Autistic spectrum disorder (pervasive developmental disorders)

- Triad of impairments in social interaction, communication and imaginative thought
  - significant genetic basis e.g. chromosome 7; high identical twin concordance
- Parental concerns around age 1-2 years - speech delay, limited eye contact / smile / emotion
  - also lack of interest in others, limited social gestures, reduced pretend play, regression
- Associations - seizures (up to 30%), visual/hearing impairment, depression, ADHD, low IQ
  - Multidisciplinary diagnosis (‘autism teams’)
- Medical management - risperidone (short-term for aggression), melatonin (sleep problems)

Asperger’s syndrome

- Primarily affects boys; later presentation than classic autism
  - language - flat tone, pedantic, literal approach to language, poor non-verbal skills
  - cognition - high IQ, obsessive, creative, low empathy, difficulty appreciating overall contexts
  - behaviour - clumsiness, sleep disturbances, solitary, eccentric, socially inappropriate

Bronchiolitis

- Common RSV infection in winter affecting young children aged 2 - 6 months
- Risks - passive smoke, prematurity / low birthweight, chronic lung disease
- Symptoms - URTI then paroxysmal cough, dyspnoea, wheeze, vomiting, apnoeas
- Investigations - NPA for RSV (for isolation)
- Management - supportive; consider bronchodilators, adrenaline, hypertonic saline
  - palivizumab - prophylaxis for high-risk e.g. chronic lung disease, cyanotic heart disease

Cerebral palsy

- Non-progressive brain abnormality causing persistent disorder of movement / posture
  - caused by damage (hypoxic, vascular, infective etc.) to immature brain up to post-natal period
  - disorder may be primarily spastic (hypertonic), athetoid (hyperkinetic), ataxic (in-coordinate)
- Risks - prematurity, IUGR, twins, traumatic birth, TORCH, high maternal age, malformation
- Symptoms - low Apgar score; FTT / missed milestones, epilepsy (30%) incontinence, poor sleep
- Management - multidisciplinary; consider baclofen (PO) for muscle spasm; botox, phenol
- Complications - GORD, aspiration pneumonia, learning difficulties, hearing loss

Congenital throat problems

- Laryngomalacia - soft, collapsible larynx causing noisy respiration / stridor (worse supine)
  - majority spontaneously resolve by age 2 years
- Congenital vocal cord paralysis - hoarse cry, feeding difficulty, aspiration, poor cough
  - associations - birth trauma / cerebral palsy, hydrocephalus, Chiari malformations
  - management - speech therapy (unilateral), surgical intervention (bilateral), nutritional support
• **Subglottic stenosis** - malformed cricoid cartilage causing, stridor, respiratory distress, GORD
  - rarely congenital; acquired in trauma, TB, syphilis, diphtheria
  - management - intubation if obstructed, endoscopic ablation, *reconstructive surgery*

### Croup

- Acute *laryngotracheobronchitis* - common viral URTI affect children aged 6 months - 3 years
  - agents - parainfluenza I-IV (majority), RSV, adenovirus, measles, *Mycoplasma pneumoniae*
  - nasopharyngeal / subglottal inflammation, vocal cord restriction, potential pseudomembrane
- Symptoms - URTI then **barking cough**, hoarseness, **stridor** - worse at night; sometimes fever
  - **steeple sign** on CXR - subglottal narrowing due to air in upper trachea (non-specific)
- Management - supportive; oral **prednisolone** (or dexametasone / nebulised budesonide)
  - consider nebulised adrenaline (1 in 1000) if respiratory distress
- Complications - RDS requiring intubation, bacterial superinfection

### Epiglottitis

- Symptoms - **sore throat**, odynophagia, **drooling**, neck tenderness (hyoid), **stridor**, tachycardia
  - associated with **tripod sign** - leaning forward to ease respiration
- Investigations - *fibre-optic laryngoscopy*, lateral neck X-ray, throat swabs, blood cultures
- Management - supportive; consider **cefotaxime**, intubation / tracheostomy in airway compromise
- Complications - abscess, meningitis, septicaemia, pneumothorax

### Febrile convulsions

- Seizures affecting children aged **6 months - 5 years** associated with **fever** (> 37.8º)
  - recur in up to 30% but normally do not recur more than once within 24 hours
- Management - if prolonged (> 15m) put in recovery position, check BM, consider PR diazepam
- Complications - **epilepsy** (roughly 1% increased risk, esp. if complex febrile seizures)

### Haemolytic uraemic syndrome (HUS)

- Triad of **haemolytic anaemia**, **thrombocytopenia**, AKI (commonest cause of AKI in children)
  - rare; affects children aged < 5 years esp. amongst rural populations and farms
  - typically caused by verotoxin-producing *E. coli* (157:H7); also *ciclosporin*, SLE, pregnancy
- Symptoms - initially **diarrhoea** that becomes **bloody**; then fever, abdominal pain, vomiting
  - HUS occurs 7 - 14 days after onset of diarrhoea leading to oliguria, oedema, weight gain
- Investigations - **LDH** (early indicator), stool culture, urinalysis (haematuria / proteinuria)
- Management (notifiable disease) - supportive, early IVT; avoid anti-diarrhoea drugs
- Complications - stricture, perforation, **intussusception**, pancreatitis, seizures, CKD, HTN
Henöch-Schönlein purpura (HSP)

- IgA-mediated autoimmune **hypersensitivity vasculitis** affecting children aged 3 - 10 years
  - usually *post-URTI*; associated with Group A *Strep.*, EBV, vaccinations, allergens, insect bites
- Symptoms - fever, malaise, **abdominal pain**, bloody diarrhoea, knee / ankle arthralgia, **orchitis**
  - characteristic **symmetrical erythematous macular rash** on back of legs / buttocks / ulnar arms
  - erythema progresses to **purpuric rash** within 24 hours
- Investigations - urinalysis (nephritis), WCC (eosinophilia), ESR (high), serum IgA (high)
- Management - self-limiting; consider NSAIDs / prednisolone; immunosuppressants (clinical trials)
  - **follow-up urinalysis** for at least 6 months to screen for nephropathy
  - may persist for several months; 30% will have at least one recurrence
- Complications - nephritis (late; microscopic haematuria, nephrotic syndrome, renal failure - 1%)
  - also **intussusception**, GI bleed, bowel infarction, pleural effusion, MI, mononeuropathy

Hip problems

**Transient synovitis (irritable hip)**

- **Post-URTI** self-limiting viral / autoimmune synovitis; commoner in boys
- Symptoms - acute onset pain on movement with reluctance to weight-bear; child is well
- Management - rest, analgesia

**Developmental dysplasia of the hip (DDH)**

- Affects *first-born females*; left hip more commonly affected than right; 20% of cases bilateral
  - risks - FH, multiple pregnancy, prematurity, breech delivery, oligohydramnios, CP
  - most stabilise spontaneously by age 6 weeks
- Screening - *Ortolani* test - anterior reduction of femoral head; *Barlow* test - posterior dislocation
  - in older children - **Galeazzi** sign - supine hip / knee flexion to identify unilateral shortening
  - **USS** if under 6 months old; pelvic X-rays in older children; consider MRI / CT
- Management - **bracing** (dynamic flexion-abduction orthosis); consider surgery if age > 6 months
- Complications - degenerative joint disease requiring THR, lower back pain

**Perthes’ disease**

- **AVN** of femoral head leading to abnormal epiphyseal growth, subchondral collapse, fracture
  - grade I - superior lateral head; grade II - 50% of head; grade III - subtotal; grade IV - total
- Affects young boys aged < 12 years; risk factors as for DDH; usually no history of trauma
- Symptoms - limp, hip / knee pain / effusion, antalgic gait, limited hip ROM; painful ‘roll test’
- Investigations - X-ray (joint space widening), MRI, arthrogram
- Management - conservative if age < 8 years; **physiotherapy** improves function
  - surgery (age > 6 years) - **proximal varus osteotomy**; arthrodesis if severe
Slipped upper femoral epiphysis (SUFE)

- Commonest adolescent hip disorder; primarily affects \textit{teenage boys}, usually chronic slippage
- Risks - minor local trauma, \textit{obesity}, hypothyroidism, hypopituitarism, pelvic radiation
- Symptoms - ambulatory hip / groin pain, worse on running; limp, limited hip ROM e.g. \textit{int. rotation}
  - acutely - unable to walk / stand; chronically - \textit{knee pain} may be only symptom
- Management - rest / immobilise hip, analgesia, \textbf{surgical closure} (percutaneous)
- Complications - chondrolysis, AVN

Hirschprung’s disease (congenital megacolon)

- Congenital absence of \textit{rectal myenteric parasympathetic ganglia} - affects boys aged < 2 years
- Associations - \textit{Down’s}, MEN-II, congenital deafness; may be X-linked component
- Symptoms - \textit{delayed meconium} (> 48hrs), abdominal distension, vomiting
  - later - chronic constipation, soiling / overflow (uncommon), early satiety, abdominal discomfort
- Investigations - barium enema (transition zone), anorectal manometry, \textbf{rectal biopsy} (definitive)
- Management - decompression of obstruction, \textbf{Swenson} resection and anastomosis
- Complications - enterocolitis (fever, diarrhoea, necrosis - 30% mortality), incontinence

Hydrocephalus

- \textbf{Bickers-Adams syndrome} - aqueduct of Sylvius stenosis; learning difficulties, thumb deformity
- Also Chiari malformations, mass lesions, haemorrhage, meningitis, achondroplasia, excess vit. A
- Symptoms - FTT, developmental delay, head circumference increase, dilated scalp veins
- Signs - ‘setting sun’ (downward-facing eyes, lid retraction), Macewen’s (‘cracked pot’ percussion)

Inborn errors of metabolism (IEM)

Phenylketonuria (PKU)

- \textbf{Autosomal recessive} - commonest IEM - insufficient \textit{phenylalanine hydroxylase} (PAH) activity
  - classically due to multiple mutations on chromosome 12 - carrier frequency 1 in 50
  - PAH converts \textit{phenylalanine} into \textit{tyrosine}; high phenylalanine gives \textit{neurotoxic byproducts}
  - insufficient tyrosine impairs \textit{melatonin} production leading to \textit{hypopigmentation}
- Symptoms - \textit{mental retardation} by age 1 year, developmental delay, fair hair, blue eyes
  - also \textit{recurrent vomiting}, ‘musty’ odour, skin lesions, seizures, behavioural disturbances
- Management - \textbf{dietary} (protein restriction / high tyrosine, vitamin / iron supplementation)
- Complications - demyelination, \textit{epilepsy}, congenital abnormalities if pregnant, agoraphobia

Medium chain acyl-CoA dehydrogenase deficiency (MCAD)

- \textbf{Autosomal recessive} - chromosome 1 mutation leading to \textit{inadequate gluconeogenesis}
  - \textit{fasting states} lead to \textbf{hypoglycaemia} - inability of fat metabolism / ketogenesis
- Symptoms (age > 3 months) - preprandial irritability, drowsiness, sweating, seizures, coma
- Management - \textit{avoid fasting}, high protein / carbohydrate but low fat diet
- Complications - \textit{sudden infant death}, hypoglycaemic brain damage
Homocystinuria

- **Autosomal recessive** - rare *cystathionine beta synthase* (CBS) deficiency (chromosome 21)
  - CBS converts *methionine* to *cysteine*; high methionine / homocysteine deforms collagen
  - hyperhomocysteinaemia also caused by advanced age, hypothyroidism, renal failure, drugs
- Symptoms - *Marfanoid habitus*, brittle hair, hypopigmentation, *lens dislocation*, glaucoma
  - also *mental retardation*, seizures, stroke / TIA, psychiatric disorders
- Management - *pyridoxine* (vitamin B6) / vitamin B12 / folate supplements, methionine restriction
- Complications - *thromboembolism*, MI, mitral valve prolapse, osteoporosis, pancreatitis

Reye’s syndrome

- Rare fatal encephalopathy - *mitochondrial dysfunction* due to aspirin / post-viral in under 14s
- Symptoms - acute deterioration with *vomiting*, diarrhoea, lethargy, *delirium*, *seizures*, coma
  - in infants - tachypnoea / respiratory distress, fever, hypoglycaemia, diarrhoea
- Investigations - LFTs (high ALT/AST), ammonia (high), amylase (high), urinalysis (ketones)
- Management - correct hypoglycaemia, consider ondansetron, specialist referral to lower ICP

Infectious mononucleosis

- Commonly affects adolescents - 90% due to *EBV*; 10% due to *CMV*; subclinical infection in many
- Associations - *Burkitt’s lymphoma* (NHL), pharyngeal carcinoma, immunodeficiency (Duncan’s)
- Symptoms - fever, malaise, nausea, sore throat, exudative tonsillitis, palatal petechiae, rash
  - also *cervical lymphadenopathy*; later hepatosplenomegaly, jaundice
- Investigations - *Monospot* (equine RBCs detecting *heterophile antibodies*) - best after 4 weeks
  - also VCA-IgM ELISA, EBV nuclear antigen (EBNA) - delayed; ESR (high), lymphocytosis
- Management - avoid alcohol / contact sports (*splenic rupture*), consider prednisolone
- Complications - splenic rupture, haemolytic anaemia, nephritis, encephalitis, Bell’s palsy, CFS?

Intussusception

- Affects children age < 1 year; commoner in boys; fatal if untreated
- Aetiology - *viral infection*, Meckel’s diverticulum, HSP, lymphoma, Peutz-Jeghers, CF, nephrosis
- Symptoms (acute) - *colicky abdominal pain*, vomiting, irritability, fever, mucoid / ‘redcurrant’ stools
  - also ‘sausage-shaped mass’ (RUQ), absent bowel in RLQ (Dance’s sign)
- Investigations - USS (doughnut sign), contrast enema
- Management - radiological reduction / air enema, laparotomy if peritonitis / perforated / prolonged

Lactose intolerance

- Lactose is found exclusively in milk; depends on *lactase* (from small intestine) for absorption
  - stimulates calcium, magnesium, zinc absorption; creates *galactose* - aids brain development
- Lactase deficiency may be *primary* (autosomal recessive) or *secondary* (e.g. transient post-viral)
- Symptoms (post-lactose load) - abdominal pain, bloating, flatulence, loose stools, pruritis ani
- Investigations - stool tests (pH < 5.5, carbohydrate malabsorption), hydrogen breath test
Kawasaki’s disease
- Idiopathic self-limiting systemic vasculitis affecting children aged < 5 years esp. of Asian origin
  - potential infective aetiology - seasonal peaks in winter/spring with 3-year cycle
- Symptoms - > 5 days fever, irritability, erythema of extremities, conjunctivitis, lymphadenopathy
  - also strawberry tongue, lip fissures, urethritis, diarrhoea, vomiting, abdominal pain, arthritis
  - associated with aortic/mitral regurgitation, proteinuria, aseptic meningitis, anterior uveitis
- Investigations - FBC (anaemia), CRP/ESR (high), echocardiogram (CAD)
- Management - aspirin (normally banned in under 16s), IV-Ig
- Complications - coronary artery aneurysm, MI, IHD, cardiac failure / sudden death

Muscular dystrophy
- Investigations - CK (very high, also carriers), genetic analysis, muscle biopsy (dystrophin assay)

Duchenne’s (DMD)
- X-linked recessive - abnormal dystrophin gene resulting in no dystrophin production
- Symptoms - progressive proximal muscular dystrophy; inability to run, Gower’s sign, cannot jump
  - also calf muscle hypertrophy, developmental delay, FTT - symptomatic by age 3 years
  - mobility lost around age 10 years; most patients survive to adulthood
- Management - physiotherapy, orthoses, ankle casting, regular prednisolone, vit. D / calcium
- Complications - respiratory failure, LRTIs, DCM, arrhythmias, learning difficulties

Becker’s (BMD)
- X-linked recessive - abnormal dystrophin gene resulting in abnormal dystrophin production
  - less common and milder / slower course than Duchenne’s
- Symptoms (around age 10 years) - muscle cramps, proximal myopathy / wasting
  - mobility lost around age 40 years; most survive to late adulthood
- Complications - DCM (main survival determinant)

Otitis media
- Common - majority have at least one episode by age 3 years; peak incidence age 1 - 2 years
  - typically post-URTI; pathogens usually Strep. pneumoniae, HiB
- Risks - male sex, dummy usage, URTIs, parental smoking
- Symptoms - otalgia, malaise, high fever, vomiting
- Management - supportive; consider 5 days of amoxicillin if persisting, bilateral, discharging
- Complications - perforation, hearing impairment, cholesteroloma, mastoiditis, meningitis (rare)

Otitis media with effusion
- Cause of hearing loss (20-50dB); affects children aged 1 - 6 years
- Management - mostly self-limiting within 3 months; consider surgery if persistent / bilateral
  - grommets first-line; adenoidectomy if recurrent URTI
- Complications - adverse speech / language development (though mostly temporary)
Pyloric stenosis

- Affects infants aged 2 - 8 weeks; commoner in first-born males at around 1 in 500 live births
- Symptoms - increasing frequency and intensity of vomit until projectile; visible peristalsis
  - also hunger, FTT, lethargy, infrequent bowel movements
- Investigations - USS, barium swallow, U&Es (hypokalaemia, metabolic alkalosis)
- Management - Ramstedt’s / laparoscopic pyloromyotomy

Scarlet fever

- Affects children aged 4 - 8 years esp. in overcrowding; notifiable disease
- Exotoxin-mediated Strep. pyogenes infection (group A) typically arising from pharyngitis
- Symptoms (sudden onset) - fever, scarlatiniform rash on day 2 (facial sparing, ‘punctuate’)
  - pharynx - haemorrhagic palatal spots, exudative tonsillitis, cervical lymphadenopathy
  - tongue - initially ‘white strawberry’, then sheds white precipitate leaving raw ‘raspberry’
- Management - penicillin / erythromycin for 10 days
- Complications - peritonsillar abscess, mastoiditis, cellulitis, pneumonia, rheumatic fever, AKI

Tonsillitis

- Commonly affects children aged 5 - 10 years; may be associated with atopy
- Group A Strep. suggested by fever, exudates, no cough, tender lymphadenopathy (Centor)
- Symptoms - sore throat, odynophagia, ear / abdominal pain, headache, voice changes / loss
  - if caused by Coxsackie - blisters on tonsils and roof of mouth leading to painful scabs
- Management - 10 days of penicillin V (only if GABS / severe, limited benefit)
  - consider tonsillectomy only if 5 or more severe disabling episodes per year
- Complications - otitis media, sinusitis, glomerulonephritis, rheumatic fever, guttate psoriasis flare

Whooping cough (pertussis)

- Severe URTI caused by Bordetella pertussis lasting > 6 weeks; primarily affects infants
  - cyclical outbreaks every 3 - 4 years; incubation of 1 week; dangerous in aged < 6 months
- Symptoms - catarrhal phase (coryza), then paroxysmal coughing (dry, choking, flailing)
  - severe persistent coughing (without drawing breath) may induce vomiting, eye haemorrhage
- Investigations - pernasal swab (to nasopharynx), FBC (lymphocytosis), CXR (perihilar infiltrates)
- Management - supportive; erythromycin may curtail infectivity but does not alter clinical course
- Complications - pneumonia, encephalitis, inguinal hernia, rectal prolapse
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<tr>
<th>Age</th>
<th>Vaccine</th>
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<tr>
<td>2 months</td>
<td>DTP/IPV/HiB - diphtheria, tetanus, pertussis; polio; HiB</td>
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<td>PCV - pneumococcus conjugate</td>
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<td><em>rotavirus (PO) - from Sept. 2013</em></td>
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<td>3 months</td>
<td>DTP/IPV/HiB</td>
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<td><strong>Men. C</strong></td>
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<td><em>rotavirus (PO)</em></td>
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<td>4 months</td>
<td>DTP/IPV/HiB</td>
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<td><strong>Men. C</strong></td>
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<td><strong>PCV</strong></td>
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<td>12 months</td>
<td>HiB/Men. C (booster)</td>
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<td><strong>MMR</strong></td>
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<td>3 - 5 years</td>
<td>DTP/IPV (booster)</td>
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<td><strong>MMR</strong></td>
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<td>12 years (girls)</td>
<td>HPV (3 injections at 0, 2, 6 months)</td>
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<td>13 - 18 years</td>
<td>DT/IPV (booster)</td>
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<td>&gt; 65 years</td>
<td><strong>influenza</strong> (annually)</td>
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<td><strong>PPV</strong> - pneumococcus polysaccharide</td>
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