer, osteoporosis, diabetes and anxiety.

TURNERS SYNDROME

- Occurs when a female has a single X chromosome, making them 45 XO

Clinical Presentation

- **Short stature**
- **Webbed neck**
- **Widely spaced nipples with broad chest**
- High arching palate
- Downward sloping with ptosis
- **Underdeveloped ovaries with reduced function**
- Late or incomplete puberty
- Most women are infertile

Associated conditions

- Recurrent otitis media
- Recurrent UTI
- Hypothyroidism
- Hypotension
- Obesity
- Diabetes

Management

- **Growth hormone therapy** can help with short stature
- Oestrogen and progesterone replacement can help establish female sex characteristics, regulate menstrual cycle and prevent osteoporosis
- Regular monitoring is needed for the associated conditions

DOWNS SYNDROME

- 3 copies of chromosome 21

Clinical Presentation

- **Hypotonia**
- Small head with flat back
- **Short neck**
- **Short stature**
- Flattened face and nose
- Low set ears
- **Single palmar crease**
- Prominent epicanthic folds (folds of skin covering the medial portion of the eye and eyelid)
- Upward sloping palpebral fissures (gaps between the upper and lower eyelid)

1. Antenatal screening
    - This is offered to screen for chances of Down's syndromes where further investigations will take place if necessary. Older mothers generally have a higher risk.
2. Combined test
    - Performed at 11-14 weeks gestation and combines ultrasound results looking at thickness on the back of the neck of the foetus (thickened) and beta-HCG (raised) and PAPPA (reduced)
3. Triple Test
    - Performed at 14-20 weeks and looks at beta-HCG (raised), AFP (low) and serum oestriol (low)
4. Quadruple Test
    - Same as triple test but also includes inhibin-A (high)

Management

- MDT approach involves every medical professional and regular screening of the complications include echo, regular thyroid checks, regular eye checks and audiometry.

Complications

- Learning disability
- Recurrent otitis media
- Deafness
- Visual problems such as strabismus
- Hypothyroidism
- Cardiac defects such as ASD, VSD
- Leukaemia
- Dementia

NOONAN SYNDROME

- A genetic condition which is mainly autosomal dominant

Clinical Presentation

- **Short stature**
- Broad forehead
- Downward sloping eyes with ptosis
- **Wide space between the eyes**
- Low set ears
- **Webbed neck**
- **Widely spaced nipples**

Associated conditions

- Congenital heart disease, particularly pulmonary stenosis, hypertrophic cardiomyopathy and ADD
- **Undescended testes leading to infertility in males**
- Learning disability
- Bleeding disorders
- Increased risk of leukaemia

Management

- Supportive management with the MDT
- Often corrective heart surgery is needed for the congenital heart disease

FRAGILE X SYNDROME

- Caused by a mutation in the FMR1 gene on the X chromosome which codes for fragile X mental retardation protein which plays a role in cognitive development in the brain.
- X linked but unknown if dominant or recessive
- Males are always affected and females vary in how they are affected due to the other X chromosome

Clinical Presentation

- Delay in speech and language development
- Intellectual disability
- **Long, narrow face**
- **Large ears**
- **Large testicles after puberty**
- Hypermobile joints (particularly in the hands)
- ADHD
- Autism
- Seizures

Management

- Supportive for treating the symptoms and complications

PRADER-WILLI SYNDROME

- A genetic condition caused by loss of functional genes on the proximal arm of **chromosome 15** inherited from the father which can be due to deletion of this portion or when both copies are inherited from the mother.

Clinical Presentation

- Constant insatiable hunger that leads to **obesity**
- **Hypotonia as an infant**
- **Learning disability**
- Fairer, softer skin that is prone to bruising
- Mental health problems such as anxiety
- Dysmorphic features
- Narrow forehead
- Almond shaped eyes
- Strabismus
- Thin upper lip
- Hypogonadism

Management

- No cure
- Dietician management to control weight
- Growth hormone is indicated by NICE to improve muscle development and body composition
- Psychologists/Psychiatrists can help tackle mental health problems

ANGELMAN SYNDROME

- Loss of function of the UBE3A gene on the copy inherited from the mother caused by a **deletion on chromosome 15.**

Clinical Presentation

- Delayed development and learning disability
- Severe delay or absence of speech development
- **Fascination with water**
- Happy demeanour
- Widely spaced teeth
- Inappropriate laughter
- Abnormal sleep patterns
- Epilepsy
- ADHD
- Dysmorphic features
- **Fair skin, light hair and blue eyes**

Management

- MDT approach to managing individual problems

WILLIAM SYNDROME

- Deletion of epigenetic material on one copy of chromosome 7 resulting in just a single copy of the genes - usually due to a random deletion during conception.

Clinical Presentation

- Broad forehead
- **Starburst eyes (star like pattern on the iris)**
- Flattened nasal bridge
- **Very sociable, trusting personality**
- **Wide mouth with widely spaced teeth**
- Small chin

Management

- MDT approach to managing individual problems

Complications

- **Supravalvular aortic stenosis**
- ADHD
- Hypertension
- **Hypercalcaemia**

EDWARDS SYNDROME

- **Trisomy 18**

Clinical Presentation

- Low birthweight
- Small mouth and chin
- Short sternum
- **Flexed, overlapping fingers**
- **‘Rocker-bottom feet’**
- Cardiac and renal malformations

PATAU SYNDROME

- Trisomy 13

Clinical Presentation

- Structural defects of the brain
- **Scalp defects**
- **Small eyes and other eye defects**
- **Cleft lip and palate**

- Polydactyly
- Cardiac and renal malformations

DUCHENNE MUSCULAR DYSTROPHY

- Affecting around 1 in 4000 male infants, this is an inherited X linked recessive disorder resulting from a deletion on the short arm of the X chromosome.

Pathophysiology

- This site codes for a protein called dystrophin which normally connects the cytoskeleton of muscle fibres to the extracellular matrix through the cell membrane therefore deficiency results in myofiber necrosis and raised serum CPK.

Clinical Presentation

- Waddling gait + slow at running + mount stairs one by one
- Language delay
- **Gower's sign - they need to turn prone to rise up from the floor**
- Pseudohypertrophy of the calves as the muscular tissue is replaced by fat/fibrous tissue
- **Boys will be slower and clumsier than their peers**
- They will no longer be able to walk by around 10-14 years due to the progressive muscular atrophy

Management

- Exercises to help maintain muscle power and mobility
- Night splints and passive stretching
- Good sitting posture helps reduce the likelihood of scoliosis
- Corticosteroids can be given to help preserve mobility and prevent scoliosis

Becker Muscular Dystrophy - Some functional dystrophin is produced therefore the features are similar as DMD however the disease progresses more slowly with age on onset being later.

Musculoskeletal

OSTEOGENESIS IMPERFECTA

- An autosomal dominant genetic condition that results in brittle bones that are susceptible to fractures.
- A genetic mutation that affects the formation of collagen which is needed to maintain the structure and function of bone, skin, tendons and other connective tissues.

Clinical Presentation

- **Recurrent and inappropriate fractures**

- Blue/grey sclera
- **Hypermobility**
- **Triangular face**
- Deafness from early adulthood
- Dental problems
- Bone deformities

Investigations

- Mainly a clinical diagnosis
- X-rays can be helpful for diagnosing fractures and deformities
- Genetic testing is done rarely

Management

- **Bisphosphonates** to increase bone density
- **Vit D supplementation**
- Physio and occupational therapy input to maximise strength and function
- Management of fractures

RICKETS

- A condition where there is defective bone mineralization causing ‘soft’ and deformed bones

Aetiology

- Vitamin D deficiency - produced by the body in response to sunlight or through food such as eggs, oily fish
- Calcium deficiency - found in dairy products and some green vegetables
- Hereditary hypophosphatemic rickets - an X-linked dominant condition

Pathophysiology

- Vit D is a hormone created from cholesterol by the skin in response to UV radiation.
- Those with malabsorption disorders such as IBD are more likely to have Vit D deficiency as well as those with CKD.
- Vitamin D is essential in calcium and phosphate absorption from the intestines and kidneys as well as regulating bone turnover and promoting bone reabsorption.
- Inadequate vit D leads to a lack of calcium and phosphate which are needed for bone formation therefore there is defective bone mineralisation.
- Low calcium causes secondary hyperparathyroidism as the parathyroid gland tries to raise calcium levels by secreting PTH which stimulates increased reabsorption of calcium and causes further bone mineralisation problems.

Risk factors

- Darker skin
- Low exposure to sunlight
- Colder climates

- Spending majority of time indoors

Clinical Presentation

- Lethargy
- Bone pain
- Poor growth
- Dental problems
- Muscle weakness

Bone deformities

1. **Bowing of the legs - legs curve outwards**
2. Knock knees - legs curve inwards
3. Rachitic rosary - the ends of the ribs expand at the costochondral junctions causing lumps along the chest
4. Craniotabes - soft skull with delayed closure of the sutures and frontal bossing
5. Delayed teeth

Investigations

- Serum 25-hydroxyvitamin D - <25 nmol/L establishes deficiency
- X-rays
- Serum calcium and phosphate may be low
- Serum ALP and PTH may be high
- Full blood tests to rule out other pathology including FBC, ESR, CRP, LFTs, TFTs, Malabsorption screen

Management

- Prevention is the best management - NICE recommend **400 IU supplements** for children and young people
- Children with deficiency can be treated with ergocalciferol (vit D)
- For those with diagnosed rickets, vit D and calcium supplementation is needed.

TRANSIENT SYNOVITIS

- Irritable hip - most common cause of hip pain in children aged 3-10
- Temporary irritation and inflammation in the synovial membrane - often associated with a viral URTI

Clinical Presentation

- **Symptoms usually occur within a few weeks of a viral illness**
- Limp
- **Refusal to weight bear**
- Groin or hip pain
- Mild low grade temperature
- Otherwise well - no signs of systemic illness

Management

- Symptomatic management
- **Exclusion of other diagnoses particularly septic arthritis**
- Generally good prognosis with recovery within 1-2 weeks without any long term effects

SEPTIC ARTHRITIS

- Infection inside a joint - most common in children under 4 years

Aetiology

- Staphylococcus aureus
- Neisseria gonorrhoea in sexually active teenagers
- Group A Strep - Strep Pyogenes
- Haemophilus influenzae
- E coli

Clinical Presentation

- Only affects a single joint - knee or hip
- **Hot, red, swollen and painful joint**
- Refusal to weight bear
- Stiffness and reduce range of motion
- Fever, lethargy and sepsis

Management

- Admission to hospital with involvement of the orthopaedic team
- **Joint aspiration** prior to antibiotics -> gram staining, crystal microscopy, culture and antibiotic sensitivities
- **Empirical IV antibiotics** followed by specific antibiotics once sensitivities are received
- Surgical drainage and washout may be needed

OSTEOMYELITIS

- Infection of the bone and bone marrow - typically in the metaphysis of the long bones
- **Staph aureus** is the most common organism
- Chronic osteomyelitis is a deep seated, slow growing infection with slowly developing symptoms
- Infection can be introduced directly into the bone e.g open fracture or travelled to the bone from the blood after entering through another medium

Risk Factors

- Males under 10
- Open bone fractures
- Orthopaedic surgery

- Immunocompromised
- Sickle cell anaemia
- HIV
- TB

Clinical Presentation

- Systemic symptoms such as fever
- **Refusing to use the limb or weight bear**
- Pain
- Swelling
- Tenderness

Investigations

- X-rays are first line investigation
- **MRI** are the gold standard imaging investigation
- Bloods including CRP, ESR and white cells
- Blood cultures
- Bone marrow aspiration

Management

- Extensive and prolonged antibiotic therapy
- Surgery may be needed for drainage and debridement of the infected bone

PERTHES DISEASE

- Disruption of blood flow to the femoral head causing **avascular necrosis of the bone** - affecting the epiphysis of the femur
- Most common between 5-8 year boys but can affect all those from 4-12 years of age
- Mainly idiopathic
- Over time, there is revascularization or neovascularization and healing of the femoral head with remodelling of the bone as it heals

Clinical Presentation

- Slow onset of pain in the hip or groin
- Limp
- Restricted hip movements
- Referred pain to the knee
- No history of trauma

Investigations

- X-ray which can be normal
- Blood tests can typically normal, particularly inflammatory markers
- Technetium bone scan
- MRI scan

Management

- Initial management is conservative to maintain healthy position and alignment in the joint and reduce the risk of damage or deformity to the femoral head including bed rest, traction, analgesia and crutches
- Physiotherapy is used to retain movement in the muscles and joints
- Regular x-rays
- Surgery may be used in severe cases, older children or those that are not healing

SLIPPED FEMORAL EPIPHYSIS

- Head of the femur is displaced along the growth plate
- More common in males and typically between ages of 8-15 years old
- **More common in obese children**

Clinical Presentation

- Adolescent, obese male undergoing a growth spurt
- History of minor trauma which may trigger the onset of symptoms
- Hip, groin, thigh or knee pain
- Restricted range of hip movement
- **Painful limp**
- Restricted movement in the hip
- **Wanting to keep the hip in external rotation with restricted internal rotation**.

Investigations

- X-ray is first line
- Blood tests are normal, but inflammatory markers may be used to exclude other causes of joint pain
- CT/MRI scan

Management

- **Surgery** is needed to return the femoral head to correct position and fix it in place

OSGOOD-SCHLATTER DISEASE

- Caused by inflammation at the **tibial tuberosity** where the patellar ligament inserts - a common cause of anterior knee pain in adolescents
- Typically occurs in male patients aged 10-15 years
- Normally is unilateral but can be bilateral

Pathophysiology

- Patella tendon inserts into the tibial tuberosity which is at the epiphyseal plate
- Stress from running, jumping and other movements at the same time as growth in the plate results in inflammation on the tibial epiphyseal plate

- There are small avulsion fractures where the patellar ligament pulls away tiny pieces of the bone, leading to growth of the tibial tuberosity, causing a visible lump below the knee
- Initially, the bump is tender due to inflammation but as the bone heals and inflammation settles, it becomes hard and non-tender.

Clinical Presentation

- Gradual onset of symptoms
- Visible or palpable hard and tender lump at the tibial tuberosity
- Pain in the anterior aspect of the knee
- **Pain is exacerbated by physical activity, kneeling and on extension of the knee**

Management

- Reduction in physical activity
- Ice
- **NSAIDs for symptomatic relief**
- Stretching and physiotherapy can be used to strengthen the joint once symptoms settle.
- Symptoms will generally resolve over time but the patient is usually left with a hard, boney lump on their knee.

DEVELOPMENTAL DYSPLASIA OF THE HIP

- Structural abnormality in the hips caused by abnormal development of the foetal bones during pregnancy leading to instability in the hips and a tendency for dislocation
- Usually picked up during the newborn examinations or later when the child presents with hip asymmetry, reduced range of movement in the hip or limp.

Risk Factors

- First degree family history
- **Breech presentation from 36 week onwards**
- **Breech presentation at birth if 28 week onwards**
- Multiple pregnancy

Screening

- NIPE examination usually picks this up - this may be suggested with
    1. Different leg lengths
    2. Restricted hip abduction on one side
    3. Significant bilateral restriction in abduction
    4. Difference in the knee level when the hips are flexed
    5. Clunking of the hips on special tests - Ortolani and Barlow tests

Investigations

- If suspected, **ultrasound** will establish the diagnosis

- X-rays can also be helpful in older infants

Management

- **Pavlik harness** if the baby presents at less than 6 months of age which is fitted and kept on permanently and adjusts for the growth of the baby.
- The aim of this is to hold the femoral head in the correct position to allow the hip socket to develop a normal shape and helps to keep the baby's hips flexed and abducted - reviewed after 6-8 weeks
- Surgery may be needed when the harness fails or if the diagnosis is made after 6 months of age

JUVENILE IDIOPATHIC ARTHRITIS

- Autoimmune inflammation which occurs in the joints
- Diagnosed when there is arthritis without any other cause, lasting more than 6 weeks in a patient under the age of 16
- Key features are joint pain, swelling and stiffness

1. Systemic JIA

Clinical Presentation

- Subtle **salmon-pink rash**
- High swinging fever
- **Enlarged lymph nodes**
- Weight loss
- Joint pain and inflammation
- Splenomegaly
- Muscle pain

Investigations

- ANA and RF will be negative
- Raised CRP, ESR, platelets and serum ferritin.

2. Polyarticular JIA

- Idiopathic inflammatory arthritis in **5 joints or more** which tends to be symmetrical and can affect small or large joints.
- **Minimal systemic symptoms** but can have mild fever, anaemia and reduced growth
- Most children will be RF negative but in older children and adolescents, they can be RF positive - similar pattern to RA.

3. Oligoarticular JIA

- Involves **4 joints or less, often only a single joint**
- More common in girls under the age of 6 and affects the larger joints
- Classic feature is **anterior uveitis** for which patients should be referred to ophthalmology

- No systemic symptoms, inflammatory markers may be normal or mildly elevated, ANA positive but RF is usually negative.

4. Enthesitis-Related Arthritis

- More common in male children over 6 years
- The paediatric version of the seronegative spondyloarthropathies such as ankylosing spondylitis
- Enthesitis is inflammation at the point where the muscle inserts into a bone - can be caused by traumatic stress or an autoimmune process.
- Majority of patients will have **HLA-B27 gene**
- They may have signs and symptoms of psoriasis and IBD
- Prone to anterior uveitis

5. Juvenile Psoriatic Arthritis

- Seronegative inflammatory arthritis associated with psoriasis
- Can be symmetrical polyarthritis affecting the small joints or an asymmetrical arthritis affecting the large joints in the lower limbs
- May have nail pitting, dactylitis, enthesitis, plaques of psoriasis on the skin

Management of JIA

- NSAIDs such as ibuprofen
- Steroids either oral, IM or intra-articular in oligoarthritis
- DMARDS such as methotrexate
- Biologicals such as TNF inhibitors such as infliximab/adalimumab