Paediatrics

Made from Zero to Finals, PassMedicine, Oxford Handbook of Paediatrics, NICE guidelines, etc (as of 2021)

Emergencies

Reduced Level of Consciousness

Causes

Infectious	Autoimmune	Trauma
• Menignitis/encephalitis • Toxic shock • Subdural empyema • Cerebral abscess	• Acute disseminated encephalomyelitis	• Concussion/contusion • Intracranial haemorrhage
Vascular	**Metabolic**	**Other**
• AVM • Aneurysm • Venous sinus thrombosis	• Hypoglycaemia • DKA • Inborn errors of metabolism • Hepatic encephalopathy • Uraemic encephalopathy • Endocrine (thyroid, adrenal, pituitary)	• Seizures & post-ictal state • Hypertension • Hydrocephalus • Hypoxia/ischaemia • CNS tumours • Toxins

Assessment

ABCD Approach
- Ensure breathing & patent airway
- Adjuncts (suction, oropharyngeal/nasopharyngeal airway, BVM) if appropriate
- Check SpO2
- Provide oxygen if available and required
- Assess HR, BP, & CRT
- IV access and fluid bolus if required
- Calculate GCS

Collateral History/Environment Inspection
- Onset & progression
- Possible toxic ingestion/exposure
- Recent trauma/illness/infection exposure
- History of previous episodes/seizures
- Family history

General Exam
- Vital signs
- Alert bracelets indicating any known condition
- Skin (trauma, rash, petechiae, jaundice, needle tracks)
- Breath odour

Head & Neck
- Fontanelle (if open): tense/flat/sunken
- CSF leakage from nose/ears or raccoon sign indicate basal skull fracture

Pupils
- Small and reactive: suggests metabolic cause
- Mid-size & unreactive: suggests midbrain lesion
- Pinpoint: suggests pontine lesion or opioids
- Unequal with one fixed & dilated: suggests lesion on side of the dilated pupil
- Bilateral fixed & dilated: poor prognosis unless associated with barbiturate poisoning/hypothermia

Fundi
- Retinal haemorrhages/papilloedema

Signs of Raised ICP
- Abnormal respiratory pattern
- Uneuqal or unreactive pupils
- Hypertension with bradycardia and apnoea (Cushing's triad)
- Tense fontanelle
- Decerebrate/decortical posturing

Investigation

Bloods
- FBC, coagulation, glucose, U+E, LFTs, TFTs, ammonia, lactate
- ABG

Microbiology
- Blood & urine cultures
- Viral respiratory panel

Imaging
- CT/MRI if suspected raised ICP or structural brain disease

Lumbar Puncture
- Defer if signs of raised ICP, focal neurology, or active bleeding

Management
- As per underlying cause
- Raised ICP with GCS </=8 requires rapid intubation and ICU management

Anaphylaxis

- Anaphylaxis is a sudden onset, severe & life-threatening IgE mediated hypersensitivity reaction.

Triggers

- Food most commonly
  - Milk in infants
  - Nuts/shellfish in older children
- Drugs
  - Penicillin

Risk Factors for Fatal Anaphylaxis

- Not lying child supine
- Delayed administration of adrenaline
- Poorly controlled asthma
- Adolescence
- Nut/shellfish allergy

Presentation

- Shortly following ingestion of allergen

Skin symptoms in 80%

- Itch, urticaria, angioedema, flushing

Airway/respiratory involvement

- Airway oedema and secretions leading to stridor/wheeze/respiratory distress

Cardiovascular involvement

- Widespread vasodilation and oedema leading to hypotension and tachycardia

Gastrointestinal involvement:

- Severe & persistent abdominal pain/vomiting/diarrhoea

Management

1. ABC assessment
   - Airway patent?
   - Give immediate high flow oxygen
   - Quality & rate of respirations
   - Pulse & CRT
   - 2x wide bore IV access

Immediate senior management – anaesthetics if airway obstruction, ENT if surgical airway likely to be required

2. Administer IM adrenaline
   - 10mcg/kg (1ml/kg of 1:1,000 adrenaline)
   - Autoinjectors: green (.15mg) for age 1-5, yellow (.3mg) for age > 5

If there is complete cardiac arrest or a complete airway obstruction, give adrenaline IV 10mcg/kg and start APLS

3. Repeat IM adrenaline every 3-5 minutes if upper/lower airway obstruction or shock persist
4. Consider 20ml/kg NaCl fluid bolus

If unresolved:

5. Consider nebulised adrenaline (5ml of 1:1,000)
6. Consider adrenaline infusion
   - 0.03mg/kg/ made up to 50ml with NaCl
   - Start at 10ml/hour
7. Nebulised salbutamol & ipratropium
8. IV aminophylline & hydrocortisone if wheeze

On Resolution:

- Cetirizine PO for itch
  - 2.5mg < 2 years
  - 5mg 2-5 years
  - 10mg 6+ years

Acute Asthma

- Rapid deterioration of the symptoms of asthma, triggered by any of the typical triggers

Presentation

- Progressively worsening shortness of breath
- Signs of respiratory distress
- Tachypnoea
- Widespread expiratory polyphonic wheeze
- Tight chest with reduced entry
  - Silent chest is an ominous sign

Severity

Moderate	Severe	Life Threatening
Peak flow > 50% predicted	Peak flow < 50% predicted	Peak flow < 33% predicted
Normal speech	Saturations < 92%	Saturations < 92%
	Unable to complete sentences in one breath	Exhaustion and poor respiratory effort
	Signs of respiratory distress	Hypotension
	Respiratory rate: > 40 in 1-5 years > 30 in > 5 years	Silent chest
	Heart rate: > 140 in 1-5 years > 125 in > 5 years	Cyanosis
		Altered consciousness / confusion

Management

Principles

- Oxygen if required (SpO2 < 94%)
- Bronchodilators
- Steroids to reduce airway inflammation
  - Should be given to all children with acute asthma for 3-5 days
- Antibiotics only if bacterial cause is suspected
- Monitor potassium when giving high doses of salbutamol

Mild Cases

- Outpatient care
- Regular salbutamol via spacer
  - 4-6 puffs every 4 hours

Moderate-Severe Cases (Stepwise)

1. Salbutamol via spacer (start with 10 puffs every 2hrs)
2. Nebulised salbutamol/ipratropium bromide
3. Oral prednisolone (1mg/kg for 3 days)
4. IV hydrocortisone
5. IV MgSO4
6. IV salbutamol
7. IV aminophylline
8. Call anaesthetics & ICU

Weaning

- Look for:
  - Cyanosis
  - Tracheal tug
  - Subcostal recessions
  - Hypoxia
  - Tachypnoea
  - Wheeze
- If well, step down the ladder

Discharge

- Considered when child is well on 6 puffs 4 hourly of salbutamol
- Step-down regime:
  - 6 puffs 4 hourly for 48 hours
  - 4 puffs 6 hourly for 48 hours
  - 2-4 puffs as required
- Finish steroid course
- Provide safety net information & personalised asthma plan

Meningitis

- Inflammation of the meninges due to infection

Organisms

< 3 Months

- GBS
- E. coli
- L. monocytogenes

< 6 Years

- N. meningitidis
- S. pneumoniae
- H. influenzae

> 6 Years

- N. meningitis
- S. pneumoniae

Viral

- Typically less severe than bacterial

Presentation

- Fever
- Nick stiffness
- Vomiting
- Headache
- Photophobia
- Altered consciousness
- Seizures
- Non-blanching petechial rash (meningococcal septicaemia & DIC)
- Kernig's/Brudzinski's tests positive

Non-specific features in neonates/young babies

- Hypotonia, lethargy
- Poor feeding
- Hypothermia
- Bulging fontanelle

Lumbar Puncture

Contraindications

- Focal neurological signs
- Papilloedema
- Significant bulging of fontanelle
- DIC
- Signs of cerebral herniation
- GCS < 9
- Haemodynamically unstable
- Suspected meningococcal septicaemia
  - Do blood cultures & meningococcus PCR

Results

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

Management of Bacterial Meningitis

Community

- Suspected meningitis + non blanching rash:
  - Immediate IM benzylpenicillin & transfer to hospital
  - Transfer is priority if any issue with antibiotics (eg allergy)

Hospital

- Ideally CSF sample should be taken before antibiotics but should not delay them if urgent
- Bloods should be sent for meningococcal PCR

1. Typical **antibiotics:**
   - **< 3 months:** IV cefotaxime + amoxicillin
   - **> 3 months:** IV ceftriaxone
   - Vancomycin added if risk of resistant pneumococcal infection
2. **Steroids**
   - Not in children < 3 months
   - Dexamethasone 4 times daily for 4 days if CSF suggests bacterial meningitis
3. **Fluids**
   - Resuscitate if shocked
4. **Cerebral Monitoring**
   - Assisted/invasive ventilation/ICU may be required
5. **Post-exposure prophylaxis**
   - Inform public health
   - Single dose of ciprofloxacin for those with close contact in last 7 days

Management of Viral Meningitis

- Viral PCR testing of CSF sample
  - HSV, VZV, enterovirus
- Typically require only supportive treatment
- Acyclovir can be used for suspected/confirms HSV/VZV infection

Complications

- Hearing loss
- Seizures & epilepsy
- Cognitive impairment & learning disability
- Memory loss

Cerebral palsy with focal deficits

Sepsis

- Systemic inflammatory response caused by infection resulting in hypotension, systemic hypoperfusion, tissue anaerobic respiration, and organ failure (septic shock)

Causative Organisms

< 3 months

- GBS
- E. coli
- L. monocytogenes

> 3 months

- N. meningitidis
- S. pneumoniae
- S. aureus
- E. coli

Presentation

- Seriously unwell appearance
- Deranged vital signs
- Prolonged capillary refill time
- Fever or hypothermia
- Deranged behaviour
- Poor feeding
- Inconsolable/high-pitched crying
- Weak cry
- Reduced consciousness
- Reduced tone
- Skin colour changes (cyanotic/pale/mottled/ashen)
- Signs of organ hypoperfusion

Immediate (Emergency) Management

- **Oxygen** if signs of shock or SpO2 < 94%
- **IV access**
- **Bloods**
  - FBC, CRP, U+E,
  - Clotting screen
  - Blood gas (lactate & acidosis)
  - Cultures (ideally before antibiotics)
- **IV Antibiotics**
  - As per local guidelines
- **IV Fluids**
  - 20ml/kg bolus if lactate > 2mmol/L or signs of shock
  - Can be repeated
- Monitor **urine output**

Further Investigations (depending on suspicions)

- CXR (pneumonia)
- Abdominal & pelvic US (intra-abdominal infection)
- Lumbar puncture (meningitis)
  - Unless contraindicated
- Meningococcal PCR (meningococcaemia)
- Serum cortisol (adrenal crisis)

NICE Feverish Illness Under 5 Risk Assessment

	Green – low risk	Amber – intermediate risk	Red – high risk
Colour (of skin, lips or tongue)	• Normal colour	• Pallor reported by parent/carer	• Pale/mottled/ashen/ blue
Activity	• Responds normally to social cues • Content/smiles • Stays awake or awakens quickly • Strong normal cry/not crying	• Not responding normally to social cues • No smile • Wakes only with prolonged stimulation • Decreased activity	• No response to social cues • Appears ill to a healthcare professional • Does not wake or if roused does not stay awake • Weak, high-pitched or continuous cry
Respiratory		• Nasal flaring • Tachypnoea: – RR >50 breaths/ minute, age 6–12 months – RR >40 breaths/ minute, age >12 months • Oxygen saturation ≤95% in air • Crackles in the chest	• Grunting • Tachypnoea: RR >60 breaths/minute • Moderate or severe chest indrawing
Circulation and hydration	• Normal skin and eyes • Moist mucous membranes	• Tachycardia: – >160 beats/minute, age <12 months – >150 beats/minute, age 12–24 months – >140 beats/minute, age 2–5 years • CRT ≥3 seconds • Dry mucous membranes • Poor feeding in infants • Reduced urine output	• Reduced skin turgor
Other	• None of the amber or red symptoms or signs	• Age 3–6 months, temperature ≥39°C • Fever for ≥5 days • Rigors • Swelling of a limb or joint • Non-weight bearing limb/not using an extremity	• Age <3 months, temperature ≥38°C* • Non-blanching rash • Bulging fontanelle • Neck stiffness • Status epilepticus • Focal neurological signs • Focal seizures

Green

- Manage at home with appropriate care & advice

Amber

- Provide information & safety net or refer to paediatric specialist

Red

- Refer urgently to paediatric specialist

Development & Growth

Normal Development

Age	Gross Motor	Fine Motor / Vision				Speech / Language	Social
		Draw	Brick	Cut	Beads		
6 weeks	▪ Good head control – raises head to 45° when on tummy, ▪ Stabilises head when raised to sitting position	▪ Tracks object/ face				▪ Stills, startles at loud noise	▪ Social smile (visual problem if not)
6 months	▪ Sit without support, rounded back ▪ Rolls tummy (prone) to back (supine) . Vice versa slightly later.	▪ Palmar grasp (5m) ▪ Transfer hand to hand				▪ Turns head to loud sounds ▪ Understands "bye bye" / "no" (7m) ▪ Babbles (monosyllabic)	▪ Puts objects to mouth (stops at 1yr) ▪ Shakes rattle ▪ Reaches for bottle / breast
9 months	▪ Stands holding on ▪ Straight back sitting (7 ½ m)	▪ Inferior pincer grip ▪ Object permanence				▪ Responds to own name ▪ Imitates adult sounds	▪ Stranger fear (6-9 mths – 2yrs) ▪ Holds and bites food
12 months	▪ Walks alone (9-18m) → 18m is threshold for worry – i.e. Duchenne's MD, hip problems, cerebral palsy etc	Draw	Brick	Cut	Beads	▪ **Shows understanding of nouns** ("where's Mummy?") ▪ 3 words (50% at 13m) ▪ Points to own body parts (15m), doll (18m)	▪ Waves "bye bye" ▪ Hand clapping ▪ Plays alone if familiar person nearby ▪ Drinks from beaker with lid
			2				
		▪ Neat pincer grip (10m) ▪ Casting bricks (should disappear by 18m – persistence beyond this = abnormal)					
18 months	▪ Runs (16m) ▪ Jumps (18m)	To and fro (15m)	4			▪ **Shows understanding of nouns** ("show me the xxxxx") ▪ 1 to 6 different words	▪ Imitates every day activities
2 Years	▪ Runs tiptoe ▪ Walks upstairs, both feet / each step. ▪ Throws ball at shoulder level	Vertical line	8			▪ **Shows understanding of verbs** ("what do you *draw* with, what do you *eat* with?") ▪ 2 words joined together (50+ words)	▪ Eats skilfully with spoon (2½ years)
		▪ Puzzles – shape matching is >2 yr skill. Random effort <2 yr ▪ Turns several pages of book at a time					
2 ½ Years	▪ Kicks ball	Horizontal line				▪ **Shows understanding of prepositions in/on** ("put the cat on the bowl") ▪ 3 – 4 words joined together	
3 Years	▪ Hops on one foot for 3 steps (each foot) ▪ Walks upstairs, one foot per step; downstairs two feet per step.	Circle	Bridge □ □ □ (or train)	Single cuts	Griffiths beads	▪ **Understands negatives** ("which of these is NOT an animal?") ▪ **Understands adjectives** ("which one is red?")	▪ Begins to share toys with friends ▪ Plays alone without parents ▪ Eats with fork and spoon ▪ Bowel control
		▪ Turns one page of book at a time					
3 ½ Years				Cuts pieces		▪ **Understands comparatives** ("which boy is bigger than this one?" while pointing to middle-sized boy! Or draw circles to illustrate point)	
4 Years	▪ Walks upstairs / downstairs in adult manner	Cross Square (4.5 yrs) Triangle / person (5 yrs)	12 blocks Steps □ □□ □□□ Big steps (5y) □ □□ □□□ □□□□	Cuts paper in half	Small beads	▪ **Understands complex instructions** ("Before you put *x* in *y*, give *z* to Mummy") ▪ Uses complex narrative / sequences to describe events.	▪ Concern/sympathy for others if hurt ▪ Has best friend ▪ Bladder control (4½ years) ▪ Engages in imaginative play, observing rules (4½ to 5 years old) ▪ Eats skilfully with little help ▪ Handles knife (at 5 yrs) ▪ Dressing and undressing

Failure to Thrive

- Poor physical growth and development in a child
- Fall in weight across:
  - 1 or more centile spaces if birth weight was below the 9th centile
  - 2 or more centile spaces if birth weight was between 9th and 91st centile
  - 3 or more centile spaces if birth weight was above 91st centile

Causes

Inadequate Nutritional Intake

- Maternal malabsorption (if breastfeeding)
- IDA
- Family/parental problems/neglect
- Poverty

Difficulty Feeding

- Poor suck (eg due to cerebral palsy)
- Cleft lip/palate
- Genetic syndromes/conditions
- Pyloric stenosis

Malabsorption

- Cystic Fibrosis
- Coeliac Disease
- Cow's milk intolerance
- Chronic diarrhoea
- IBD

Increased Energy Requirements

- Hyperthyroidism
- Chronic disease
- Malignancy
- Chronic infections (HIV, immunodeficiency)

Metabolic

- Inborn errors of metabolism
- Type 1 diabetes

Investigations

- Urine dipstick
- Coeliac screen (anti-TTG/anti-EMA)
- Other investigations only if specific clinical concerns

Management

- MDT input
- Regular mealtimes and snacks
- Reduce milk intake, encourage other foods
- Dietician review
- Additional energy dense foods/supplements
- Enteral tube feeding may be necessary in severe situations

Short Stature

- 2 standard deviations below the average height for age and sex (<2nd centile)

Predicted Height

- Boys: (Mother's height + father's height + 14)/2
- Girls: (Mother's height + father's height - 14)/2

Causes

- Familial
- Constitutional delay in growth and puberty
- Malnutrition
- Chronic diseases
- Endocrine disorders
- Genetic conditions
- Skeletal dysplasias

Constitutional Delay in Growth and Puberty (CDGP)

- Short stature in childhood but normal adult height
  - Delayed puberty but prolonged growth spurt in puberty
- Delayed bone age based on bones and growth plate of wrist

Diagnosis

- History & exam
- X-ray of hand and wrist for bone age

Management

- Exclude other causes
- Reassure and monitor

Delayed Puberty

Normal Puberty

- Starts age 8-14 in girls, 9-15 in boys
- 4 years from start to finish

Girls

- Growth spurt comes earlier than boys
- First breast buds, then pubic hair, then menstruation ~2 years after puberty onset

Boys

- First enlargement of the testes, then penis, darkening of scrotum, development of pubic hair, deepening of the voice

Causes

Hypogonadotropic Hypogonadism (Lack of LH/FSH)

- Damage to hypothalamus/pituitary
  - Previous surgery or radiation
- GH deficiency
- Hypothyroidism
- Hyperprolactinaemia
- Serious chronic conditions
- Excessive exercise/dieting
- CDGP
- Kallman syndrome

Hypergonadotropic Hypogonadism (No response to LH/FSH)

- Damage to the gonads
  - Torsion
  - Cancer
  - Infections
- Congital absence of gonads
- Androgen insensitivity syndrome
- Klinefelter's Syndrome
- Turner's Syndrome

Investigations

- Investigate when there is no signs of puberty at 13 in girls/14 in boys

Bloods

- FBC & ferritin, U&E
- Anti-TTG/anti-EMA
- Early morning LH & FSH
- TFTs, IGF-1, prolactin

Microarray

- Klinefelter's/Turner's syndrome

Imaging

- X-ray of wrist for bone age
- Pelvic ultrasound in girls
- MRI of the brain to look for pituitary pathology/olfactory bulbs in Kallman syndrome

Management

- Treat underlying condition
- Reassure and monitor constitutional delay
- Hormone replacement in certain circumstances

Precocious Puberty

- Development of secondary sexual characteristics before the age of 8 in girls and 9 in boys

Classification

Gonadotropin Dependent/Central/True

- Premature activation of HPG axis
- FSH & LH raised

Gonadotropin Independent/Pseudo/False

- Excess sex hormones
- FSH & LH low

In Males

- Rare, usually has an organic cause

Testes

- Bilateral enlargement – Release from intracranial lesion
- Unilateral enlargement – Gonadal tumour
- Small testes – Adrenal cause (tumour/hyperplasia)

In Females

- Usually idiopathic/familial
- Organic causes (eg McCune Albright Syndrome) are rare and associated with rapid onset and neuro symptoms

Developmental Delay

Global Developmental Delay

- Slow development in 2 or more domains

Causes

- Down's syndrome
- Fragile X syndrome
- Fetal alcohol syndrome
- Rett syndrome
- Metabolic disorders
- Etc etc

Gross Motor Delay

- Cerebral palsy
- Congenital ataxia
- Myopathy
- Spina bifida
- Visual impairment

Fine Motor Delay

- Dyspraxia
- Cerebral palsy
- Muscular dystrophy
- Visual impairment
- Congenital ataxia

Language Delay

- Specific social circumstances
  - Multiple languages
  - Talkative siblings
- Hearing impairment
- Learning disability
- Neglect
- Autism
- Cerebral palsy

Personal & Social Delay

- Emotional and social neglect
- Parenting issues
- Autism

Learning Disability

Types

- **Dyslexia:** Difficulty in reading, writing and spelling
- **Dysgraphia:** Specific difficulty in writing
- **Dyspraxia/Developmental Co-ordination Disorder:** Difficulty in physical co-ordination, presents with motor delays and clumsiness
- **Auditory Processing Disorder:** Difficulty processing auditory information
- **Non-Verbal Learning Disability:** Difficulty processing non-verbal information, such as facial expression or body language
- **Profound and Multiple Learning Disability:** Severe difficulties across multiple areas, requiring assistance with activities of daily living

Classification (based on IQ)

- **55-70:** Mild
- **40-55:** Moderate
- **25-40:** Severe
- **<25:** Profound

Causes

- Often no clear cause
- Family history increases risk
- Abuse, neglect, psychological trauma, toxins increase risk
- Specific causes:
  - Genetic conditions
  - Antenatal/intrapartum/neonatal problems
  - Autism
  - Epilepsy

Capacity

- Is decision-specific
- Must demonstrate ability to:
  - Understand the decision to be made
  - Retain the information needed
  - Weigh up the options and implications
  - Communicate their decision

Genetic Syndromes

Down's Syndrome (Trisomy 21)

Dysmorphic Features

- Face
  - Upslanting palpebral fissures
  - Epicanthic folds
  - Brushfield iris spots
  - Protruding tongue
  - Small low-set ears
  - Round/flat face
- Short neck
- Flat occiput
- Short stature
- Hypotonia
- Single palmar crease
- Saddle-gap deformity

Complications

Cardiac

- Endocardial cushion defect 40%
- Ventricular septal defect 30%
- Secundum atrial septal defect 10%
- Tetralogy of Fallot 5%
- Isolated PDA 5%

GI

- Duodenal atresia
- Hirschsprung's disease

ENT

- Eustachian tube abnormalities
  - Recurrent otitis media
  - Glue ear
- Deafness

Ophthalmological

- Cataracts
- Myopia
- Strabismus

Other

- Learning disability
- Hypothyroidism
- Atlanto-axial instability
- Leukaemia (ALL)
- Dementia
- Down's Arthritis

Antenatal

Screening

- Screening tests give a probability of trisomy 21
- Combined test 1st line
  - 11-14wks
  - Ultrasound measurement of nuchal translucency (↑)
  - Maternal bloods
    - B-hCG (↑)
    - PAPP-A (↓)
- Triple test
  - 14-20wks
  - Maternal bloods
    - B-hCG (↑)
    - PAPP-A (↓)
    - Oestriol (↓)
- Quadruple test
  - As above + inhibin A (↑)

Definitive Testing

- Performed if risk based on screening is >1/150
- Chorionic villous sampling (before 15wks)
- Amniocentesis (later, when there is enough amniotic fluid)
- Cells obtained undergo karyotyping

Non-Invasive Prenatal Testing

- New testing method, analyses fragments of fetal DNA in maternal blood
- Not definitive but potential non-invasive alternative to chorionic villous sampling/amniocentesis

Management

MDT Approach

- OT, SALT, physio, dietetics, social services
- Paediatrics, GP, ENT, cardiology, audiology
- Optician

Screening for complications

- Echocardiogram
- Regular TFTs (2 yearly)
- Regular audiometry
- Regular eye checks

Other Trisomies

Edward's Syndrome (Trisomy 18)

Features

- Learning disability
- Micrognathia
- Low-set ears
- Rocker-bottom feet
- Overlapping of fingers

Patau Syndrome (Trisomy 13)

- Learning disability
- Rocker-bottom feet
- Microcephaly
- Small eyes
- Polydactyly
- Scalp lesions

Klinefelter Syndrome

- Males with karyotype 47XXY
- Features may not be noticed until puberty
- Rare, more severe karyotypes can occur (48XXXY, 49XXXXY)

Features

- Tall height
- Lack of male secondary sexual characteristics
- Wide hips
- Gynaecomastia
- Weaker muscles
- Small testes
- Reduced libido
- Shyness
- Infertility
- Potential subtle learning difficulty
  - Particularly affecting speech/language
- Hypergonadotropic hypogonadism

Diagnosis

- Karyotype

Management

- Testosterone injections can improve features
- Advanced IVF techniques can allow fertility
- Breast reduction surgery
- MDT input

Prognosis

- Normal life expectancy
- Slight increased risk of:
  - Breast cancer (relative to other males only)
  - Osteoporosis
  - Diabetes
  - Anxiety & depression

Turner Syndrome

- Females with karyotype 45XO

Features

- Short stature
- Webbed neck
- Broad “shield” chest
- Widely spaced nipples
- High arched palate
- Downward sloping eyes & ptosis
- Short 4th metacarpal
- Multiple pigmented naevi
- Cubitus valgus
- Underdeveloped ovaries
- Late/incomplete puberty
- Infertility

Associated Conditions

- Recurrent otitis media
- Recurrent urinary tract infections
- Coarctation of the aorta
- Hypothyroidism
- Hypertension
- Obesity
- Diabetes
- Osteoporosis
- Autoimmune conditions
- Crohn’s disease
- Horseshoe kidney
- Learning disabilities

Diagnosis

- Karyotype

Management

- Growth hormone therapy helps prevent short stature
- Progesterone & oestrogen replacement help establish secondary sexual characteristic, regulate menstruation and prevent osteoporosis
- Fertility treatment
- Monitoring for associated conditions

Fragile X Syndrome

- Trinucleotide repeat in FMR1 (Fragile X Mental Retardation 1) gene on the X chromosome
  - Protein plays a role in cognitive development in brain
- Males more severely affected due to single X chromosome
  - Females affecting to varying degrees
- X-linked, may be inherited from mother (even if phenotypically normal) or a de novo mutation

Features

- Intellectual disability
- Long narrow face
- Large ears
- High-arched palate
- Macroorchidism
- Hypermobile joints
- ADHD
- Autism
- Seizures
- Mitral valve prolapse

Diagnosis

- Can be made antenatally by chorionic villus sampling or amniocentesis
- Analysis of the number of CGG repeats using restriction endonuclease digestion and southern blot analysis

Management

- Symptomatic
- MDT involvement

Noonan Syndrome

- Autosomal dominant condition with normal karyotype
- Though to be lots of causes, prominently a defect on chromosome 12

Features

- Short stature
- Webbed neck
- Low-set ears
- Widely-spaced nipples
- Broad forehead
- Downward sloping eyes & hypertelorism
- Prominent nasolabial folds

Associated Conditions

- Congenital heart disease (pulmonic stenosis)
- Cryptorchidism → infertility in males
- Learning disability
- Bleeding diatheses (factor XI deficiency)
- Lymphoedema
- Increased risk of leukaemia & neuroblastoma

Management

- Supportive
- MDT involvement
- Surgery for congenital cardiac defects

William Syndrome

- Microdeletion on chromosome 7
- Usually a random deletion occurring at conception rather than an inherited condition

Features

- Broad forehead
- Starburst eyes
- Flattened nasal bridge
- Long philtrum
- Wide mouth & widely spaced teeth
- Small chin
- Sociable trusting personality
- Mild learning disability

Associated Conditions

- Supravalvular aortic stenosis
- ADHD
- Hypertension
- Hypercalcaemia

Management

- MDT involvement
- Echocardiograms and blood pressure monitoring
- Low calcium diet & avoid calcium/vitamin D supplements

Prader-Willi Syndrome

- Example of genetic imprinting where there is a deletion on the long arm of chromosome 15
  - Prader-Willi syndrome if deleted from father
    - Loss of Prader-Willi gene
  - Angelman syndrome if deleted from mother
    - Loss of UBE3A gene
- Prader-Willi may be caused by:
  - Microdeletion of paternal 15q11-13 (70%)
  - Maternal uniparental disomy of Chr 15

Features

- Insatiable hunger
  - Childhood obesity
- Hypotonia
- Mild-moderate learning disability
- Hypogonadism
- Fair, soft skin prone to bruising
- Mental health issues
  - Particularly anxiety
- Narrow forehead
- Almond shaped eyes
- Strabismus
- Thin upper lip
- Downturned mouth

Management

- Dietetics and strict restriction of dietary intake
  - This can be extreme and require locking of cupboards, not leaving uneaten food in bins, etc
  - Caloric intake when controlled can be slightly less than normal due to reduced activity as a result of poor muscle tone & growth
- NICE recommends growth hormone to treat poor muscle tone & growth
- Other MDT involvement
  - Parental education
  - Social services & support
  - OT
  - Educational support
  - Physio
  - CAMHS

Angelman Syndrome

- Caused by loss of UBE3A gene of Chr 15
  - Deletion on maternal Chr 15
  - Paternal uniparental disomy of Chr 15
  - Mutations within UBE3A gene

Features

- Developmental delay & learning disability
- Severe speech delay/absence of speech development
- Ataxia
- Fascination with water
- Happy demeanour
- Inappropriate laughter
- Hand flapping
- Abnormal sleep patterns
- Epilepsy
- ADHD
- Microcephaly
- Fair skin, light hair, blue eyes
- Wide mouth & widely spaced teeth

Management

- MDT involvement
  - Parental education
  - Social services & support
  - OT
  - Educational support
  - Physio
  - CAMHS
- Anti-epileptic medication as required

Neonatology

Neonatal Resuscitation

Issues

- Hypoxia – may lead to HIE
- Temperature
  - Babies have a large surface area to weight ratio and are born wet, so lose heat rapidly
- Meconium – may be present in mouth or airway

Principles

Warm the Baby

- Vigorous drying
- Resuscitation under heat lamp
- Babies under 28wks are placed in a plastic bag while still wet

Calculate APGAR Score

- 1, 5 & 10 minutes

Finding	0	1	2
Appearance (skin colour)	Blue / pale centrally	Blue extremities	Pink
Pulse	Absent	< 100	> 100
Grimmace (response to stimulation)	No response	Little response	Good response
Activity (muscle tone)	Floppy	Flexed arms and legs	Active
Respiration	Absent	Slow/irregular	Strong/crying

Stimulate Breathing

- Vigorous drying
- Place head in a neutral position (eg towel under shoulders)
- If gasping/not breathing, check airway for obstruction and consider aspiration

Inflation Breaths

- If neonate is gasping/not breathing despite adequate stimulation
  - 2 cycle of 5 inflation breaths lasting 3 seconds each
  - If no response and heart rate low, 30 seconds of ventilation breaths
  - If still no response, chest compressions coordinated with ventilation breaths

Chest Compressions

- Performed if heart rate remains below 60 despite resuscitation and inflation breaths
- 3:1 ratio

Severe Situations

- Intubation and IV drugs may be needed
- Babies with potential HIE may benefit from therapeutic cooling

Delayed Cord Clamping

- Uncompromised neonates: 1 minute
- "Placental transfusion" reduces anaemia, hypotension, intraventricular haemorrhage and necrotising enterocolitis
- ↑ neonatal jaundice & need for phototherapy

Normal Care After Birth

Immediate

- **Skin-skin contact**
  - Helps warm baby
  - Improves mother and baby interaction
  - Calms baby
  - Improves breast feeding
- Clamp the cord
- Dry the baby
- **Vitamin K**
  - Babies born deficient
  - Standard practice is IM injection immediately after birth
  - Oral is possible, requires doses at birth, 7 days and 6 weeks
- Label the baby
- Measure and weigh

Later

- Initiate breast/bottle feeding as soon as baby is alert enough
- First bath after baby is warm and stable (can wait a few days)
- Newborn exam within 72hrs
- Bloodspot test
- Hearing test

Bloodspot Screening

- Day 5 (8 at the latest)
- Heel prick and 4 drops on screening card
- Screens for 8 congenital conditions:
  - Cystic fibrosis
  - Congenital hypothyroidism
  - Phenylketonuria
  - Medium chain acyl-CoA dehydrogenase deficiency (MCADD)
  - Maple syrup urine disease (MSUD)
  - Classical galactosaemia (C Gal)
  - Glutaric aciduria type 1 (GA1)
  - Homocystinuria

Birth Injuries

Caput Succedaneum

- Scalp oedema outside the periosteum
- Due to instrumental (particularly ventouse) or prolonged delivery
- Crosses suture lines
- No/mild discolouration
- Resolves in a few days

Cephalhaematoma

- Bleeding between periosteum and skull
- Commonly over parietal region
- Due to instrumental or prolonged delivery
- Doesn't cross suture lines
- Skin discolouration
- May cause anaemia or jaundice
- Resolves in a few weeks

Facial Paralysis

- Facial nerve injury is associated with forceps delivery
- Function normally returns after a few months
- Neurosurgical input if not resolved spontaneously

Erb's Palsy

- Damage to C5/C6 nerves in brachial plexus
- Associated with shoulder dystocia, instrumental delivery and high birth weight
- Weakness of shoulder abduction, external rotation, arm flexion, and finger extension
  - Adducted internally rotated shoulder
  - Extended elblow
  - Pronated flexed wrist
  - Lack of movement
- Function normally returns after a few months
- Neurosurgical input if not resolved spontaneously

Fractured Clavicle

- Associated with shoulder dystocia, instrumental delivery and high birth weight
- Presents with:
  - Noticeable lack/asymmetry of movement of affected arm
  - Asymmetry of shoulders (affected shoulder lower)
  - Pain and distress on movement of affected arm
- Confirmed with ultrasound or x-ray
- Managed with immobilisation
- May cause brachial plexus injury

Hypoxic Ischaemic Encephalopathy

- Prolonged/severe hypoxia causing brain ischaemia during birth
- Can cause cerebral palsy or death
- Suspect if:
  - Perinatal/intrapartum complications
  - Acidosis (pH<7) on umbilical artery blood gas
  - Poor Apgar scores
  - Evidence of multi-organ failure

Causes

- Maternal shock
- Intrapartum haemorrhage
- Prolapsed cord
- Nuchal cord

Features (Sarnat Staging)

Mild

- Poor feeding, irritability, hyper-alert
- Resolves within 24hrs
- Normal prognosis

Moderate

- Poor feeding, lethargy, hypotonia, seizures
- Can take weeks to resolve
- 40% develop cerebral palsy

Severe

- Reduced consciousness, apnoeic episodes, flaccidity, reduced/absent reflexes
- Up to 50% mortality
- 90% develop cerebral palsy

Management

- Full supportive care
- Therapeutic cooling may be appropriate
- Follow up with paediatrics and MDT

Therapeutic Hypothermia

- Controlled cooling in NICU to prevent secondary neuronal death in HIE
- 33-34° monitored by rectal probe
- 72hrs duration, followed by gradual re-warming over 6hrs

Criteria

- Gestational age >36wks and birth weight >1800g
- History of perinatal period of hypoxia/Apgar score ≤5 at 10 minutes/positive pressure ventilation for 10 minutes
- Severe metabolic acidosis on cord gas or blood gas within 1hr of birth
- Evidence of moderate-severe HIE
  - Seizures
  - Evidence of poor tone/reflexes/consciousness level etc
  - Abnormal EEG

Intraventricular Haemorrhage

- Can occur spontaneously in premature neonates
- Majority occur in the 1st 72 hours
- Aetiology poorly understood, associated with birth trauma and cellular hypoxia
- May clot and cause hydrocephalus

Management

- Supportive
- Shunting if hydrocephalus causes ↑ ICP

Retinopathy of Prematurity

- Typically affects babies born before 32wks

Pathophysiology

- Normal hypoxia during gestation stimulates retinal blood vessel growth until 37-40wks
- This stimulation is removed by premature birth, especially with supplemental oxygen
- If hypoxia recurs excessive neovascularisation and scarring occurs, and can lead to retinal detachment

Assessment

Zones

1. Optic nerve and macula
2. Zone 1 to ora serrata (pigmented border of retina and ciliary body)
3. Outside ora serrata

- Location of disease described by clock face position

Stages

- **Stage 1:** slightly abnormal vessel growth
- **Stage 5:** complete retinal detachment

Plus Disease

- Additional findings (tortuous vessels, vitreous haze)

Screening

- All babies under 32wks or 1.5kg
- Starts at:
  - 30-31wks gestational age if born <27wks
  - 4-5wks of age if born >27wks
- Examination by ophthalmologist every 2 weeks until vessels enter zone 3

Management

- Transpupillary laser photocoagulation to stop neovascularisation is first line
- Cryotherapy and anti-VEGF injections are alternative options
- Retinal detachment requires surgery

Respiratory Distress Syndrome

- AKA Surfactant Deficient Lung Disease/Hyaline Membrane Disease
- Associated with prematurity
  - 50% born at 26-28wks
  - 25% born at 30-31wks
- Insufficient surfactant production by type 2 pneumocytes and immature lungs leading to distal airway collapse

Other Risk Factors

- Male sex
- Diabetic mothers
- Caesarean section
- Second born of premature twins

Features

- Tachypnoea
- Intercostal recessions
- Expiratory grunting
- Cyanosis

Chest X-Ray

- Ground glass appearance with indistinct heart border
- Usually bilateral & symmetrical
- Air bronchograms
- Exclude RDS if there is a normal CXR at 6 hours post birth

Management

- Antenatal steroids for mothers with expected preterm delivery
- Supplementary oxygen
- CPAP
- Intubation
  - Allows for endotracheal surfactant
- Respiratory support stepped down as lungs mature

Complications

Short-Term

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

Long-Term

- Chronic lung disease of prematurity
- Retinopathy of prematurity
- Neurological/visual/hearing impairment

Transient Tachypnoea of the Newborn

- Commonest cause of respiratory distress in the neonate
- Caused by delayed resorption of fluid in the lungs
- More common following Caesarean sections, particularly elective (due to less release of cortisol)

Chest X-Ray

- Lung hyperinflation
- Fluid in the horizontal fissure

Management

- Supplementary oxygen may be needed
- Usually resolves in 1-2 days

Apnoea of Prematurity

- Periods where breathing stops spontaneously for 20 seconds/in association with desaturation and bradycardia
- Very common in premature neonates
  - Almost all <28wks
  - Usually indicates underlying pathology in term babies

Causes

- Usually due to immature autonomic nervous system
- Can be a sign of developing illness:
  - Infection
  - Anaemia
  - Airway obstruction
  - CNS pathology
  - Gastro-oesophageal reflux
  - Neonatal abstinence syndrome

Management

- Apnoea monitors in neonatal units
- Tactile stimulation to prompt breathing
- IV caffeine can prevent recurrent apnoea and bradycardia

Meconium Aspiration Syndrome

- Most common cause of respiratory distress in the term/post-dates neonates

Neonatal Sepsis

- Sepsis in the first 28 days of life
- **Early (EOS):** First 72 hours
- **Late (LOS):** Later than 72 hours

Organisms

- Group B Strep and E. coli are the most common

EOS

- GBS (75%)

LOS

- Coagulase -ve Staphylococci & Gram -ves

Other causes

- Staph aureus
- Enterococcus
- Listeria
- Viruses (herpes simplex, enterovirus)

Risk Factors

- Maternal GBS colonisation
- Previous baby with GBS infection
- Maternal sepsis/chorioamnionitis/fever >38°
- Premature/prolonged rupture of membranes
- Prematurity
- Low birth weight

Features

- Fever
- Reduced tone/activity
- Poor feeding
- Respiratory distress/apnoea
- Vomiting
- Tachy/bradycardia
- Hypoxia
- Jaundice within 24hrs
- Seizures
- Hypoglycaemia

Red Flags

- Confirmed/suspected sepsis in the mother
- Signs of shock
- Seizures
- Term baby needing mechanical ventilation
- Respiratory distress starting after 4hrs
- Presumed sepsis in other baby in multiple pregnancy

Management

- Give antibiotics if ≥2 risk factors/clinical features or ≥1 red flag
  - Benzylpenicillin and gentamycin
- Blood cultures before antibiotics
- Baseline FBC & CRP, LP if meningitic features
- Recheck CRP at 24hrs, cultures at 36hrs, CRP at 5d
  - LP if any CRP >10
  - Consider stopping antibiotics when clinically well and CRP/cultures/LP are negative

Neonatal Hypoglycaemia

- <2.6mmol/L
- Transient hypoglycaemia in the first few hours is common

Causes of Persistent Hypoglycaemia

- Preterm birth
- Maternal DM
- IUGR
- Hypothermia
- Neonatal sepsis
- Inborn errors of metabolism
- Nesidioblastosis
- Beckwith-Wiedemann syndrome

Features

- May be asymptomatic

Autonomic Symptoms

- "Jitteriness"/irritability
- Tachypnoea
- Pallor

Neuroglycopenic Symptoms

- Poor feeding/sucking
- Weak cry
- Drowsiness
- Hypotonia
- Seizures

Other Symptoms

- Apnoea
- Hypothermia

Management

Asymptomatic/Mild

- Encourage normal feeding (breast/bottle)
- Monitor blood glucose

Symptomatic/Severe

- Admit to the neonatal unit
- 10% dextrose IV infusion

Neonatal Jaundice

Physiological Jaundice

- Normal from days 2-14
- Due to fast breakdown of fetal red blood cells and slower processing of bilirubin by the immature liver

Pathological Jaundice

- Jaundice before 24hrs
- Jaundice after 14 days in term babies/21 days in preterm babies
- If severe can lead to **kernicterus:**
  - Excess bilirubin crosses the blood-brain-barrier and causes direct damage to the CNS
  - Floppy/drowsy baby/poor feeding
  - Results in cerebral palsy, learning difficulty and deafness

Increased Bilirubin Production

- Sepsis/DIC
- Haemolytic disease of the newborn
- ABO incompatibility
- Haemorrhage/intraventricular haemorrhage/cephalhaematoma
- Polycythaemia
- G6PD deficiency

Decreased Bilirubin Clearance

- Prematurity
- Breast milk jaundice
- Neonatal cholestasis
- Extrahepatic biliary atresia
- Endocrine disorders
- Gilbert syndrome

Investigations

- Conjugated & unconjugated bilirubin
- FBC & blood film
- Blood typing of baby & mother
- Direct Coombs test
- TFTs, U+Es, LFTs
- Urine MCS & blood cultures
- G6PD levels
- Exclude galactosaemia if baby is very unwell

Management

- Plot age of baby and total bilirubin on gestational age specific treatment threshold chart

Phototherapy

- Usually sufficient to control jaundice
- Blue light converts unconjugated bilirubin in the skin to photoisomers which can be excreted in bile
- Rebound bilirubin should be measured after 12-18hrs if coombs positive
- Skin may blister in response to phototherapy in porphyria

Exchange Transfusion

- Removal of blood and replacement from a donor
- Rarely needed, most severe cases only

Necrotising Enterocolitis

- Affects premature neonates, especially with REDF
- Part of bowel becomes necrotic which can lead to perforation, peritonitis, shock and death
- Cause unclear

Risk Factors

- Very low birth weight/very premature baby/REDF
- Formula feeds (less common in breastfed babies)
- Respiratory distress/assisted ventilation
- Sepsis
- Patent ductus arteriosus/other congenital heart disease

Presentation

- Intolerance to feeds
- Blood in stool
- Vomiting (particularly green bile)
- Distended/tender/discoloured abdomen
- Absent bowel sounds
- Peritonitis and shock if perforated

Investigations

Bloods

- FBC & CRP
- Blood gas (metabolic acidosis)
- Blood culture

Imaging

- Abdominal x-ray (supine ± lateral/lateral decubitus)
  - Dilated bowel loops (often asymmetrical)
  - Bowel wall oedema
  - Pneumatosis intestinalis (intramural gas)
  - Pneumoperitoneum
  - Portal venous gas
  - Air both inside and outside the bowel wall (Rigler sign)
  - Air outlining falciform ligament (football sign)

Management

- Immediate surgical referral
- Graded by Bell's criteria
- Some recover with medical treatment alone
  - Nil by mouth
  - NG tube drainage
  - IV fluids, TPN, IV antibiotics
- Some need surgical removal of affected bowel
  - May be left with temporary stoma

Complications

- Peritonitis, sepsis, death
- Strictures
- Abscess formation
- Recurrence
- Long term stoma/short bowel syndrome (surgery)

Neonatal Cyanosis

- Peripheral cyanosis is common in first 24hrs, can occur due to crying or illness of any type
- Central cyanosis is recognised when concentration of deoxygenated haemoglobin exceeds 5g/dl

Causes

Airway

- Choanal atresia
- Micrognathia/retrognathia
- Pierre Robin sequence
  - Underdeveloped jaw, posterior tongue displacement
- Laryngomalacia
- Vocal cord paralysis
- External/intrinsic tracheal compression
  - Tracheal stenosis/complete tracheal rings
  - Mediastinal mass/ vascular slings

Breathing (lungs)

- Pneumonia
- RDS
- Congenital diaphragmatic hernia
- Pulmonary hypoplasia
- Phrenic nerve palsy
- Hypoventilation

Circulation

- Polycythaemia
- Anaemia
- Congenital heart disease

Nitrogen Washout Test

- AKA hyperoxia test
- ABG taken after 10 minutes of 100% oxygen
- pO2 of <15kPa indicates cyanotic congenital heart disease

Cyanotic Congenital Heart Disease

- Tetralogy of Fallot
  - Most common, but typically presents at 1-2 months
- Transposition of the Great Arteries
  - Most commonly diagnosed at birth
- Tricuspid Atresia

Initial Management

- Supportive care
- Prostaglandin E1 to maintain PDA in duct-dependent cyanotic disease

Acrocyanosis

- Benign peripheral cyanosis around mouth & extremities
- Peripheral vasoconstriction
- Differentiated from pathological causes of cyanosis as it occurs immediately after birth
- Persists for 24-48hrs

Neonatal Hypotonia

Acute Illness

- Neonatal sepsis

Associated with Encephalopathy

- HIE

Central Causes

- Down's syndrome
- Prader-Willi syndrome
- Hypothyroidism
- Cerebral palsy (hypotonia may precede spasticity)

Neurological/Muscular Causes

- Spinal muscular atrophy
- Spina bifida

Maternal Causes

- Drugs
  - Benzodiazepines
- Myasthenia Gravis

Classical Galactosaemia

- AR deficiency of galactose-1-phosphate uridyl transferase
- Inability to breakdown down galactose (component of lactose)
  - Leads to liver toxicity
  - Jaundice & coagulopathy
- Much higher incidence in Traveller populations

Screening

- Bloodspot for non-Traveller neonates
- Beutler test for Traveller neonates
  - Day 1 of life
  - Galactose-free (soy-based) feeds only until test result is available

Management

- Lactose free diet

Conditions Arising in Pregnancy

Fetal Alcohol Syndrome

- Effects of alcohol greatest in 1st trimester
- Alcohol can lead to miscarriage, preterm delivery, and small for dates

Features

- Microcephly
- Thin upper lip & smooth philtrum
- Short palpebral fissure
- Learning disability
- Behavioural difficulties
- Hearing & vision problems
- Cerebral palsy

Congenital Rubella Syndrome

- Greatest risk during 1st trimester
- MMR cannot be given during pregnancy as it is a live vaccine, non-immune mothers can be immunised after delivery

Features

- Congenital cataracts
- Congenital heart disease (PDA, pulmonic stenosis)
- Learning disability
- Hearing loss

Congenital Cytomegalovirus

- Most cases of CMV in pregnancy do not cause congenital features

Features

- Fetal growth restriction
- Microcephaly
- Hearing loss
- Vision loss
- Learning disability
- Seizures

Congenital Toxoplasmosis

- Toxoplasma gondii transmitted from cat faeces
- Usually asympomatic
- Greater risk of congenital features later in pregnancy

Features

- Intracranial calcification
- Hydrocephalus
- Chorioretinitis

Congenital Varicella Syndrome

- Occurs in 1% of cases of chickenpox exposure in pregnancy
- Infection in 1st 28wks

Exposure to Chickenpox

- If mother has previously had chickenpox they are safe
- If unsure about levels, check VZV IgG
  - If positive, they are safe
  - If negative, VZV IVIg should be given within 10 days of exposure
- If mother develops chickenpox rash, they should be treated with oral acyclovir within 24hrs if more than 20wks gestation

Features

- Growth restriction
- Microcephaly
- Hydrocephalus
- Learning disability
- Scars and skin changes along dermatome distribution
- Limb hypoplasia
- Cataracts & chorioretinitis

Congenital Zika Syndrome

- Spread by Aedes mosquito or by sex with an infected individual
- No symptoms or mild flu-like illness in infected adults
- Pregnant women who may have been in contact should be tested viral PCR and Zika antibodies
  - Referral to fetal medicine if positive
  - No treatment

Features

- Microcephaly
- Fetal growth restriction
- Ventriculomegaly
- Cerebellar atrophy

Neonatal Abstinence Syndrome

- Withdrawal from substances used/abused by the mother

Causes

- Opiates
- Methadone
- Benzodiazepines
- Cocaine
- Amphetamines
- Nicotine
- Cannabis
- Alcohol
- SSRIs

Features

CNS

- Irritability
- Increased tone
- High-pitched cry
- Not settling
- Tremors
- Seizures

Vasomotor & Respiratory

- Yawning
- Sweating
- Unstable temperature/pyrexia
- Tachypnoea

Metabolic & GI

- Poor feeding
- Regurgitation/vomiting
- Hypoglycaemia
- Loose stools & sore nappy area

Management

Monitoring

- Kept in hospital with NAS monitoring chart for 3 days (48hrs for SSRIs) if NAS expected
- Dim room with gentle handling
- Urine can be tested for substances

Medical Management (moderate-severe symptoms)

- Oral morphine sulphate for opiate withdrawal
- Oral phenobarbitone for non-opiate withdrawal
- SSRI withdrawal does not require treatment

Additional Considerations

- Test for hepatitis B/C and HIV
- Safeguarding & social work involvement
- Follow-up from paediatrics, social work, & GP
- Assistance for mother to stop using substances
- Check suitability of breastfeeding

Gastroenterology

Vomiting

Patterns

Acute

- Discrete episode of high intensity
- Commonest, usually associated with acute illness

Chronic

- Low-grade daily pattern

Cyclic

- Severe discrete episodes associated with pallor & lethargy ± abdominal pain
- Well between episodes

Causes

Acute

- GI infection
  - Gastroenteritis – most common
- Non-GI infection
- GI obstruction
  - Congenital (eg pyloric stenosis, malrotation)
  - Acquired
- Adverse food reaction
- Poisoning
- Raised ICP
- Metabolic/endocrine cause (eg DKA)
- Surgical cause (especially if bilious)

Chronic

- PUD, GORD, gastritis
- Chronic infection
- Gastroparesis
- Food allergy
- Psychogenic
- Bulimia nervosa

Cyclic

- Idiopathic
- CNS disease
- Abdominal migraine
- Endocrine (eg Addison's)
- Metabolic (eg Acute Intermittent Porphyria)

Investigations (if severe/chronic)

- FBC
- ESR/CRP if chronic
- U+E, glucose, creatinine
- Stool for culture & virology
- Imaging as appropriate/indicated

Management

- Manage dehydration as needed
- Manage underlying causes if necessary (eg pyloric stenosis)

Diarrhoea

Causes

- Gastroenteritis – most common
- HUS
- Non-enteric infection
- Food hypersensitivity
- Drugs (eg antibiotics)
- HSP
- Intussusception
- Pseudomembranous colitis

Assessing Dehydration

Clinical dehydration	Clinical shock
Appears to be unwell or deteriorating	Decreased level of consciousness
Decreased urine output	
Skin colour unchanged	Pale or mottled skin
Warm extremities	Cold extremities
Altered responsiveness (for example, irritable, lethargic)	
Sunken eyes	
Dry mucous membranes	
Tachycardia	Tachycardia
Tachypnoea	Tachypnoea
Normal peripheral pulses	Weak peripheral pulses
Normal capillary refill time	Prolonged capillary refill time
Reduced skin turgor	
Normal blood pressure	Hypotension

Investigations

- As per vomiting
- Stool culture indicated if:
  - Sepsis suspected
  - Blood/mucus in stool
  - Immunocompromised child
  - Child recently been abroad
  - Not resolved by day 10
  - Uncertain about diagnosis of gastroenteritis

Management

No Dehydration

- Continue breastfeeding/other milk feeds
- Encourage fluid intake
- Discourage fruit juices & carbonated drink

Clinical Dehydration

- 50ml/kg Oral Rehydration Solution over 4hrs + maintenance
- Continue breastfeeding

Clinical Shock

- Admission for IV rehydration

Acute Gastroenteritis Causes

Viral Gastroenteritis

- Most common
- Highly contagious
- Rotavirus/Norovirus
- Adenovirus less common and presents subacutely

E. Coli

- Normal intestinal bacteria, only certain strains cause gastroenteritis
- Spread through contact with faeces, unwashed salads or contaminated water
- Common amongst travellers
- Watery diarrhoea, cramps, and vomiting

E. coli O157:H7

- Produces Shiga toxin and causes bloody diarrhoea
- Causes HUS
  - Antibiotics increase risk of HUS and should not be used

Campylobacter Jejuni

- Gram -ve bacteria, cause of traveller's diarrhoea
- Most common bacterial gastroenteritis worldwide
- 2-5 day incubation, resolution after 3-6 days
- **Spread:**
  - Raw/improperly cooked poultry
  - Untreated water
  - Unpasteurised milk
- **Features:**
  - Fever
  - Abdominal cramps
  - Vomiting
  - Diarrhoea ± blood
- May mimic appendicitis
- Can cause Guillan-Barre syndrome
- Antibiotics may be considered if C. jejuni is isolated and patients have severe symptoms/risk factors such as HIV
  - Azithromycin/ciprofloxacin

Shigella

- Bloody diarrhoea, abdominal cramps, and fever
- Shiga toxin - can cause HUS
- Severe cases treated with azithromycin/cipro

Bacillus Cereus

- Gram +ve rod
- Grows well on food not immediately refrigerated after cooking
  - Typically rice left out after cooking
- **Features:**
  - Abdominal cramping and vomiting within 5 hours and/or
  - Watery diarrhoea within 8 hours of ingestion
  - Resolution after 24hrs

Salmonella

- **Spread:**
  - Raw eggs/pulty
  - Foods contaminated with small animal faeces
- **Features:**
  - 12hrs-3 days incubation, resolution after 1 week
  - Watery diarrhoea ± blood/mucous
  - Abdominal pain
  - Vomiting
- Antibiotics in severe cases as guided by stool culture & sensitivity

Yersinia Enterocolitica

- Gram -ve bacillus
- **Spread:**
  - Raw/undercooks pork
  - Contamination with cat/rabbit faeces/urine
- **Features:**
  - Incubation 4-7 days, resolution after ≤3wks
  - Watery/bloody diarrhoea
  - Adbo pain, fever, lymphadenopathy
  - Mesenteric lymphadenitis in older children/adults
- Antibiotics in severe cases as guided by stool culture & sensitivity

Giardiasis

- Intestinal parasite
- **Spread:**
  - Cysts in stools of infected mammals can contaminate food/water
- **Features:**
  - Asymptomatic
  - Chronic, malabsorption-like diarrhoea
- Diagnosis by stool microscopy
- Treatment with metronidazole

Staph Aureus Toxin

- Causes by enterotoxin, not bacteria
- Produced by bacteria growing on eggs/dairy/meat
- Diarrhoea, vomiting, cramps, fever
- Usually settles within 12-24hrs

Acute Gastroenteritis Assessment & Management

- Principle of management is isolation and treatment of dehydration as appropriate

History

- Sudden onset vomiting & diarrhoea
- Abdominal pain
- Fever
- Assess dehydration risk
- Sick contacts
- Known infection source
- Recent travel

Examination

- Weight
- Vital signs
- Abdominal exam

Hydration status

- **Mild Dehydration (<5%):**
  - Alert
  - CRT <2 seconds
  - Moist mucous membranes
  - Skin turgor normal
  - Decreased urine output
  - Mildly increased HR
  - Normal BP & pulse pressure
- **Moderate Dehydration (6-10%):**
  - Lethargic
  - CRT 2-4 seconds
  - Dry mucous membranes
  - Slow skin turgor
  - Depressed fontanelle (infants)
  - Sunken eyes
  - Oliguria
  - Moderately increased HR
  - Thready pulse
  - Normal or low BP
- **Severe Dehydration (>10%):**
  - Obtunded
  - CRT >4 seconds
  - Parched mucous mebranes
  - Tenting skin turgor
  - Sunken fontanelle (infants)
  - Very sunken eyes
  - Anuria
  - Severely decreased HR
  - Faint pulse
  - Low BP

Investigations

- Often not needed
- Stool culture indicated if:
  - Sepsis suspected
  - Blood/mucus in stool
  - Immunocompromised child
  - Child recently been abroad
  - Not resolved by day 10
  - Uncertain about diagnosis of gastroenteritis

Rehydration

Oral (Mild Dehydration)

- ORS given by spoon every 15 minutes over 3-4 hours

NG/IV

- Indicated by:
  - Moderate-severe dehydration
  - Persistent vomiting with inability to tolerate oral fluids

Hospital Admission

- Need for NG/IV fluids
- Severe abdominal pain/tenderness
- Bilious vomiting

Post-Gastroenteritis Complications

- Lactose intolerance
- Irritable bowel syndrome
- Reactive arthritis
- Guillain-Barré Syndrome (Campylobacter)
- Haemolytic Uraemic Syndrome (E. coli O157:H7 or Shigella)

Abdominal Pain

Causes

Medical

- Constipation
- UTI
- Coeliac disease
- IBS
- IBD
- **Mesenteric adenitis**
  - Features similar to appendicitis
  - Follows viral infection
  - Needs no treatment
- Abdominal migraine
- Pyelonephritis
- HSP
- DKA
- Infantile

Gynaecological

- Dysmenorrhoea
- Mittelschmerz
- Ectopic pregnancy
- PID
- Ovarian torsion
- Pregnancy

Surgical

- Appendicitis
- Intussusception
  - Colicky pain with redcurrant jelly stools
- Bowel obstruction
- Testicular torsion

Red flags for serious underlying pathology

- Persistent/bilious vomiting
- Severe chronic diarrhoea
- Fever
- Rectal bleeding
- Weight loss/faltering growth
- Dysphagia
- Nighttime pain
- Abdominal tenderness

Investigations for serious underlying pathology

- Anaemia can indicate IBD/coeliac
- ESR/CRP can indicate IBD
- Anti-TTG/anti-EMA indicate coeliac
- Raised faecal calprotectin indicates IBD
- Positive urine dipstick indicates UTI

Recurrent Abdominal Pain

- Non-organic/functional recurrent episodes
- Corresponds with stressful life events
- May be caused by inappropriate signals from visceral nerves in response to normal stimuli

Management

- Explanation & reassurance
- Distracting the child
- Encourage parents not to ask/focus on pain
- Probiotics may help
- Avoid NSAIDs
- Address psychosocial factors & triggers
- Support from school counsellor/child psychologist

Abdominal Migraine

- May occur before development of traditional migraines
- Central abdominal pain lasting more than 1 hour ± associated:
  - Nausea and vomiting
  - Anorexia
  - Pallor
  - Headache
  - Photophobia
  - Aura
- Normal examination

Treating Acute Attack

- Low stimulus environment
- Paracetamol
- Ibuprofen
- Sumatriptan

Prevention

- Pizotifen
  - Serotonin agonist
  - First line
  - Has to be withdrawn slowly
- Propranolol
  - Non-selective beta-blocker
- Cyproheptadine
  - Antihistamine
- Flunarazine
  - Calcium channel blocker

Constipation

- Most cases are idiopathic constipation/functional obstruction
- Associated with **lifestyle factors:**
  - Habitually not opening bowels
  - Low fibre diet
  - Poor fluid intake/dehydration
  - Sedentary lifestyle
  - Psychosocial problems

Features

- Less than 3 stools per week
- Hard stools that are difficult to pass
- Rabbit dropping stools
- Straining/painful passage of stools
- Abdominal pain
- Abnormal posture (retentive posturing)
- Rectal bleeding associated with hard stools
- Overflow soiling
- Hard stools palpable in abdomen
- Loss of sensation of need to open bowels

Encopresis

- Faecal incontinence
- Pathological after 4yrs
- Usually a result of chronic constipation/overflow soiling

Other causes (rare)

- Spina bifida
- Hirschsprung's disease
- Cerebral palsy
- Learning disability
- Psychosocial stress

Secondary Causes of Constipation

- Hirschsprung's disease
- Cystic fibrosis (meconium ileus)
- Coeliac disease
- Hypothyroidism
- Spinal cord lesions
- Sexual abuse
- Intestinal obstruction
- Anal stenosis
- Cow's milk intolerance

Red Flags for Secondary Causes

- Meconium ileus (CF/Hirschsprung's)
- Neurological signs
- Vomiting (Hirschsprung's)
- Ribbon stool (anal stenosis)
- Abnormal anus (anal stenosis, IBD, sexual abuse)
- Abnormal lower back/buttocks (spina bifida, spinal cord lesion, sacral agenesis)
- Failure to thrive (coeliac disease, hypothyroidism)
- Acute severe abdominal pain/bloating (obstruction/intussusception)

Complications

- Pain
- Desensitisation, impaction, overflow soiling
- Anal fissures
- Haemorrhoids
- Psychosocial morbidity

Management

- Diagnosis of functional constipation can be made without investigations in the absence of red flags

Conservative

- Increased fluid intake
- Bowel retraining (PooPassport)

Faecal Impaction

- Movicol Paediatric Plan with escalating dose regime first line
- Stimulant laxative added second line
- Stimulant laxative alone or in combination with osmotic laxative if Movicol is not tolerated
- Soiling and pain may increase initially

Maintenance

- As above with reduced starting doses
- Encourage and praise toilet visits
  - Scheduled visits
  - Toilet diary
  - Star charts
- Dose weaned as bowel habits become more regular

Gastro-Oesophageal Reflux

- Normal regurgitation occurs in up to 50% of infants due to immature lower oesophageal sphincter
- Not problematic if there is normal growth and child is well
- Resolves by 1 year in 90%

Problematic Features

- Chronic cough
- Hoarse cry
- Reluctance to feed
- Distress/crying/unsettled after feeding
- Pneumonia
- Poor weight gain
- Frequent otitis media
- Dental erosion

Management

Conservative

- Small, frequent meals
- Burping regularly
- Not over-feeding
- Keeping upright/30° head up

Problematic Cases

- Trial of feed thickeners (carobel) or thickened formula (Enfamil AR or SMA Staydown)
- Trial of alginates if breast fed
  - Gaviscon
  - Not at the same time as thickened formula
- PPI trial if there is ≥1 of:
  - Unexplained feeding difficulties (refusal/gagging/choking)
  - Distressed behaviour
  - Faltering growth
- **Ranitidine withdrawn due to presence of carcinogen N-nitrosodimethylamine (NDMA)**

Severe Cases

- Investigation with barium meal/endoscopy
- Surgical fundoplication (rarely needed)

Sandifer's Syndrome

- Abnormal movements associated with reflux
- **Torticollis:** Forceful contraction of neck muscles causing neck twisting
- **Dystonia:** Abnormal contractions causing twisting movements/back arching/abnormal postures
- Resolves as reflux improves but should be referred to exclude infantile spasms/seizures

Pyloric Stenosis

- Hypertrophy of circular muscles of pylorus causing outflow obstruction
- Powerful peristalsis against obstructed pylorus → projectile vomiting

Features

- 4-6wks
- Thin, pale, hungry baby failing to thrive
- Projectile vomiting ~30mins after feeds
- Constipation, dehydration
- Palpable, olive-shaped mass in upper abdomen
- Hypochloraemic hypokalaemic metabolic acidosis

Diagnosis

- Abdominal ultrasound

Management

- Ramstedt pyloromyotomy

Coeliac Disease

- Autoimmune condition in response to gluten exposure
- Autoantibodies target duodenum and jejunum causing villous atrophy, crypt hyperplasia and malabsorption

Autoantibodies

- Anti-tissue transglutaminase (anti-TTG)
- Anti-endomysial (anti-EMA)
- Anti-deaminated gliadin peptides (anti-DGP)
- Anti-TTG and anti-EMA are IgA
  - Total IgA levels must be checked as test will be negative in IgA deficiency, IgG can be requested alternatively

Features

- Failure to thrive
- Diarrhoea
- Fatigue
- Weight loss
- Abdominal pain/cramping/bloating
- Mouth ulcers
- Anaemia/B12/folate deficiency
- Dermatitis herpetiformis
- Neurological symptoms (rare)
  - Peripheral neuropathy
  - Cerebellar ataxia
  - Epilepsy

Associations

Genetic

- HLA-DQ2 (95%)
- HLA-DQ8 (80%)

Conditions (warrant testing for coeliac)

- Autoimmune thyroid disease
- Type 1 DM
- IBS

Diagnosis

- Investigations only while patient's diet includes gluten
- Total IgA levels and:
  - Anti-TTG
  - Anti-EMA
- Endoscopy and intestinal biopsy:
  - Crypt hyperplasia
  - Villous atrophy

Management

- Gluten free diet
- Offer pneumococcal & influenza vaccines (functional hyposplenism)

Biliary Atresia

Types

1. Proximal ducts patent, common duct obliterated
2. Atresia of cystic duct, cystic structures found in porta hepatis
3. Atresia of left and right ducts at level of porta hepatis (>90%)

Presentation

Symptoms

- Jaundice beyond physiological 14 days
- Dark urine, pale stools
- Appetite and growth disturbance

Signs

- Jaundice
- Hepatosplenomegaly
- Abnormal growth
- Cardiac murmurs of associated cardiac abnormalities

Management

- Surgery is definitive management
  - Kasai portoenterostomy
- Medical:
  - Antibiotic coverage and bile acid enhancers following surgery

Complications

- Unsuccessful anastomosis
- Liver transplantation
- Progressive liver disease
- Cirrhosis and HCC

Inflammatory Bowel Disease

Common Presentation

- Periods of remission and exacerbation
- Perfuse diarrhoea
- Abdominal pain
- Rectal bleeding
- Weight loss
- Anaemia
- Systemic upset during flares

Extraintestinal Manifestations

- Finger clubbing
- Erythema nodosum
- Pyoderma gangrenosum
- Episcleritis & uveitis
- Inflammatory arthritis

Differentiating Features

Crohn's Disease	Ulcerative Colitis
Blood & mucous less common	Blood & mucous common
Entire GI tract	Limited to rectum & colon
Skip lesions Transmural inflammation Goblet cells Granulomas	Continuous inflammation Superficial inflammation Pseudopolyps Crypt abscesses
Smoking is a risk factor	Smoking is protective
Peri-anal disease common Mouth ulcers Strictures Fistulas	Primary sclerosing cholangitis Leadpipe colon

Investigation

Bloods

- FBC, CRP, U+E, LFT, TFT
  - Raised CRP indicates active inflammation

Faecal Calprotectin

- Released by inflamed intestine
- 90% sensitive & specific

Endoscopy & biopsy

- Gold standard investigation

Imaging

- Barium enema/CT/MRI/US
- Identify complications
  - Strictures, fistulas, abscesses

General Management of IBD

- Specialist management
- MDT follow-up
- Monitor growth and pubertal development, especially if being treated with steroids

Management of Crohn's Disease

Inducing Remission

1. Steroids 1st line
   - Oral prednisolone
   - IV hydrocortisone
2. Immunosuppression may be added (specialist guidance)
   - Azathioprine
   - Mercaptopurine
   - Methotrexate
   - Infliximab
   - Adalimumab

Maintaining Remission

1. Azathioprine/mercaptopurine 1st line
2. Alternatives:
   - Methotrexate
   - Infliximab
   - Adalimumab

Surgery

- Resection of distal ileum possible if flares only affect this area
- Fistulas/strictures can be treated surgically

Management of Ulcerative Colitis

Inducing Remission

- **Mild-moderate disease**
  - Aminosalicylate (**rectal**/oral mesalazine) 1st line
  - Corticosteroids 2nd line
- **Severe Disease**
  - IV steroids 1st line
  - IV ciclosporin 2nd line

Maintaining Remission

1. Aminosalicylates (**rectal**/oral mesalazine)
2. Azathioprine
3. Mercaptopurine

Surgery

- Panproctocolectomy will remove disease
  - Leaves either a permanent ileostomy or ileo-anal anastomosis (J-pouch)

Intestinal Obstruction

Causes

- **Meconium ileus**
  - Delayed passage of meconium
  - Majority have CF
  - Fluid level on X-ray
  - PR contrast studies/N-acetyl-cysteine may dislodge meconium plug, otherwise surgery is required
- Hirschsprung's Disease
- **Oesophageal atresia**
  - Associated with trachea-oesophageal fistula and polyhydramnios
  - Choking and cyanosis following aspiration
  - Vomiting of undigested food
- **Duodenal atresia**
  - Associated with Down's syndrome
  - Double bubble sign on AXR
- Intussusception
- Imperforate anus
- Malrotation & volvulus
- Strangulated hernia

Presentation

- Persistent bilious vomiting
- Abdominal pain and distension
- Absolute constipation & failure to pass wind
- Abnormal bowel sounds
  - High-pitched/tinkling early
  - Absent late

Investigations

- AXR
  - Dilated bowel loops proximal to obstruction
  - Collapsed bowel distal to obstruction
  - Absence of air in rectum

Management

- Emergency paediatric surgical referral
- Nil by mouth & nasogastric drainage
- IV fluids
- Surgical management of underlying cause

Malrotation

- High caecum at midline
- Associated with exomphalos, congenital diaphragmatic hernia, intrinsic duodenal atresia
- Complicated by volvulus formation → obstruction

Investigations

- Upper GI endoscopy
- USS

Management

- Laparotomy if volvulus present
- Ladd's procedure if high risk of volvulus

Hirschsprung's Disease

Pathophysiology

- Failure of neural crest cells to migrate to distal colon & form parasympathetic Meissner's and Auerbach plexuses
- Uncoordinated peristalsis and functional obstruction, proximal dilatation
- Amount of colon affected varies
  - Whole colon: total colonic aganglionosis

Associations

- M:F 3:1
- Positive family history
- Down's syndrome
- Neurofibromatosis
- MEN2

Presentation

Neonatal

- Meconium ileus

Older children

- Chronic constipation since birth
- Abdominal pian & distension
- Vomiting
- Failure to thrive

Hirschsprung-Associated Enterocolitis (HAEC)

- Affects 20% of neonates with Hirschsprung's
- Presents at 2-4wks
  - Fever ± sepsis
  - Abdominal distension & diarrhoea ± blood
  - Can cause toxic megacolon/perforation
- Urgent antibiotics, fluid resuscitation and decompression

Investigation

- AXR demonstrates obstruction/HAEC features
- Rectal biopsy gold standard for diagnosis
  - Absence of ganglionic cells

Management

- Fluid resuscitation/management of obstruction in unwell children
- Treat HAEC
- Surgical removal of affected colon

Congenital Diaphragmatic Hernia

- 1/2,000 newborns
- Herniation of abdominal viscera into the chest cavity due to incomplete formation of the diaphragm
  - Failure of pleuroperitoneal cavity to close properly
- Results in pulmonary hypoplasia and hypertension → respiratory distress shortly after birth
- ~50% mortality

Intussusception

- Invagination/telescoping of section of bowel into the adjacent section
- Most common in ileo-caecal region
- Typically 6-18 months
- Boys > girls 2:1

Associations

- Concurrent viral illness
- HSP
- Intestinal polyps
- CF
- Meckel's diverticulum

Presentation

- Sudden severe colicky abdominal pain
  - Infant will characteristically draw knees up and become pale
- Vomiting/intestinal obstruction
- Redcurrant jelly stool
  - Late sign
- Sausage-shaped RUQ mass

Investigations

- Ultrasound is diagnostic
  - Target-shaped mass

Management

- Air insufflation under radiological control is first line
- Therapeutic barium enema was used previously
- Surgical reduction required if:
  - Insufflation fails
  - Signs of peritonitis/perforation

Complications

- Obstruction
- Gangrenous bowel
- Perforation
- Death

Appendicitis

- Inflammation of appendix due to infection trapped by obstruction
- Can lead to rupture and peritonitis
- Peak incidence 10-20yrs

Presentation

- Atypical presentations common, particularly with retrocaecal/pelvic appendix
- Central abdominal pain later localising to RIF
- Nausea & vomiting, anorexia
- Tenderness at McBurney's point
  - 1/3 from ASIS to umbilicus
- Rovsing's sign
  - Palpation of LIF causes RIF pain
- Guarding
- Rebound/percussion tenderness
  - Suggest peritonitis

Investigation

- Diagnosed clinically & based on inflammatory markers
- CT can confirm diagnosis if another cause is likely
- USS can rule out gynaecological causes
- Diagnostic laparoscopy visualises appendix directly
  - Directly precedes removal

Differentials

- Ectopic pregnancy
- Ovarian cysts
- Meckel's diverticulum pathology
- Mesenteric adenitis
- Appendix mass

Management

- Surgical removal
- Laparoscopy preferred to laparotomy

Appendicectomy Complications

- Bleeding, infection, pain, scars, VTE
- Damage to bowel/bladder/other organs
- Removal of a healthy appendix

Meckel's Diverticulum

- Congenital diverticulum of small intestine
- Remnant of omphalomesenteric (AKA vitellointestinal) duct
- Contains ectopic ileal/gastric/pancreatic mucosa

Rule of 2's

- 2% of population
- 2 inches long
- 2 feet from ileocaecal valve

Presentation

- Usually asymptomatic
- Abdominal pain mimicking appendicitis
- Rectal bleeding
  - Most common cause of massive GI bleed in children aged 1-2
- Obstruction
  - Omphalomesenteric band
  - Volvulus
  - Intussusception

Management

- Removal if narrow neck/symptoms
- Wedge excision/small bowel resection with anastomosis

Cardiology

Fetal Circulation

- Fetal blood needs to take on oxygen and nutrients and lose carbon dioxide and other waste (eg lactate) at the placenta
- Fetal blood does not need to enter the lungs

Shunts

- **Ductus Venosus** connects the umbilical vein to the IVC to allow blood from the placenta to bypass the liver
- **Foramen Ovale** connects the right atrium to the left atrium to allow blood to bypass the right ventricle and pulmonary circulation
- **Ductus Arteriosus** connects the pulmonary artery to the aorta to allow blood to bypass the pulmonary circulation

At Birth

- First breath and alveolar expansion cause a reduction in pulmonary vascular resistance, decreasing the pressure in the pulmonary circulation and right atrium
  - Left atrial pressure, now greater than the right atrium, causes closure of the foramen ovale
  - Closed foramen ovale seals and becomes fossa ovale after a few weeks
- Increased blood oxygenation cause a drop in prostaglandin levels, which are needed to keep the ductus arteriosus open
  - Closure of ductus arteriosus, which becomes ligamentum arteriosum
- Ductus venosus stops functioning after clamping of the umbilical cord due to lack of umbilical venous return
  - Closes after a few days and becomes ligamentum venosum

Murmurs

Innocent/Flow/Still's Murmur

Features

- Soft
- Short
- Systolic
- Symptomless
- Situation dependent (varies with posture/illness)
- Site (LLSE)

Features Requiring Investigation/Referral

- Louder than 2/6
- Diastolic murmur
- Louder on standing
- Other symptoms (FTT, cyanosis, SOB, feeding issues)

Investigations

- ECG
- CXR
- Echo

Pansystolic Murmur

- Mitral regurgitation heard in mitral area
- Tricuspid regurgitation heard in tricuspid area
- VSD heard at LLSE

Systolic Ejection Murmur

- Aortic stenosis heard in aortic area
- Pulmonic stenosis heard in pulmonic area
- HOCM heard at 4th intercostal space on LSE

Atrial Septal Defect

- Mid-systolic crescendo-decrescendo murmur
- Loudest at ULSE
- Associated with fixed splitting of S2

Patent Ductus Arteriosus

- Can be silent if small
- Continuous machinery-like murmur

Tetralogy of Fallot

- Pulmonic stenosis

Patent Ductus Arteriosus

- Failure of closure of ductus arteriosus
- Acyanotic congenital cardiac defect
  - Uncorrected can cause late cyanosis in lower extremities (**differential cyanosis**)
- Occasionally asymptomatic in childhood and presents in adults with heart failure
- Left to right shunt can cause pulmonary hypertension, RVH and resultant LVH

Risk Factors

- Prematurity
- Birth at high altitude
- Maternal rubella infection in 1st trimester

Presentation

- Murmur heard during newborn exam
  - Continuous machinery-like murmur
- Shortness of breath
- Difficulty feeding
- Poor weight gain
- LRTIs

Diagnosis

- Echocardiogram confirms PDA and assesses extend of L-R shunt and effects of PDA on the ventricles

Management

- Indomethacin/ibuprofen IV given to symptomatic neonate
- If picked up later asymptomatic children are monitored until 1 year
  - Highly unlikely to close spontaneously after this
- Trans-catheter or surgical closure if symptomatic/after 1 year

Atrial Septal Defect

- Most common congenital cardiac defect found in adulthood
  - 50% mortality at 50yrs if not found/treated earlier

Types

Ostium Secundum (70%)

- Septum secundum fails to fully close
- **ECG**: RBBB with RAD

Ostium Primum

- Septum primum fails to fully close
- Associated with atrioventricular valve defects/atrioventricular septal defects
- **ECG**: RBBB with LAD & PR interval prolongation

Presentation

- May be found on antenatal scans/neonatal exams
- Mid-systolic crescendo-decrescendo murmur
  - Loudest at ULSE
  - Associated with fixed splitting of S2

Symptoms in Childhood

- Shortness of breath
- Difficulty feeding
- Poor weight gain
- LRTIs

Symptoms in Adulthood

- Dyspnoea
- Heart failure
- Stroke

Complications

- Paradoxical emboli – stroke resulting from DVT
- Atrial fibrillation/flutter
- Pulmonary hypertension
- RHF
- Eisenmenger's syndrome

Management

- Referral to paediatric cardiologist
- Watchful waiting appropriate if small & asymptomatic
- Closure
  - Transvenous catheter closure
  - Open surgery
- Antiplatelets/anticoagulants can reduce stroke risk in adults

Ventricular Septal Defect

- Most common cause of congenital heart disease
- Close spontaneously in 50%

Aetiology

- Chromosomal disorders
  - Down's
  - Edward's
  - Patau
  - Cri-du-chat
- Congenital infections
- Acquired (post-MI)

Presentation

- May be found on antenatal scans/neonatal exams
- Pan-systolic murmur
  - Lower left sternal edge at 3rd & 4th intercostal spaces
  - Associated systolic thrill
  - Louder in smaller defects
- Poor feeding
- Failure to thrive
- Hepatomegaly
- Tachypnoea
- Tachycardia
- Pallor

Complications

- Aortic regurgitation
  - Due to poorly supported right coronary cusp
- Infective endocarditis
  - Antibiotic prophylaxis should be considered during surgical procedures
- Pulmonary hypertension
- RHF
- Eisenmenger's syndrome

Management

- Led by paediatric cardiologist

Small & Asymptomatic

- Watchful waiting, may close spontaneously

Moderate-Large/Symptomatic

- Nutritional support
- Diuretics for HF symptoms
- Closure
  - Transvenous catheter closure
  - Open surgery

Eisenmenger Syndrome

- Reversal of left to right shunt in acyanotic cardiac defects
- Prolonged L-R shunt causes pulmonary hypertension and right ventricular hypertrophy, which can reverse flow direction in the shunt
- Causes cyanosis from an originally acyanotic defect & polycythaemia
- Develops:
  - After 1-2 years with large defects
  - In adulthood with small defects
  - Accelerated in pregnancy – women with defects need close monitoring

Causes

- Patent Ductus Arteriosus
- Atrial Septal Defect
- Ventricular Septal Defect

Presentation

- Potential disappearance of original murmur

Features of Pulmonary Hypertension

- Right ventricular heave
- Loud P2
- Raised JVP
- Peripheral oedema

Features of R-L Shunt & Hypoxia

- Cyanosis
- Clubbing
- Dyspnoea
- Plethoric complexion (due to polycythaemia)

Prognosis

- Life expectancy reduced by ~20 years
- Mortality up to 50% in pregnancy
- Irreversible once the syndrome develops

Management

Medical

- Oxygen helps symptoms, not outcomes
- Treatment of PHT (sildenafil)
- Treatment of arrhythmias
- Treatment of polycythaemia (venesection)
- Thromboprophylaxis
- Endocarditis prophylaxis

Surgical

- Heart-lung transplant is the only definitive treatment

Coarctation of the Aorta

- Congenital narrowing of the aortic arch/descending aorta, usually around the level of the ductus arteriosus
- Reduced arterial pressure distal to narrowing and increased pressure proximal to narrowing

Associations

- Turner syndrome, yet still more common overall in boys
- Bicuspid aortic valve
- Berry aneurysms
- Neurofibromatosis

Presentation

Infancy

- Weak/absent femoral pulses on neonate exam
- Systolic murmur below left clavicle and below left scapula
- Tachypnoea/increased work of breathing
- Poor feeding
- Grey/floppy baby

Later

- Radio-femoral delay
- Left ventricular heave
- Underdevelopment of legs ± left arm
- Inferior notching of ribs on CXR
- Four-limb blood pressure:
  - Increased BP before narrowing
  - Decreased BP after narrowing

Management

- Mild cases may not need surgery until adulthood
- Severe cases need surgery shortly after birth
  - Prostaglandin E2 given to maintain ductus arteriosus until surgery
  - Surgical correction of the coarctation and ligation of ductus arteriosus

Congenital Aortic Stenosis

Features

Symptoms

- May be asymptomatic
- Features will typically be worse on exertion:
  - Fatigue, dyspnoea, dizziness/fainting
- Severe cases will cause heart failure within months of birth

Signs

- Ejection systolic murmur
  - Loudest in aortic area
  - Crescendo-decrescendo
  - Radiates to carotids
  - ± ejection click immediately before murmur
- Palpable systolic thrill
- Slow rising pulse, narrow pulse pressure

Investigations

- Echocardiogram is gold standard
- Follow up with echocardiogram, ECG, & exercise tests

Complications

- Left ventricular outflow tract obstruction
- Heart failure
- Ventricular arrhythmia
- Bacterial endocarditis
- Sudden death (especially on exertion)

Management

1. Percutaneous balloon aortic valvuloplasty
2. Surgical aortic valvotomy
3. Valve replacement

Congenital Pulmonic Stenosis

Associations

- Tetralogy of Fallot
- William syndrome
- Noonan syndrome
- Congenital rubella syndrome

Features

- May be asymptomatic
- Features will typically be worse on exertion:
  - Fatigue, dyspnoea, dizziness/fainting
- Ejection systolic murmur in pulmonic area
- Thrill in pulmonic area
- Right ventricular heave
- Raised JVP

Management

- Echocardiogram is gold standard for diagnosis

1. Percutaneous balloon valvuloplasty
2. Surgical valve replacement

Tetralogy of Fallot

- Most common cyanotic congenital cardiac defect
- Normally diagnosed after 1-2 months
- Caused by anterior malalignment of aorticopulmonary septum resulting in:

  - Ventricular septal defect
  - Right ventricular outflow tract obstruction/pulmonic stenosis
  - Right ventricular hypertrophy
  - Overriding aorta
- Results in blood being shunted from the right to the left ventricle & into the aorta
- Degree of RV outflow tract obstruction determines severity

Associations
- Rubella
- Alcohol consumption in pregnancy
- Older age of mother
- Diabetic mother

Presentation
- May be found on antenatal scans/neonatal exams
- Severe cases: heart failure before 1 year
- Pulmonic stenosis murmur
- Cyanosis, clubbing
- FFT, feeding issues
- Tet spells

Investigations
- Echo with doppler flow studies is diagnostic
- CXR: boot-shaped heart due to RVH

Management
- Neonatal prostaglandin E2 to maintain PDA
- **Open surgical correction** (often in 2 parts)

Tet Spells
- Cyanotic episodes caused by ↑ PVR/↓ SVR
  - Eg due to increased CO_2 levels
- Precipitated by waking/activity/crying

Features
- Cyanosis
- Dyspnoea
- Seizures
- Reduced consciousness
- Death

Management
- Position – squat/knees up to chest
- Supplementary oxygen
- Beta-blockers (relax RV/infundibular spasm)
- IV fluids (increase preload and pulmonary flow)
- Morphine (reduces respiratory drive)
- Sodium bicarbonate (buffers metabolic acidosis)
- Phenylephrine infusion (increases SVR)

Transposition of the Great Arteries
- Most common cyanotic congenital cardiac defect *diagnosed at birth*
- Failure of aorticopulmonary septum to spiral during septation
  - Aorta arises from RV, pulmonary trunk arises from LV
- Two separate circulations which don't mix
- Increased risk in diabetic mothers
- Immediately life-threatening at birth and survival is dependent on one of:
  - PDA
  - VSD
  - ASD

Associations
- VSD
- Coarctation of the aorta
- Pulmonic stenosis

Presentation
- Often diagnosed on antenatal scans
  - Needs close monitoring and arrangement of appropriate care immediately after birth
- Cyanosis within a few days of birth
- Decompensation of PDA/VSD after a few weeks:
  - Respiratory distress
  - Tachycardia
  - Poor feeding/weight gain
  - Sweating
- Loud single S2
- Prominent right ventricular impulse
- "Egg-on-side" appearance on CXR

Management
- Prostaglandin E2 infusion to maintain PDA
- Balloon septostomy
  - Catheter insertion to foramen ovale via umbilicus
  - Dilation to create large ASD
- **Open surgical correction**
  - Bypass & arterial switch
  - Simultaneous correction of VSD/ASD

Ebstein's Anomaly

- Lower insertion of tricuspid valve in right side of heart
  - Large RA & small RV
  - "Atrialisation" of RV
- Can be caused by lithium exposure in utero

Associations

- PFO/ASD in 80% → R-L shunt & cyanosis
  - Symptoms appear/worsen after closure of ductus arteriosus
- Wolff-Parkinson-White syndrome

Presentation

- Cyanosis
- Evidence of heart failure
  - Oedema
  - Hepatomegaly
- Shortness of breath & tachypnoea
- Poor feeding
- Collapse/cardiac arrest
- Elevated JVP with prominent A wave
- Gallop rhythm

Investigations

Echo

- Diagnostic & assessment of severity

ECG

- Arrhythmias
- Right atrial enlargement
- RBBB
- LAD

CXR

- Cardiomegaly
- Right atrial enlargement

Management

- Treat arrhythmias/heart failure medically
- Infective endocarditis prophylaxis
- **Surgical correction**

Causes

Airway

- Choanal atresia
- Micrognathia/retrognathia
- Pierre Robin sequence
  - Underdeveloped jaw and posterior tongue displacement
- Larygnomalacia
- Vocal cord paralysis
- External/intrinsic tracheal compression
  - Tracheal stenosis/complete tracheal rings
  - Mediastinal mass
  - Vascular slings

Breathing (lungs)

- Pneumonia
- RDS
- Congenital diaphragmatic hernia
- Pulminary hypoplasia

ENT & Respiratory

Acute Otitis Media

- Bacterial infection often preceded by viral URTI, most common from 6 months to 3 years

Organisms

- Streptococcus pneumoniae
- Haemophilus influenzae
- Moraxella catarrhalis

Features

Symptoms

- Otalgia (children may rub/tug ears)
- Fever (50%)
- Hearing loss
- Ear discharge

Signs

- Bulging TM -> loss of light reflex
- Opacification/erythema of TM
- Perforation of TM with purulent discharge

Diagnosis Requires:

1. Acute onset
2. Presence of middle ear effusion
   - Bulging TM/otorrhoea/reduced mobility on pneumatic otoscopy
3. Inflammation of TM
   - Erythema

Management

- Paracetamol
- No role for decongestants
- Antibiotics should not be given routinely

Indications for antibiotics:

- Symptoms lasting 4 days/not improving
- Systemically unwell (not requiring admission)
- Perforated TM/discharge in canal
- Younger than 2 with bilateral AOM
- Immunocompromise/significant heart/lung/kidney/liver/neuromuscular disease
- **Antibiotics:** Amoxicillin 5-7/7

Referral to ENT

- >4 episodes in 6 months
- Any complication
- OME with speech development problems
- Surgical treatment is grommets ± adenoidectomy

Complications

- OME
- Conductive hearing loss (usually temporary)
- Perforated eardrum/CSOM
- Recurrent infection
- Facial nerve palsy
- Acute mastoiditis (rare)
- Intracranial spread
  - Subdural/extradural abscess (rare)
  - Meningitis (rare)
  - Venous sinus thrombosis (rare)

Otitis Media with Effusion

- Fluid in the middle ear – “glue ear”

Pathophysiology

- Swollen adenoids/nasopharynx following URTI causes eustachian tube blockage
- Middle ear mucosa absorbing air causes negative pressure
- Interstitial fluid is drawn into the middle ear
- Bacterial colonisation

Risk factors

- Cleft palate
- Down’s syndrome
- Cystic fibrosis
- Primary ciliary dyskinesia
- Radiotherapy for nasopharyngeal carcinoma

Presentation

- Hearing loss

Otoscopy

- Dull tympanic membrane with air bubbles/fluid level

Management

- Observe for 2-3 months
- No role for antibiotics, steroids, mucolytics or decongestants
- Auto insufflation may be of benefit

Referral to ENT

- >3 years with persistent bilateral OME for >3 months
- Hearing loss >25dB
- Speech/language/developmental problems
- Surgical treatment is grommets (± adenoidectomy if recurrent)

Grommets

- Ventilate middle ear instead of ET
- Prevents negative pressure
- Last 6-12 months
- Complications:
  - Otorrhoea 20-40%
  - Bleeding
  - Infections
  - Ossicular damage
  - TM perforation/scarring
  - Cholesteatoma
- Need to prevent dirty/bath/chlorinated water entering ear canal

Tonsillitis

- Peaks at 5-10 & 15-20

Causes

- Viral (most common)
- Streptococcus pyogenes (most common bacterial)
- Streptococcus pneumoniae
- Haemophilus influenzae
- Moraxella catarrhalis
- Staphylococcus aureus

Features

- Fever
- Sore throat
- Dysphagia
- Non-specific (particularly in young children)
  - Poor oral intake, headache, vomiting, abdominal pain
- Red, inflamed tonsils ± exudates

Assessing Probability of Bacterial Tonsillitis

Centor Criteria

- Presence of ≥3 indicates 40-60% chance of bacterial tonsillitis
  - Fever over 38°C
  - Tonsillar exudates
  - Absence of pain
  - Tender anterior cervical lymphadenopathy

FeverPAIN Score

- 2-3: 34-40%
- 4-5: 62-65%
  - Fever in previous 24hrs
  - Purulence
  - Attended within 3 days of onset
  - Inflamed tonsils
  - No cough/coryza

Management

Conservative

- Paracetamol ± NSAID
- Delayed prescription

Antibiotics

- Centor ≥3/FeverPAIN ≥4
- Immunocompromised
- Significant comorbidity
- History of rheumatic fever

Admission

- Systemically unwell/dehydrated
- Stridor/respiratory distress
- Quinsy/cellulitis
- Immunocompromised

Complications

- Quinsy, otitis media
- Post-streptococcal syndromes/scarlet fever

Peritonsillar Abscess/Quinsy

Development

- Complication of suppurating tonsillitis
- Can spread through fascial neck planes if untreated

Features

Symptoms

- Severe throat pain localising to one side
- Trismus
- "Hot potato voice"

Signs

- Uvula deviation away from affected side
- Swelling of soft palate around tonsil
- Tender cervical lymphadenopathy

Management

- Needle aspiration or incision and drainage + IV antibiotics
- Consider tonsillectomy to prevent recurrence

Tonsillectomy

Indications

Recurrent Tonsillitis

- 7 episodes in 1 year
- 5 episodes per year for 2 year
- 3 episodes per year for 3 years

Other

- Quinsy x2
- OSA

Complications

Peri-operative

- Dental
- TMJ subluxation

Post-operative

- Pain
- Bleeding
  - Primary/reactionary in first 24hrs
    - Mostly due to impaired haemostasis
  - Secondary after the first 24hrs
    - Mostly due to infection

Management of post-tonsillectomy haemorrhage

1. ABC
2. IV access
3. Bloods including group & cross-match
4. Resuscitate with fluids ± blood
5. Cautery of tonsil fossa if possible
6. Cautery under GA

Infectious Mononucleosis

Causes

- EBV (HHV-4) in 90% of cases
- CMV
- HHV-6

Features

Classic Triad

- Sore throat (membranous tonsillitis)
- Tender lymphadenopathy in anterior and posterior triangles of neck
  - Contrasts to tonsillitis which affects upper anterior cervical chain
- Fever

Others

- Malaise, anorexia, headache
- Palatal petechiae
- Splenomegaly in 50%, may rarely predispose to rupture
- Hepatitis
- Lymphocytosis
- Cold haemolytic anaemia
- Rash in response to ampicillin/amoxicillin

Diagnosis

- Heterophil antibody (Monospot) test
- NICE recommends FBC and Monospot in 2nd week of illness

Management

- Rest, hydration, avoid alcohol
- Simple analgesia for aches and pains
- Steroids if severe illness with airway compromise
- Avoid contact sports for 8 weeks

Hearing Loss in Children

Causes

Congenital

- Rubella/CMV
- Inherited deafness (AR/AD)
- Associated syndromes
  - Down's

Perinatal

- Prematurity
- Hypoxia

After Birth

- Jaundice
- Meningitis/encephalitis
- Otitis media/glue ear
- Chemotherapy

Presentation

- Neonatal screening
- Parental concerns/behavioural changes
  - Ignoring calls/sounds
  - Frustration/poor behaviours
  - Delayed speech/language development
  - Poor school performance

Investigation – Audiogram

Normal Hearing

- All sounds < 20dB (high on the chart)

Sensorineural Hearing Loss

- Both air and bone readings > 20dB (low on the chart)
- May affect one or both sides

Conductive Hearing Loss

- Bone conduction readings < 20dB
- Air conduction readings > 20dB

Mixed Hearing Loss

- Both air and bone readings > 20dB
- Difference of > 15dB between air and bone readings

Management

- MDT input
  - SALT
  - Educational psychology
  - ENT specialist
  - Sign language
  - Hearing aids if some hearing is retained

Cleft Lip/Palate

- **Cleft Lip:** Split/open section in the upper lip
- **Cleft Palate:** Defect in hard/soft palate allowing communication between oral and nasal cavities
- Mostly random with slight association with family history
- 3/10 associated with underlying syndrome

Complications

- Difficulties with feeding & speech
- Psychosocial implications
- Cleft palate:
  - Hearing problems
  - Ear infections/glue ear

Management

- MDT cleft lip services
  - Specialist nurses
  - Plastic, maxillofacial, ENT surgeons
  - Dentists
  - SALT
  - Psychologists
  - GP
- Priority: ensure baby can eat and drink
- Surgery (definitive)
  - Cleft lip at 3 months
  - Cleft palate at 6-12 months

Tongue-Tie/Ankyloglossia

- Congenital short/tight lingual frenulum
- Limited extension of tongue

Presentation

- Noticed on neonate exam
- Difficulty feeding/latching

Management

- Monitor if mild
- Frenotomy if problematic
  - Can be done on ward without anaesthetic
  - Complications rare
    - Excessive bleeding
    - Scar
    - Infection

Neck Lumps in Children

Cystic Hygroma

- Lymphatic malformation, most commonly congenital
- Typically in left posterior triangle of neck
- Found on antenatal scans/neonatal exams

Features

- Can be large
- Soft
- Non-tender
- Transilluminable

Complications

- Interference with feeding
- Haemorrhage into hygroma
- Infection

Management

- Do not resolve spontaneously
- Aspiration gives temporary improvement
- Surgical removal/sclerotherapy

Thyroglossal Cyst

- Remnant of thyroglossal duct
- Can be complicated by infection – hot, red, painful lump

Features

- Midline neck lump
- Mobile
- Soft
- Non-tender
- Fluctuant
- Moves with protrusion of tongue

Management

- CT/USS to diagnose
  - Anechoic – echogenicity suggests infection
- Surgical removal to confirm diagnosis/prevent infection

Branchial Cyst

- Remnant of improperly developed 2nd branchial cleft
- Space surrounded by epithelial tissue ± fluid

Features

- Between angle of jaw and SCM
- Transilluminable
- Anechoic unless infected

Fistulas/sinuses

- **Branchial Cleft Sinus:** Cyst is connected via tract to skin ± discharge
- **Branchial Pouch Sinus:** Cyst is connected via tract to oropharynx
- **Branchial Fistula:** Tract connecting oropharynx to skin via branchial cyst

Management

- Surgical excision if recurrent infections, diagnostic doubt or cosmetic/functional problems

Croup

- URTI causing oedema and secretions in the larynx
- 6 months – 2 years
- More common in autumn
- Majority caused by parainfluenza viruses

Presentation

- Stridor
- Increased work of breathing
- Barking cough occurring in clusters
- Hoarse voice
- Fever
- Coryzal symptoms

X-Ray

- Steeple sign

Management

- Oral dexamethasone regardless of severity
  - 150mcg/kg
  - Can be repeated after 12hrs
  - Prednisolone is an alternative

Admission

- Moderate (audible stridor, frequent cough, recessions) to severe (Prominent stridor, marked retractions, agitation, tachycardia) croup
- <6 months
- Known upper airway abnormalities

Emergency Treatment

- High-flow oxygen
- Nebulised adrenaline

Laryngomalacia

- Congenital abnormality of the supraglottic larynx, allowing it to cause partial airway collapse during inspiration
- Shortened aryepiglottic folds

Presentation

- Intermittent inspiratory stridor peaking at 6 months
  - Worsened when feeding, upset, lying on back, or during URTI

Management

- Usually resolves as the larynx matures without intervention
- May rarely require tracheostomy
- Surgery can alter laryngeal tissue to improve symptoms

Epiglottitis

- Infection of the epiglottis, usually by Haemophilus influenzae type B
  - Now rare due to vaccinations
- Life threatening emergency due to airway obstruction

Presentation

- Rapid onset
- Fever & generally unwell
- Drooling
- Stridor
- Tripod position
- Muffled voice

Investigations

- Not performed if acutely unwell
- Lateral X-Ray: Thumb sign
- Diagnosed on direct visualisation of epiglottis by senior airway trained staff

Management

- Don't distress child
- Alert senior paediatrics & anaesthetics
- Priority is airway control
  - Intubation may be needed with little warning
  - Tracheostomy may be needed if airway completely closes
- IV antibiotics (ceftriaxone)
- Dexamethasone

Whooping Cough

- URTI caused by Bordetella pertussis (Gram -ve)
- Vaccination protects neonates & infants but does not last for life

Presentation

- 2-3 days of coryza & fever ± dry cough precede:
- Severe paroxysmal coughing bouts
  - Worse at night/after feeding
  - Inspiratory "whoop"
  - ± fainting, vomiting, apnoea, pneumothorax

Diagnosis

- Nasopharyngeal swab for Bordetella PCR/culture
- Anti-pertussis IgG if cough present for >2wks

Management

- Notifiable disease
- Admit infants <6 months
- Macrolide antibiotics in 1st 21 days
  - Reduce spread but do not alter course
- Prophylactic antibiotics for vulnerable contacts
- Can last weeks-months and cause bronchiectasis/pneumonia

Bronchiolitis

- Acute bronchiolar inflammation typically caused by respiratory syncytial virus (RSV) (75%)
- Peak incidence 3-6 months, but can be diagnosed up to 2 years, especially in ex-premature babies with chronic lung disease
- Higher incidence in winter
- Breast feeding is protective against severe disease

Presentation

- Coryzal symptoms
- Respiratory distress
  - Dyspnoea, tachypnoea
  - Accessory muscles, head bobbing, nasal flaring, tracheal tugging, sub/intercostal recessions
  - Cyanosis
  - Abnormal airway noises
- Dry cough
- Poor feeding
- Mild fever (<39°)
  - Consider another diagnosis with fever >39°
- Apnoeas
- Audible wheeze, wheeze & crackles on auscultation
- Post-bronchiolitis wheeze is common

Risk Factors for Severe Disease

- Prematurity <35wks
- Congenital heart disease
- Chronic lung disease of prematurity
- Immunodeficiency
- Down's syndrome
- Severe hypotonia
- Parental smoking

Investigations

- Pulse oximetry
- NPA for RSV/respiratory panel
- CXR only if severe:
  - Visible thickening of bronchial wall causing tram-track or doughnut sign
  - Associated atelectasis

Admission

Urgent (Ambulance)

- Apnoea
- Child looks seriously unwell to health professional
- Signs of severe respiratory distress
- Resp rate > 70
- Central cyanosis
- Persistent oxygen saturations < 90%

Consider Admission

- Resp rate > 60
- Difficulty feeding
- Clinical dehydration
- Age < 3 months
- Pre-existing condition such as Down's, prematurity, CF

Management

- Nutritional support: NG, IV fluids
- Upper airway suction, saline nasal drops
- Supplementary oxygen if SpO_2 < 92%

Ventilatory Support

1. High-flow humidified oxygen via Airvo/Optiflow
2. CPAP
3. Invasive ventilation

- Ventilation monitored with capillary blood gas
  - Rising pCO_2/respiratory acidosis (type 2 failure) indicates airway collapse

Palivizumab

- Monoclonal antibody to RSV
- Considered for infants <12 months old with:
  - Extreme prematurity
  - Acyanotic congenital heart disease
  - Congenital or acquired significant orphan lung disease
  - Immunodeficiency
- Dose every month

Viral-Induced Wheeze

- Acute wheezy illness caused by viral infection resulting in small amounts of inflammation and oedema
- Typically in children under 3 years of age due to narrower airways
  - **Poiseuille's Law:** Flow rate in a tube is proportional to the radius to the power of 4
- Hereditary element
- Greater risk of later developing asthma

Differentiating from Asthma

Viral-Induced Wheeze	Asthma
< 3 years	> 3 years
No atopic history	Atopic history
Associated with viral illness only	Can be triggered by viral & bacterial illness, as well as exercise, cold weather, dust, strong emotions

Presentation

- Evidence of viral illness (coryza/cough/fever) for 1-2 days before:
  - Shortness of breath
  - Respiratory distress
  - Expiratory wheeze through the chest

Management

- Smoking parents should be encouraged to quit
- Manage symptoms as per acute asthma

Chronic Asthma

- Chronic inflammatory airway disease due to type 1 hypersensitivity leading to bronchospasm and reversible airway obstruction

Presentation

- Episodic symptoms with intermittent exacerbations
- Diurnal variation (worse at night & early morning)
- **Typical triggers**
  - Infection
  - Exercise
  - Cold
  - Dust
  - Emotion
  - Food allergens
  - Animals
- **History/family history of atopy**
  - Asthma
  - Eczema
  - Hay fever
  - Food allergies

Symptoms

- Cough
- Wheeze
- Dyspnoea

Signs

- Widespread expiratory polyphonic wheeze
- Reduced PEFR

Features suggesting alternative diagnosis

- Wheeze related only to coughs/colds
- Focal/unilateral wheeze
- Isolated/productive cough
- No response to treatment
- Normal investigations

Diagnosis/Investigations

Age 2/3-5

- Clinical diagnosis

Age 5-17

- Spirometry with bronchodilator reversibility (BDR) testing
- If inconclusive:
  - Direct bronchial challenge (histamine/methacholine)
  - FeNO
  - Peak flow variability diary

Spirometry

- Obstructive picture (FEV1:FVC < 70%)
- BDR: improvement in FEV1 of > 12%

Fractional Exhaled Nitric Oxide (FeNO)

- Inducible Nitric Oxide Synthase (iNOS) is expressed in inflammatory cells (esp eosinophils) and so levels correlate to inflammation
- In children ≥ 35 parts per billion (ppb) is positive

Management

Principles

- Stepwise ladder
  - Enter ladder as appropriate for symptoms
  - Review regularly & step up/down based on symptoms
- Aim for no symptoms/exacerbations at lowest dose and number of treatments
- Check inhaler technique at every review
- **ICS:** Inhaled corticosteroids, safe in children but associated with reduction in final adult height of up to 1cm
  - Effect of poorly controlled asthma would likely be greater
- **MART:** Maintenance and reliever therapy, combination of ICS and LABA with fast-acting element (eg formoterol)

Under 5 years

1. SABA as required
2. SABA + paediatric low dose ICS **or** leukotriene receptor antagonist (LTRA – montelukast)
3. SABA + paediatric low dose ICS + LTRA
4. Stop LTRA and refer to specialist

Age 5-16

1. SABA as required
2. SABA + paediatric low dose ICS
3. SABA + paediatric low dose ICS + LTRA
4. SABA + paediatric low dose ICS + LABA
   - NICE recommend stopping LTRA if it hasn't helped
5. SABA + MART with paediatric low dose ICS
6. SABA + MART with paediatric moderate dose ICS
7. Consider:
   - Increase to MART with paediatric high dose ICS
   - Trial of additional drug (eg theophylline)
   - Specialist advice

Inhaler Technique

- Remove the cap
- Shake the inhaler (depending on the type)
- Sit or stand up straight
- Lift the chin slightly
- Fully exhale
- Make a tight seal around the inhaler between the lips
- Take a steady breath in whilst pressing the canister
- Continue breathing for 3 – 4 seconds after pressing the canister
  - If using spacer, breathe in & out 5 times
- Hold the breath for 10 seconds or as long as comfortably possible
- Wait 30 seconds before giving a further dose
- Rinse the mouth after using a steroid inhaler

Acute Asthma

- Rapid deterioration of the symptoms of asthma, triggered by any of the typical triggers

Presentation

- Progressively worsening shortness of breath
- Signs of respiratory distress
- Tachypnoea
- Widespread expiratory polyphonic wheeze
- Tight chest with reduced entry
  - Silent chest is an ominous sign

Severity

Moderate	Severe	Life Threatening
Peak flow > 50% predicted	Peak flow < 50% predicted	Peak flow < 33% predicted
Normal speech	Saturations < 92%	Saturations < 92%
	Unable to complete sentences in one breath	Exhaustion and poor respiratory effort
	Signs of respiratory distress	Hypotension
	Respiratory rate: > 40 in 1-5 years > 30 in > 5 years	Silent chest
	Heart rate: > 140 in 1-5 years > 125 in > 5 years	Cyanosis
		Altered consciousness / confusion

Management

Principles

- Oxygen if required (SpO2 < 94%)
- Bronchodilators
- Steroids to reduce airway inflammation
  - Should be given to all children with acute asthma for 3-5 days
- Antibiotics only if bacterial cause is suspected
- Monitor potassium when giving high doses of salbutamol

Mild Cases

- Outpatient care
- Regular salbutamol via spacer
  - 4-6 puffs every 4 hours

Moderate-Severe Cases (Stepwise)

1. Salbutamol via spacer (start with 10 puffs every 2hrs)
2. Nebulised salbutamol/ipratropium bromide
3. Oral prednisolone (1mg/kg for 3 days) 1

4. IV hydrocortisone
5. IV MgSO4
6. IV salbutamol
7. IV aminophylline
8. Call anaesthetics & ICU

Weaning

- Look for:
  - Cyanosis
  - Tracheal tug
  - Subcostal recessions
  - Hypoxia
  - Tachypnoea
  - Wheeze
- If well, step down the ladder

Discharge

- Considered when child is well on 6 puffs 4 hourly of salbutamol
- Step-down regime:
  - 6 puffs 4 hourly for 48 hours
  - 4 puffs 6 hourly for 48 hours
  - 2-4 puffs as required
- Finish steroid course
- Provide safety net information & personalised asthma plan

Pneumonia

- Infection & inflammation of lung tissue with sputum filling alveoli & airways

Prevention

- Vaccines
  - PCV, pertussis, haemophilus
- Smoking cessation in the home

Presentation

- Productive cough
- High fever ± toxicity
- Tachypnoea & tachycardia
- Increased work of breathing
- Hypotension
- Lethargy
- Delirium

Characteristic Chest Signs

- Accessory muscles, grunting, recessions/tracheal tug
- Bronchial breath sounds
- Focal coarse crackles
- Dullness to percussion
- NOT wheeze

Causes

Viral

- Most common, over 90% in children <2 years
- RSV (most common viral)
- Parainfluenza, influenza

Bacterial

- Streptococcus pneumoniae (most common)
- Group A/B strep
- Moraxella
- Staphylococcus aureus
  - Associated with penumatocoeles and multiple lobe consolidation on CXR
- Haemophilus influenzae (particular un-/pre-vaccinated)
- Mycoplasma/other atypicals

Investigations

- CXR is investigation of choice but not routinely required, indicated by:
  - Significant respiratory distress
  - Requiring hospitalisation
  - Suspected complications – effusion, abscess
  - Failure to resolve/respond
  - Follow up
- CXR can show "round pneumonia" – multiple small, circumscribed, round opacities
  - Common around 5 years
  - Most commonly strep
  - Treat as standard pneumonia
- Repeat CXR after 4-6 weeks if:
  - Round pneumonia
  - Collapse
  - Ongoing symptoms
- All patients should have blood cultures
- Capillary blood gas useful in monitoring unwell patients
- Sputum cultures/throat swabs for culture/PCR can help diagnose organism & guide treatment

Management

CAP < 2 months

- IV amoxicillin + IV gentamycin + IV cefotaxime

CAP > 2 months

- PO amoxicillin if well
- IV amoxicillin if unwell
  - +/- azithromycin PO

Sepsis/pleural effusion

- IV cefuroxime/cefotaxime + PO azithromycin

Aspiration Pneumonia

- IV co-amoxiclav

Empyema

- Diagnosis with chest US ± PE
- Chest drainage ± urokinase

Investigations in Recurrent LRTIs

- FBC (levels & differential of white cells)
- CXR (structural abnormalities/scarring)
- Serum Ig's (selective antibody deficiency)
- IgG to previous vaccines (immunoglobulin class-switch recombination deficiency)
- Sweat test (CF)
- HIV test

Chronic Lung Disease of Prematurity

- AKA bronchopulmonary dysplasia
- Typically occurs in babies born before 28wks who suffer from respiratory distress syndrome & require oxygen therapy/intubation
- Diagnosed based on CXR changes & when infant requires oxygen therapy at 36wks gestational age

Features

- Low oxygen saturations
- Increased work of breathing
- Poor feeding & weight gain
- Crackles & wheezes on auscultation
- Increased susceptibility to infection

Prevention

Antenatal

- Steroids to mothers who are likely to enter premature (<36wks) labour

Postnatal

- Using CPAP over invasive ventilation where possible
- Using caffeine to stimulate respiratory effort
- Not over-oxygenating with supplementary oxygen

Management

- Formal sleep study to assess oxygen saturations
- Discharge with low dose home oxygen
  - Eg 0.01L/minute via nasal cannula
  - Weaned over 1st year of life
- Protection against RSV (palivizumab)

Primary Ciliary Dyskinesia/Kartagener's

- Autosomal recessive condition affecting motile cilia (dynein protein)
- Mucous build-up in respiratory tract → chronic infections & bronchiectasis (similar picture to CF)
- Associated with consanguinity

Features

Kartagener's Triad (not all patients will have all 3)

- Paranasal sinusitis
- Bronchiectasis
- Situs inversus
  - 25% with situs inversus have PCD
  - 50% with PCD have situs inversus

Other

- Infertility (fallopian tube cilia/sperm flagella)

Diagnosis

- Sample of ciliated epithelium via nasal brushing or bronchoscopy

Management

- Daily physio, high calory diet, antibiotics

Cystic Fibrosis

Pathophysiology

- Autosomal recessive defect in cystic fibrosis transmembrane conductance regulator (CFTR) gene on chromosome 7
- Most common mutation is delta-F508
- Gene encodes a chloride channel, defects result in low volume, thick, biliary, pancreatic, and respiratory secretions
  - Also congenital absence of the vas deferens in males
- 1 in 25 are carriers

Presentation

- Vast majority of cases of meconium illness are caused by CF

Symptoms

- Chronic cough with thick sputum production
- Recurrent respiratory tract infections
- Steatorrhoea
- Abdominal pain & bloating
- Failure to thrive
- Parents may report salty taste to the skin
- Male infertility & female subfertility in adults

Signs

- Nasal polyps
- Finger clubbing
- Crackles & wheezes on auscultation
- Abdominal distension

Complications

- DM
- Rectal prolapse

Microbial Colonisers

- **Staphylococcus aureus**
- **Pseudomonas aeruginosa**
  - Significant morbidity and mortality
  - Very difficult to get rid of & develops multidrug resistance
  - Treated with long term nebulised antibiotics (eg tobramycin)
- Burkholderia capecia
- Aspergillus
- Haemophilus influenzae
- Klebsiella pneumoniae
- Escherichia coli

Prognosis

- Median life expectancy of 47 years
- 90% of adults with CF develop pancreatic insufficiency
- 50% develop CF related diabetes treated with insulin
- 30% develop liver failure

Diagnosis

- Neonatal bloodspot test

Sweat Test

- Gold standard for diagnosis
- Pilocarpine applied to skin & electric current passed between electrodes causing skin to sweat
- Sweat absorbed and tested for chloride concentration
- Cut off: > 60mmol/L
- **Causes of false positive sweat test:**
  - Malnutrition
  - Adrenal insufficiency
  - Glycogen storage disease
  - Nephrogenic diabetes insipidus
  - Hypothyroidism/hypoparathyroidism
  - G6PD
  - Ectodermal dysplasia

Genetic Testing of CFTR Gene

- Chorionic villous sampling/amniocentesis
- Blood

Management

- Twice daily chest physio & postural drainage
- Minimise contact with other CF patients
- High calory diet with high fat intake
- Pancreatic enzyme supplements (CREON) taken with meals
- Vitamin supplementation
- Antibiotic prophylaxis (flucloxacillin)
- Bronchodilators
- Nebulised DNase
- Nebulised hypertonic saline
- Fertility treatment

Vaccinations

- Pneumococcal
- Influenza
- Varicella

Transplantation

- Lung transplant in end stage respiratory failure
- Liver transplant in liver failure

Lumacaftor/ivacaftor (Orkambi)

- Treats CF if homozygous for delta-F508 mutation
- Lumacaftor increases CFTR transportation to cell surface
- Ivacaftor potentiates CFTR already at the cell surface

Monitoring (6 Monthly)

- Sputum cultures
- DM
- Osteoporosis
- Vitamin D deficiency
- Liver failure

Endocrinology & Metabolic Disorders

Type 1 Diabetes Mellitus

- Pancreas loses ability to produce insulin typically due to autoimmune destruction of β cells of the islets of Langerhans
- May be triggered by viruses such as Coxsackie B/enterovirus

Normal Glucose Levels

- Concentrations typically kept between 4.4 and 6.1 mmol/L

Presentation

- 25-50% are first diagnosed presenting in DKA
- Earlier symptoms occur 1-6 weeks before development of DKA
  - Polyuria
  - Polydipsia
  - Weight loss
  - Enuresis
  - Recurrent infection

Diagnosis

If symptomatic

- Fasting blood glucose ≥ 7mmol/L
- Random blood glucose/75g OGTT ≥ 11.1mmol/L

If asymptomatic

- As above on 2 separate occasions

Other Investigations

- Baseline bloods including FBC, U+E
- Blood cultures in suspected infection
- HbA1c (limited use in new diagnoses)
- TFTs & anti-TPO antibodies
- Anti-TTG antibodies
- Insulin, anti-GAD, & islet cell antibodies

Short Term Complications

Hyperglycaemia (& DKA)

- If not taking enough insulin to match carbohydrate intake

Hypoglycaemia

- Not taking in enough carbohydrates to match insulin dose/carbohydrates not being processed properly
- Awareness of hypoglycaemia at different blood sugar levels varies between patients
- **Symptoms:**
  - Hunger
  - Tremor, sweating, irritability
  - Pallor
  - Dizziness
  - Coma
- Treated with fast & slow acting oral glucose (eg Lucozade + buiscuits) if conscious or IM glucagon/IV dextrose if unconscious

Long Term Complications

Microvascular

- Peripheral neuropathy
- Autonomic neuropathy & gastroparesis
- Retinopathy
- Nephropathy

Macrovascular

- Coronary artery disease
- Peripheral vascular disease
- Hypertension
- Stroke

Treatment Related

- Lipohypertrophy at injection sites
  - Affects absorption of insulin

Infection Related

- UTIs, pneumonia, skin & soft tissue infection
- Fungal (oral/vaginal candidiasis)

Management

- Subcutaneous insulin

Basal-Bolus Regimes

- Most commonly initiated in new diagnoses
- Long-acting (basal) insulin such as Lantus in the evening giving 24hr background insulin
- Short-acting (bolus) insulin such as Actrapid 3 times a day ~30 minutes before meals
  - Amount of bolus insulin is tailored to the amount of carbohydrates to be eaten
- Injection sites rotated to avoid lipohypertrophy

Insulin Pump

- Continuously infuses insulin at different rates via a cannula sitting under the skin
  - Cannula is replaced every 2-3 days and injection sites are rotated
- Patients > 12 with difficulty controlling HbA1c
- **Advantages:** Better glucose control, fewer injections, more flexibility with eating
- **Disadvantages:** Learning to use, attached at all times, blockages, small infection risk

Monitoring

HbA1c

- Glycated haemoglobin; reflects glucose levels over previous 3 months
- Used to monitor long term control of glucose level

Capillary Glucose Monitoring

- Fingerprick & glucometer – gives instant blood glucose level
- Used to guide insulin on injection-injection basis

Flash Glucose Monitoring

- Eg Freestyle Libre – sits on skin & measures glucose levels in interstitial fluid
- Lags 5 minutes behind blood, so capillary glucose must also be checked if a hypo is suspected

Diabetic Ketoacidosis (DKA)

- Life-threatening condition caused by a lack of sufficient insulin or precipitant such as infection or MI in type 1 (and rarely type 2) diabetes

Pathophysiology

Ketoacidosis

- Body cannot utilise glucose due to lack of insulin, so ketogenesis occurs for fuel

- Ketones are acidic and the kidneys compensate by producing bicarbonate
- Glucose and ketone levels rise until bicarbonate buffer is overwhelmed and blood becomes acidotic

Dehydration

- Hyperglycaemia overwhelms the kidneys' ability to reabsorb glucose and massive osmotic diuresis occurs
- **Fluid deficit:**
  - Mild-moderate DKA (pH > 7.1) 5%
  - Severe DKA (pH < 7.1): 10%

Potassium Imbalance

- Insulin normally drives potassium into cells
- In DKA, blood potassium can be normal or high but total body potassium is low due to diuresis & lack of transport into cells

Presentation

- Polyuria & polydipsia
- Weight loss
- Nausea & vomiting
- Abdominal pain
- Acetone breath smell
- Dehydration & hypotension
- Altered consciousness
- Symptoms of underlying trigger

Diagnosis

- Requires:
  - Hyperglycaemia (glucose > 11mmol/L)
  - Ketonaemia (> 3mmol/L or ++ on dipstick)
  - Acidosis (pH <7.3)
- Also, bicarbonate <15mmol/L

Management

Fluids & Electrolytes

- If shocked: 10ml/kg NS fluid challenge (x3 maximum)
- Maintenance fluids + (deficit - boluses given) to be replaced over 48hrs
  - Slower if risk of cerebral oedema
  - Normal saline with 20mmol KCl in 500ml
    - Monitor serum potassium closely

Insulin & Glucose

- Patient's long-acting insulin should be continued
- No insulin infusion in the first hour
- 0.05-0.1U/kg/hour
- When glucose < 14mmol/L, add dextrose to fluid regime

General

- Treat underlying trigger (eg sepsis)
- Monitor for signs of cerebral oedema
- Monitor glucose, ketones & pH
- Plan insulin regime going forward & commence/recommence SC regime before stopping infusion

Cerebral Oedema

- Fluid shift from extracellular to intracellular space in the brain following rapid correction of dehydration & hyperglycaemia
- Patients are at risk during treatment of DKA

Presentation

- Headaches
- Bradycardia
- Altered behaviour
- Altered consciousness

Management

- Slow IV fluids
- IV mannitol
- IV hypertonic saline

Adrenal Insufficiency

Types

Primary

- Addison's Disease
- Autoimmune (usually) damage to adrenal glands

Secondary

- Lack of ACTH from the pituitary
- Damage or congenital hypoplasia of the pituitary

Tertiary

- Lack of CRH from the hypothalamus
- Usually due to HPA axis suppression

Presentation

Babies

- Lethargy
- Vomiting, poor feeding, failure to thrive
- Hypoglycaemia
- Jaundice

Older Children

- Nausea & vomiting
- Reduced appetite & poor weight gain/weight loss
- Abdominal pain
- Muscle cramps
- Developmental delay
- Bronze hyperpigmentation (Addison's)

Investigations

- U+E, blood glucose
- **Primary:**
  - Cortisol low
  - ACTH high
  - Aldosterone low
  - Renin high
- **Secondary:**
  - Cortisol low
  - ACTH low
  - Aldosterone & renin normal

Short Synacthen Test

- Synthetic ACTH given in the morning
- Cortisol measured at baseline, 30, & 60 minutes
- Failure of cortisol to rise to 2x baseline: primary adrenal insufficiency

Management

- Glucocorticoid replacement (hydrocortisone)
- Mineralocorticoid replacement (fludrocortisone)
- Steroid card & emergency ID tag

Monitoring

- Growth & development
- Blood pressure
- Bloods: U+Es, glucose, bone profile, vitamin D

Sick Day Rules

- Increased steroid dose until illness is resolved
  - IM/IV may be required if vomiting or diarrhoea
- Blood sugar monitored closely

Addisonian/Adrenal Crisis

- Life threatening absence of steroids
- May be first presentation of adrenal insufficiency or triggered by acute illness

Features

- Hypotension
- Reduced consciousness
- Hypoglycaemia, hyponatraemia & hyperkalaemia

Management

- Parenteral hydrocortisone
  - No fludrocortisone required as high dose glucocorticoids have weak mineralocorticoid effect
- Fluid resuscitation
- Correction of hypoglycaemia
- Return to oral replacement after 24 hours

Congenital Adrenal Hyperplasia

Deficiency of

21-hydroxylase (<90%)

- Can be complete absence or variable deficiency
- No conversion of progesterone to mineralocorticoids/glucocorticoids → increased conversion to testosterone
- No negative feedback of ACTH → hyperplasia

11-beta hydroxylase (5%)

17-hydroxylase (rare)

Presentation

Severe cases of 21-hydroxylase deficiency present shortly after birth:

- Ambiguous genitalia in females
- Hyponatraemia, hyperkalaemia, hypoglycaemia
  - Poor feeding
  - Vomiting
  - Dehydration
  - Arrhythmias

Mild cases present later due to raised androgen levels:

- Females
  - Tall for their age
  - Virilisation
  - Hirsuitism
  - Amenorrhoea
- Males
  - Tall for their age
  - Precocious puberty
  - Small testes

Management

- Mineralo/glucocorticoid replacement
- Surgery for females with virilised genitalia

Growth Hormone Deficiency

- **Congenital:** Genetic mutations (GH1, GHRHR)/empty sella syndrome
- **Acquired:** Infection/trauma/surgery
- Can occur in isolation or in combination with other pituitary deficiencies (panhypopituitarism)

Presentation

Neonatal

- Micropenis
- Hypoglycaemia
- Severe jaundice

Older Children

- Poor growth
  - Stops/severely slows at 2-3
- Slow development of movement & strength
- Delayed puberty

Investigations

Growth Hormone Stimulation Test

- GH levels measured in response to exogenous stimulant (glucagon/insulin/arginine/clonidine)
- Poor response indicated GH deficiency
- Test for other deficiencies (thyroid, adrenal)
- MRI brain
- Genetic testing for associated syndromes (Turner, Prader-Willi)
- X-Ray wrist/DEXA scan to determine bone age

Management

- Daily SC growth hormone (somatropin)
- Treatment of any associated deficiencies
- Monitor height & development

Hypothyroidism

Congenital

- Affects ~ 1/4,000 babies
- Underdeveloped gland (dysgenesis) or gland that does not produce hormones (dyshormonogenesis)
- Can cause permanent cognitive impairment if not treated in the first 4 weeks

Presentation

- Usually picked up on bloodspot test
- Prolonged jaundice
- Poor feeding
- Constipation
- Increased sleeping
- Puffy face & macroglossia
- Hypotonia & reduced activity
- Slow growth & development

Acquired

- Most commonly causes by autoimmune (Hashimoto's) thyroiditis
  - Anti-TPO and anti-thyroglobulin antibodies
  - Associated with other autoimmune conditions (T1DM, coeliac)

Presentation

- Fatigue & low energy
- Poor growth
- Weight gain
- Poor school performance
- Constipation
- Dry skin & hair loss

Investigations

- TFTs
- Thyroid ultrasound
- Thyroid antibodies

Management

- Levothyroxine orally once daily
  - Titrated based on TFTs & symptoms

Phenylketonuria

- Autosomal recessive defect in phenylalanine hydroxylase (responsible for converting phenylalanine to tyrosine) gene located on chromosome 12
- 1/10,000 live births

Presentation

- (If missed by bloodspot)
- Developmental delay by 6 months
- Fair hair, blue eyes (classically)
- Learning difficulties
- Infantile spasms
- Eczema
- Musty odour to urine & sweat (phenylacetate)

Diagnosis

- Neonatal bloodspot test
- Hyperphenylalaninaemia
- Phenylpyruvic acid in urine

Management

- Early dietary restriction of phenylalanine

Homocystinuria

- Autosomal recessive deficiency in cystathionine-beta-synthase resulting in severe elevations in plasma/urine homocysteine levels

Presentation

- Fine, fair hair (classically)
- Learning difficulties
- Seizures
- Downward dislocation of lens
- Severe myopia
- Marfanoid habitus
- Osteoporosis
- Kyphosis
- Increased risk of venous/arterial thromboembolism
- Livedo reticularis
- Malar flush

Diagnosis

- Neonatal bloodspot test
- Increased homocysteine levels in blood & urine
- Cyanide-nitroprusside test

Management

- Vitamin B6 (pyridoxine) supplements

Nephrology & Urology

Nephrotic Syndrome

- Peak in children from 2-5 years
- GBM becomes permeable to proteins

Presentation

Classic Triad

- Proteinuria (> 1g/m^2/24hrs)
- Hypoalbuminaemia (< 25g/L)
- Oedema

Other Features

- Frothy urine
- Pallor
- High cholesterol, triglycerides, & LDL
- Hypertension
- Hypercoagulability (loss of ATIII)
- Infection susceptibility (loss of Igs)

Causes

Minimal Change Disease

- 90% of cases of nephrotic syndrome in children
- Majority are idiopathic, 10-20% have underlying cause:
  - Drugs (rifampicin, NSAIDs)
  - Hodgkin's lymphoma, thymoma
  - Infectious mononucleosis
- No changes on light microscopy, electron microscopy shows foot process effacement
- 80% respond to steroids, cyclophosphamide is 2nd line
- Prognosis
  - 1/3 have 1 episode
  - 1/3 have infrequent relapses
  - 1/3 have frequent relapses which stop before adulthood

Others

- Intrinsic kidney disease
  - Focal segmental glomerulosclerosis
  - Membranoproliferative glomerulonephritis
- Systemic illness
  - HSP
  - Diabetes
  - Infection (HIV, hepatitis, malaria)

Management

Steroids

- 80% are steroid sensitive
  - 80% will relapse & need further steroids

Steroid Resistant Patients

- ACE inhibitors & immunosuppression
- Cyclophosphamide/cyclosporin/tacrolimus/ rituximab

Other Mx

- Low salt diet, diuretics
- Albumin infusions in severe hypoalbuminaemia
- Antibiotic prophylaxis

Nephritic Syndrome

- Inflammation within the nephron causing:
  - Reduction in renal function
  - Haematuria (visible or invisible)
  - Proteinuria (typically not as severe as nephrotic syndrome)
- Main causes in children are post-streptococcal glomerulonephritis and IgA nephropathy

Post-Streptococcal Glomerulonephritis

- Occurs 1-3 weeks after a group A strep infection (such as tonsillitis)
- Caused by immune complex (IgG, IgM, C3) deposition in the glomeruli

Presentation

- Headache, malaise
- Haematuria
- Proteinuria ± oedema
- AKI (oliguria)
- Hypertension

Investigations

- Streptococcal throat swab
- Raised ASO titres
- Low complement levels
- Renal biopsy:
  - Diffuse glomerulonephritis with endothelial proliferation & neutrophils
  - EM: subepithelial immune complex deposition

Management

- Supportive, 80% make full recovery
- Antihypertensives/diuretics may be needed to treat hypertension/oedema

IgA Nephropathy (Berger's Disease)

- Related to HSP (IgA vasculitis)
- Typically occurs in teenagers (M:F) following URTI

Presentation

- Macroscopic haematuria 1-2 days following URTI
- Nephrotic level proteinuria & AKI are rare

Investigations

- Complement levels normal
- Renal biopsy:
  - IgA deposition & glomerular mesangial proliferation

Management

- Supportive treatment
- ACE inhibitors
- Steroids/immunosuppression
  - Effectiveness of these is questionable
- 25% develop ESRF

Haemolytic Uraemic Syndrome

- Typically occurs 5 days after gastroenteritis from E. coli O157:H7 infection
  - Use of antibiotics or anti-motility drugs during gastroenteritis increases risk of HUS
- Triad of:
  - Microangiopathic haemolytic anaemia (MAHA)
  - Thrombocytopenia
  - Acute kidney injury

Presentation

- Follows gastroenteritis
- Reduced urine output
- Haematuria/dark brown urine
- Hypertension
- Oedema
- Bruising
- Lethargy/irritability
- Confusion

Management

- Supportive management
  - Dialysis if required
  - Antihypertensives if required
  - Blood transfusions
  - Careful fluid balance
- No role for antibiotics
- Plasma exchange in severe cases
- 70-80% make full recovery

Wilms' Tumour (Wilms' Nephroblastoma)

- Typically affects children under 5 (median age 3)
- 1/3 associated with LOF mutation of WT1 gene on chromosome 11

Presentation

- Abdominal mass (may be noticed by parents)
- Painless haematuria
- Flank/abdominal pain
- Anorexia, fever, lethargy, weight loss
- Hypertension
- 95% unilateral
- 20% metastasised (usually lung)

Investigations

- US is initial investigation
- Biopsy is needed for diagnosis
- CT/MRI for staging

Management

- Nephrectomy
- Adjuvant chemo/radiotherapy depending on stage & histology
- 80% cure rate (worse if metastases are present)

Autosomal Recessive Polycystic Kidney Disease

- Much rarer than ADPKD
- Presents in neonatal/antenatal period rather than in adulthood

Pathophysiology

- Mutation of polycystic kidney & hepatic disease 1 (PKHD1) gene on chromosome 6
- Encodes fibrocystin/polyductin protein complex (FPC) which plays a role in renal tubules & hepatic epithelium

Results in:

- Cystic enlargement of renal collection ducts
- Oligohydramnios, pulmonary hypoplasia & Potter syndrome
- Congenital liver fibrosis

Outcomes

Potter Syndrome

- Dysmorphic features due to oligohydramnios
- Underdeveloped ear cartilage, low set ears, flat nasal bridge & abnormalities of the skeleton

Pulmonary Hypoplasia

- Respiratory distress shortly after birth
- Chronic lung disease

Hepatic

- Congenital fibrosis
- Portal hypertension & varices

Renal

- Hypertension
- ESRF before adulthood

Prognosis

- 1/3 do not survive neonatal period
- 1/3 survive to adulthood

Multicystic Dysplastic Kidney (MCKD)

- 1 kidney is cystic while the other is normal
- Rarely bilateral (inevitably causes death in infancy)
- Usually diagnosed on antenatal scans

Outcomes

- 1 kidney sufficient, cystic kidney usually atrophies and disappears by age 5
- Increased risk of UTIs, hypertension, CKD

Urinary Tract Infection

Causes

Organisms

- E. coli (80%)
- Proteus
- Pseudomonas

Predisposing Factors

- Incomplete bladder emptying
  - Hurried micturition
  - Infrequent voiding
  - Constipation
  - Neuropathic bladder
- Vesicoureteric reflux
  - Present in 35% of children with a UTI
- Poor hygiene
  - Not wiping front to back (girls)

Presentation

Babies

- Poor feeding
- Vomiting
- Irritability
- Fever
- Frequency

Older Infants & Children

- Fever
- Abdominal/suprapubic pain
- Dysuria
- Haematuria
- Frequency/incontinence
- Vomiting

Acute Pyelonephritis

- Fever > 38°
- Loin pain/tenderness

Investigation

Urine dipstick in any child with:

- Signs/symptoms suggestive of UTI
- Unexplained fever > 38°
- An alternative site of infection but are remaining unwell

Urine Sample

- Clean catch if possible
- If nitrites/leukocytes are present, send sample to lab for MSU
- If neither are present, UTI is unlikely

Management

Oral Antibiotics

- > 3 months & otherwise well
- Trimethoprim/nitrofurantoin/cefalexin/amoxicillin

IV Antibiotics

- < 3 months or acute pyelonephritis

Investigating Recurrent UTIs

Ultrasound

- All children under 6 months with a 1st UTI should have an abdominal ultrasound within 6wks
  - During illness if there are recurrent UTIs/atypical bacteria
- Children with recurrent UTIs should have an ultrasound within 6wks
- Children with atypical UTIs should have an ultrasound during the illness

Dimercaptosuccinic Acid Scan (DMSA)

- Injection of radioactive DMSA & imaging with gamma camera
- Areas of poor uptake in kidneys indicate potential scarring

Performed

- 4-6 months after illness to assess damage from recurrent or atypical UTIs

Micturating Cystourethrogram (MCUG)

- Catheterisation & contrast injection into bladder followed by x-ray imaging
- Used to diagnose VUR
- Prophylactic antibiotics given for 3 days

Performed

- Atypical/recurrent UTIs in infants < 6 months
- Family history of VUR
- Dilatation of ureters on US
- Poor urinary flow

Vesicoureteric Reflux

- Ureters are displaced laterally entering bladder at a more perpendicular angle
- Short intramural course & impaired VUJ function
- Backflow of urine from bladder to ureters/kidneys

Presentation

- Antenatal: hydronephrosis on US
- Recurrent childhood UTIs
- Reflux nephropathy

Diagnosis

- MCUG

Grade:

I: Reflux into ureter only, no dilatation
II: Reflux into renal pelvis, no dilatation
III: Mild/moderate dilatation of ureter/pelvis/calyces
IV: Dilatation with moderate ureteral tortuosity
V: Gross dilatation with ureteral tortuosity

Management

- Avoid constipation/excessively full bladder
- Prophylactic antibiotics
- Surgical input depending on severity

Posterior Urethral Valve

- Present in newborn boys
- Tissue at proximal urethra prevents urine outflow, leading to hydronephrosis, incomplete voiding, and predisposition to UTIs

Presentation

Antenatal

- Bilateral hydronephrosis & oligohydramnios
  - Severe cases only
  - Results in pulmonary hypoplasia & respiratory distress

Children

- Difficulty urinating
- Poor stream
- Chronic retention
- Palpable bladder
- Recurrent UTIs
- Impaired renal function

Investigations

- Severe cases picked up on antenatal scans
- Abdominal ultrasound: Enlarged, thickened bladder with bilateral hydronephrosis
- MCUG shows location of extra tissue & poor flow/reflux of urine
- Cystoscopy can visualise/ablate the extra tissue

Management

- Mild cases can be monitored
- Temporary catheters can be used while waiting for definitive treatment
- Definitive treatment is ablation during cystoscopy

Enuresis

- Involuntary urination
- Most children have control of daytime urination by 2 years and nighttime urination by 3-4 years

Types

Primary Nocturnal Enuresis

- Child has never consistently been dry at night
- Causes:
  - Overactive bladder
  - Excessive nighttime fluid intake
  - Failure to wake (deep sleep, underdeveloped bladder signals)
  - Psychological distress
  - Chronic constipation
  - Learning difficulty
  - Cerebral palsy

Secondary Nocturnal Enuresis

- Child had previously been dry at night for at least 6 months
- Causes:
  - UTI
  - Constipation
  - T1DM
  - New psychosocial problems/maltreatment
- Can often be managed by treating underlying cause

Diurnal Enuresis

- Daytime incontinence
- More commonly affects girls
- Causes:
  - Urge incontinence
  - Stress incontinence
  - Recurrent UTI
  - Psychosocial problems
  - Constipation

Management

- Look for & treat underlying cause/trigger
- Advise on fluid intake & toileting behaviour
- Reward systems (eg star charts) for agreed behaviour, not for dry nights
- Enuresis alarms first line for children under 7
- Desmopressin first line for children over 7, particularly if:
  - Short-term control is needed
  - Enuresis alarm has been ineffective
- Other pharmacological options
  - Oxybutinin
  - Imipramine

Undescended Testes

- Occurs in 2-5% of term boys
  - Much more common in preterm boys
- 25% of cases are bilateral
- Undescended testes later in childhood/adulthood are at increased risk of:

- Torsion
- Infertility
- Testicular cancer
- Psychological issues

Risk Factors
- Family history
- Low birth weight/SGA
- Prematurity
- Maternal smoking

Retractile Testes
- Testes may move up into inguinal canal when it is cold/due to cremasteric reflex before puberty
- Usually resolves as they go through puberty
- Occasionally fully retract or fail to descend & require orchidopexy

Management
Unilateral
- Referral by 3 months, ideally seeing urologist by 6 months
- Orchidopexy by 1 year

Bilateral
- Senior paediatric review within 24hrs of birth
  - May need urgent endocrine/genetic investigation

Hypospadias
- Congenital ventral displacement of the urethral meatus on the penis
  - Bottom of the glans (subcoronal) (90%)
  - Mid-shaft
  - Base of shaft/scrotal (penoscrotal)
- Usually diagnosed on neonatal exam

Management
- Referral to paediatric urologist
- Child cannot be circumcised unless parents told otherwise by a urologist
- Mild cases may not require surgery
- Surgery usually performed at 3-4 months

Complications
- Difficulty directing urination
- Cosmetic/psychological concerns
- Sexual dysfunction

Hydrocele
- Collection of fluid in the tunica vaginalis

Simple/Non-communicating Hydrocele
- Fluid is trapped in the tunica vaginalis
- Common in newborn boys
- Typically reabsorbed over time

Communicating Hydrocele
- Tunica vaginalis remains connected to peritoneal cavity via processus vaginalis
- Fluid moves between peritoneum and hydrocele allowing it to fluctuate in size

Features
- Smooth non-tender swelling in front of and below testicle
- Transilluminable
- Can “get above” a simple hydrocele

Management
- Ultrasound if clinical diagnosis uncertain

Simple
- Usually resolve in 2 years
- May require surgery if associated with other problems (eg hernia)

Communicating
- Surgical removal/ligation of processus vaginalis

Haematology

Anaemia

Features

Symptoms

- Tiredness
- Dyspnoea
- Tachycardia/palpitations
- Headaches/dizziness
- Pica (iron deficiency)
- Hair loss (iron deficiency)

Signs

- Pallor (skin, conjunctiva, palmar creases)
- Tachycardia, tachypnoea
- Koilonychia, angular cheilitis, atrophic glossitis, brittle hair & nails (iron deficiency)
- Jaundice (haemolytic anaemia)
- Bone deformities (thalassaemia)

Causes of Anaemia in Infancy

- Physiological anaemia of infancy
- Anaemia of prematurity
- Blood loss
- Haemolysis
  - Haemolytic disease of the newborn
  - Hereditary spherocytosis
  - G6PD deficiency
- Twin-twin transfusion syndrome

Physiological Anaemia of Infancy

- Most common cause of anaemia in infancy
- Occurs around 6-9 weeks of age in healthy term babies
- Due to negative feedback of erythropoietin caused by high oxygen delivery by high haemoglobin levels at birth

Anaemia of Prematurity

- Premature babies are more prone to anaemia
- Likelihood of needing a transfusion scales with level of prematurity
- Especially likely if unwell at birth (eg neonatal sepsis)
- Factors:
  - Less time receiving iron from mother in utero
  - Erythropoiesis can't keep up with rapid growth in first few weeks
  - Reduced erythropoietin levels
  - Blood samples remove a significant proportion of circulating volume

Haemolytic Disease of the Newborn

- Rhesus D -ve mother with Rhesus D +ve baby who was sensitised in a previous pregnancy
- Anti-D antibodies cross the placenta in subsequent pregnancies causing:
  - Haemolytic anaema
  - Neonatal jaundice
- Direct Coombs Test +ve

Causes of Anaemia in Older Children (& Adults)

Microcytic Anaemia (TAILS)

- Thalassaemia
- Anaemia of chronic disease
- Iron deficiency
- Lead poisoning
- Sideroblastic anaemia

Normocytic Anaemia (3 As, 2 Hs)

- Acute blood loss
- Anaemia of chronic disease
- Aplastic anaemia
- Haemolytic anaemia
- Hypothyroidism

Megaloblastic Anaemia

- B12 deficiency
- Folate deficiency

Normoblastic Macrocytic Anaemia

- Alcohol
- Reticulocytosis (usually due to HA or blood loss)
- Hypothyroidism
- Liver disease
- Drugs (eg azathioprine)

Investigations

- FBC
- Blood film
- Reticulocyte count
- Ferritin
- B12 & folate
- Bilirubin
- Direct Coombs Test
- Haemoglobin electrophoresis

Hereditary Spherocytosis

- Autosomal dominant defect in RBC cytoskeleton
- Spherical fragile cells are destroyed passing through the spleen
- Most common inherited haemolytic anaemia in northern Europeans

Presentation

- FTT
- Anaemia
- Jaundice
- Gallstones
- Splenomegaly

Haemolytic Crisis

- Triggered by infections
- Anaemia & jaundice are worsened

Aplastic Crisis

- Triggered by Parvovirus B19 infection
- No reticulocyte response to haemolysis

Diagnosis

- Family history
- Typical clinical features
- Typical lab features
  - Spherocytes on blood film
  - Raised MCHC

Management

Acute Haemolytic/Aplastic Crisis

- Supportive, transfusion may be necessary

Long Term

- Folate replacement
- Splenectomy

G6PD Deficiency

- X-linked recessive deficiency of enzyme protective against reactive oxygen species (ROS)
- More common in Mediterranean, African, & Middle Eastern patients
- RBCs are particularly vulnerable to damage from ROS
- Periods of stress/increased ROS production lead to acute haemolytic anaemia

Features

Neonatal

- Jaundice

Later

- Intermittent jaundice, particularly in response to **triggers:**
  - Infection
  - Drugs
    - Primaquine
    - Ciprofloxacin
    - Nitrofurantoin
    - Trimethoprim
    - Sulfonylureas
    - Sulfasalazine
  - Fava (broad) beans
- Anaemia
- Gallstones
- Splenomegaly

Investigations

- Heinz bodies, bite cells, blister cells on blood film
- Diagnosed by G6PD enzyme assay
  - Must be performed at least 3 months after a haemolytic crisis

Management

- Avoid triggers
- Supportive care in acute crises

Sickle Cell Disease

Pathophysiology

- Autosomal recessive condition of beta-globin gene on chromosome 11 resulting in formation of abnormal haemoglobin chain (HbS)
  - One copy (HbAS): sickle cell trait
  - Two copies (HbSS): sickle cell disease
- HbS is insoluble and crystalises (causing the cell to "sickle") when deoxygenated
  - HbAS sickles at pO2 2.5-4kPa
  - HbSS sickles at pO2 5-6kPa
- Does not present until ~ 6 months when adult haemoglobin replaces fetal haemoglobin
- Common in patients of African/Indian/Middle-Eastern/Caribbean descent as the trait is protective against malaria

Presentation/Complications

- Anaemia
- Increased infection risk
- Avascular necrosis of large joints
- Stroke
- Pulmonary hypertension
- Priapism
- Chronic kidney disease
- Crises

Diagnosis

- Haemoglobin electrophoresis
- Pregnant women at risk of being carriers are offered testing during pregnancy

General Management

- Avoid dehydration/other crisis triggers
- Up to date vaccines
  - NICE recommend polysaccharide pneumococcal vaccine every 5 years
- Antibiotic prophylaxis
  - Usually penicillin V
- Hydroxycarbamide (AKA hydroxyurea)
  - Stimulates production of HbF instead of HbS
- Transfusion for severe anaemia
- Bone marrow transplant can be curative

General Management of Crises

- Analgesia
- Hydration
- Oxygen
- Keep warm
- Antibiotics if evidence of infection
- Blood transfusion if severe
- Exchange transfusion if very severe (eg neurological complications)

Thrombotic/Vaso-Occlusive/"Painful" Crises

- Precipitated by infection/dehydration/ deoxygenation
- Sickled cells block capillaries and cause distal ischaemia/micro-infarcts

Features

- Pain
- Fever
- Features of infarcts
  - Brain
  - Lung
  - Hip (avascular necrosis)
  - Bones of hands/feet
- Priapism

Diagnosis

- Clinical

Management

- Supportive
- Aspiration of priapism

Sequestration Crises

- Sickled cells block flow and cause pooling in spleen (or lungs)

Features

- Severe anaemia
- Hypovolaemic shock
- Splenic infarction & later hyposplenism

Management

- Fluid resuscitation & blood transfusion
- Splenectomy in recurrent cases

Acute Chest Syndrome

- Triggered by infection (pneumonia/bronchiolitis) or other cause (pulmonary thrombotic crisis, embolus)
- Most common cause of death in SCD after childhood

Features/Diagnosis

- Fever/respiratory symptoms AND
- New pulmonary infiltrates on CXR

Management

- Antibiotics/antivirals for infections
- Blood transfusion
- Incentive spirometry
- Invasive ventilation may be necessary

Aplastic Crises

- Triggered by Parvovirus B19

Features

- Sudden severe anaemia
- Resolves within 1 week

Management

- Supportive with transfusions if necessary

Acute Lymphoblastic Leukaemia

- Most common (80%) childhood malignancy
- Peaks at age 2-5, boys slightly more than girls

Risk Factors

- Radiation exposure (eg x-ray during pregnancy)
- Down's/Klinefelter/Noonan Syndrome

Types

- Common ALL (75%): Pre-B phenotype (CD10 present)
- T-cell ALL (20%)
- B-cell ALL (5%)

Features

Bone Marrow Failure

- Anaemia
- Neutropenia: frequent/severe infections
- Thrombocytopenia: bleeding, petechiae, purpura

Other

- Lymphadenopathy
- Persistent fever (constitutional or concurrent infection)
- Weight loss
- Night sweats
- Bone pain
- Hepatosplenomegaly
- Testicular swelling

Investigations

Establishing Diagnosis

- FBC & blood film
  - Abnormal white cells/blast cells
- Bone marrow/lymph node biopsies

Staging

- CXR
- CT
- Lumbar puncture
- Genetic analysis & immunophenotyping abnormal cells

Management

- Primarily chemotherapy
- Supportive – blood products, allopurinol
- Infections – gentamycin & tazocin
- Bone marrow transplant, radiotherapy

Prognosis

- 85% survival in children

Poor Prognostic Factors

- Age < 2 or > 10 at diagnosis
- WBC > 20 at diagnosis
- Mature T or B cell surface markers
- Non-Caucasian
- Male

Idiopathic Thrombocytopenic Purpura

- AKA Immune Thrombocytopenia (ITP either way)
- Destruction of platelets by antibodies against glycoprotein IIb/IIIa or Ib-V-IX complex
- Spontaneous or triggered (eg by a viral infection)

Presentation

- Typically children under 10 yrs with history of recent viral illness
- Onset of symptoms over 24-48hrs:
  - Bleeding (from gums, epistaxis, menorrhagia, etc)
  - Bruising
  - Petechial/purpuric rash
- Otherwise perfectly well

Investigations

- Urgent FBC (to rule out leukaemia)
  - Isolated thrombocytopenia

Management

- Most cases require no treatment
  - Typical self-limiting course in 1-2 weeks
- Avoid contact sports
- Avoid IM injections, LPs
- Avoid NSAIDs/aspirin/other blood-thinning drugs

Active bleeding/severe thrombocytopenia (< 10)

- Prednisolone
- IVIg
- Transfusion if required
- Platelet transfusions only work temporarily as new platelets are destroyed

Complications

- Chronic ITP
- Anaemia
- Intracranial & subarachnoid haemorrhage
- GI bleeding

Neurology

Epilepsy Types

- Group of conditions characterised by recurrent unprovoked seizures
- Seizures are periods of abnormal activity in the brain

Generalised Tonic-Clonic Seizures

- Most common
- **Tonic:** Sustained contraction & stiffness
- **Clonic:** Jerking of one limb/side/whole body
- May be associated with tongue biting, incontinence, groaning, or irregular breathing
- **Post-ictal phase:** ~ 15 minute period of drowsiness, tiredness & irritability following seizure

Management

1. Valproate
2. Lamotrigine/carbamazepine

Focal Seizures

- Start in one area of the brain
- May become secondarily generalised

Temporal (most common)

- Memory flashbacks
- Déjà vu
- Automatisms
- Contralateral dystonic movements

Occipital

- Coloured bright lights spreading from on area in homonymous visual fields

Centroparietal

- Sensorimotor phenomena

Frontal

- Dystonic posturing, strange guttural noises

Management

1. Lamotrigine/carbamazepine
2. Valproate/levetiracetam

Absence Seizures

- 10-20s episodes of psychomotor arrest – "staring into space"
- Child won't be aware of surroundings or respond
- Mostly (90%) stop as the child gets older

Management

1. Ethosuximide/valproate

Myoclonic Seizures

- Sudden shock-like movements of part of body
- Patient typically remains awake
- Typically occur in Juvenile Myoclonic Epilepsy

Management

1. Valproate
2. Lamotrigine/levetiracetam/topiramate

Infantile Spasms (West Syndrome)

- Typically presents at 4-8 months
- **"Salaam" attacks:** Flexion of head, trunk, & arms followed by extension of arms
  - Lasts 1-2 seconds
  - May occur up to 50 times

Investigation

- EEG: hypsarrhythmia in 2/3
- CT/MRI: diffuse/localised brain disease in 2/3 (eg Tuberous Sclerosis)

Management

- ACTH/prednisolone & vigabatrin
- 1/3 die by 25, 1/3 seizure free

Lennox-Gastaut Syndrome

- Onset 1-5 years
- 50% have history of infantile spasms
- 90% have moderate-severe mental handicap
- **Drop attacks:** atonic seizures (brief lapses in muscle tone)
  - Last up to 3 minutes

Investigations

- EEG: slow spike waves

Management

- Poor prognosis
- Ketogenic diet may help

Status Epilepticus

- Seizure lasting more than 5 minutes OR
- 2 seizures within 5 minutes without returning to normal between them

Community Management

- Buccal midazolam/rectal diazepam

Hospital Management

1. Secure airway
2. High-flow oxygen
3. Assess breathing & circulation
4. Check blood glucose
5. IV access
6. IV lorazepam
   - Repeat after 10 minutes if persistent
7. Infusion of phenytoin/phenobarbitone if still persistent
8. Consider GA

Epilepsy Investigations

- Patients with clear febrile convulsion or syncope with no indications for investigations do not need investigating

EEG

- Used to classify epilepsy
- EEG may be normal between seizures & children without epilepsy/seizures may show epileptiform features on EEG

Indications

- After second afebrile seizure
- After first complex febrile seizure
- Monitoring efficacy of treatment
- Acute encephalopathy of unknown origin
- Unexplained deterioration of cognitive performance
- When a new epilepsy syndrome type becomes apparent

Imaging (MRI)

Indications

- First seizure < 2 years
- Focal seizures
- Epilepsy resistant to treatment

Others

ECG

- Exclude cardiac problems
- Long QT syndrome & associated syncope may mimic seizure

Bloods

- Electrolytes
- Glucose
- Cultures if sepsis/meningitis/encephalitis suspected
  - + urine cultures, LP

Side Effects of Anti-Epileptic Drugs

Valproate

- Teratogenic
  - Associated with NTDs & neurodevelopmental delay
- Liver damage & hepatitis
- Hair loss
- Tremor
- P450 inhibition

Carbamazepine

- Agranulocytosis
- Aplastic anaemia
- Dizziness/ataxia
- SJS
- SIADH
- P450 induction

Phenytoin

- Folate & vitamin D deficiency
- Aplastic anaemia
- Neurological (confusion, ataxia, peripheral neuropathy)
- Hepatitis
- Gingival hyperplasia
- Teratogenic

Ethosuximide

- Night terrors
- Rashes

Lamotrigine

- SJS/DRESS
- Leukopenia

Febrile Seizures

- Seizures provoked by fever in an otherwise healthy child
- Occur between 6 months and 5 years old in the context of a high fever (≥ 39°)

Classification

Simple	Complex
< 15 minutes	15+ minutes
Generalised seizure	Focal seizure
No recurrence	Occur multiple times in the same febrile illness
Complete recovery within 1 hour	Delayed recovery

Differentials to be excluded

- Epilepsy
- Meningitis/encephalitis
- Space occupying lesion
- Syncopal episode
- Electrolyte abnormalities
- Trauma/NAI

Management

- Identify & manage infection source
- Simple analgesia/antipyresis
- Complex febrile seizures warrant referral and investigation

Prognosis

- 1/3 will have another seizure in a further febrile illness
- 2.5% with no risk factors will develop epilepsy
- 50% with 3 risk factors will develop epilepsy
  - Family history of epilepsy
  - Complex febrile seizure
  - Background of neurodevelopmental disorder

Breath Holding Spells

- Involuntary episodes in response to upsetting/frightening stimulus
- Typically occur between 6 & 18 months and resolve by 4/5 years

Cyanotic Breath Holding Spells

- Child is upset or crying
- Stop breathing after long cry/expiratoin, become cyanotic & lose consciousness
- Regain consciousness & restart breathing within ~1 minute
- May be tired/lethargic following episode

Reflex Anoxic Seizures

- Syncope in response to noxious stimulus & strong vagal response
- Child loses consciousness, heart stops beating, & may have seizure-like activity
- Rapid recovery after ~30 minutes

Management

- Exclude other pathology & reassure
- Link to iron deficiency anaemia – identify & treat if present

Syncope

- Vasovagal attacks common in children – vagal overstimulation & parasympathetic overactivity causing hypotension and brain hypoperfusion

Causes

Primary (Simple Fainting)

- Dehydration
- Prolonged standing
- Warm environment
- Missed meals
- "Vasovagal stimuli"
  - Fright
  - Pain
  - Sight of blood

Secondary

- Hypoglycaemia, anaemia
- Infection, anaphylaxis
- Arrhythmias, valvular heart disease, HOCM

Differentiation from Seizure

Syncope	Seizure
Prolonged upright position before the event Lightheaded before the event Sweating before the event Blurring or clouding of vision before the event Reduced tone during the episode Return of consciousness shortly after falling No prolonged post-ictal period	Epilepsy aura (smells, tastes or deja vu) before the event Head turning or abnormal limb positions Tonic clonic activity Tongue biting Cyanosis Lasts more than 5 minutes Prolonged post-ictal period

Investigations

- ECG (particular attention to QT interval)
  - 24hr if paroxysmal arrhythmia suspected
- Echo
- FBC, electrolytes, glucose

Management

- Rule out/refer seizures/underlying pathology
- Avoid dehydration, missing meals, prolonged standing
- Sit down & eat/drink in response to prodromal symptoms

Cerebral Palsy

- Permanent neurological deficits caused by damage to brain at/around birth

Causes

Antenatal

- Maternal infections
- Trauma

Perinatal

- Birth asphyxia
- Pre-term birth

Postnatal

- Meningitis
- Severe jaundice
- Head injury

Types

- **Spastic/pyramidal:** Hypertonia caused by damage to UMNs
- **Dyskinetic/athetoid/extrapyramidal:** Problems controlling tone, hyper & hypotonia, athetoid movements caused by basal ganglia damage
- **Ataxic:** Problems with coordination caused by damage to cerebellum
- **Mixed**
- Can result in monoplegia, hemiplegia, diplegia or quadriplegia

Presentation

- Failure to meet milestones
- Increased/decreased tone in specific limbs
- Hand preference before 18 months
- Problems with coordination, speech, walking, feeding, swallowing, learning difficulties

Neurological Exam

- Gait
  - Hemi/diplegic (extended legs swung in wide arc, UMN)
  - Broad-based/ataxic (cerebellar)
- **UMN lesion:** Increased tone, brisk reflexes, reduced power
- **Extrapyramidal lesion:** Athetoid movements
- **Cerebellar lesion:** Ataxia

Management

MDT approach

- Physio, OT, SALT, dietetics
- Paediatrics, GP
- Orthopaedics (tenotomies)

Medical

- Muscle relaxants (baclofen, diazepam, botox) for spasticity
- Anti-epileptic drugs for seizures
- Glycopyrronium bromide for excessive drooling

Headache

Headache Red Flags

- Persistent/recurrent vomiting
- Persistent/recurrent vomiting
- Balance/coordination/focal neurological issues
- Abnormal eye movements
- Behavioural changes
- Seizures
- Increasing head circumference in younger children
- Delayed/arrested puberty

Migraine

- Most common primary cause of headache in children
- May have a history of abdominal migraine features

Types

- Migraine with aura
- Migraine without aura
- Abdominal migraine
- Silent migraine
- Hemiplegic migraine

IHS Diagnosis of Paediatric Migraine Without Aura

- > 5 attacks fulfilling each of:
  - Headache lasts 4-72hrs
  - Headache fulfils at least 2 of:
    - Bilateral or unilateral (frontal/temporal)
    - Pulsating
    - Moderate-severe intensity
    - Aggravated by routine physical activity
  - Headache is accompanied by at least 1 of:
    - Nausea/vomiting
    - Photophobia & phonophobia

Aura

- May or may not be present
- Typically visual & progressive lasting 5-60 minutes
- Transient hemianopic disturbance or spreading scintillating scotoma

Acute Management

- Ibuprofen & paracetamol
- Sumatriptan nasal spray (other triptans unlicensed in children)

Prophylaxis

- Propranolol (avoid in asthma)
- Pizotifen
- Topiramate (teratogenic, contraceptive precautions in girls)

Tension-Type Headache

- 2nd most common cause of primary headache in children

IHS Diagnosis of TTP in Children

- At least 10 episodes fulfilling each of:
  - Headache lasts 30 minutes – 7 days
  - Fulfils at least 2 of:
    - Pressing/tightening non-pulsating quality
    - Mild-moderate intensity
    - Bilateral location
    - Not aggravated by physical activity
  - Fulfils both of:
    - No nausea/vomiting
    - Photophobia and/or phonophobia

Triggers

- Stress/fear/discomfort
- Skipping meals
- Dehydration
- Infection

Management

- Simple analgesia, rest, reassurance

Raised ICP

Features

- Short history of vomiting, morning headache & visual disturbance
- Papilloedema
- Bulging fontanelle in young children
- May be signs of infection

Investigation

- Neuroimaging to assess possible CSF obstruction

Idiopathic Intracranial Hypertension

- Papilloedema ± visual symptoms with normal cranial imaging
- Female sex & obesity are risk factors
- Diagnosed by raised LP opening pressure (>20cmH2O)

Other Causes

Primary

- Cluster headache
- Medication overuses headache

Secondary

- Infection
  - Sinusitis/URTI
  - Meningitis
- Brain tumours
- Trauma
- Vascular causes
- Psychiatric/functional
- Cranial neuralgias

Hydrocephalus

- Imbalance of CSF production and absorption causing build-up in the ventricular system of brain and spinal cord

Congenital Causes

Non-Communicating

- Aqueductal stenosis (most common cause)
- Arnold-Chairi malformation
- Intraventricular haemorrhage
- Posterior fossa tumours

Communicating

- Arachnoid cysts
- Meningitis

Presentation

Before closure of sutures (~2 years)

- OFC crossing centiles
- Bulging fontanelle
- Poor feeding
- Poor tone
- Sleepiness
- "Sunsetting" of eyes

Older children

- Headache
- Vomiting
- Sleepiness
- Hyperreflexia/spasticity
- Papilloedema
- "Sunsetting" of eyes

Investigations

- Cranial imaging to look for enlarged ventricles & underlying cause

Ventriculoperitoneal Shunt

- Mainstay of treatment for hydrocephalus

Complications

- Infection
- Blockage
- Excessive drainage
- IVH
- Outgrowing the shunt
  - Typically needs replacement every 2 years while growing
  - Coiling the shunt in the abdomen may help

Macrocephaly

- OFC > 99.6th centile
- Majority are benign and familial but may be caused by hydrocephalus/degenerative disorder

Causes

Hydrocephalus

- Communicating
  - Achondroplasia, meningitis, choroid plexus papilloma
- Non-communitcating
  - Aqueductal stenosis, Chiari malformation, toxoplasmosis, tumours/abscess/vein of Galen aneurysm
- Hydranencephaly
  - Holoprosencephaly, prosencephaly, massive hydrocephalus

Large Brain

- Benign familial macrocephaly
- Genetic
  - Noonan syndrome, Fragile X syndrome, neurocutaneous syndromes
- Metabolic
  - Lysosomal storage disorders, organic acidurias, leukodystrophies

Large Cranium

- Increased skull bone
  - Anaemia, thalassaemia, SCD, rickets, osteoporosis, osteogenesis imperfecta
- Apparently large head
  - IUGR, skeletal dysplasias

Assessment

- Full history including development
- Static or progressive?
- Signs of raised ICP?
  - Bulging fontanelle, sunset gaze, dilated scalp veins
  - Cushing's triad warrants urgent neurosurgical involvement
- Skin signs of neurofibromatosis
- Features of autism or neurodegenerative disorders
- Plot OFC on growth chart

Investigation/Management

- If OFC is crossing centiles, brain imaging to look for hydrocephalus/other intracranial causes
- Compare to parents OFC centiles
  - If similar, likely familial
  - If larger but no abnormal findings or crossing of centiles, likely benign
    - Follow measurements & review
- Further specific investigation/referral depending on specific history/exam findings

Microcephaly

- OFC < 0.4th centile
- Majority have coexisting developmental/neurological abnormalities

Causes

Congenital

- TORCH/Zika infection
- Toxin mediated (maternal alcohol, cocaine, PKU)
- Primary brain malformations (lissencephaly, hollow prosencephaly)
- Genetic syndrome (Cri-du-chat, Edward's, Patau)

Acquired

- HIE/Stroke
- Meningoencephalitis
- Trauma/NAI
- Metabolic (glucose transporter deficiency)
- Genetic (Rett's syndrome)
- Craniosynostosis

Consequences

- Cerebral palsy
- Developmental delay
- Epilepsy
- Cortical visual difficulties
- Feeding difficulties
- Behavioural issues

Assessment

- Full history including development, antenatal history, & infection during pregnancy
- Enquire about newborn screening (PKU)
- Plot OFC on growth chart
- Look for features of craniosynostosis

Investigation/Management

- Repeat PKU screening
- Karyotype, plasma lactate, TORCH screen, urine for CMV, plasma & urine for amino acids & organic acids
- MRI

Craniosynostosis

- Premature fusion of skull sutures resulting in raised ICP

Types

Type of Synostosis	Affected Suture	Head Shape
Saggital Synostosis	Saggital suture	Long and narrow from front to back
Coronal Synostosis	Coronal suture	Bulging on one side of the forehead
Metopic Synostosis	Metopic suture	Pointy triangular forehead
Lambdoid Synostosis	Lambdoid suture	Flattening on one side of the occiput

Presentation

- Abnormal head shape as above
- Anterior fontanelle closure before 1yr

If Untreated

- Developmental delay
- Cognitive impairment
- Vomiting
- Irritability
- Visual impairment
- Seizures

Investigations

- Specialist referral
- Skull X-Ray first line
- CT head to confirm diagnosis/if there is doubt

Management

- Mild cases can be monitored
- More severe cases need surgical reconstruction of the skull

Plagiocephaly/Brachycephaly

- Plagiocephaly refers to flattening of one side of the head
- Brachycephaly refers to shortening of the head by flattening of the back of the head
- Occur due to tendency to rest head on a particular side/point (positional plagiocephaly)
  - Brachycephaly is more common since campaigns to have babies sleeping on their backs to avoid SIDS
- Typically presents at 3-6 months

Management

- Exclude craniosynostosis by history and palpation
- Look for congenital muscular torticullus (CMT)
  - Requires referral to physiotherapy
- Reassurance & conservative measures:
  - Position on rounded side for sleep
  - Supervised tummy time
  - Using rolled towels etc to prop up head
  - Minimising time in pushchairs and car seats
- Plagiocephaly helmets may help but are not routinely used

Muscular Dystrophy

- Genetic conditions causing gradual wasting of muscles

Gower's Sign

- Specific way of standing up in children with proximal weakness
- Hands and knees → straightened knees with hips in air (downward dog yoga pose) → weight shifted backwards with hands on knees → hands used to "walk" up legs until upright

General Management

- No cure
- OT, physio, wheelchairs, braces
- Surgical & medical management of complications such as scoliosis, heart failure

Duchenne Muscular Dystrophy

- X-linked recessive defect of dystrophin gene (muscle cell structural protein)

Presentation

- 3-5 years
- Progressive proximal weakness
- Gower's sign
- Calf pseudohypertrophy
- Associated with dilated cardiomyopathy
- 30% have intellectual impairment

Prognosis

- Progressive
- Wheelchair by teenage years
- Life expectancy 25-35yrs due to cardiac/respiratory failure

Specific Management

- Oral steroids can slow progression by up to 2 years
- Creatine can help muscle strength

Becker Muscular Dystrophy

- Milder defects of dystrophin gene
- Presents at 8-12 years
- Patients may require wheelchair by late 20s/30s

Myotonic Dystrophy

- Typically presents in adulthood with:
  - Progressive muscle weakness
  - Prolonged muscle contractions
  - Cataracts
  - Cardiac arrhythmias

Others

- Facioscapulohumeral Muscular Dystrophy
- Oculopharyngeal Muscular Dystrophy
- Limb-girdle Muscular Dystrophy
- Emery-Dreifuss Muscualr Dystrophy

Spinal Muscular Atrophy

- Rare autosomal recessive condition
- Loss of spinal lower moto neurons
  - **LMN signs:**
    - Fasciculations
    - Reduced muscle bulk
    - Reduced tone
    - Reduced power
    - Reduced/absent reflexes

Categories

SMA Type 1

- Onset in first few months and death by 2 years

SMA Type 2

- Onset in first 18 months
- Most never walk but survive into adulthood

SMA Type 3

- Onset after first year
- Most walk unassisted but later lose that ability
- Life expectancy close to normal

SMA Type 4

- Onset in 20s
- Retain ability to walk short distances but need wheelchairs for mobility
- Everyday tasks lead to significant fatigue

Management

- MDT input
- Physio & splints, braces, wheelchairs
- Non-invasive ventilation
- SMA Type 1 may require tracheostomy & mechanical ventilation
- PEG feeding if swallowing muscles are affected

Neural Tube Defects

- Failure of neural tube closure in week 4 of gestation
- Incidence reducing due to antenatal recognition & termination & periconceptual folate therapy

Anencephaly

- Absence of skull bones, forebrain, upper brainstem
- Invariably lethal

Encephalocele

- Midline skull defect with brain tissue herniation
- Lesion covered in skin
- Requires surgical excision & closure
- Associated with poor neurodevelopment

Spina Bifida

- Failure of midline fusion of dorsal vertebral bodies

Spina Bifida Occulta

- Ne herniation of neural tissue
- Overlying dermal sinus/dimple/hairy naevus

Meningocele

- Herniation of meninges & fluid only with skin covering
- Requires surgical repair
- Excellent prognosis

Myelomeningocele

- Herniation of spinal neural tissue
  - May be covered by skin/meninges or open
- Adjacent spinal cord always abnormal
- Thoraco-lumbar, thoracic, or thoraco-sacral
- **Problems**
  - Flaccid paralysis below lesion
  - Faecal & urinary incontinence
  - Urinary tract dilatation
  - Bulbar paresis (secondary to Chiari malformation)
  - Vertebral abnormalities
- **Management**
  - Surgical closure & hydrocephalus drainage
  - Prognosis related to size & location
  - Palliative care may be appropriate

Infectious Disease

Sepsis

- Systemic inflammatory response caused by infection resulting in hypotension, systemic hypoperfusion, tissue anaerobic respiration, and organ failure (septic shock)

Causative Organisms

< 3 months

- GBS
- E. coli
- L. monocytogenes

> 3 months

- N. meningitidis
- S. pneumoniae
- S. aureus
- E. coli

Presentation

- Seriously unwell appearance
- Deranged vital signs
- Prolonged capillary refill time
- Fever or hypothermia
- Deranged behaviour
- Poor feeding
- Inconsolable/high-pitched crying
- Weak cry
- Reduced consciousness
- Reduced tone
- Skin colour changes (cyanotic/pale/mottled/ashen)
- Signs of organ hypoperfusion

Immediate (Emergency) Management

- **Oxygen** if signs of shock or SpO2 < 94%
- **IV access**
- **Bloods**
  - FBC, CRP, U+E,
  - Clotting screen
  - Blood gas (lactate & acidosis)
  - Cultures (ideally before antibiotics)
- **IV Antibiotics**
  - As per local guidelines
- **IV Fluids**
  - 20ml/kg bolus if lactate > 2mmol/L or signs of shock
  - Can be repeated
- Monitor **urine output**

Further Investigations (depending on suspicions)

- CXR (pneumonia)
- Abdominal & pelvic US (intra-abdominal infection)
- Lumbar puncture (meningitis)
  - Unless contraindicated
- Meningococcal PCR (meningococcaemia)
- Serum cortisol (adrenal crisis)

NICE Feverish Illness Under 5 Risk Assessment

	Green – low risk	Amber – intermediate risk	Red – high risk
Colour (of skin, lips or tongue)	• Normal colour	• Pallor reported by parent/carer	• Pale/mottled/ashen/ blue
Activity	• Responds normally to social cues • Content/smiles • Stays awake or awakens quickly • Strong normal cry/not crying	• Not responding normally to social cues • No smile • Wakes only with prolonged stimulation • Decreased activity	• No response to social cues • Appears ill to a healthcare professional • Does not wake or if roused does not stay awake • Weak, high-pitched or continuous cry
Respiratory		• Nasal flaring • Tachypnoea: – RR >50 breaths/ minute, age 6–12 months – RR >40 breaths/ minute, age >12 months • Oxygen saturation ≤95% in air • Crackles in the chest	• Grunting • Tachypnoea: RR >60 breaths/minute • Moderate or severe chest indrawing
Circulation and hydration	• Normal skin and eyes • Moist mucous membranes	• Tachycardia: – >160 beats/minute, age <12 months – >150 beats/minute, age 12–24 months – >140 beats/minute, age 2–5 years • CRT ≥3 seconds • Dry mucous membranes • Poor feeding in infants • Reduced urine output	• Reduced skin turgor
Other	• None of the amber or red symptoms or signs	• Age 3–6 months, temperature ≥39°C • Fever for ≥5 days • Rigors • Swelling of a limb or joint • Non-weight bearing limb/not using an extremity	• Age <3 months, temperature ≥38°C* • Non-blanching rash • Bulging fontanelle • Neck stiffness • Status epilepticus • Focal neurological signs • Focal seizures

Green

- Manage at home with appropriate care & advice

Amber

- Provide information & safety net or refer to paediatric specialist

Red

- Refer urgently to paediatric specialist

Meningitis

- Inflammation of the meninges due to infection

Organisms

< 3 Months

- GBS
- E. coli
- L. monocytogenes

< 6 Years

- N. meningitidis
- S. pneumoniae
- H. influenzae

> 6 Years

- N. meningitis
- S. pneumoniae

Viral

- Typically less severe than bacterial

Presentation

- Fever
- Nick stiffness
- Vomiting
- Headache
- Photophobia
- Altered consciousness
- Seizures
- Non-blanching petechial rash (meningococcal septicaemia & DIC)
- Kernig's/Brudzinski's tests positive

Non-specific features in neonates/young babies

- Hypotonia, lethargy
- Poor feeding
- Hypothermia
- Bulging fontanelle

Lumbar Puncture

Contraindications

- Focal neurological signs
- Papilloedema
- Significant bulging of fontanelle
- DIC
- Signs of cerebral herniation
- GCS < 9
- Haemodynamically unstable
- Suspected meningococcal septicaemia
  - Do blood cultures & meningococcus PCR

Results

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

Management of Bacterial Meningitis

Community

- Suspected meningitis + non blanching rash:
  - Immediate IM benzylpenicillin & transfer to hospital
  - Transfer is priority if any issue with antibiotics (eg allergy)

Hospital

- Ideally CSF sample should be taken before antibiotics but should not delay them if urgent
- Bloods should be sent for meningococcal PCR

6. Typical **antibiotics:**
   - **< 3 months:** IV cefotaxime + amoxicillin
   - **> 3 months:** IV ceftriaxone
   - Vancomycin added if risk of resistant pneumococcal infection
7. **Steroids**
   - Not in children < 3 months
   - Dexamethasone 4 times daily for 4 days if CSF suggests bacterial meningitis
8. **Fluids**
   - Resuscitate if shocked
9. **Cerebral Monitoring**
   - Assisted/invasive ventilation/ICU may be required
10. **Post-exposure prophylaxis**
    - Inform public health
    - Single dose of ciprofloxacin for those with close contact in last 7 days

Management of Viral Meningitis

- Viral PCR testing of CSF sample
  - HSV, VZV, enterovirus
- Typically require only supportive treatment
- Acyclovir can be used for suspected/confirms HSV/VZV infection

Complications

- Hearing loss
- Seizures & epilepsy
- Cognitive impairment & learning disability
- Memory loss
- Cerebral palsy with focal deficits

Encephalitis

Causes

- HSV
  - HSV-2 in neonates
  - HSV-1 in older children
- VZV (associated with chickenpox)
- CMV (associated with immunodeficiency)
- EBV
- Enterovirus
- Adenovirus
- Influenza virus
- Polio, mumps, measles, rubella (if unvaccinated)

Features

- Altered consciousness/cognition
- Unusual behaviours
- Acute onset focal neurology
- Acute onset focal seizures
- Fever

Investigation

- LP for viral PCR
  - CT if contraindicated
- MRI scan to visualise brain in detail
- EEG can be useful if ambiguous/uncertain
- Throat/vesicle swabs
- HIV testing

Management

- **IV Antivirals**
  - Acyclovir for HSV/VZV
  - Ganciclovir for CMV
- Acyclovir typically started empirically
- Repeat LP before stopping antivirals
- Follow-up & rehab

Complications

- Lasting fatigue/prolonged recovery
- Changes to personality/mood
- Changes to memory/cognition
- Learning disability
- Headaches
- Chronic pain
- Sensorimotor disturbance
- Seizures
- Hormonal imbalance

Mumps

- Viral infection spread by respiratory droplets
- RNA paramyxovirus
- 14-25 days incubation, symptoms last 1 week

Presentation

- Prodrome:
  - Fever
  - Muscle aches
  - Lethargy
  - Reduced appetite
  - Headache
  - Dry mouth
- **Parotitis**
  - Painful
  - Unilateral → bilateral
- Abdominal pain (pancreatitis)
- Testicular pain (orchitis)
- Confusion/neck pain etc (meningitis/encephalitis)

Management

- Diagnosed by PCR testing of saliva swab
  - Can also be tested for antibodies
- Notifiable disease
- Supportive management
  - Rest
  - Fluids
  - Analgesia

Complications

- Pancreatitis
- Orchitis
- Meningitis
- SNHL

Infections Characterised by Rash

Measles/Rubeola

- 6-19 day incubation
- Infectious until 4 days after rash appears

Features

- Prodromal (2-4 days) high fever, URTI, conjunctivitis
- Maculopapular rash spreading from face
- Koplik spots rare but pathognomonic

Management

- Supportive
- MMR is protective
- Notifiable

Scarlet Fever

- Group A strep
- 2-5 day incubation

Features

- Fever lasting 24-48hrs
- Malaise, headache, nausea/vomiting
- Sore throat
- Lymphadenopathy
- Strawberry tongue
- Rash
  - Fine punctate erythema (sandpaper rash)
  - Appears on torso first, spares palms and soles
  - Flushing with circumoral pallor
  - Late desquamation

Diagnosis

- Throat swab (don't wait for results)

Management

- Oral penicillin V for 10 days
- IV penicillin G for invasive disease
- Notifiable disease
- Return to school 24hrs after starting antibiotics

Complications

- Otitis media
- Rheumatic fever
- Acute glomerulonephritis
- Invasive infection

Parvovirus B19/Fifth Disease/Erythema Infectiosum

- 4-7 days incubation
- Isolation not required

Features

- 3-7 day prodrome
- Rash for 1-4 days
  - Slapped cheek appearance
- Evanescent rash for 1-3wks followed by arthropathy
- Congential: red cell aplasia & hydrops

Management

- Supportive
- Fetal transfusion/IVIg in neonatal severe disease

Sixth Disease/Roseola Infantum/Exanthem Subitum

- HHV-6
- 10 day incubation

Features

- Temperature for ~3 days, improves with appearance of rash
- Maculopapular rash

Management

- Supportive

Rubella/German Measles

- 14-21 day incubation
- Infective from 7 days before until 7 days after rash

Features

- Postauricular & posterior cervical lymphadenopathy
- Maculopapular rash rapidly spreading from face & fading in 3-5 days

Management

- Supportive
- Prevented by MMR vaccine
- Screening in pregnancy

Lyme Disease

- Borrelia burgdorferi transmitted by Ixodes tick bite
- 3-20 day incubation

Features

- Erythema migrans
  - Painless, non-pruritic, circular
  - Spreads outward from bite with central clearing
- Early localised disease
  - EM
  - Fever
  - Headache
  - Myalgia
  - Arthralgia
  - Lymphadenopathy
- Early disseminated disease
  - Multiple EM lesions
  - Facial nerve palsy
  - Meningits
  - Papilloedema
  - Myocarditis
- Late disease
  - Large joint arthritis
  - Peripheral neuropathy
  - CNS manifestations

Management

- **< 8 years:** Amoxicllin (azithromycin if allergic)
- **> 8 years:** Doxycycline
- **Cardiac/CNS:** Ceftriaxone

Infections Characterised by Rash Ctd

Chickenpox

- Varicella Zoster Virus primary infection
- 1-6 years
- Spread by respiratory droplets/vesicle contact
- 10-21 day incubation
- Infective until all vesicles are crusted

Features

- Rash
  - Starts at head & trunk
  - Red macules to popular, vesicular, pustular, & crusting stages
  - Itchy
- Fever
  - High-grade fever uncommon & may indicate secondary bacterial infection
- Headache
- Anorexia
- Signs of URTI

Management

- Supportive
- VZIG used in exposed at-risk individuals
- Acyclovir used in encephalitis, pneumonia, neonatal infection, & immunocompromised patients

Complications

- Bacterial infection of lesions
  - Cellulitis
  - Necrotising fasciitis
  - Toxic shock syndrome
- Pneumonitis
- Encephalitis

Infectious Mononucleosis

- EBV
- 4-6 week incubation
- Infects B-lymphocytes

Features

- Flu-like illness
- Lethargy
- Exudative pharyngitis
- Lymphadenopathy
- Hepatosplenomegaly
- Generalised maculopapular rash
  - Especially if taking an aminopenicillin

Diagnosis

- Atypical lymphocytes & low platelets
- Hepatitis
- Paul Bunnell/Monospot test

Management

- Supportive
- Avoid contact sport if splenomegaly present

Complications

- Splenic rupture
- Glomerulonephritis
- Haemolytic anaemia
- Thrombocytopenia
- Chronic fatigue
- Meningoencephalitis
- Increased risk of certain cancers
  - Burkitt's lymphoma

Immunisations

2 Months

- 6-in-1 Vaccine
  - Diphtheria
  - Tetanus
  - Pertussis
  - Hib
  - Polio
  - Hepatitis B
- PCV (pneumococcal conjugate vaccine)
- MenB Vaccine
- Rotavirus Oral Vaccine

4 Months

- 6-in-1
- MenB Vaccine
- Rotavirus Oral Vaccine

6 Months

- 6-in-1
- PCV
- MenC Vaccine

12 Months

- MMR
- MenB

13 Months

- Hib/MenC
- PCV

Rheumatology

Juvenile Idiopathic Arthritis

- Diagnosed where there is arthritis with no other cause lasting longer than 6 weeks in a child younger than 16

Systemic JIA (Still's Disease)

Features

- Salmon-pink rash
- High swinging fevers
- Lymphadenopathy
- Anorexia & weight loss
- Arthritis
- Uveitis
- Splenomegaly
- Muscular pain
- Pleuritis & pericarditis

Investigations

- ANA & RF typically negative
- Elevated CRP, ESR, platelets & ferritin

Macrophage Activation Syndrome

- Complication of systemic JIA
- Massive immune system activation & inflammatory response
- Acutely unwell child with:
  - DIC
  - Anaemia
  - Thrombocytopenia
  - Bleeding
  - Non-blanching rash
  - Low ESR

Polyarticular JIA

- Idiopathic inflammatory arthritis in 5 joints or more
- Symmetrical, affecting small & large joints

Other Features

- Mild fever
- Anaemia
- Reduced growth

Link to Rheumatoid Arthritis

- Equivalent of RA in children
- Most children are seronegative for RF
- Seropositive children tend to be older and have similar course to RA in adults

Oligoarticular/Pauciarticular JIA

- Affects less than 4 joints (typically one large joint – monoarthritis)
- More common in girls under 6

Other Features

- Classically associated with anterior uveitis/iridocyclitis and needs ophthalmology review (even if asymptomatic)

Investigations

- ANA often positive, RF usually negative

Enthesitis-Associated Arthritis

- Childhood equivalent of seronegative spondyloarthropathies
- Strong association with HLA-B27

Features

- Inflammatory arthritis
- Enthesitis
- Associations with:
  - IBD
  - Psoriasis
  - Anterior uveitis
    - Needs ophthalmology review even if asymptomatic

Juvenile Psoriatic Arthritis

- Seronegative inflammatory arthritis associated with psoriasis

Joint Features

- Symmetrical small joint arthritis OR
- Asymmetrical large joint arthritis of lower limbs

Psoriasis Features

- Skin plaques
- Nail pitting
- Onycholysis
- Dactylitis
- Enthesitis

Management of Juvenile Idiopathic Arthritis

- Coordinated by paediatric rheumatology specialist with input from specialist MDT
- Medical treatment depends on severity & response:
  - NSAIDs
  - Steroids
  - DMARDs
  - Biologics

Ehlers-Danlos Syndrome

- Group of genetic defects in collagen causing joint hypermobility and connective tissue abnormalities

Types

Hypermobile

- Most common & least severe
- Joint hypermobility & stretchy skin
- No single gene/mode of inheritance identified

Classical

- Markedly stretchy skin with velvety feel
- Severe joint hypermobility & joint pain
- Impaired wound healing
- Prone to hernias, prolapses, mitral regurgitation, aortic root dilatation
- Autosomal dominant inheritance

Vascular

- Most dangerous
- Thin, translucent skin
- Skin, organs, & blood vessels prone to rupture
- Patients monitored for vascular abnormalites and told to seek urgent medical attention in response to any sudden pain/bleeding
- Autosomal dominant inheritance

Kyphoscoliotic

- Poor tone as a neonate/infant
- Kyphoscoliosis as they grow
- Patients tend to be tall & thin
- Risk of rupture in medium-sized arteries
- Autosomal dominant inheritance

Multi-System Features

- Easy bruising
- Poor wound healing
- Headaches
- Dizziness & syncope
- GORD
- Abdominal pain, IBS
- Menorrhagia
- Urinary incontinence
- Pelvic organ prolapse
- TMJ dysfunction

Beighton Score for Hypermobility

- Both hands flat on floor with knees straight (+1)
- Elbows hyperextend (+1 each)
- Knees hyperextend (+1 each)
- Thumb can bend to touch forearm (+1 each)
- Little finger can extend past 90° (+1 each)

Management

- Diagnose with Beighton score & rule out Marfan's
- Multiple disciplinary management of joint pain & complications

Marfan Syndrome

- Autosomal dominant defect in fibrillin gene resulting in abnormal connective tissue

Features

- Tall stature
- Long neck
- Long limbs
- Arachnodactyly
- High arched palate
- Hypermobility
- Pectus carinatum/excavatum
- Downward sloping palpebral fissures

Associated Conditions

- Lens dislocation
- Joint dislocation/pain
- Scoliosis
- Pneumothorax
- GORD
- Mitral valve prolapse & regurgitation
- Aortic valve prolapse & regurgitation
- Aortic aneurysms

Management

- Physiotherapy
- Genetic counselling
- Prevention of cardiac complications
  - Beta-blockers
  - ACE inhibitors
  - Counselling re risk of pregnancy
- Surgical correction of cardiac complications

Henoch-Schonlein Purpura (HSP)

- IgA vasculitis of the skin, kidneys and GI tract affecting children typically under the age of 10 following an URTI/gastroenteritis

Features

Purpura (100%)

- Palpable purple rash
- Localised oedema
- Start on legs & spread to buttocks, may involve upper limbs/trunk
- Ulceration & necrosis can occur in severe cases

Joint Pain (75%)

- Polyarthritis affecting knees/ankles

Abdominal Pain (50%)

- Involvement of GI mucosa
- Severe cases can cause GI haemorrhage, intussusception, or bowel infarction

Renal Involvement (50%)

- IgA nephropathy

Differentials

- Meningococcal septicaemia
- Leukaemia
- HUS
- ITP

Investigation

- FBC & blood film
- U+E
- Serum albumin
- CRP, blood cultures
- Urine dipstick & PCR
- Blood pressure

Diagnostic Criteria (EULAR/PRINTO/PRES)

- Palpable purpura + at least one of:
  - Diffuse abdominal pain
  - Arthritis/arthralgia
  - IgA deposits on renal biopsy histology
  - Proteinuria/haematuria

Management

- Supportive
- Steroids
  - Use debated
  - Specialist prescription in severe GI/renal involvement
- Monitoring
  - Urine dipstick (proteinuria)
  - Blood pressure

Prognosis

- No renal involvement: recovery in 4-6wks
- 1/3 will have recurrence in ~ 6 months
- Small proportional will develop ESRF

Kawasaki Disease

- AKA mucocutaneous lymph node syndrome
- Systemic medium vessel vasculitis typically affecting children younger than 5

Features

- Persistent high fever (> 39°) lasting > 5 days
- Unwell child
- Widespread erythematous maculopapular rash with
- Erythematous palms & soles with later desquamation between fingers & toes
- Strawberry tongue
- Cracked lips
- Bilateral conjunctivitis
- Cervical lymphadenopathy

Disease Course

Acute Phase

- Lasts 1-2 weeks
- Child is most unwell with fever & rash

Subacute Phase

- Lasts 2-4 weeks
- Acute symptoms settle
- Desquamation & arthralgia
- Risk of coronary artery aneurysms

Convalescent Phase

- Lasts 2-4 weeks
- Remaining symptoms fade
- Blood tests return to normal
- Coronary artery aneurysms may regress

Investigations

- **FBC** can show anaemia, leukocytosis, thrombocytosis
- **LFTs** can show hypoalbuminaemia & raised enzymes
- **Inflammatory markers** raised (especially ESR)
- **Urinalysis** can show white cells without infection
- **Echocardiogram** can demonstrate coronary artery aneurysms

Management

- High-dose aspirin
  - To reduce thrombosis risk
  - One of the few scenarios when aspirin can be prescribed to children despite the risk of Reye's syndrome
- IV immunoglobulins
  - Reduce aneurysm risk

Monitoring

- Close follow-up with echocardiograms

Rheumatic Fever

- Multi-system autoimmune disorder triggered by recent (2-6wks ago) Streptococcus pyogenes infection

Pathophysiology

- Immune response to streptococcal M protein results in type II hypersensitivity to similarly structured self-antigens (molecular mimicry)
- Antibodies to M protein target myosin & vascular smooth muscle
- **Aschoff bodies** are granulomatous nodules found in rheumatic heart fever

Presentation

Joint Involvement

- Migratory arthritis of large joints
- Hot, swollen, painful joints

Heart Involvement

- Carditis (pericarditis + myocarditis + endocarditis):
  - Tachy/bradycardia
  - Murmurs of valvular disease (typically mitral)
  - Pericardial rub
  - Heart failure

Skin Involvement

- Subcutaneous nodules
  - Firm painless nodules over extensor surfaces
- Erythema marginatum
  - Pink rings of varying sizes on torso & proximal limbs

Nervous System Involvement

- Sydenham Chorea/St Vitus' Dance
  - Irregular, uncontrolled, rapid movement of the limbs

Investigation

- Throat swab for bacterial culture
- Anti-streptolysin O (ASO) titres
  - Antibodies against streptococcus
  - After acute infection, typically:
    - Rise for 2-4 weeks
    - Peak at 3-6 weeks
    - Gradually fall over 2-3 months
  - Usually repeated after 2 weeks to:
    - Confirm a negative test
    - Assess whether levels are rising/falling
- Echo, ECG, & CXR for cardiac involvement

Jones Criteria for Diagnosis

- Evidence of recent streptococcal infection +
  - 2 major criteria
  - 1 major & 2 minor criteria

Recent Streptococcal Infection

- Positive throat swab
- Raised ASO titres
- Positive rapid group A strep antigen test

Major Criteria (JONES)

- Joint pain (migratory arthritis)
- Organ involvement (carditis)
  - Must be evident as new regurgitant murmur
- Nodules
- Erythema marginatum
- Sydenham chorea

Minor Criteria (FEAR)

- Fever
- ECG changes (prolonged PR without carditis)
- Arthralgia (without arthritis)
- Raised inflammatory markers (CRP & ESR)

Management

- Oral penicillin V for 10 days
- NSAIDs
- Aspirin & steroids can help carditis
- Prophylactic antibiotics against further streptococcal infection & recurrence of RF continues into adulthood
- Monitor for & manage complications

Complications

- Recurrence
- Valvular heart disease
  - Particularly valvular stenosis
  - Chronic heart failures

Orthopaedics

Fractures

Paediatric Bone Physiology

- Growth plates remain open until adolescence and are typically stronger than the surrounding bone
- Children's bones contain more cancellous bone and less cortical bone than adults
  - More flexible, less strong

Types of Fracture

Complete fracture

- Both sides of cortex breached
- Transverse
- Oblique
  - Toddler's fracture
- Spiral
- Segmented
  - Multiple usually transverse breaks
- Comminuted
  - Complicated fracture with multiple bone fragments

Greenstick Fracture

- Unilateral cortical breach only

Buckle/Torus Fracture

- Incomplete cortical disruption leading to periosteal haematoma
- Weakness against compression

Plastic Deformity

- Deformity without cortical disruption

Salter-Harris

- Fractures involving growth plates
- Growth plate tenderness can indicate fracture even when XR appears normal
- Type 3, 4 & 5 usually require surgery
- Type 5 is associated with growth disruption
- **Type 1:** Fracture through physis only (XR often normal)
- **Type 2:** Fracture through physis and metaphysis
- **Type 3:** Fracture though physis and epiphysis including the joint
- **Type 4:** Fracture involving metaphysis, physis, epiphysis
- **Type 5:** Rare, crush injury of physis (XR may resemble type 1 or appear normal)

Features Suspicious of Non-Accidental Injury

- Delayed presentation
- Developmental delay
- Inconsistency between history and actual injury
- Multiple injuries
- Injuries at sites not commonly exposed to trauma

Management

- Safeguarding if there are concerns of NAI

Mechanical Alignment

- Closed reduction via joint manipulation if possible
- Open reduction (ORF) via surgery may be necessary

Fixation

- External casts
- Surgical methods
  - K wires
  - Intramedullary wires/nails
  - Screws
  - Plates

Pain Management

- Paediatric pain ladder:
  - 1. Paracetamol or ibuprofen
  - 2. Morphine
- Codeine/tramadol are not used due to unpredictable metabolism
- Aspirin is not used due to risk of Reye's syndrome

Hip Pain/Acute Limp

Causes

0-4 Years

- Septic arthritis
  - Unwell child with acutely inflamed joint
- Developmental dysplasia of the hip
  - Usually detected in neonates
- Transient synovitis
  - Acute onset & accompanied by viral illness
- Trauma

5-10 years

- Septic arthritis
- Transient synovitis
- Perthe's disease
  - Avascular necrosis of the femoral head
- Trauma

10+ Years

- Septic arthritis
- Slipped upper femoral epiphysis (SUFE)
  - Postero-inferior displacement of femoral head
- Juvenile idiopathic arthritis
  - Limp may be painless
- Trauma

Red Flags for Investigation/Referral

- Child < 3
- Child > 9 with restricted/painful hip
- Unable to bear weight
- Evidence of neurovascular compromise
- Severe pain or agitation
- Red flags for serious pathology
- Suspicion of abuse

General Investigations

- FBC & inflammatory markers for septic arthritis/JIA
- X-ray can diagnose fractures/SUFE/etc
- Ultrasound can demonstrate joint effusion
- Joint aspiration diagnoses or excludes septic arthritis
- MRI is used to diagnose or exclude osteomyelitis

Septic Arthritis

- Infection within a joint which can rapidly lead to joint destruction and/or systemic illness
- Twice as common in boys
- Most commonly affects hip, knee, and ankle

Presentation

- Acute onset of:
  - Hot, red, tender joint
  - Limp or refusal to bear weight
  - Stiffness and reduced range of motion
- Systemic features:
  - Fever
  - Lethargy
  - Sepsis

Bacteria

- Staph aureus (most common)
- Neisseria gonorrhoea (common in sexually active teenagers)
- Group A strep
- H. influenzae
- E. coli

Investigations

- FBC and inflammatory markers (raised WCC & ESR)
- Blood cultures
- Joint aspiration for gram stain, culture & sensitivity, & microscopy

Diagnosis

- Purulent joint aspiration/.positive culture
- **Kocher Criteria:**
  - Fever > 38.5
  - Non-weight bearing
  - Raised ESR
  - Raised WCC

Management

- Empirical IV antibiotics later narrowed to sensitivities
- Some patients may require surgical drainage/washout

< 3 months

- Cefotaxime + flucloxacillin + gentamycin

3 months – 5 years

- Cefazolin

> 5 years

- Flucloxacillin or cefazolin

Transient Synovitis

- Joint pain due to irritation inflammation of synovial membrane in association with viral URTI
- Commonest cause of hip pain in children

Presentation

- Follows viral illness by a few weeks
- Limp
- Pain
- Refusal to weight bear
- Low grade fever and clinically well
  - High fever or unwell child should prompt suspicion for septic arthritis

Course

- Symptoms peak at 48 hours and should resolve in 1-2 weeks
- Will recur in 20% of patients

Management

- Analgaesia & rest
- Investigate for septic arthritis if suspicious
- Safety net – patients should return if fever or joint inflammation gets worse

Slipped Upper Femoral Epiphysis

- Postero-inferior displacement of the femoral head along the growth plate
- More common in boys and obese children
- Average age 11-12
- May be triggered by minor trauma

Presentation

- Pain out of proportion with trauma
  - Hip/knee/groin/medial thigh
- Painful limp
- Restricted range of movement
  - Loss of internal rotation, held externally rotated

Investigation

- AP and lateral (frog leg) X-rays of the hip are diagnostic

Management

- Surgical internal fixation with cannulated screw through growth plate

Perthe's Disease

- Idiopathic avascular necrosis of the femoral head
- Common from 5-8, 5x more common in boys
- Bilateral in 10%

Presentation

- Hip pain developing over a few weeks ± referred pain to the knee
- Limp
- Restricted hip movements

Investigations

- Inflammatory markers are normal
- X-ray
  - Early: widening of joint space
  - Late: flattening of the femoral head
  - May be normal
- Technetium bone scan/MRI if X-ray is normal and symptoms persist

Catterall Staging

1. Clinical/histological features only
2. Sclerosis ± cystic changes with articular surface intact
3. Loss of structural integrity of femoral head
4. Loss of acetabular integrity

Management

- Maintain joint alignment
  - Bed rest, traction, braces, crutches
  - Physiotherapy
- Surgical management of older (>6) patients or on severe deformities
- Regular X-rays to assess results

Complications

- Osteoarthritis
- Premature fusion of growth plates

Osteomyelitis

- Infection in bone & bone marrow
- Most commonly Staph aureus

Classification

Haematogenous

- Spread of infection to bone via bacteraemia, usually monomicrobial
- Most common form in children
- Most commonly presents in metaphyses of long bones in children, vertebral osteomyelitis in adults

Non-haematogenous

- Results from direct spread from adjacent infected soft tissue or from direct trauma to bone
- Usually polymicrobial
- Most common form in adults

Risk Factors

- Open fracture
- Orthopaedic surgery
- Immunosuppression
  - Iatrogenic
  - HIV
- Sickle cell disease
- Tuberculosis
- Infective endocarditis
- Diabetes mellitus

Presentation

- May be acute or chronic with more subtle features
- Refusal to use limb/weight bear
- Pain & tenderness
- Swelling
- Fever
  - May be afebrile/low grade fever with chronic osteomyelitis
  - High grade fever with osteomyelitis/associated secondary arthritis

Investigation

- MRI is diagnostic investigation of choice
- Raised WCC, ESR, CRP
- Blood cultures will establish organism
- Bone marrow aspiration for culture may be necessary

Management

- Surgical drainage/debridement may be necessary

< 3 months

- Cefotaxime + flucloxacillin + gentamycin

3 months – 5 years

- Cefazolin

> 5 years

- Flucloxacillin or cefazolin

Osgood-Schlatter Disease

- Tibial traction apophysitis (inflammation at insertion of patellar ligament) from repeated small avulsion fractures
- More common in males aged 10-15 years
- Usually unilateral

Presentation

- Gradual onset of symptoms
  - Visible & palpable hard & tender lump at tibial tuberosity
  - Pain in anterior aspect of knee
    - Exacerbated by activity, kneeling, & extension of joint

Management

- Rest, ice, NSAIDs
- Stretching & physiotherapy to improve joint function after symptoms settle

Complications

- Large avulsion fracture may require surgery
- Bony lump may remain indefinitely

Talipes

- Abnormal fixed ankle position present at birth
- Twice as common in males, 50% bilateral
- Diagnosis is made clinically & no investigations are needed

Talipes Calcaneovalgus

- Ankle dorsiflexed and pronated (everted)

Talipes Equinovarus

- Ankle plantarflexed and (inverted)

Associations

- Spina bifida
- Cerebral palsy
- Edward's syndrome
- Oligohydramnios
- Arthrogryposis

Management

Ponseti method:

- Manipulation and progressive casting from birth
- Deformity usually corrected by 6-10 weeks
- Achilles tenotomy under LA required in 85% of cases
- Night-time braces worn until 4 years

Developmental Dysplasia of the Hip

- Congenital instability in the hip joint leading to dislocation & subluxation
- Leads to abnormal gate & degenerative changes in adulthood

Risk Factors

- Female sex
- First degree relative
- Breech presentation
- Multiple pregnancy
- Oligohydramnios
- Birth weight > 5kg

Screening

General

- Barlow & Ortolani tests for all newborns & 6-8 week checks

High-Risk

- Ultrasound + 6 month X-ray for the following:
  - First degree relative with DDH
  - Multiple pregnancy
  - Breech presentation at or after 36wks (regardless of presentation at birth)

Management

- Pavlik harness for 6-8 weeks if diagnosed before 4.5 months
- Surgery if diagnosed later

Rickets

- Defective bone mineralisation resulting from insufficient vitamin D or calcium leading to soft/deformed bones

Causes

Insufficient Vitamin D

- Dietary
  - Eggs
  - Oily fish
  - Fortified cereals/supplements
- Low UV sunlight exposure/dark skin
- Malabsorption
- Chronic kidney disease

Insufficient Calcium

- Dietary
  - Dairy
  - Green leafy vegetables

Genetic (rare)

- Hereditary hypophosphataemic ricketts
  - X-linked dominant

Presentation

- Lethargy
- Bone pain
- Poor growth
- Dental problems
- Muscle weakness
- Pathological/abnormal fractures

Bone Deformities

- Outward bowing of legs
- Knock knees
- Rachitic rosary
  - Ends of ribs expand at costochondral junctions causing lumps along chest
- Craniotabes
  - Soft skull with delayed suture closing & frontal bossing

Investigations

- Serum 25-hydroxy-vitamin D < 25 nM diagnoses vitamin D deficiency
- X-ray diagnoses rickets (osteopenia & translucent bands)

Prevention

- Vitamin D supplements for all children and breastfeeding women

Management

- Vitamin D and calcium supplementation

Achondroplasia

- Skeletal dysplasia which is the most common cause of disproportionate short stature (dwarfism)

Genetics

- Defective Fibroblast Growth Factor Receptor 3 (FGFR3) gene on chromosome 4 resulting in abnormal cartilage and growth plate function
- Sporadic mutation (70%) or inherited in an autosomal dominant fashion
- Homozygous mutations are fatal in the neonatal period

Features

- Average full grown height of 4 feet
- Spine & trunk not affected, proximal limb bones more affected than distal
- Brachydactyly
- Genu varum
- Large head with frontal bossing & narrow foramen magnum
- Midface hypoplasia with flattened nasal bridge
- Trident hands
- Lumbar lordosis

Associations

- Recurrent otitis media
- Kyphoscoliosis
- Spinal stenosis
- Obstructive sleep apnoea
- Obesity
- Cervical cord compression & hydrocephalus

Management

- Multidisciplinary support
  - Paediatrics, specialist nurses, physiotherapists, occupational therapists, orthopaedic surgeons etc
- Limb lengthening surgery may have some benefit
  - Targeted osteotomies and separation with Ilizarov frames
  - Controversial due to risk of chronic pain and reduced functionality

Osteogenesis Imperfecta

- Disorders of collagen metabolism resulting in brittle bones and frequent fractures
- Type 1 is the more common and milder form
  - Autosomal dominant
  - Decreased synthesis of pro-alpha 1 or pro-alpha 2 collagen polypeptides

Presentation

- Frequent fractures following minor trauma
- Hypermobility
- Blue/grey sclera
- Deafness in late adolescence/early adulthood
  - Otosclerosis
- Short stature
- Triangular face
- Dental problems
- Bone deformities & bone/joint pain

Diagnosis

- Clinical
- X-rays may help diagnose fractures
- Bony bio profile generally normal
- Genetic testing may be performed

Management

- Bisphosphonates & vitamin D supplementation
- Multidisciplinary support
  - Paediatrics, specialist nurses, physiotherapists, occupational therapists, orthopaedic surgeons etc