Notes on Paediatrics

Accidents

Most common cause of death in children.
Toddlers – falls, scalds, drowning, choking.
Children – falls, RTAs.

Burns
Superficial – red, no blisters, only epithelium.
Partial – pink or mottled, blisters, some dermal damage.
Full thickness – painless, white, full dermal and nerve damage.

Admit if over 5% full thickness, difficult area e.g. face (scarring, airway involvement), hands and feet (contractures, functional loss), perineum (infection, difficult to nurse), or inhalation injury.
Calculate surface area with Lund and Browder chart.
Give IV morphine, treat shock with fluids, dress wounds.

Near-drowning
Problems are hypoxia (due to laryngospasm) and hypothermia.
Need resuscitation and slow warming. Hospitalise for 24hrs due to risk of pneumonia and pulmonary oedema.
Poor prognosis – submerged for long time, no gasp after 40mins, low temperature, persisting coma, low pH, low pO2.

Poisoning
Mostly accidental by toddlers. Sometimes self-harm in adolescents or deliberate poisoning.
Establish – what ingested, amount ingested, time ingested.
Inspect oropharynx and vomit. Assess level of consciousness and features relating to specific poisons (small pupils – opiates, tachpnoea – salicylate, arrhythmias – TCAs, digoxin).
Measure blood levels.
Activated charcoal binds most drugs (except iron and lithium) and is useful if given early.

Paracetamol – liver failure, IV N-acetylcysteine.
Iron – shock and haemorrhage, liver failure, treat with IV desferrioxamine.
Aspirin – hyperventilation, tinnitus, metabolic acidosis, alkaline diuresis.
TCA – tachycardia and arrhythmias, dry mouth, blurred vision, convulsions.
Household substances
Plants and berries
Paediatric Cardiology

**Foetal circulation**
In utero, R pressure > L so blood flows RL through foramen ovale and DA and bypasses lungs. Foramen ovale allows oxygenated blood from placenta to enter LA and systemic circulation. DV allows blood returning from placenta to bypass liver. At birth, cord clamping and lung expansion cause pressures to change, closing foramen ovale and DV and umbilical veins collapse. DA closes by muscular contraction in response to rising oxygen tension, then anatomically over months.

**Murmurs**
Mostly innocent.
**Innocent murmurs** – turbulent flow in structurally normal system, may appear when febrile due to increased CO. Asymptomatic child, normal CVS examination, systolic, no radiation, changes with posture. Ejection murmurs – soft, blowing, systolic, 2nd or 4th L IC space. Venous hums, head and neck veins – continuous rumble, disappear on lying flat.

**Significant murmurs** – symptoms e.g. syncope, cyanosis, CVS signs, diastolic, radiates, thrill. Can be caused by PS or PDA (continuous machine like sound under clavicle).

**Cyanosis**
Cyanosis occurs when [deoxy Hb]>5g/dl. Peripheral cyanosis (acrocyanosis) is normal in cold, crying and unwell babies.
Distinguish central cyanosis due to respiratory disease from cardiac disease by change in artery PO2 after breathing 100% oxygen. Differential cyanosis between limbs is cardiac disease.
Due to RL shunt or abnormal mixing (increased pulmonary vascularity, systemic venous return bypasses lungs) or inadequate pulmonary blood flow (oligaemic lung fields).

**Congenital heart disease**
8/1000 live-born infants. 15% have multiple abnormalities, 15% also have non-cardiac abnormality. In most cases cause is unknown. Some causes recognised are chromosomal disorders: Down (AVSD), Turner (AS, coarctation) and teratogens: rubella (PDA, PS), alcohol (ASD, VSD), lithium (Ebsteins anomaly – high tricuspid valve).
All (except ostium secundum defects) are at risk of endocarditis so need prophylactic antibiotics for surgery and dental procedures.

Investigate initially by CXR (cardiac shadow, plethora with LR shunt, oligaemia with reduced pulmonary blood flow) and ECG, usually diagnoses by echo and Doppler.
Most are treated by surgery – mortality has fallen to about 5%.

Acyanotic – lesions which allow blood to shunt LR or which obstruct the flow of blood by narrowing a valve or vessel.
Cyanotic – decreased pulmonary blood flow with RL shunt (TOF) or increased pulmonary blood flow with abnormal mixing (TGA).

**Acyanotic heart defects**
PDA – usually closes by 4th day of life, diagnosed if not closed after 1month. Common in preterm infants, Down’s and high altitudes. Due to immaturity and hypoxia. Presents when pulmonary vascular resistance falls causing LR shunt as tachycardia, murmur (systolic then as pulmonary resistance falls continuous machinery-like), bounding pulse and difficulty weaning ventilated infants.
CXR and ECG changes are the same as a large VSD. Usually closes spontaneously but if persists treat as high risk of endocarditis and pulmonary hypertension. If baby is ventilated or developing heart failure treat. Treatment is with PG inhibitor e.g. indomethacin or surgical closure (division, ligation, transvenous umbrella occlusion).
ASD – distinguish from PFO as large and always open rather than open only if raised atrial pressure or volume. Ostium secundum: most common, high in septum, usually asymptomatic, pulmonary hypertension uncommon, but can develop to AF and heart failure if persists. Ostium primum – lower, associated with mitral regurg, found in Down’s. Get abnormal RV impulse, widely split second sound, tricuspid (mid diastolic, LSE) and pulmonary flow murmurs, RV hypertrophy and pulmonary plethora due to increased R sided flow. Surgical treatment to prevent R cardiac failure in later life at about 3yrs.

Coarcatation of the aorta – male preponderance, key feature is weak femoral pulses. Preductal – sick neonate with absent pulses, whilst duct is open RV supplies systemic circulation, cardiac failure when duct closes. Give PG infusion to maintain duct patency and transfer for surgery. Postductal – asymptomatic children, may get leg pains or headache. Hypertension in the arm and weak or absent femoral pulses. May have ejection click (if bicuspid aortic valve associated) and systolic ejection murmur radiating to back. Needs surgical correction by balloon dilatation or resection.

Severe form is interrupted aortic arch – no connection between aorta proximal and distal to ductus arteriosus.

Aortic stenosis – male preponderance. Varying signs depending on severity: mild (<30mmHg gradient) – asymptomatic murmur, severe – heart failure in infant or syncope and chest pain in older child. Avoid strenuous exercise. Surgical treatment – balloon or surgical valvotomy, aortic valve replacement. Often associated with mitral valve stenosis or coarctation.

Pulmonary stenosis – mostly valvular, also subvalvular (TOF) or supravalvular. Usually mild (<30mmHg gradient) and asymptomatic with widely split S2, systolic ejection click if valvular and systolic ejection murmur radiating to back. If severe develop heart failure. Treat with balloon dilatation or valvotomy if moderate or severe.

Hypoplastic left heart syndrome – very sick neonates with duct dependent circulation. Weakness of all pulses. Start PG infusion to keep duct open. Needs neonatal heart transplant, otherwise Norwood’s procedure is palliative.

VSD
Most common cause of CHD.
Acyanotic unless pulmonary hypertension and RV hypertrophy cause flow to become RL (Eisenmenger’s syndrome, rare until 2nd decade). This causes irreversible increase in pulmonary vascular resistance so needs heart-lung transplant.
Problems are R heart failure and pulmonary hypertension.

Small VSD – asymptomatic, pansystolic murmur. Need antibiotic prophylaxis against endocarditis for dental extractions, but otherwise no treatment. Most close spontaneously.

Medium VSD – slow weight gain, difficulty feeding and chest infections in infancy. Pansystolic murmur, thrill and increased cardiac impulse. CXR shows cardiac enlargement and pulmonary plethora. ECG show RV hypertrophy. Treat heart failure, spontaneous closure often occurs, if still present treat at about 4yrs.

Large VSD – early heart failure. Medical treatment of heart failure and surgery. Banding PA to increase RV pressure and decrease shunt gives respite until child is big enough for correction.

Tetralogy of Fallot
Aorta over-riding ventricular septum, large VSD, RV hypertrophy, pulmonary stenosis.
Get RL shunt so blood in aorta is mostly deoxygenated from R ventricle. Oxygenation in lungs depends on blood flowing LR across patent DA, as this closes get severe cyanosis and no blood through lungs. Usually present in first months with cyanotic spells, clubbing, loud S2, ejection systolic murmur. Get RV hypertrophy on CXR and ECG and oligaemic lung fields. Don’t get congestive cardiac failure.
Treat prolonged cyanotic spells with morphine (relieves pain and hyperpnoea), sodium bicarbonate (to correct acidosis) and propanolol (to cause peripheral vasoconstriction and stop spasm). Treat surgically from about 4 months – close VSD and widen RV outflow tract. Palliative procedures in infants – shunt between subclavian and pulmonary arteries to increase pulmonary blood flow.

**Transposition of great vessels**
Aorta arises from RV and PA from LV. Depends on mixing via foramen ovale, ductus arteriosus or VSD to be compatible with life.
Severe early cyanosis, worsened with closure of DA. Profound hypoxaemia, unresponsive to inhaled oxygen. Immediate management is to improve mixing of blood – PG infusion to open DA, emergency balloon atrial septostomy (Rashkind procedure). Definitive surgical repair at a few weeks.

**Arrhythmias**
Sinus arrhythmia is more pronounced in children.
Arrhythmias are very rare except paroxysmal supraventricular tachycardia. In this HR >220, usually asymptomatic but infants may get heart failure. Most have accessory connection between atria and ventricles (WPW syndrome). Stop acute episode by vagal stimulation (carotid sinus massage, ice cold compress), IV adenosine or DC cardioversion if these fail. Most will have no further episodes, if it frequently recurs, ablate bypass tract.
1\textsuperscript{st} degree heart block (prolonged QT interval) – ASD, Ebstein’s anomaly, myocarditis.
Complete heart block – antiRo antibodies.
On ECGs P wave morphology is rarely significant, normal children can get partial RBBB.

**Heart failure**
**Infants** – poor feeding, cough, breathlessness, tachypnoea and tachycardia, hepatomegaly, 3\textsuperscript{rd} heart sound, rapid weight gain from oedema.
Either due to pressure overload (obstructive lesions e.g. coarctation of the aorta, aortic valve stenosis, hypoplastic left heart syndrome, often depend on RL flow through duct), or volume overload (LR shunt which increases as pulmonary resistance falls e.g. VSD, PDA).
**Children** – dilated cardiomyopathy, HOCM (AD with incomplete penetrance), rarely CHD, valvular heart disease, Kawasaki disease, CAD in FH.
Get breathlessness on exertion, cyanosis, finger clubbing, growth retardation.
Usually treat medically.

**Myocarditis**
Mostly viral (coxsackie B).
Get cardiomegaly, hepatomegaly, tachypnoea, cyanosis and shock.
Treat with rest, oxygen, diuretics, vasodilators, steroids.
Can become chronic – dilated cardiomyopathy.
Community Paediatrics

Child health promotion
Neonatal check – congenital abnormalities, otoacoustic emission, Guthrie test (PKU, hypothyroidism to avoid irreversible brain damage, sometimes CF, MD). Register birth within 6wks by anyone present at birth.
6 weeks - congenital abnormalities, hips, growth, development, vision and hearing, family’s adjustment to child, maternal depression, advice on feeding.
6-9 months – squint, distraction hearing, growth and development, guidance on diet, safety, sleep habits.
18-24 months – development, growth, toilet training and behaviour advice.
36-42 months – detect health problems before school. Vision and hearing test.

Primary prevention – prevent disease starting e.g. education, immunisation.
Secondary prevention – treat asymptomatic abnormalities e.g. Guthrie test.
Tertiary prevention – limit disease and disability.

Consent – if older than 16yrs can give consent. If below 16, need parental consent unless: emergency treatment required or child consents and doctor believes child can make informed decision and will not consent to parent being asked (Gillick). Either parent can give consent and has rights and responsibilities (if on birth certificate).

Adoption – rights and responsibilities pass to adopting parents irreversibly after 3 months. Original parents have no right of access and adopted child has no right to claim maintenance from them.

Impaired hearing
Conductive – otitis media with effusion (affects over half pre-school children), foreign body, wax, otosclerosis.
Sensorineural – congenital infection (rubella, CMV), drug toxicity, prematurity (esp hypoxia and jaundice, <32wks), meningitis, genetic (either isolated or part of syndrome). Most congenital deafness is sensorineural (about half inherited), most in children is due to glue ear (this usually resolves by adolescence).

Tests: newborn screening otoacoustic emission, 8 months distraction test, up to 2 years threshold audiometry and impedance tympanometry if suspected problems, school entry sweep test to screen all children.

Speech delay
Normal speech development needs hearing others speaking, understanding the meaning of words, being able to make sounds, putting words together.

Refer to speech therapist. Assess comprehension and possibility of global delay. Assess hearing.

Abuse and child protection
Risk factors for abuse – young, immature isolated parent, low socio-economic group, stress, drug abuse, parental psychiatric disorder, child that is difficult to look after (e.g. medical condition), child that is unwanted (e.g. step child), young children (can’t complain, need more care), premature babies, parents who were abused.

Child in need – unlikely to achieve a reasonable standard of health or development without provision of services or disabled (e.g. deaf, blind, mental handicap). Impairment is loss of structure or function, disability is restriction of ability to perform, handicap is the resultant disadvantage.

Physical abuse – NAI. Features in history that raise concern – delay in seeking help, implausible explanation, changing story, trauma inappropriate for age, previous unexplained injury, unconcern about injury. Examination – neglect, withdrawn.
Suspicious injuries – frenulum laceration, post rib fractures, bruises behind ear, genital injuries, old
injuries, bruises in a non-mobile baby, bruises on face, back and buttocks, bruises in shape of hand
or belt, multiple bruises of different ages. Shaken baby syndrome – subdural haematoma, retinal
haemorrhage, rib and limb fractures, fractured skull, torn frenulum, bruises.

**Neglect and emotional abuse** – neglect = failure to meet child’s needs. Emotional abuse =
persistent emotional ill treatment. Present with failure to thrive, developmental delay, poor hygiene,
inappropriate behaviour.

**Management** – record carefully with diagrams and photographs (with consent). Date, time and sign.
Exclude bruising disorders, skeletal survey.
Section 47 is the local authorities duty to investigate if child abuse is suspected.
Can get emergency protection order from social services (8 days) or police protection order (72hrs).
Behavioural Paediatrics

Enuresis
Involuntary discharge of urine at an age where continence is expected (5yrs, slightly later in boys, older for nocturnal). Average child is dry by day at 2 and by night at 3. Classify as primary or secondary (previously continent) and nocturnal (most) or diurnal. A delay can be due to a global developmental delay.

Nocturnal enuresis – multifactorial with genetic, emotional and cultural factors. Rarely organic cause – UTI, diabetes, chronic renal failure, neuropathic bladder, faecal retention causing bladder neck dysfunction, congenital abnormality e.g. ectopic ureter. Management – support and advice. Discourage parental intolerance (problems is very common) and incentives (e.g. sleeping in parent’s bed). Diary of wetting. If under 5 years, should resolve. If over, star charts, alarms. Rarely drugs – ADH, tricyclic antidepressants.

Encopresis
Faecal continence should be achieved by 4 years. Either retentive (overflow incontinence, most cases) or non-retentive (sphincter problem or psychiatric). Related to harsh toilet training and constipation (rectum becomes distended so child is unaware of need to empty it). Psychological problems can be a cause and result. Secondary encopresis with no constipation suggests a psychiatric cause.

Management – investigate bowel history, family’s functioning, neurological system and rectum. If due to retention, empty rectum as soon as possible (enema, stool softener or laxative). Encourage regular defaecation by habit and high fibre diet.

Hyperkinetic disorder and ADHD
Inattention, hyperactivity and impulsiveness for at least 6 months in more than one situation such that function is impaired. More common in boys, often family history. Usually present when starting school. Physical examination to exclude developmental delay, problems with hearing or vision, syndromes and organic causes e.g. hyperthyroidism. Can attempt to quantify with Conners questionnaire. Manage with behaviour modifying and education – structured environment, positive reinforcement, cognitive training to improve self-control and relaxation. Drug treatment if this fails – methylphenidate (slow growth and hypertension so need to monitor height, weight, pulse and BP), atomoxetine.

Conduct disorder
This has a prevalence of 4-8% and is more common in urban areas and boys. It involves persistent dissocial, aggressive and defiant behaviour.
Management involves behaviour modification, parent training, family therapy, support for schools. Medication is only used if there is comorbidity e.g. ADHD. 40% of sufferers are antisocial as adults.

Sleep disorder
The average baby sleeps for 15hrs per day. At about 4months night-time feeds can often be discontinued.

Toddlers: Difficulty in getting to sleep – separation anxiety, fear of darkness, erratic bedtime routine, use of bedroom as punishment, reward for getting up (majority, e.g. a story). Create a predictable routine and leave child to settle for lengthening periods.
**Nightmares and terrors** – nightmares are bad dreams that can be recalled by children, normal unless very frequent or stereotyped, need reassurance. Night terrors are parasomnia when child emerges from deep sleep into state of high arousal and confusion. Child is often found sitting with open eyes but is disorientated and unresponsive and settles with no recall. Waking child before terror is expected to occur may break pattern.

**Autism**
Involves delayed and abnormal communication, impaired social interaction e.g. eye contact and restricted interests and activities.
More common in males and usually evident by 3 years e.g. solitary play, withdrawn, poor communication, ritualistic, compulsive, poor eye contact, head banging, rocking.
Asperger's syndrome is on the autism spectrum but language is preserved.
Strong genetic component, may be found in isolation or with fragile X, tuberous sclerosis etc.
2/3s of children also have a learning disability and ¼ have epilepsy.
Management involves giving support and advice and behavioural techniques.

**Eating disorders**
**Food refusal** – toddlers become fussy but are well nourished. Advise to avoid snacks between meals, irregular meals, unsuitable food and punitive methods.

**Anorexia nervosa** – low weight, fear of being fat, disturbed body image, amenorrhoea. Mainly girls. Emaciated, lanugo hair, bradycardia, endocrine abnormalities. Admit if severe, aiming for gradual weight gain and psychological support.

**Psychosomatic disorders**
**Recurrent pain without organic cause** – rare in children. Strict divide between organic and psychological causation is unhelpful. Explain as dysfunctional – child is more vulnerable to pain in response to stress. For abdominal pain, the further away from the umbilicus, the more likely to be organic (Apley's rule).
Encourage normal activity and offer symptomatic relief.
**Chronic fatigue syndrome** – persistent subjective fatigue, often with non-specific pain. Exclude other causes e.g. anaemia, hypothyroidism. Support with symptom relief and psychological support.

**Separation reaction**
If separated from mother in strange surroundings – protest and mounting anxiety, withdrawn state and despair then cheers up but detachment when mother reappears.
Congenital Abnormalities

Cleft lip and palate
Failure of fusion of frontonasal and maxillary processes.
Can be cleft lip alone, cleft palate alone, or cleft lip and palate – bilateral or unilateral.
Polygenic inheritance but may be associated with maternal anticonvulsants.
Can breastfeed some, others may need special long teats or other feeding devices, esp if cleft palate.
Surgical repair – 6-12 months on palate, either 1st week or 3 months on lip.
Most will need speech therapy.

Pierre-Robin anomaly – cleft palate, micrognathia, posterior displacement of the tongue.
Leads to breathing difficulties. Position prone to maintain airway until mandible grows.

Neural tube defects
Failure of fusion of neural tube in first 4 weeks.
Incidence reduced by prophylactic folic acid supplements and antenatal screening.
Spina bifida occulta – absent posterior vertebral arch as hasn’t fused, hairy patch on skin, no neurological deficits but may appear with growth and cord tethering.
Meningocele – herniation of pia and arachnoid filled with CSF. No neurological deficit, can be surgically closed.
Myelomeningocele – 90% of overt spina bifida. Usually open with exposed meninges and leaking CSF. Neurological deficits e.g. motor and sensory loss in lower limbs, neuropathic bladder and bowel. Often also get scoliosis and hydrocephalus due to Arnold-Chiari malformation (herniation of cerebellar tonsils through foramen magnum). Surgery prevents infection but cannot restore neurological function.

Hypospadias
Spectrum of congenital abnormalities of position of urethral meatus.
Have proximal ventral urethral orifice (due to failure of fusion of inner genital folds), dorsal hooded foreskin (hasn’t fused ventrally) and in severe forms chordee.
If severe and no palpable gonads, need to investigate as intersex.
Carry out surgery before 2yrs.

Developmental dysplasia of the hip
Perinatal hip instability leads to progressive malformation of joint. Spectrum from dislocated hips to acetabular dysplasia (shallow acetabulum). Mostly congenital but can develop after birth. Either typical or tetralogical (neurological and genetic conditions).

Clinically screen all babies at birth and 6wk check but about 40% missed. May have asymmetrical skin creases (though this can be normal) present with delayed walking, painless limp or waddling gait. USS is controversial. Risk factors are congenital muscular torticollis, congenital foot abnormalities, breech or caesarean delivery, family history, neuromuscular disorders, these babies should have USS.

Diagnose from examination with limited abduction, shortened femur and definitively by USS (Xrays not useful as femoral head not ossified).
90% resolve spontaneously. Can fix hip with harness, must adjust for growth and keep on at all times. If after 8months then open reduction and femoral osteotomy.

Talipes
Foot is fixed inverted and supinated.
Multifactorial inheritance, associated with oligohydramnios and hip dysplasia.
Positional – mild deformity which can be corrected by manipulation.
If more severe, treat with strapping, passive manipulation and boots. If not improving then surgery.
Renal anomalies
May present on antenatal scan, or with UTIs, recurrent abdominal pain, a mass, haematuria or failure to thrive.

Kidney anomalies: Renal agenesis – causes Potter sequence, if bilateral always fatal. Duplex kidney – 2 ureters, the upper may be ectopic, draining to urethra or vagina, and the lower often refluxes. Horseshoe kidney. Cystic disease – AR infantile polycystic kidney disease, AD adult polycystic kidney disease, tuberous sclerosis. Ectopic kidney – pain arising from the kidney may be misleading, prone to UTI and reflux.

Obstructive lesions: Pelviureteric obstruction – this gives congenital hydronephrosis, due to a vessel or fibrous band, if almost complete lose renal tissue, mostly boys. Often resolves spontaneously if mild but may need pyeloplasty. Vesicoureteric obstruction – stenosis or uterocoele, gives hydrourereter and hydronephrosis. Posterior urethral valves – pain in pales and block urethra, can cause death in utero or soon after birth if severe, gives bilateral hydronephrosis.

Abdominal wall defects
Gastroschisis – no covering sac, adjacent to umbilicus, usually isolated anomaly.
Exomphalos - covering sac, through abdominal ring.
Patent urachus – tube remains from ant abd wall to bladder, discharges urine.
Vitello-intestinal fistula – tract remains from ant abd wall to gut.
Meckel’s diverticulum - partial remnant of vitello-intestinal duct, 2% of population, 2 inches long, 2 feet proximal to ileocaecal valve. May contain ectopic pancreatic or gastric mucosa. Usually asymptomatic but may cause painless rectal bleeding, peptic ulceration, diverticulitis, apex of volvulus if attached to umbilicus, focus of intussusception.
Umbilical hernia – common. Should resolve by 2. If not needs surgery.

Diaphragmatic hernia
Abdominal contents herniated into chest. Usually diagnoses antenatally, otherwise get respiratory distress and scaphoid abdomen.
Initially resuscitate by intubation (not bag and mask) and NG aspiration, then surgical repair.
High mortality due to pulmonary hypoplasia.

Oesophageal atresia
Get polyhydramnios, dribbling coughing newborn.
Usually has tracheo-oesophageal fistula.
Diagnose by attempting to pass tube before attempting first feed to avoid aspiration.
About half have other malformations e.g. VACTERL.

Imperforate anus
Some have a fistula to vagina or bladder.
High or low depending on relation to levator ani. If high, often fistula to bladder or vagina.
A proportion have other congenital abnormalities.

Ambiguous genitalia
Normal development – Mullerian and Wollfian duct systems. Gonad differentiates depending on chromosomes (SRY region on Y makes a testis). Testes produce Mullerian inhibitory factor to stop female organs developing and testosterone to make male external genitalia. In the absence of these get a female. Testes migrate down to scrotum guided by gubernaculum.

Sex is determined at many levels – chromosomal, gonadal, anatomical, hormonal, psychological.
Most common is CAH causing virilised female.
46XY with testis but incomplete masculinisation – defect in testosterone synthesis, defects in androgen action (5alpha reductase deficiency), androgen resistance (testicular feminisation syndrome).
46XX with ovaries but masculinised (fused labia, clitoromegaly) – CAH, maternal androgens.

**Investigations** - karyotype, USS of pelvic organs and adrenal glands, electrolytes (to exclude salt losing crisis), endocrine investigations (LH and testosterone high in androgen insensitivity, mother for source of androgens).

**Management** – don’t name or register until gender assigned. Normally, virilised females can be surgically feminised. Genetic males may respond to testosterone, otherwise (e.g. if testicular feminisation) may be better to raise as female as external genitalia is right. Remove intra-abdominal gonads due to risk of malignancy. Ensure adequate hormones.

**Associations**
Collection of malformations that occur together more often than expected by chance but in different combinations.
VACTERL – vertebral, anorectal, cardiac, tracheo-oesophageal, renal, limb.
CHARGE – coloboma, heart defect, choanal atresia, retardation of growth, genital abnormalities, ear abnormalities.
Paediatric Dermatology

**Eczema**
Common, affecting up to 20% of children. Usually history of atopy. Dry red itchy rash on extensor surfaces and face in infants then flexures in older children. Diagnose clinically, though investigating allergies may help management. Management – avoid synthetic fabrics, biological detergents or fabric conditioners, furry bets, house dust. Use emollients and stroids.

**Seborrhoeic dermatitis**
Mild, first 2 months of life with non-itchy rash on scalp, face and nappy area. Treat with emollients and steroids and keratolytic for cradle cap.

**Nappy rash**
Common – due to contact dermatitis, candidiasis, seborrhoeic dermatitis. Usually prolonged contact of urine and faeces with skin. Prevent by frequent nappy changes and barrier creams.

**Skin infection**
Impetigo, herpes simplex, varicella, scabies, head lice, molluscum, tinea.

**Birth marks**
*Pigmented naevus* – normal mole.
*Strawberry naevus* – common, raised soft red black which regresses by late childhood. If massive can get thrombocytopenia and cardiac failure.
*Capillary haemangioma* – port wine stain. Usually isolated but can be part of Sturge-Weber syndrome.
Growth and Endocrinology

**Diabetes mellitus**

**Type 1** – usually presents between 7 and 15, increasingly recognised early with polyuria, polydipsia and weight loss. Aetiology – genetic susceptibility (2-5% in first degree relatives, 30% concordance, HLA DR3/4), autoimmune (antibodies, association with other autoimmune diseases), environmental (triggers may include virus infections). Diagnosis – random glucose >11 or fasting >8mmol/l. Insulin replacement – about 1u/kg/day, often increase during puberty. HbA1c reflects last 3m.

**Hypoglycaemia** – usually at below 4mmol/l, children often not aware, tend to get less autonomic symptoms.

**DKA** – dehydration, metabolic acidosis, abdominal pain, hyperventilation.

**Short stature**

Significant – below 0.4 centile, predicted height < mid parental target range, abnormal growth velocity.

If normal growth velocity, investigate but don’t need to treat. If abnormal growth velocity, investigate and treat.

**Thyroid dysfunction**

Thyroid hormone is needed for normal growth and development.

Thyroid dysfunction is more common in Downs.

**Hypothyroidism** – can be congenital or acquired. Leads to disruption of growth.

Congenital – thyroid agenesis, inborn errors of hormone synthesis (usually get goitre), transient (maternal goitrogens), maternal iodide deficiency. Get prolonged jaundice, constipation, coarse facies, hypotonia, goitre. Neonatal screening for all babies, then give oral thyroxine.

**Hyperthyroidism:** Neonatal – transfer of maternal thyroid stimulating Ig if maternal Graves’ disease. Juvenile – usually Graves’ disease, mostly girls, may present with deteriorating school performance or disturbed puberty. Treat with carbimazole, surgery or radioiodine.

**Adrenal disorders**

**CAH** – inherited error of metabolism in pathway synthesising cortisol, usually 21-hydroxylase, get androgen excess (female virilisation), cortisol deficiency and sometimes mineralocorticoid deficiency (rare with 11-hydroxylase deficiency, causes salt losing crisis and volume depletion).

Manage salt losing crisis with volume replacement and systemic steroids. Long term treatment – cortisol replacement with hydrocortisone (also suppresses ACTH and androgen overproduction), mineralocorticoid replacement with fludrocortisone, surgical correction of female genital abnormalities.

**Primary adrenal insufficiency** – Addison’s disease. Due to autoimmune disease, haemorrhage and infarction (Waterhouse-Friderichsen syndrome) and rarely TB. Get postural hypotension, increased pigmentation. Intercurrent illness causes adrenal crisis (vomiting, dehydration, shock).

**Secondary adrenal insufficiency** – mostly due to prolonged glucocorticoid use e.g. in asthma. Reduction of steroid doses should be sow.

**Cushing syndrome** – glucocorticoid excess due to overproduction of cortisol (adrenal tumour, pituitary tumour, ectopic ACTH production) or exogenous cortisol administration. Get short stature, truncal obesity, round face, virilisation, striae, hypertension. Investigation shows high cortisol with loss of diurnal rhythm. Dexamethasone suppression test will suppress cortisol production due to high ACTH but not that from adrenal tumour. CT or MRI shows adrenal or pituitary tumours.
Pituitary disorders
Anterior – usually deficiency due to cranial defects (e.g. agenesis of corpus callosum), tumours (e.g. craniopharyngoma), idiopathic, trauma, surgery or radiation. Get growth retardation with thyroid, adrenal or gonadal dysfunction. GH deficiency is usually idiopathic and isolated.

Posterior: Diabetes insipidus – lack of ADH (sometimes nephrogenic – unresponsive kidneys) leads to polydipsia and polyuria, diagnose with water deprivation test. SIADH – hypoosmolality and hyponatraemia, waterintoxication leads to vomiting and seizures. Common stress response and may be due to meningitis, brain tumours, head injury or pneumonia. Treat cause and fluid restrict.

Calcium metabolism
Hypocalcaemia – neonatally if high phosphate milk, vitamin D deficiency, hypoparathyroidism (usually as part of Di George syndrome), pseudohypoparathyroidism (end organ resistance, also obesity, nodules, LDs), pseudopseudohypoparathyroidism (physical characteristics of pseudohptism but calcium, phosphate and PTH normal).

Hypercalcaemia – vit D overdose, hyperparathyroidism.
Disorders of puberty

Male – testicular growth, then penis enlargement, then growth spurt (on avg 2 yrs later than females).

Female – breast development, then hair, menarche later.
Investigate bone age, blood tests for systemic disease, hormone levels.

Precocious puberty – secondary sexual characteristics before 8yrs in girls and 9 in boys.
Differentiate from premature adrenarche – isolated early appearance of pubic hair, benign self-limiting due to early maturation of adrenal glands often due to obesity, may need to exclude tumour.
Premature thelache is isolated breast development, self-limiting.
Causes – idiopathic, CNS tumours, McCune-Albright syndrome (gonadotrophin independent, fibrous dysplasia of bone), adrenal or gonadal tumours.
Girls – much more common, usually early onset of normal puberty.
Boys – usually pathological.
If very early, stop with GnRH to prevent final height compromise and social difficulties of early menarche.

Delayed puberty – absence of secondary sexual characteristics in girls of 13 or boys of 14. More common in boys, most are normal. Exclude systemic disease (hypothalamic suppression), chromosomal analysis, measure gonadotrophins. Hormone therapy can be used to accelerate growth and induce secondary sexual characteristics in boys.
Causes with low gonadotrophins – constitutional, panhypopituitarism, intracranial tumours (e.g. craniopharyngioma), Kallman’s syndrome (check sense of smell), severe systemic disease e.g. CF, asthma.
Causes with high gonadotrophins – Klinefelter and Turner syndrome.

Obesity
Usually overeating, often overweight parents offer child too much food and overeating becomes a habit, low activity.
Endocrine abnormalities – hypothyroidism, Cushing’s, PCO, Klinefelters.
Hypothalamic lesion.
Prader-Willi.

Complications – social, early menarche, irregular menstruation, long-term medical.
Treat by diet and increasing activity.
Inborn errors of metabolism
Usually autosomal recessive, some X linked.
Clinical effects due to accumulation of precursors, toxic metabolites or energy insufficiency.
Symptoms usually during periods of metabolic stress e.g. neonatal, weaning, infections.
Can cause progressive learning difficulties, seizures, failure to thrive, hepatosplenomegaly.
Generally stop feed and give dextrose to stop metabolic load. May need to correct metabolic disturbances and give renal support.
Prognosis is usually poor, but make diagnosis for future pregnancies.

Ornithine transcarbamylase deficiency – X linked recessive, mutation of urea-cycle enzyme leading to hyperammonaemia. Serious illness when protein containing feeds are given. Affects some female carriers – learning difficulties, illness after high protein meals.

PKU – AR, defect in phenylalanine hydroxylase so phenylalanine builds up and creates toxic biproducts. Screened for with Guthrie test, need infant to have been fed. Infants are clinically normal at birth but develop neurological problems e.g. retardation and seizures, growth retardation and hypopigmentation (no tyrosine so no melanin). Treat by excluding phenylalanine from diet.

Galactosaemia – liver dysfunction, coagulopathy and cataracts. Diagnose by reducing substances in urine (galactose) and red cell enzyme analysis. Causes severe neonatal jaundice. Treat with lactose free diet and no breastfeeding.

Glycogen storage diseases – abnormal accumulation of glycogen in tissues and can’t mobilise glucose from it. Children get growth failure, hypoglycaemia and hepatomegaly. Treat by frequent feeds.

Mucopolysaccharidoses – progressive disorders causing developmental delay and coarse facies. Many different types e.g. Hurler syndrome. Treat with marrow transplant.
**ENT**

**Acute otitis media**
Very common – viral e.g. RSV or bacterial (H inf, pneumococcus).
Present with fever, vomiting and distress so need to examine eardrums.
See red eardrum with no light reflex, may bulge or perforate.
Symptomatic treatment and amoxicillin.

**Glue ears**
Recurrent otitis media. Mild ear fluid persists causing conductive hearing loss and susceptibility to re-infection. May need grommets to drain ear.

**Sleep apnoea**
Can cause failure to thrive, daytime somnolence and poor school performance. Long-term complications of cor pulmonale.
May be due to large tonsils and adenoid or craniofacial abnormalities.
Do adenotonsillectomy and nocturnal CPAP.

**Allergic rhinitis**
Nasal lining has hypersensitivity IgE mediated reaction e.g. to pollen, house dust, animal dander, feathers, eggs, milk.
Seasonal allergic rhinitis is hayfever.
Avoid allergen, drug therapy e.g. steroids, antihistamine, sodium cromoglycate (mast cell stabilisers), desensitising immunotherapy.

**Nose bleeds**
Usually due to nose picking or coryza, rarely trauma, teleangiectasia, bleeding tendency, neoplasm, hypertension.
Get bleeding from Little’s area.
Eyes

**Squints**
When one eye has an abnormal position so both eyes cannot focus on a target at the same time. The vast majority of childhood squints are concomitant.
Input to one eye is reduced, which may lead to ambylopia and binocular vision. Must be treated by patching before 8 years of age, whilst the brain retains elasticity, so vision develops.

**Aetiology** – idiopathic (usually convergent, majority), severe hypermetropia (convergent), severe myopia (divergent), unilateral refractive error, blindness in one eye, brain damage.
Can also get pseudosquint (wide nasal bridge), latent squint (only when tired or unwell).

**Visual impairment**
Familial, congenital infection, prematurity are risk factors.
Ocular malformations e.g. coloboma.
Congenital cataracts (familial, rubella, CMV) or glaucoma (idiopathic, familial, retinoblastoma, neurofibromatosis, Sturge-Weber syndrome).
Choroidoretinitis or ROP.
Pituitary tumour or compression.
Cortical blindness.

**Red eye**
Subconjunctival or retrobulbar haemorrhage, conjunctivitis, anterior uveitis, keratitis, acute glaucoma, episcleritis.


**Congenital abnormalities**
Sclerocornea
Coloboma – iris doesn’t fuse properly.
Cataracts – either part of syndrome, inherited, due to infection e.g. rubella or cryptogenic.
Capillary hemangioma – only a problem if on eyelid as causes stimulus deprivation.
Glaucoma – causes extreme photophobia, eye watering, globe enlargement.
Aniridia – related to Wilm’s tumour.
Optic nerve hypoplasia – not genetic, due to pre-natal insult.
Genetics

Chromosomal disorders
Usually multiple congenital abnormalities and learning difficulties. Most arise de novo.

Do chromosomal analysis (usually T cells stimulated to divide) if – phenotype of known disorder, multiple congenital abnormalities, dysmorphic features, recurrent pregnancy losses.

Single gene defects
AD – several generations with affected individuals, equal male and female, male to male transmission occurs. Often high proportion of new mutations. Can be complicated by variable expression, non-penetrance and sporadic cases. Mostly structural proteins e.g. myotonic dystrophy, Marfan syndrome, neurofibromatosis type 1, tuberous sclerosis, achondroplasia, Ehler Danlos, FH, Noonan’s syndrome, osteogenesis imperfecta.

AR – usually enzymes, risk increased by parental consanguinity. CF, thalassaemia, sickle cell, CAH, inborn errors of metabolism.

X-linked recessive – only affects males, females may have slight effects depending on lyonisation pattern, no male to male transmission. E.g. haemophilia A and B, Duchenne muscular dystrophy, fragile X syndrome, G6PD deficiency, colour blindness.

X-linked dominant – very rare e.g. familial rickets.


Multifactorial disorders
Combination of polygenic genetic susceptibility and the environment. Recurrence risk increased with severely affected proband, multiple affected family members, if proband is of sex less often affected. E.g. NTDs, facial clefts, pyloric stenosis, talipes, asthma, diabetes, epilepsy.
<table>
<thead>
<tr>
<th>Genetic basis</th>
<th>Clinical features</th>
<th>Prognosis</th>
<th>Medical associations</th>
</tr>
</thead>
<tbody>
<tr>
<td><strong>Down’s</strong></td>
<td>Trisomy 21, mostly nondysjunction (translocation and mosaicism).</td>
<td>Characteristic appearance – oblique palpebral fissures, single transverse palmar crease, hypotonia.</td>
<td>IQ 20-55. Gap to normal widens through childhood. Most survive to middle age then decline due to Alzheimer’s.</td>
</tr>
<tr>
<td><strong>Patau’s</strong></td>
<td>Trisomy 13.</td>
<td>Cleft palate, polydactyly, cardiac and eye defects.</td>
<td>Almost all die by age 3.</td>
</tr>
<tr>
<td><strong>Prader-Willi</strong></td>
<td>Lack of paternal 15q.</td>
<td>Obesity, hypogonadism, hypotonia, violent temper sleep apnoea</td>
<td>Mild LD. Expressive language disorder.</td>
</tr>
<tr>
<td><strong>Angelman’s</strong></td>
<td>Lack of maternal 15q.</td>
<td>Microcephaly, ataxic jerky movements.</td>
<td>Severe LD.</td>
</tr>
<tr>
<td><strong>Cri du Chat</strong></td>
<td>5p.</td>
<td>Round flat face, hypertelorism, micrognathia, characteristic cry.</td>
<td>Severe LD.</td>
</tr>
<tr>
<td><strong>XY Y</strong></td>
<td>XXY</td>
<td>Assertive, impulsive.</td>
<td>May have LD.</td>
</tr>
<tr>
<td><strong>Neurofibromatosis</strong></td>
<td>AD</td>
<td>Neurofibromata.</td>
<td>10% severe LD.</td>
</tr>
<tr>
<td><strong>Apert</strong></td>
<td>AD, mainly sporadic.</td>
<td></td>
<td>High mortality in 1st year. Learning disabilities.</td>
</tr>
<tr>
<td><strong>PKU</strong></td>
<td>AR Phenylalanine hydroxylase def.</td>
<td></td>
<td>Microcephalic mental impairment. Autism. Epilepsy.</td>
</tr>
<tr>
<td><strong>Rett</strong></td>
<td>X linked dominant.</td>
<td>Fits, agitation, truncal ataxia, depression.</td>
<td>Normal dev for 1 year then lose speech mobility etc.</td>
</tr>
</tbody>
</table>
GI and Liver

**Gastroenteritis**
Present as diarrhoea and vomiting, sometimes prodromal illness, abdominal pain and blood or mucus in stool suggests invasive bacterial pathogen.
Usually mild and self-limiting.
Cause - usually viral (esp rotavirus), but also bacteria (shigella, salmonellae, camplybacter, E coli) and protozoa (entamoeba, giardia, cryptosporidia).
Most are faeco-oral transmission.
Most important thing to assess is dehydration, must exclude pyloric stenosis and intussusception.

Management – rehydration and correction of any electrolyte imbalance.

**Infantile colic**
Recurrent inconsolable crying accompanied by drawing up legs. Especially in evenings.
Common and benign, but need to include serious conditions.
Usually resolves by 4 months.

**Lactose intolerance**
Due to intestinal disaccharidase deficiency. Most commonly transient phenomenon post gastroenteritis but can by congenital.
Accumulation of sugar causes watery diarrhoea and bacterial production of organic acid, which excoriates perianal region.
Diagnosis - positive test for reducing substance (lactose is a reducing sugar).
Treat by a lactose free diet.

**Food intolerances**
Mostly immune mediated reactions, usually to proteins. Common foods are cow's milk, soya, wheat, fish, egg, nuts. Diagnose when symptoms are relieved by removal of a food and recur on reintroduction.
More common in infants with atopy or IgA deficiency.
Usually present with protracted diarrhoea and failure to thrive.

Avoid food until 2 yrs. Most patients grow out of intolerance by then so conduct dietary challenge.

Cow’s milk protein intolerance can lead to protein-losing enteropathy and blood loss. Manage with a casein-hydrolysate based formula as infants likely to become intolerant to soya.

**Coeliac disease**
Malabsorption due to gluten-mediated damage to the mucosa of proximal intestine causing atrophy of villi and loss of absorptive surface.
Familial predisposition and association with HLA DQ2.

Clinical features – failure to thrive when gluten is introduced, with abdominal distension and buttock wasting. Most present more subtly with diarrhoea, irritability, anaemia due to iron or folate deficiency.

Diagnosis – specific IgA antigliadin or antiendomysial antibodies for screening. Definitively by flat mucosa on jejunal biopsy which improves on gluten free diet and recurs on gluten challenge.

Management – diet free of gluten for life. Associated with other autoimmune disorders and increased risk of small bowel lymphoma (probably reduced by gluten free diet).
IBD
One quarter of cases have onset in childhood.
Present with lower abdominal pain, bloody diarrhoea and failure to thrive or weight loss.
Crohns – steroids, elemental diet, immunosuppressives, surgery.
UC – aminosalicylates, steroids, colectomy.

GO reflux
Common in infants due to functional immaturity of lower oesophageal sphincter.
In most is mild e.g. regurgitation, no treatment is required and resolves spontaneously. Can help by
thickening feeds, early weaning, alginate and antacid medication and positioning head up. In a
minority (esp CP, preterm, congenital oesophageal abnormalities and chronic lung disease), may be
severe leading to failure to thrive, oesophagitis (irritability, pain after feeding, blood in vomit),
aspiration pneumonia. Treat with prokinetic drugs (e.g. domperidone) and drugs to reduce gastric
acid secretion and surgery if very severe (Nissen fundoplication).
Usually diagnose clinically, but can confirm and assess severity by 24hr ambulatory oesophageal
pH monitoring, barium studies (for anatomical abnormality), endoscopy (if oesophagitis), nuclear
milk scan.

Worm infection
Threadworms are very common.
Young children may also get dog or cat worms.

Mesenteric adenitis
Non-specific inflammation of lymph nodes provokes a peritoneal reaction mimicking appendicitis.
Common, often associated with fever, headache, pharyngitis and cervical lymphadenopathy.
May need observation in hospital due to difficult diagnosis, self-limiting so manage conservatively. If
persists, may need surgical exploration.

Toddler’s diarrhoea
Well thriving child with persistent loose stools. Due to maturational delay in intestinal mobility
causing reduced transit time and undigested vegetables.

Pyloric stenosis
Persistent projectile vomiting in a hungry baby, with reduced stool output, visible peristalsis and
palpable pyloric mass. Typically boys between a few weeks and 3 months, often family history.
Develop weight loss, constipation, mild jaundice and dehydration, risk of hypochloraemic metabolic
alkalosis.
Due to hypertrophy of smooth muscle of pylorus.
Diagnose clinically by feeling hypertrophied pylorus during test feed (olive, RUQ), USS can confirm
Correct dehydration and electrolyte imbalance first then manage surgically by Ramstedt’s
procedure – divide musculature.

Constipation
Simple – febrile illness or dehydration cause hard stool. This causes discomfort or pain on
defaecation and maybe an anal fissure leading to retention of faeces. Rectal capacity increases
leading to further retention and increased discomfort in a vicious cycle. May get overflow
incontinence as child gets used to full rectum.
Need to empty rectum first. Treat by improving diet, encouraging regular toileting and giving stool
softeners (e.g. lactulose) or if severe drugs to increase intestinal mobility (e.g. senna, picosulphate).
Organic causes – Hirschprungs, hypercalcaemia, hypothyroidism.
**Hirschprung’s**
Congenital aganglionic megacolon. Aganglionic segment is narrow and contracted. More common in boys, often family history.
Usually presents neonatally with failure to pass meconium followed by abdominal distension and bile-stained vomiting. Older children may present with chronic severe constipation from birth, abdominal distension, narrow rectum.
Show with unprepped barium enema. Definitive diagnosis is by lack of ganglion cells on suction rectal biopsy.
Differentiate from simple constipation as presents earlier (first month of life), delayed passage of meconium, soiling is rare, distended abdomen, narrow empty rectum rather than loaded.

**Management** – usually surgical. Temporary colostomy, followed by anastamosis of normal bowel to anus (pull-through procedure).

**Biliary atresia**
Rare cause of persistent conjugated neonatal jaundice due to destruction of absence of extra or intra hepatic biliary tree.
Diagnose by abdo USS, radionucleide scan (uptake by liver but no excretion to bowel), liver biopsy (fibrosis) and intraoperative cholangiography.
Treat surgically by hepatoportoenterostomy (Kasai procedure), ideally before 6wks to prevent liver disease. The earlier the treatment the better the prognosis.

The biliary system can also be obstructed by choledochal cysts. Diagnose by USS and treat by surgical excision. Risk of malignancy.

**Hepatitis**
Neonatal – congenital infection (TORCH), drugs, A1AT deficiency, galactosaemia, TPN cholestasis, obstructive jaundice. May have IUGR.
In childhood – hepatitis viruses, EBV. May not develop jaundice.
Haematology and Oncology

Anaemia
Symptoms rare above 7-8g/dl but causes anorexia and impaired development and growth so treat. Severe anaemia can cause congestive cardiac failure.
Infants are born with about 17g/dl (depending on cord clamping and positioning) and physiologically falls as low as 9.5g/dl around 2 months due to erythroid hypoplasia and switch to adult Hb (should have about 5% HbF at 3 months, none by 1yr). Lower limit of normal from 1yr until puberty is 11g/dl. Nutritional iron deficiency is the most common cause.

Causes:
Decreased red cell production – iron deficiency (nutritional, occult blood loss, malabsorption), haemoglobinopathy (thalassaemia), marrow failure (malignant disease, aplasia), chronic disease (renal failure, inflammation).
Reduced red cell life span (haemolytic, normal is 120 days) – intrinsic red cell defects (spherocytosis, sickle cell, thalassaemia, G6PD), immune (e.g. incompatibility, autoimmune), bacterial infections, malaria, hypersplenism, HUS. Haemolysis either occurs in reticulo-endothelial system or intravascularly (get haemosiderin or haemoglobin in urine).
Excessive blood loss – GI (hookworm infestation, Meckel’s diverticulum), iatrogenic, epistaxis, menstruation.

Investigations:
Peripheral blood film – FBC, sickle cells, spherocytes, reticulocytes (increased – haemolysis, haemorrhage, decrease – marrow aplasia).
Red cell indices – MCV: microcytic in iron deficiency, thalassaemia; macrocytic in normal neonates, folate or B12 deficiency. MCHC: hypochromic in iron deficiency and thalassaemia. Pancytopenia – marrow failure or hypersplenism.
Serum iron, ferritin and TIBC to investigate iron deficiency.
Coomb’s test (immune reactions).
Haemoglobin electrophoresis.
Red cell enzyme estimation (G6PD, pyruvate kinase).
Bone marrow aspiration.

Nutritional iron deficiency anaemia – term infants have reserves for 4months, preterm for less (less stores and faster growth). Need about 1mg/kg/day. Breast and cow’s milk are low in iron, though more is absorbed from breast milk. Most common problem is too much cows milk so not enough solid food. If diet seems adequate or fail to respond to therapy, consider absorption problems esp Crohns and coeliac disease. May lead to inappropriate eating of non-food materials (pica). Treat with oral iron – usually sodium iron edentate as more palatable than ferrous sulphate. Continue for at least 3months to correct anaemia then build up iron stores.

Hereditary spherocytosis – AD due to abnormalities in spectrin. May be a sporadic mutation. Severity is variable, usually mild anaemia, jaundice and splenomegaly. Complications are aplastic crisis if parvovirus infection and gallstones. See spherocytes on blood film and confirm by osmotic fragility test (higher SA so rupture more easily in hypotonic solutions). Most cases need only folic acid supplements, if severe can do splenectomy.

Aplastic anaemia – usually idiopathic or inherited e.g. Fanconi. Need bone marrow transplant or sometimes immunosupression if autoimmune. If no donor, then immunosupression or support with blood product transfusions.

G6PD deficiency – X linked recessive disorder. Don’t generate sufficient glutathione so at risk from oxidant agents. Causes neonatal jaundice and intravascular haemolysis and haemoglobinuria induced by infection or oxidant compounds (e.g. sulphonamides, fava beans).
Pyruvate kinase deficiency – AR. Get infection induced haemolysis and low Hb levels. Can manage by splenectomy.

Haemoglobinopathies
Fetal haemoglobin HbF – alpha2 gamma2 (higher oxygen affinity)
Adult haemoglobin HbA – alpha2 beta2, HbA2 – alpha2 delta2
Sickle haemoglobin HbS – alpha2 betaS2
3 alpha genes deleted HbH – 4beta alpha

Generally present around 6 months due to switch from HbF. Diagnose by Hb electrophoresis.

Sickle cell disease – AR mutation in B globin gene. HbS is insoluble in deoxygenated state so aggregates and distorts RBCs. Cells have a reduced life span and get trapped in microcirculation causing ischaemia. Get progressive anaemia with jaundice. Initially, have splenomegaly, there is then autosplenectomy due to repeated infarction and fibrosis. Hyposplenism means patients are at risk of infection, esp by H inf and pneumococcus, so vaccinate and give penicillin. Give folate supplements to support erythropoiesis.

Vaso-occlusive crises – precipitated by infection, dehydration or cold. Mostly painful crises in long bones also dactylitis and priapism (painful prolonged erection). Treat with analgesia, oxygenation and hydration. Hydroxyurea may reduce crises by increasing HbF. Bone pain may also be due to osteomyelitis, esp due to salmonella. Can also get cerebral or pulmonary infarction, treat with exchange transfusion.

Aplastic crises – may need transfusion.
Sickle cell trait is only a problem if hypoxic stress e.g. during GA.

Thalassaemia – defects in globin chain synthesis. Excess unpaired chains produce insoluble tetramers that cause cell death in bone marrow (ineffective erythropoiesis) or premature removal of red cell (haemolytic anaemia).

B-thalassaemia – either homozygous or heterozygous (trait). If homozygous, complete lack of B chain synthesis (some mutations allow partial synthesis), so can’t make HbA. Infants present around 6m with severe anaemia, jaundice and hepatosplenomegaly. Get bone marrow hyperplasia leading to maxillary hypertrophy and skull bossing. Severity depends on amount of HbF. Treat by regular blood transfusion, though problems with iron overload which accumulates in heart, liver, pancreas and skin. Give chelation therapy, but ultimately patients get cardiomyopathy and congestive heart failure around 30. Splenectomy and bone marrow transplantation may help. In b-thalassaemia trait, get mild hypochromic microcytic anaemia, which is mostly asymptomatic, have raised HbA2 and HbF.

A-thalassamia – usually due to gene deletion. A-thalassaemia major is when all 4 genes deletes so can’t make HbF and get death in utero (Bart’s haemoglobin, 4 gamma). If 3 deleted, get severe anaemia and HbH. A-thalassaemia trait if 2 deleted – get normal Hb and mild anaemia. Silent carrier if one deleted.

Bleeding disorders
Coagulation cascade - injury to endothelium, tissue factor activation, factors VII, X, IX (PT, PTT) to factor Xa, activates thrombin, fibrinogen to fibrin (TT, fibrinogen), platelet fibrin plug.
Also need platelet activation and vasoconstriction by vessels.
PT – prolonged in vit K deficiency.
PTT – prolonged in haemophilia.
TT – prolonged in DIC.

Bleeding into skin is usually a platelet or vascular disorder. Bleeding into muscles or joints is usually a coagulation disorder.

Blood vessel disorders – Ehler-Danlos syndrome (excessive capillary fragility), hereditary haemorrhage telangiectasia, scurvy, HSP.
Henoch Schonlein purpura – multisystem vasculitis of small blood vessels. Usually follows URTI, assumed to be mediated by IgA. Get purpuric rash, colicky abdo pain and bleeding, pain in large joints, glomerulonephritis (rarely severe). Diagnose clinical and treat symptomatically, very good prognosis.

Haemophilia – X-linked recessive, 1/3 spontaneous mutations. Prolonged APTT time.
Haemophilia A – reduced or absent factor VIII (has 2 components, antihaemophilic factor and von Willendbrand factor). Clinical features vary greatly depending on level of factor (>5% is mild, severe is none) but get spontaneous or traumatic bleeding into skin, muscles and joints. Long-term, main problems are joint disease (haemarthroses) or life-threatening internal or intracranial haemorrhage. Treat with IV infusion of factor VIII concentrate (produced by recombinant technology so no risk of infection) or desmopressin (releases factor from tissue stores) for mild disease.
Haemophilia B - factor IX. Clinically similar to haemophilia A but much rarer. Treat with prothrombin complex concentrate.

Thrombocytopenia – purpura usually occur when count falls to 20x10^9/l. Decreased production due to bone marrow failure or reduced survival due to ITP, hypersplenism, giant haemangioma or DIC.

ITP - immune-mediated, platelets mostly destroyed in spleen. Presents with purpura and superficial bleeding, usually after a viral infection. On FBC, have low platelets, megakaryocytes (platelet precursors) but other blood cells are normal. Need bone marrow aspiration to exclude infiltration or aplasia if uncertainty about diagnosis or considering steroids. In most children, is benign and self-limiting, serious bleeding rare as platelets function more effectively. Platelet infusions are rapidly destroyed so only useful in life-threatening emergency. IVGG increases platelet counts in severe disease and a short course of oral steroids inhibits platelet destruction and reduce capillary fragility. Splenectomy if this fails, most common in teenage girls.

Von Willenbrand disease – deficiency of von Willenbrand factor (carrier for factor VIII and facilitate platelet adhesion). Usually AD with variable penetrance, affects about 1%. Bleeding into skin and mucous membranes. Treat with desmopressin for mild disease, von W factor concentrate for severe disease.

Thrombotic disorders – rare in children as thrombin is more inhibited and generated less readily. Factor V Leiden (abnormal factor V), protein C or S deficiency, antithrombin III deficiency.
Childhood Cancer

Mostly leukaemia, CNS tumours and lymphomas. Affects 1 in 600 by age 16, 70% are curable.

Aetiology – mostly sporadic though there may be a genetic predisposition. Some genetic syndrome: retinoblastoma – deletion of tumour suppressor gene (C13), Li Fraumeni – p53 mutation, ataxia-teleangiectasia – DNA repair defect, Down syndrome. Some infections: EBV – Burkitt’s lymphoma due to c-myc oncogene, HIV. Environmental and toxin factors are rare in childhood cancers, though previous treatment of malignancy is an important risk factor.


Investigations – biopsy or bone marrow aspiration, imaging, tumour markers (AFP for liver, urinary catecholamines for neuroblastoma).

Management:
Surgery - for biopsy, removal of solid tumours, debulking, palliative.
Radiotherapy - esp for CNS tumours, has more effect on growth and normal tissue in children.
Chemotherapy – primary therapy for leukaemias, to shrink bulky disease before local treatment, to treat micrometastases. Mainly limited by bone marrow toxicity, patients may need bone marrow transplantation after (bone marrow rescue). Rapid lysis of tumour may cause metabolic disturbance. Typical stages – preparation (treat infection, blood transfusion, allopurinol to protect kidneys), induction (intensive to reduce tumour load), early CNS directed therapy, consolidation (therapy after remission), intensification if risk of relapse, maintenance (up to 2 years, 3 in boys). Long term consequences are decreased growth, second primary malignancies (about 5%), infertility, missed schooling and psychological.

Leukaemia
Proliferation of immature WBCs (blasts). Vast majority are acute, chronic myeloid leukaemia is very rare. Classified by cell as lymphoctic (lymphoid cells) or myeloid (granulocytic or monocytic)

Clinical features - present due to bone marrow failure with anaemia or bruising, hepatosplenomegaly, lymphadenopathy, infection due to neutropenia or bone pain. Bone marrow aspiration shows replacement of normal elements by blast cells.

Acute lymphocytic leukaemia – most common form. Lymphoblasts remain frozen at early stage of development – most is common ALL (before differentiation to T or B cell), also get T cell (older children) or B cell. More than 65% now cured, best prognosis if common subtype, low WBC count, >1yr. Chemotherapy and CNS directed radiotherapy, bone marrow transplantation for high risk groups only.


Lymphoma
May present with systemic symptoms e.g. fever, weight loss or lymph node enlargement.

Non-Hodgkin’s lymphoma – younger children. Heterogenous group with different cells of origin. Can develop in immunocompromised children. Tend to be aggressive and rapidly growing – most present with mass, intrathoracic usually T cell, peripheral or abdominal usually B cell. Treat mainly with chemotherapy but may need surgical debulking for abdominal tumours.
Hodgkin’s disease – Reed Sternberg cell, mainly adolescents. Usually painful cervical or supraclavicular lymphadenopathy. Metastasis to lungs, liver and bone marrow. Diagnose and classify by lymph node biopsy – lymphocyte predominant (best prognosis), nodular sclerosing (most common), lymphocyte depleted (rare, worst prognosis). Stage by imaging and treat with chemotherapy or radiotherapy if localised. About 80% are cured.

Neuroblastoma
Malignancy of neural crest cells (normally from sympathetic ganglia and adrenals).
Unusual as can regress spontaneously in very young children (stage IVs).
Present with a mass in the abdomen, usually around 2 yrs. May also have systemic signs, hepatomegaly, unilateral proptosis (metastases to eye), opsoclonus-myoclonus (dancing eye, immune response), watery diarrhoea (VIP secretion). Diagnose and monitor by raised urinary catecholamines (VMA and HVA), can also do biopsy or tumour specific scanning.
Treat by surgical resection, chemotherapy and radiation. Worse prognosis if older, metastatic or high N-myc expression.

Wilms tumour
Arises from embryonal renal cells, usually in toddlers.
Familial forms exist – associated with aniridia and C11 deletion.
Present with asymptomatic mass in the abdomen that does not cross midline. May get abdo pain, haematuria or hypertension. 5% are bilateral.
Diagnose from CT (intrinsic renal mass with solid and cystic areas) and biopsy. Look for metastases, mostly lung and liver.
Treatment – surgical resection, chemotherapy depending on stage and grade and radiotherapy if advanced. Good prognosis if no metastases with 80% cure.

Brain tumours
Most common solid tumour in childhood. Most are infratentorial. Metastasis is rare.
Surgery is usually first treatment if possible.

Astrocytomas – cerebellar astrocytomas are usually low-grade slow-growing and present with raised ICP (headache, vomiting), cerebellar dysfunction (ataxia, nystagmus, uncoordination) or 6th nerve palsy. Supratentorial astrocytomas are usually high-grade and present with focal neurological signs and seizures. Usually diagnose with MRI, prognosis is poor despite radiotherapy.

Primitive neuroectodermal tumours – usually arise in midline and invade 4th ventricle and cerebellum. Unusual as seed through CNS and have spinal metastases. Present with headache, vomiting and ataxia. Treat by surgical removal and CNS irradiation. 50% cure.

Craniopharyngoma – from remnant of Rathke’s pouch, locally invasive causing visual field loss and pituitary dysfunction. Most are calcified so see on radiograph. Surgical excision and radiotherapy. Good prognosis but sequelae of visual disturbance and pituitary insufficiency.

Bone tumours
Common in adolescents, esp boys.
Present with local pain and swelling.
Osteogenic sarcoma (most around knee joint) and Ewing’s sarcoma.
Need surgery and aggressive chemotherapy for micrometastatic disease.
Immunology and infectious diseases

**Allergy and anaphylactic shock**

**Allergy** – hypersensitivity due to immune mechanisms (mainly IgE, in biphasic response – 20mins then 3-6 hrs). In early childhood mainly food allergies, then inhaled allergens. Tend to improve as child grows older. Cross reactions if share epitope e.g. grass and peanut, latex and banana.

**History** – age of onset, day and seasonal variation, family history, dietary history, time between exposure and reaction, reproducibility, house and school conditions.

**Examination** – respiratory (wheeze, angio-oedema, rhinoconjunctivitis), GI (nausea, vomiting, diarrhoea, abdo distension, failure to thrive), CV (hypotension, shock, dizziness), skin (pruritis, urticaria, atopy).

**Allergy testing** – severe, persistent or recurrent symptoms. Skin prick testing – cheap, quick and safe but high false +. Specific serum IgE levels – RAST assay, high false +. Food challenge – gold standard, risk of severe reaction.

**Anaphylaxis** – must be sensitised first. Typically food, insect venom, latex, drugs. Treat by IM adrenaline (ionotropic, peripheral vasoconstriction and bronchodilation), IV hydrocortisone and antihistamines. Allergen avoidance is only preventative measure.

**Immunodeficiency**

Suspect if recurrent, severe, atypical infections or failure to thrive. Primary (inherited intrinsic defect) are rare, secondary more common.

**Primary:**

- **Common variable antibody deficiency** – heterogenous, low levels of IgG and IgA, late childhood with recurrent bacterial lung infection.
- **X-linked agammaglobulinaemia** – failure of B cell development, early bacterial infections.
- **IgG subclass** - susceptible to encapsulated organisms e.g. pneumococcus and H inf.
- **Selective IgA** - common, often asymptomatic, associated with autoimmune diseases and IgG deficiency.
- **Severe combined immunodeficiency** - presents early, cellular and humoral immunity affected. Treat with bone marrow transplantation.
- **Chronic granulomatous disease** – usually Xlinked, phagocytic cells fail to produce superoxide anion. Repeated baceria and fungal infections which form granulomas and abscesses. Confirm with NBT test (failure to reduce).

**Secondary:**

- **Malnutrition**
- **Infections** – HIV (test children by viral culture or detection of viral genome, not antibody as may have mother’s, shorter incubation period than adults), measles.
- **Immunosupression** – steroids (esp chickenpox), cytotoxic drugs (marrow suppression, neutrophil counts fall), immunosuppressive drugs for organ transplant (CMV).
- **Hyposplenism** - sickle cell, splenectomy.

**Antibody defects** – pneumococcus, staphylococcus, streptococcus, h inf, enteroviruses.
**Cell mediated** – herpes, measles, candida, pneumocystis carinii, mycobacteria, listeria.
**Neutrophils** – bacteria, candida.

**Basic screening** – immunoglobulins (trough between 3 and 6 months as no more maternally transferred and not yet fully synthesised), WCC and differential.

**Management options** – antibiotic prophylaxis (e.g. co-trimoxazole for PCP), aggressive antibiotic therapy for infections, Ig replacement therapy, bone marrow transplantation, gene therapy.
Septicaemia
Serious suggested by - <3 months, bulging fontanelle, high or low WCC, shock, decreased consciousness, tachcardia, non-blanching rash.
Strep pneumoniae, meningococcus, staph aureus, salmonella, Hib.

Pyrexia of unknown origin
Protacted fever (>7 days) with no diagnosis. Mostly infectious. 40-60% resolve spontaneously.
Supportive management - treat temperature symptomatically, give fluids.

Kawasaki disease
Rare systemic vasculitis. Problem as causes acquired heart disease. Much more common in Japanese.
Presentation – fever often unresponsive to antipyretics, miserable child, conjunctivitis, rash, cracked lips, reddening of palms and soles with oedema and peeling of skin. Get vasculitis affection coronary arteries leading to aneurysm formation in 30% cases. Scars then cause vessel narrowing and myocardial ischaemia.
Treat with single dose of IVIG to reduce risks of cardiac complications and aspirin.

Specific infections
Measles - morbillivirus infection.
Rates reduced by vaccine.
Get fever, cough, coryza, conjunctivitis, followed by Koplik’s spots on buccal mucosa, then maculopapular rash spreading downwards.
Very dangerous in immunocompromised children, they get giant cell pneumonia and encephalitis.
Very rare long term complication is subacute sclerosing panencephalitis (immune mediated neurodegenerative disease) many years later.

Rubella – low grade fever, with pink maculopapular rash. Main problem is effect on fetus and rare complications e.g. arthritis and encephalitis. Rare due to vaccine.

Parvovirus – often asymptomatic. Rash on face, extremities and trunks. Suppressed erythropoeisis so may cause aplastic crisis in children with haemolytic anaemia.


Chicken pox – varicella zoster infection. Common and highly infectious by droplet infection. Brief coryza followed by itchy vesicular rash. Complications rare unless immunocompromised, include secondary bacterial infection, encephalitis. Serious illness if immunosupressed. Treat symptomatically unless immunosupressed then give VZIG and acyclovir.

Ebstein Barr virus – infectious mononucleosis. Transmission by droplets or saliva. Generally asymptomatic but may get glandular fever – fever, malaise, pharygitis, cervical lymphadenopathy, splenomegalgy. Monospot and Paul-Bunnell tests or EBV serology.

Diphtheria – rare. Exotoxin causes local necrosis and adherent pseudomembrane in nose, tonsils and/or pharynx. If tonsils and pharynx, may get absorption of toxins leading to hypotension, paralysis and myocarditis. Treat with penicillin and antitoxin.

Mumps – paramyxovirus, routine vaccination has reduced incidence. Fever, malaise and parotitis. 10% have signs of meningoencephalitis. Complications are pancreatitis and epididymo-orchitis.

Staphylococcal – mainly staph aureus but epidermidis in neonates and ITU. Staph aureus – impetigo, folliculitis, wound infections, pneumonia, osteomyelitis, septic arthritis. Toxins cause toxic shock syndrome and scalded skin syndrome.
**Streptococcal infection** - strep pyogenes, group B strep and pneumococcus. Strep pyogenes causes pharyngitis, osteomyelitis, septicaemia and toxin mediated conditions (scarlet fever, erysipelas, toxic shock like syndrome). Post-infectious immune-mediated problems of group A strep include acute glomerular nephritis, rheumatic fever, Sydenham’s chorea (emotional lability and involuntary jerky movements).

Rheumatic fever involves polyarthritis, fever, malaise and pancarditis – pericarditis leads to effusion, myocarditis leads to heart failure, endocarditis affects valves causing murmurs and risk of long term valvular damage. Treat by aspirin, steroids and prophylactic penicillin for life. Antistreptolysin O titre – check for previous group A strep infection, useful for acute nephritis, rheumatic or scarlet fever. Strep pneumoniae causes otitis media, pneumonia, meningitis and septicaemia.

GBS is carried by 25% of women in genital tract and affects neonates causing meningitis and septicaemia.

**Worms** – enterobius vermicularis (threadworm) causes perianal pruritis and vulvovaginitis.

**TB** – most are asymptomatic. Culturing TB is difficult. Treat with triple therapy.

**Lyme disease** – tick transmission. Get skin expanding skin lesion with malaise, fever, arthralgia and lymphadenopathy. Late disease causes meningitis, neuropathies, arthritis, carditis. Treat with antibiotics.
Locomotor

**Reactive arthritis**
Usually following enteritis, sometimes chlamydia or rheumatic fever.
In Reiter’s syndrome, there is also urethritis, conjunctivitis or oral ulceration.
Treat with NSAIDS and steroid injections to refractory joints. Usually settles within a year.

**Juvenile idiopathic arthritis**
Various different types – diagnosed if arthritis for >6wks excluding other causes.
More common in girls.
Manage with physiotherapy and medication (NSAIDs, systemic steroids if severe, methotrexate).

**Systemic (Still's disease)** – mainly young children, primarily affecting knees, wrist and ankles.
Also systemically unwell with fever, rash, hepatosplenomegaly, pericarditis.

**Polyarticular**: Rheumatoid factor negative - all ages and all joints except MCPs, good prognosis.
Positive - mainly older females, small joints of hand, hip and knee. May get systemic vasculitis, poor functional prognosis.

**Oligoarticular** – most common, usually younger girls. Assymetrical. Get antinuclear antibodies and in some chronic anterior uveitis. Good prognosis.

**Limps**
Must not miss septic arthritis or osteomyelitis.

**Perthe’s disease** – ischaemia of femoral head prior to skeletal maturity causes growth disturbance then cycle of osteonecrosis with flattening and fragmentation of femoral head. Resumption of growth (may be abnormal) leads to revascularisation and reossification. Cycle takes 3yrs. More common in boys, often family history. Get insidious onset of limp, usually around 6yrs, with intermittent pain and limited movement. Diagnose with hip Xray. Good prognosis if young and only part of femoral head involved - bracing and mild activity restriction. Older children or most of femoral head involved - about half get permanent deformity of femoral head and degenerative arthritis. Need to fix hip in abduction (plaster, osteotomy) so acetabulum shapes femoral head.

**Transient synovitis (irritable hip)** – common self-limiting condition. Get sudden onset hip pain, refusal to bear weight and limp. Diagnose of exclusion, must rule out septic arthritis (child otherwise well and afebrile and not in pain at rest). If in doubt, start antibiotics and aspirate joint. Treat supportively with analgesia and rest, resolves in 2wks.


**Osgood-Schlatter disease** – common, benign self-limiting knee pain due to periostitis of proximal tibial tuberosity, with tender swelling over insertion of patella tendon. Usually traumatic origin in active teenagers.

**Back pain**
Rare before adolescence, usually serious pathology in children.
In adolescence – muscle or soft tissue (usually sport related), Schuermann disease (osteochondritis of lower thoracic vertebrae), spondylosis (anterior shift of vertebral body, usually L4, pain worse on bending back), vertebral osteomyelitis, tumours.
Scoliosis
Lateral curvature of spine with rotational deformity (get rib hump when bend forward).
Vertebral causes – hemivertebra, osteogenesis imperfecta.
Neuromuscular – polio, CP.
Idiopathic (occur in adolescence) or dysmorphic syndromes.
Generally not painful.
Leave mild curves, brace moderate curves, surgery for severe curves (fuse spine so stop growth).

Growing pains
Generally 7-11 year olds. Occurs in evening, usually lower limbs, normal examination. Never functional disability, limp or morning symptoms.

Osteogenesis imperfecta
Due to mutations in collagen gene. Leads to brittle bones and frequent fractures.
Various different types – 1 is common with blue sclera and conductive hearing loss, 2 is lethal.

Osteomyelitis
Infection of bone, usually haematogenous.
Most common in neonates (S aureus, GBS, E coli) and late childhood (S aureus).
Present with fever and refusal to move limb. Blood cultures not always positive so may need aspiration of bone. Bone scans in early phase, Xrays may be normal.
Treat with IV antibiotics until clinical improvement, then oral antibiotics. May need surgical drainage. Can lead to growth disturbance or deformity (if affects epiphyseal plate) or septic arthritis.

Septic arthritis
Medical emergency – joint destruction within 24hrs if untreated. Xrays don’t help early diagnosis, so need aspiration of joint.
Staph aureus is most common pathogen.
Most common in young children. Get painful joint with fever and irritability, pseudoparalysis in infants.
Need early and prolonged IV antibiotics, with surgical drainage if recurrent or affecting hip.

Normal postural variants
Bow legs (genu varum) – common in infants and toddlers.
Knock knees (genu valgum) – third and fourth years.
Flat feet (pes planus) – toddlers.
Intoeing – due to metatarsus varus in infants, medial tibial torsion in toddlers and femoral anteversion in children.
Pathology likely if severe progression, family history, asymmetry.
Neonatal Medicine

**Care of the newborn**

**Skin** – high surface area and immature skin so at risk of heat and water loss.

**Respiratory system** – lung fluid removed by squeezing during delivery and increased absorption due to catecholamines. Surfactant reduces surface tension. Avg time to breath is about 30s. Lung expansion causes pulmonary vascular resistance to fall. Mainly breathe with diaphragm, obligate nose breathers. Self-limiting apnoea spells.

**Cardiovascular system** – foetal circulation changes to adult circulation.

**GI system** – at 35wks can latch on, suckle and swallow. Meconium is normally passed in 1st 6 hours, longer than 24hrs is abnormal. Changing stools replace meconium on day 3, then yellow stool of milk fed baby.

**GU system** – should void within 24hrs.

**Haematological and immune system** – HbF. Hb concentration of about 17g/dl at birth. Immune system is incomplete due to impaired neutrophil reserves, diminished phagocytosis, decreased complement and low immunoglobulins.

**CNS** – myelination continues in 1st 2 yrs of life.

**Neonatal resuscitation**

Different to adults as fluid filled lungs, brain better at withstanding hypoxia, chest compressions more effective.

Neonates can autoresucitate – in response to asphyxia have gasps, primary apnoea and bradycardia, then gasp again, then secondary apnoea (increasing brain damage and death). Heart can continue for up to 20mins.

Important to dry and warm. Clear mouth and nose.

Bag and mask if heart rate falling and does not start to breath.

Give 5 inflation breaths – longer and higher pressure.

If failing to improve then intubate.

Need cardiac compression if heart rate falls below 60.

Drugs are given through umbilical vein – mostly adrenaline.

Failure to respond – incorrect intubation or tube blocked (no chest expansion), anaemia, pulmonary hypoplasia, congenital heart defect, pneumothorax, diaphragmatic hernia.

**Prematurity**

**Temperature control** – low heat production (no brown fat, little muscle activity) and high heat loss lead to hypothermia.

**Cardiac** – hypotension, easy bruising, bleeding. PDA.

**Respiratory** – narrow airways, soft thoracic cage, poor cough, unstable respiratory drive, alveolar collapse, surfactant deficiency. Leads to RDS, pneumothorax, chronic lung disease, pneumonia, pulmonary hypertension.

**GI** – uncoordinated suck and swallow, feed intolerance, GO reflux, NEC

**Infection** – limited immunity, esp GBS, staph epidermis, fungi, gram- cocci.

**Nervous** – intraventricular haemorrhage, periventricular leukomalacia (cysts), retinopathy of prematurity, developmental delay.

**Bone** – osteopenia of prematurity (prevented by giving phosphate, calcium and vit D).

**Metabolic** - dehydration, hypoglycaemia, jaundice, poor renal function leading to hyponatraemia and oedema.

**Long term problems** – retinopathy of prematurity (due to hyperoxia, vascular proliferation leading to fibrosis and retinal detachment, treat by laser), chronic lung disease, neurodevelopmental problems (about 25% of those born at 25wks, CP, developmental delay, hearing loss, seizure, behavioural difficulties). 75% survival at 27wks, 25% at 24wks.
**Opthalmitis neonatorum**
Gonococcus, Chlamydia, S. aureus, and E. coli.
Gonococcus is the most serious as pus builds up and puts pressure on the eyes and damages the cornea.

**RDS**
Commonest cause of breathing difficulties in newborn. Due to surfactant deficiency (type 2 alveolar cells), mainly in premature babies. Hypothermia, acidosis, hypoxaemia and hypovolaemia all decrease surfactant synthesis.
This is the same as hyaline membrane disease (pathological term).
Higher surface tension means that airways collapse during expiration.

**Management** – prevent with antenatal glucocorticoids. Resuscitation at birth reduces severity.
Manage by giving artificial surfactant down ET tube (2 doses 12 hrs apart), oxygen and assisted ventilation (CPAP to prevent collapse and maintain FRC).
Ventilatory support can also involve giving nitric oxide (pulmonary vasodilatation) or ECMO.
**Complications** – pneumothorax, interstitial emphysema, infection, chronic lung disease, intraventricular haemorrhage, PDA.

**Apnoeic attacks**
Preterm infants have periodic respiration with episodes of cessation of breathing due to immaturity of respiratory centre.
Use apnoea alarms. Breathing usually restarts with physical stimulation. If frequent, prevent with oral caffeine.

**Transient tachypnoea of the newborn**
Streaky appearance on CXR with fluid in horizontal fissure.
Resolves in 48hrs.

**Chronic lung disease**
Same as bronchopulmonary dysplasia. Continued need for oxygen past 36wks CGA (sometimes 28 days of life).
Related to prolonged ventilation with high pressures and oxygen concentration. Try to avoid by not using high pressures or volumes and keeping sats lower.
Recovery of lung function usually occurs over months but increased risk of respiratory infections.

**Persistent pulmonary hypertension**
Complication of RDS, meconium aspiration, polycythaemia, pulmonary hypoplasia.
Can cause cyanosis.
Treat with vasodilators and inhaled nitric oxide.

**Intracranial haemorrhage**
Very common in premature babies, most are intraventricular haemorrhages.
Detect by USS through fontanelle.
Mild IVH does not affect prognosis, severe has 50% mortality and most survivors have neurological handicaps e.g. CP.
Can also get peri-ventricular leukomalacia – softening of white matter, which can lead to cognitive deficiency or CP.

**Hypoglycaemia**
Commonest in small and premature babies. Rarely, due to liver failure or inborn error of metabolism.
May appear jittery or have convulsions or apnoea.
Prevent with frequent feeds, treat with oral or IV dextrose.
**Haemorrhagic disease of the newborn**
Deficiency in vit K. usually affects breast fed infants, as breastmilk does not contain adequate vit K. Bleeding from GI tract, umbilical stump or intracranial. Prevent by single intramuscular dose of vit K.

**Jaundice**
**Bilirubin metabolism** - haemoglobin broken down in spleen to unconjugated bilirubin. This binds to albumin and is conjugated in liver and excreted in bile. In gut converted to urobilinogen and stercobilinogen. Some is excreted in faeces, some is resorbed by enterhepatic recycling and may be re-excreted in bile or excreted in kidneys.

Mild unconjugated jaundice is normal in neonates, esp if preterm, due to increased RBC breakdown and immaturity of liver enzymes. Breast-milk jaundice is also normal, of unknown cause and resolves in first month. Onset in first 24hrs and persistence beyond 2 weeks is abnormal.

**Management** - need to treat severe (>340) neonatal unconjugated hyperbilirubinaemia (can cross BBB) to avoid kernicterus (brain damage due to deposition of bilirubin in basal ganglia). Level at which kernicterus occurs is reduced by prematurity, hypoalbuminaemia, anoxia, hypoglycaemia, acidosis, hypothermia. Treat with phototherapy (blue light breaks down bilirubin in skin and superficial capillaries to water-soluble metabolites) or if severe exchange transfusion (reduce bilirubin and also remove cause if due to isoimmune haemolysis). Persistent conjugated hyperbilirubinaemia must also be investigated to allow treatment of biliary atresia before 6wks.

**Septicaemia**
Common causes – GBS, staph aureus, coag neg staph, Ecoli, Klebsiella. Present with lethargy, drowsiness, irritability, poor feeding, temperature, blood glucose or CV instability, pallor, acidosis. Mostly related to meningitis, pneumonia or UTI.

**Seizures**
More common as brain is mostly excitatory. Most are due to perinatal complications e.g. anoxia, ICH. Also low glucose, calcium, sodium, magnesium, meningitis. Subtle seizures – eye deviation, apnoea, oral movements. Clonic seizures (rhythmic jerking, not generalised in neonates), myoclonic seizures (rapid isolated jerks), tonic seizures. Investigate – glucose, electrolytes, CSF, cranial USS for haemorrhage. Possibly also – inborn errors of metabolism, infection screen, CT or MRI.

**Hypoxic ischaemic encephalopathy**
Due to perinatal asphyxia. 
**Mild** – lethargy followed by hyperalertness and irritability, no focal signs. Good prognosis. 
**Moderate** – mild with generalised seizures. Resolves in a few days. Variable prognosis. 

**Management** - respiratory support, anticonvulsants, fluid restriction and circulatory support to prevent further injury. MRI and EEG can predict outcome.

**Necrotising enterocolitis**
Necrosis of the intestine. 
**Predisposing factors** – preterm birth, polycythaemia, PDA, early rapid oral feeding. 
**Features** – abdominal distension, vomiting, bloody stools, pneumatosis intestinalis on Xray. 
**Management** – gastric aspiration, TPN, antibiotics, surgery.
Neurology

Epilepsy
Affects 0.5% of children.
An epileptic seizure is a transient event, whereas epilepsy is a chronic syndrome, characterised by recurrent unprovoked epileptic seizures.
A seizure is a transient episode of abnormal and excessive neuronal activity, which manifests as a motor (convulsion), sensory, autonomic, cognitive or psychic disturbance and is apparent to the subject and an observer.
A single seizure can also be provoked by fever, hypoglycaemia, trauma, infection and hypoxia.

Classification of seizures
Generalised seizures (involving both cerebral hemispheres) include absence seizures, clonic seizures, tonic seizures, tonic-clonic seizures; myoclonic and atonic seizures (worse prognosis, often neurodegenerative).
Generalised tonic-clonic seizures – tonic phase of rigidity followed by clonic movements of all four limbs, loss of consciousness, post-ictal drowsiness.
Absence seizures – brief unawareness, no loss of posture, immediate recovery, automatisms.
Partial seizures (initially involving only one hemisphere): Simple (consciousness not impaired) with motor symptoms (Jacksonian), somatosensory symptoms, autonomic symptoms or psychic symptoms. Complex (impairment of consciousness) - focal symptoms then collapse or partial with secondary generalisation. E.g. temporal lobe epilepsy (automatisms, hallucinations, collapse).

Classification of epilepsy
Various different types of epilepsy syndrome, classified by generalised or localised, then idiopathic (no apparent cause) or symptomatic (known cause).
Idiopathic generalised epilepsy – includes childhood absence epilepsy and juvenile myoclonic epilepsy.
Symptomatic generalised epilepsy syndromes – infantile spasms (West syndrome), Lennox-Gastaut syndrome, progressive myoclonic epilepsy (due to inborn errors of metabolism or neurodegenerative disease e.g. neurocutaneous syndromes, Downs, fragile X, CNS lesions).
Idiopathic partial epilepsy – e.g. benign rolandic childhood epilepsy (centrotemporal spikes).
Symptomatic partial epilepsy – due to cortical dysgenesis, CNS infection, head injury, brain tumours, AV malformations etc.

Infantile spasms (West syndrome) – rare, onset around 4 months. Get myoclonic seizures and characteristic EEG (hypsarrhythmia – slow waves with spikes and sharp waves). Usually has cause e.g. tuberous sclerosis, HIA. Poor prognosis, but improved with treatment by ACTH or vigabatrin (for tuberous sclerosis).

Lennox-Gastaut – toddlers, myoclonic and atonic seizures with neurodevelopmental problems. Poor prognosis.
Childhood absence epilepsy – relatively common. Characteristic ictal EEG with generalised synchronous 3Hz spike waves. Good prognosis with spontaneous remission in adolescence. If frequent absences, then treat with valproate.

Benign rolandic childhood epilepsy – most common, partial seizure (aura) with secondary generalisation (tonic-clonic seizure). Often have CP or mental retardation. Seizures usually stop by mid teens. Worse prognosis if severe, learning disability or structural lesion.

Management
Encourage to enjoy a full life, but caution with certain activities e.g. swimming, bathing, cycling, climbing.
Medication – valproate for generalised epilepsy, carbamazepine for partial epilepsy. Newer drugs include lamotrigine and vigabatrin (permanent visual field defects).
Prolonged convulsions – cause brain damage due to hypoxia. Maintain airway, check glucose, give PR diazepam or IV lorazepam, then PR paraldehyde, then IV phenytoin. If fails intubate and ventilate. Usually due to epilepsy, febrile, head injury, intracranial infection or metabolic.

Febrile seizures
A seizure associated with a fever in a child between 6 months and 6 yrs of age (usually 2) in the absence of intracranial infection (important to exclude) of identifiable neurological disorder. Can be familial predisposition. Generally brief tonic-clonic seizures.

Prognosis – very good, normal neurological outcome, 30% recurrence risk. Worrying features are focal, long duration, persisting deficit, more than one per illness.

Management – treat underlying infection, keep patient cool, stop prolonged convulsion, educate parents. May need LP to exclude meningitis.

Seizure-like turns
Breath holding attacks – provoked by temper, screaming toddler holds breath in expiration, raised intrathoracic pressure decreases venous return, goes blue, then limp, then makes a rapid spontaneous recovery.
Reflex anoxic seizures – provoked by pain, toddler becomes pale and loses consciousness (vagal induced bradycardia), hypoxia may induce seizure.
Rigors – exaggerated shivering with high fever.

Neurocutaneous syndromes
Affect skin and CNS (both from ectoderm).

Neurofibromatosis type 1 – AD (50% new mutations). Café-au-lait patches (>6), Lisch nodule (pigmented hamartomas on eye), neurofibromas on peripheral nerves (can compress nerves and may become sarcomatous).
Neurofibromatosis type 2 – different mutation. Get acoustic neuromata and other CNS tumours.
Tuberous sclerosis – AD (most new mutations). Causes seizures, mental retardation, facial angiofibromas, hypopigmented macules (with Wood’s light), adenoma sebaceum. Also affects other organs.
Sturge-Weber syndrome – unilateral port-wine stain in distribution of trigeminal nerve associated with abnormal vessels in the brain causing seizures and haemangiomas in spinal cord.

Meningitis
Causes – meningococcus (esp group B), pneumococcus (higher fatality and sequelae), H inf. In neonates - E coli, GBS, listeria. Viruses – mumps, enteroviruses, EBV. Rarely, fungi or TB. Men C and H inf are now vaccinated against making them rare. Bacterial is the more serious, viral is usually self-limiting. Most common in young children.
Pathogens are carried in nasal passages and invade meninges via blood stream. In infants, early signs are non-specific e.g. irritability, poor feeding, vomiting, fever, drowsiness. Specific signs – bulging fontanelle, neck stiffness and photophobia, seizures, rash (meningococcal septicemia). Diagnose by lumbar puncture (not if signs of raised ICP e.g. focal neurological signs, decreased consciousness, papilloedema, or if coagulopathy or shock). Treat with IV antibiotics e.g. ceftriaxone. Steroids may reduce incidence of complications.

Complications – subdural effusion, cerebral oedema, convulsions, SIADH, neurological sequelae (esp sensorineural hearing loss).
**Encephalitis**
Inflammation of brain substance, usually viral esp HSV and viral xanthem.
Get fever, headache and vomiting followed by altered consciousness and seizures.
Give high dose acyclovir, support ABC, control seizures, EEG and MRI may show characteristic temporal lobe abnormalities.

**Neuromuscular disorders**
Anterior horn cell – spinal muscular atrophy, poliomyelitis.
Peripheral nerve – hereditary neuropathy, Guillain-Barre syndrome, Bell’s palsy.
NMJ – MG.
Muscle – muscular dystrophies, myotonia, congenital myopathies.

**Clinical features** – weakness with hypotonia, delayed motor milestones, abnormal gait.
**Investigations** – muscle enzymes (CKase raised in muscular dystrophies), nerve conduction studies, muscle and nerve biopsies, edrophonium test (MG).

**Muscular dystrophies** – inherited disorders with progressive degeneration of muscle. Duchenne – X-linked recessive (30% new mutations) mutation of dystrophin gene. Patients are wheelchair bound by 12 and die from congestive heart failure in 20s. Get pseudohypertrophy of calves, proximal muscle weakness, Gower sign, scoliosis, dilated cardiomyopathy.
Becker – milder with later onset and prolonged survival
Myotonic – AD, hypotonia, learning difficulties, distal wasting. Also cataracts and balness. Usually death due to cardiomyopathy.
Management – supportive, prolong walking with braces.

**Spinal muscle atrophy** – lose anterior horn cells. AR. Nerve conduction studies, CKase and CSF are normal. Diagnose with muscle biopsy.

**Guillain-Barre syndrome** - acute post-infectious encephalopathy, demyelinating neuropathy following infection (esp with campylobacter, CMV, EBV, mycoplasma). Get symmetrical ascending paralysis with loss of tendon reflexes and autonomic involvement (e.g. arrhythmias, respiratory failure). Raised CSF protein. Manage supportively and with IVIG and plasma exchange. Full recovery is expected.

**Headaches**
**Tension headache** - symmetrical and band-like, gradual onset, no nausea or vomiting, common.
**Migraine** – unilateral and throbbing, visual aura, associated nausea and vomiting, last several hours. May be triggered by stress or foods e.g. cheese, chocolate. Often family history.
Common – no aura, classical – aura then headache, complex – associated neurological deficit.
Treat acute headache with paracetamol and ibuprofen as early as possible. If recurrent attacks, prophylaxis with pizotifen (5HT antagonist) or propanolol.
**Raised ICP** – worse when lying down, associated with nausea, usually mild and diffuse, often focal neurological signs.

**Cerebral palsy**
Disorder of motor function due to a non-progressive lesion (but signs evolve as nervous system develops) of the developing brain, that occurred as a result of a single insult. Generally, children have other problems reflecting more widespread damage to the brain.
**Main causes** – congenital infections (rubella, CMV, TP), birth asphyxia, intraventricular haemorrhage, HIA, hyperbilirubinaemia, hypoglycaemia, head injury, intracranial infection.
Most cases have an antenatal cause.
Risk factors – maternal pre-eclampsia, prematurity, fetal distress in labour, neonatal cyanosis, fits or hypothermia.

**Clinical features** – delayed motor milestones, abnormal tone, feeding difficulties, speech delay.
Diagnose clinically by abnormalities of tone, power, reflexes, movement or posture.
Spastic CP – damage to UMN, increased tone, brisk tendon reflexes, extensor plantar response. Divide into hemiparesis (damage to internal capsule, one side, arm worse), diplegia (periventricular infarction, legs), quadriplegia. Most cases.

Dykinetic CP – damage to basal ganglia or extrapyramidal pathways e.g. kernicterus. Get abnormal movements e.g. chorea, athetosis or dystonia.

Ataxic CP – damage to cerebellum, early hypotonia with poor balance and lack of coordination.

Management – physiotherapy (reduce hypertonia and prevent contractures), braces to allow walking, surgery e.g. for contractures, support and education, antispasmodics.

Associated problems – learning difficulty, seizures, reflux, feeding problems, recurrent pneumonia.

Hydrocephalus
Enlargement of cerebral ventricles due to accumulation of CSF. Caused by obstruction and very rarely overproduction of CSF.

Intraventricular obstruction (non-communicating) – aqueduct stenosis, Dandy Walker syndrome, intraventricular haemorrhage, ventriculitis, brain tumour.

Extraventricular obstruction (communicating) – subarachnoid haemorrhage, meningitis, Arnold-Chiari malformation (blocks outflow of 4th ventricle).

Present with disproportionately large or rapidly expanding head circumference. Also have bulging fontanelle, separated sutures and eyes deviating downwards (sun-setting). In older children, signs of increased ICP.

Diagnosed by imaging.

May be able to reduce pressure with CAse inhibitors, treat lesion if possible, otherwise manage with ventriculoperitoneal shunt.

Craniosynotosis – premature fusion of cranial sutures. Infants present early with abnormal shape skull. If generalised causes microcephaly. Can be associated with syndromes e.g. Aperts, Crouzons (exophthalmus).
Renal

**UTI and pyelonephritis**

**Presentation** - presents without specific signs or symptoms in young children. Infants may develop jaundice and septicaemia rapidly.

Possible symptoms: polyuria or enuresis (also diabetes), dysuria, fever and rigors, vomiting, failure to thrive, enuresis. Pyelonephritis causes fever and loin pain presents with fever and loin pain in older children.

**Aetiology** – more common boys in neonatal period, then girls during childhood.

Mostly due to E coli, also proteus (boys). Usually related to urinary stasis e.g. reflux, obstruction, neuropathic bladder, infrequent voiding, constipation. 90% of children under 2 with UTIs have underlying anomaly.

Ask about fluid intake, voiding pattern and constipation.

**Investigation** – culture urine in any infant with a fever, unexplained abdo pain or urinary symptoms.

Diagnose with culture and growth of more than 10⁵ colony forming units/l. Dipstick may show nitrites and leucocyte esterase with Ecoli but can get false negatives.

**Management** - important in children as potential to damage growing kidneys leading to hypertension and chronic renal failure. Treat acute infection with antibiotics: oral trimethoprim in older well child, parenteral in neonates, unwell children, children with signs of pyelonephritis, children with urinary tract anomaly. After first UTI, image urinary tract to identify predisposing underlying abnormality e.g. reflux or renal scarring – USS for all children, radioisotope scan and MCUG for infants, those with anomaly or recurrent infection. Give prophylactic antibiotics until investigation complete. Advise high fluid intake, regular voiding, good perineal hygiene.

**Ureteric reflux**

Usually a developmental abnormality of vesicoureteric junction – ureter enters bladder directly, not at an angle, or segment of ureter in bladder wall is too short.

Urine refluxes up ureter during voiding – grade 1: ureter only, grade 3: moderate dilatation of ureter and renal pelvis, grade 5: gross dilatation of pelvis and calyces.

Important as exposes the kidneys to high pressure and bacteria so risk of pyelonephritis and reflux nephropathy (loss of renal tissue due to scarring).

Diagnose by micturating cystourethrogram.

Mild reflux resolves spontaneously but give prophylactic antibiotics. Surgery if prophylaxis fails or severe reflux.

**Nephrotic syndrome**

Clinical condition with heavy proteinuria, oedema and hypoalbuminaemia.

Renal biopsy to identify rare causes needed if infant, frank haematuria, steroid resistant, persistent hypertension or renal failure.

Treat with oral steroids. Most are steroid sensitive (good outcome) and remission occurs in 10 days, may have relapses. About 10% are steroid resistant (no effect after 4wks), this can lead to hypovolaemia, thrombosis, secondary infection.

**Acute glomerular nephritis**

Inflammatory changes in the glomeruli leading to oedema, hypertension, haematuria and proteinuria.

Usually post-group A strep infection, more rarely due to HSP, IgA nephropathy, SLE.

**Management** – urine is positive for blood and protein and may have casts. Check renal function and investigate aetiology. Control fluid and electrolyte balance, use diuretic and antihypertensives as needed. Usually resolves but rarely leads to renal failure.
**Hypertension**  
Usually due to renal problems in children, typically scarring due to reflux nephropathy.

**Acute renal failure**  
Mostly prerenal in children due to hypovolaemia.  
Renal failure – mostly HUS, also HSP, acute tubular necrosis.  
Post renal failure – relief by nephrostomy, catheterisation or surgery.

May need dialysis if conservative management fails, electrolyte disturbance, pulmonary oedema.  
Usually use peritoneal dialysis for children.  
Generally very good prognosis.

**Chronic renal failure**  
Rare in children, most are congenital or familial causes.  
Get bony deformities due to renal osteodystrophy, hypertension, failure to thrive and acute on chronic renal failure.  
Optimise nutrition. Restrict phosphate and give vit D to prevent bone problems.
Respiratory

**Differences in paediatric respiratory system**
Anatomy – larger tongue, higher funnel shaped larynx, shorter trachea, narrowest at cricoid not vocal cords.
Neonates are obligate nose breathers until 5 months.
More compliant chest and ribs lead to collapse.
Respiratory muscles more fatiguable.
Increased oxygen consumption.
Children with CP etc. cannot cough well so are susceptible to RTIs.

**Common cold**
Viral infection causing coryza, cough, fever and malaise.
Mostly caused by rhinoviruses, also RSV, parainfluenza, adenovirus.
Treat symptomatically e.g. with paracetamol, no treatment affects clinical outcome.
Possible complications or URTI – difficulty in feeding in infants as blocked noses, febrile convulsions, acute exacerbation of asthma.

**Pharyngitis and tonsillitis**
Usually viral, also group A strep (suggested by purulent exudate, lymphadenopathy and severe pain). Treat symptomatically. Complications include retropharyngeal (rare, stridor, toxic) or peritonsillar abscess (very sore throat, trismus, drooling) and post strep complications.

**Chronic tonsillitis** – persistently red tonsils with malaise and recurrent acute tonsillitis. Check no other disease, may respond to a long course of antibiotics.

**Asthma**
Chronic inflammatory disorder of airways with increase in airways resistance due to inflammation and hyper-reactive bronchoconstriction in response to a number of triggers (e.g. smoking, temperature change, URTI, allergens, exercise, emotion).
15% prevalence, more common in boys.

**Risk factors** – family history, atopy, parental smoking, hospitalisation for bronchiectasis, preterm.

**Clinical features** – recurrent cough, wheeze and breathlessness, bronchial hyperreactivity (diurnal variation of PEFR, worse in morning). Chronic asthma leads to thoracic deformity e.g. hyperexpansion, pectus carinatum, Harrison sulcus (depression at muscular insertion to diaphragm). Diagnose clinically, ask about severity e.g. frequency of symptoms, missing school, playing sport, disturbing sleep, longest symptom free period. Divided into infrequent episodic (most, normal lung function, step 1), frequent episodic (more severe and frequent exacerbations and mild interval symptoms with abnormal lung function, step 2-3), persistent episodic (daily symptoms, abnormal lung function, step 4-5), also exercise induced (can be mild or severe). Generally, improves in adulthood, though some remain symptomatic, esp if severe.

**Management** – control symptoms, optimise lung function, minimise side effects of medication.
Avoid triggers e.g. house dust mite (vacuuming, change duvets and pillows), parental smoking and pets.
Medication is preventers (inhaled steroid – use minimum dose regularly) and relievers (salbutamol).
**Short acting b2 agonists** e.g. salbutamol, terbutaline – cause smooth muscle relaxation and relief of bronchospasm, side effects are tachycardia, hypokalaemia, restlessness.
**Long acting b2 agonists** e.g. salmeterol – cause smooth muscle relaxation, use for nocturnal and exercise induced asthma.
**Theophylline** – phosphodiesterase inhibitor, use orally for nocturnal asthma or IV aminophylline for acute severe asthma, can cause diuresis and arrhythmias. Have to monitor blood level and can cause vomiting and headaches so rarely used.
Anticholinergics e.g. ipratropium – inhibit cholinergic bronchoconstriction, use as add on to b2 agonists in acute attack or first-line in infants, cause dry mouth and urinary retention.

Steroids – inhibit synthesis of inflammatory mediators, reduce hyperresponsiveness, prevent irreversible airway narrowing, used for prophylaxis and treatment of acute attacks. Inhaled steroids as preventers, oral e.g prednisolone for exacerbations, IV in acute asthma. Problems are oral thrush and systemic effects e.g. decreased growth, adrenal suppression.

Leukotriene receptor antagonists – add-on therapy.

Need age appropriate deliver device for medication – metered dose inhaler (older children, much deposited in nose and pharynx), metered dose inhaler with spacer (all ages but bulky), dry powder inhaler (small but needs rapid inspiration), nebuliser (severe attack, expensive, bulky and noisy). Use stepwise medication: Step 1 – inhaled b2 agonist. Step 2 – if needing reliever regularly, low dose inhaled steroids. Step 3 – leukotriene receptor antagonists, long acting B2 agonists then increase steroid dose. Step 4 – oral theophylline, high dose inhaled steroids. Step 5 – alternate day oral steroids. At any step, give rescue course of oral prednisolone. Step down if symptoms controlled.

Management of acute asthma – if severe (tachypnoea >50, tachycardia >140, can’t talk, accessory muscles) high flow oxygen, nebulised salbutamol and ipatropium, IV steroids. If life threatening (decreased consciousness, exhaustion, silent chest, poor respiratory effort, falling sats) then continuous nebulised salbutamol, IV aminophylline or salbutamol and admit to ITU.

Bronchiolitis
Viral cause, mostly RSV (some parainfluenza).

Annual winter epidemics in infants (rare over 1yr), esp ex preterm and CHD. Present with irregular breathing, tachypnoea, hypoxia, hyperinflation, crackles and wheeze.

Supportive management – monitor sats, apnoea alarm, humidified oxygen, fluids. If mild (feeding, low respiratory rate, maintaining sats) can go home. If moderate (difficulty feeding, tachypnoea, recession, low sats), admit and give oxygen and fluid. If severe (severe tachypnoea, apnoea, hypoxia), admit to ITU with high inspired oxygen and possible ventilation.

Complications – recurrent cough and wheeze over next few years.
Can give monoclonal antibody (palivizumab) to high risk infants to prevent serious disease.

Cystic fibrosis
Autosomal recessive with carrier rate of 1:25 and incidence of 1:2500.
Mutation on ch7 of CFTR protein. Most common is delta 508 (deletion of phenylalanine). Results in defective chloride ion transport, so increased viscosity of secretions, esp respiratory tract (so lose layer of epithelial lining fluid so cilia squashed) and exocrine pancreas.
Sweat test – pilocarpine iontophoresis shows high concentrations of NaCl in sweat due to failure of reabsorption, diagnose CF. Also diagnose CF due to common mutations by DNA analysis.

Clinical features – recurrent chest infection (staph, H inf, pseudomonas, b cepacia), failure to thrive, finger clubbing, hyperinflated lungs with bronchial wall thickening and wheeze. If pancreatic insufficiency, malabsorption, steatorrhoea. In 10% infants, meconium ileus causes obstruction. Also – nasal polyps, infertility in males (absent vas deferens), diabetes.

Management – prevent progression of lung disease, promote adequate nutrition. Respiratory – physiotherapy (chest percussion, breathing exercises, positive expiratory pressure masks), prophylactic antibiotics (flucloxacillin for first 2 yrs of life, antipseudomonal nebulised antibiotics if colonised), vigorous treatment of acute infections, mucolytics e.g. nebulised DNAase. Nutritional – high calorie diet, vitamin supplements, high calorie supplements, pancreatic enzyme supplements. Prognosis depends on rigid adherence to daily treatment, genotype, exposure to environmental factors.
**Bronchiectasis**

**Aetiology** – CF, primary ciliary dyskinesia, hypogammaglobulinaemia, post pneumonia (esp pertussis, TB, measles), inhaled foreign body, TB.

Get obstruction of bronchi with distal collapse and infection which heals with dilated bronchi and chronic productive cough. Can lead to cor pulmonale, pneumothorax, empyema, haemoptysis.

**Pneumonia**

Inflammation of parenchyma and consolidation of alveoli.

Following URTI get worsening fever, cough and breathlessness with tachypnoea.

Get signs of consolidation (dull to percussion, decreased breath sounds, bronchial breathing) and chest pain due to pleural inflammation.

**Management** - CXR, FBC, blood culture and nasopharyngeal aspirate. Give respiratory support, physiotherapy, careful fluid balance and IV antibiotics (penicillin or cefuroxime, erythromycin if mycoplasma or chlamydia suspected).

**Causative organisms:** Neonates – GBS, chlamydia (also conjunctivitis). Infants – RSV, pneumococcus, H inf, pertussis. Children – pneumococcus, H inf, group A strep, mycoplasma.

In older children, bacterial infections become more common, bacterial suggested by leucocytosis, consolidation, effusion.

**Complications** – bronchiectasis, lung abscess (S aureus), meningitis (H inf).

**Croup**

Viral laryngotracheobronchitis, usually caused by parainfluenza virus.

More common in boys and toddlers.

**Symptoms** - URTI, then barking cough, stridor and hoarse voice, which is worse at night. Generally able to drink and look weller than epiglottitis.

**Management** - 10% of children need hospitalisation as severe illness, young or signs of dehydration or respiratory failure. Monitor sats and heart rate. If necessary give humidified oxygen, nebulised steroid and adrenaline.

Can also get bacterial tracheitis (staph or H inf) which is similar, but child is toxic, febrile and rapid progression.

**Epiglottitis**

Due to H inf, rare due to Hib vaccine.

Generally rapid onset in young children, with absent cough and muffled voice. Child is toxic and febrile. Swallowing hurts so drool and can’t drink.

Don’t upset child, call for help, secure airway and start antibiotics.

**Whooping cough**

Contagious clinical syndrome, mostly due to bordetella pertussis. Get paroxysmal coughing, during which child may go blue and vomit. Can get nose bleeds, subconj haemorrhage, pneumonia, convulsions and bronchiectasis. Treatment with antibiotics reduces infectivity but does not shorten disease.

Dangerous to young infants so vaccinate early.
**Surgery**

**Undescended testes**
Testes develop intra-abdominally and migrate through inguinal canal to scrotum in 3rd trimester. Normal to be undescended in preterm infants, but should be in scrotum at term. Undescended testes are either incompletely descended (normal pathway) or ectopic (deviate from path after superficial inguinal ring, majority). Need to distinguish from retractile testis (can coax down to scrotum in warm room) and atrophic testes (e.g. unrecognised torsion, mumps, Klinefelter’s, Noonan’s).

Examination in newborn, at 6wks and at 18months.
If not found check existence and location by USS, laparoscopy, endocrine investigations.
Treat by orchidopexy aged 1 – thought to reduce risk of torsion, have psychological and cosmetic benefits, reduce risk of malignancy and improve fertility. If impossible, then orchidectomy.

**Phimosis and paraphimosis**

**Phimosis** - adhesion of foreskin to glans penis after the age of 3, may cause urinary tract obstruction. Non-retractile foreskins and preputial adhesions are normal in young boys and forcible retraction may result in scarring. Ballooning during urination is also common and usually resolves as prepuce becomes retractile.
If mild manage with periodic gentle retraction, if more severe may need surgery.
Smegma deposits are very common, due to adherent foreskin, resolve spontaneously.

**Paraphimosis** - irreducible retraction of foreskin beyond coronal sulcus.

**Inguinal hernia and hydrocoele**
When testes descend, take processus vaginalis, which should be obliterated around brith. If this persists get indirect inguinal hernia or hydrocoele.

**Inguinal hernia** – more common in boys, preterm babies, infants and family history. Risk of strangulation, esp in young infants. Refer for prompt surgery, usually as day case except in infants.

**Hydrocoele** – small connection with processus vaginalis gives painless transilluminating swelling which can’t be reduced. Usually resolves spontaneously by 1yr so don’t treat unless very large.

**Varicocele** – varicosities of testicular veins, occur around puberty. Treat if symptoms.

**Intussusception**
One segment of bowel telescopes into an adjacent distal part of bowel. Usually begins just proximal to the ileocaecal valve, lead point is thought to be Peyer’s patches in a young child or Meckel’s diverticulum or a polyp in an older child.

Present as vomiting (may be bilious) with episodic severe colicky abdo pain, an abdominal mass (typically sausage shaped in RUQ) and later ‘redcurrant jelly stools’. Develop signs of intestinal obstruction and shock.
Peak age is 6 months.

Investigate by USS and diagnostic enema.
In most cases, can reduce by air enema, but if unsuccessful or if long history, signs of obstruction, perforation or shock need operative reduction.

**Appendicitis**
Appendix become obstructed (by faecolith or inflammation) leading to inflammation, infection, peritoneal irritation and ultimately rupture. Rare in infants as lumen is usually wider.

Clinical features – central abdo pain that moves to RIF with peritonism. May be atypical if retrocaecal or pelvic appendix and poorly localised pain. Anorexia, mild fever, tachycardia, dehydration, guarding.
Usually diagnose clinically. Urine microscopy, FBC, CXR and abd USS may be useful if unclear. May observe for a short time (be aware of rapid progression to peritonitis), then appendicectomy.

**Volvulus**

Usually presents with bilious vomiting in first week (can be later). Due to malrotation of mid gut. This causes intermittent symptoms and vomiting, can progress to volvulus (blood supply compromised with different degrees of ischaemia). Need rapid surgery to prevent necrosis of entire midgut. Usually widen base of mesentery.

**Testicular torsion**

Presents with acute scrotum – mostly neonatal and puberty. Inadequate fixation of tunica vaginalis allows testis to rotate and occlude vascular supply. Doppler studies may be useful, then urgent surgical exploration to prevent non-viable testes. Often bilateral defect so fix both testes.

Can also get torsion of testicular appendage, usually just prior to puberty.

**Circumcision**

Only medical indication is balanitis xerotica obliterans (thickened scarred fixed white prepuce) and possibly recurrent balantitis, UTIs or phimosis. Complications – haemorrhage, infection, damage to glans, meatal stenosis. Must not circumcise infant with hypospadias as foreskin is used for correction.
Some Paediatric Syndromes

Di George (CATCH 22) – deletion of 22q11. Abnormality in development of branchial arches leading to thymus hypoplasia and T cell deficiency and hypoparathyroidism. CATCH - cardiac defects (of outflow tracts e.g. truncus arteriosus), abnormal face, thymic hypoplasia, cleft palate and hypocalcaemia.

Alport – familial deafness and nephritis.

SIDS – sudden death of an infant under 1yr, which is unexplained after complete PM. Risk factors – sleeping prone, maternal smoking, overheating at home, bed sharing, preterm birth, low-social class, drug abuse, previous child (x10). Now have CONI scheme – regular HV visits along with option of apnoea alarm, symptom diary and Cardiff weight chart (no evidence these save lives).

Achondroplasia – AD, 80% sporadic, defect in fibroblast growth receptor leading to short limbs and large head, motor development delay.

Haemolytic uraemic syndrome – most common cause of paediatric acute renal failure. Due to toxin from E coli 0157. Causes bloody diarrhoea, microangiopathic haemolytic anaemia, thrombocytopenia and renal failure to thrombosis and infarction of kidney vasculature. Antibiotics are contra-indicated, give supportive management. 90% recover full renal function.

Friedreich’s ataxia – commonest cause of primary neuronal degeneration. AR. Get loss of spino-cerebellar tracts causing truncal ataxia, then weakness, dysarthria, cardiomyopathy. Life-limiting.

Noonan’s – unusual facies (e.g. hypertelorism, down-slanting eyes, webbed neck), congenital heart disease (esp PS), short stature and chest deformity (similar to Turner’s but normal karyotype). About 25% have mental retardation, 50% have a bleeding.

Marfan’s – AD. Long fingers and limbs, hypermobile joints, high palate, dislocated lens, scoliosis. Mortality due to CV disease esp thoracic aortic aneurysm and valve disease.

William’s – distinct facial appearance, supravalvular aortic stenosis, hypercalcaemia (sometimes), behavioural and learning disabilities.

Sotos – tall, large head, facial dysmorphism, learning difficulties.

Kartagener’s – primary ciliary dyskinesia with situs inversus and dextrocardia (50% of cases).

Fanconi syndrome – generalised proximal tubular dysfunction. Causes excessive urine loss of amino acids, glucose, phosphate, sodium etc. Due to inborn errors of metabolism or toxins.

Fanconi anaemia – inherited aplastic anaemia. AR with other malformation.

Alagilles – intrahepatic biliary hypoplasia, AD with other abnormalities.

Reyes – acute non-inflammatory encephalopathy with infiltration of liver. Thought to be associated with aspirin.

Important Note
These notes were written by Liz Tatman, as a fourth year medical student in 2006. They are presented in good faith and every effort has been taken to ensure their accuracy. Nevertheless, medical practice changes over time and it is always important to check the information with your clinical teachers and with other reliable sources. Disclaimer: no responsibility can be taken by either the author or publisher for any loss, damage or injury occasioned to any person acting or refraining from action as a result of this information.

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support@askdoctorclarke.com