

DEFINITION Chronic inflammatory dermatosis. Classified as mild, moderate, or severe.

AETIOLOGY

Infantile acne: <3 months of life; transient and usually due to maternal androgens.

Adolescent acne:

1. ↑Sebum production: androgenic stimulation of hyper-responsive pilosebaceous units.
2. Impaired flow of sebum: obstruction of the pilosebaceous duct by hyperkeratosis.
3. *Propionobacterium acnes*: gram-positive anaerobe is implicated in the inflammation.

Associations/related: Puberty, may ↑ premenstrually, POS, excess cortisol (Cushing syndrome).

EPIDEMIOLOGY

Developed world: Affects 79–95% of the adolescent population, peaking at 14–18 years; tends to recede by early twenties.

Developing world: Acne incidence is considerably lower; likely combination of environmental and genetic factors.

HISTORY Usually self-diagnosed, acute onset, greasy skin, may be painful.

EXAMINATION

Open comedones: Whiteheads; flesh-coloured papules.

Closed comedones: Blackheads; black colour is due to oxidation of the melanin pigment.

Other features: Pustules, nodules, cysts, scarring, and seborrhoea.

Distribution: Primarily affects the face, neck, chest and back (where sebaceous glands are most numerous).

PATHOPHYSIOLOGY Gross distension of the pilosebaceous follicle with neutrophil infiltration. Closed comedones may contain serous fluid. Severe acne can create fistulas between inflamed glands.

INVESTIGATIONS Not indicated unless other signs of androgen excess; prepubertal body odour, axillary/pubis hair or genital maturation; postpubertal infrequent menses, hirsutism or truncal obesity (suspect POS).

Bloods: Free testosterone, FSH, LH.

Urine: 24-hour urinary cortisol (if Cushing syndrome is suspected).

MANAGEMENT Indication for treatment is based on classification and degree of psychosocial impact. In severe acne, therapy should be commenced early to prevent scarring. General advice should include use of non-greasy cosmetics, daily face wash. Dietary restriction is not beneficial in the treatment of acne.

Single topical preparations

1. Benzoyl peroxide: keratolytic agent, encourages skin peeling, and bactericidal (S/E: irritation and bleaching of clothes).
2. Vitamin A derivatives: retinoids reduce obstruction within the follicle.
3. Antibiotics: clindamycin or erythromycin is effective but when used as single agents may result in *P. acne* resistance.

Combination topical preparations

1. Topical retinoids and antibiotics are synergistic.
2. Topical benzoyl peroxide and antibiotics reduce the likelihood of *P. acne* resistance and are also more effective.

Systemic preparations

1. Antibiotics: tetracyclines (doxycycline, minocycline). S/E: discoloration of teeth. Macrolides (erythromycin or azithromycin) or trimethoprim-sulphamethoxazole (Septrin).
2. Vitamin A derivative: isotretinoin (Roaccutane PO): 4–6-month course only by specialist prescription for severe acne (S/E: potent teratogen, hyperlipidaemia).
3. Hormones: oestrogen-containing OCP, antiandrogens (spironolactone S/E: hyperkalaemia), cyproterone acetate in females only.

Physical treatments: Short-term efficacy from optical treatments such as lasers, light sources and photodynamic therapy.

COMPLICATIONS

Physical: Facial scarring (atrophic/keloid), hyperpigmentation, 2° infection and fistulas.

Psychosocial: Lack of self-confidence.

PROGNOSIS Generally improves spontaneously over months/years. At 25 years, persists in 12% of women and 5% of men.

DEFINITION Inflammation of the membrane lining the nose. ARIA definition; intermittent (<4 days/week or <4 weeks) versus persistent (>4 days/week and >4 weeks). Impact on quality of life: mild, moderate or severe. 'Seasonal' (spring/summer) versus 'perennial' definition is also important in the UK.

AETIOLOGY Parental atopy. AR is more likely to occur in first-born children (hygiene hypothesis).

ASSOCIATIONS/RELATED Children with AR have an increased risk of developing asthma, sinusitis or otitis media. Allergic conjunctivitis frequently co-exists.

EPIDEMIOLOGY Seasonal AR is found in 10% and perennial AR in 10–20% of the population. Prevalence is increasing.

HISTORY Itchy nose, rhinorrhoea, sneezing, persistent nose blowing, congested nose, snoring, throat clearing, morning halitosis (post-nasal drip), chronic cough.

EXAMINATION Allergic nasal crease, allergic 'salute', allergic 'shiners' (dark circles under eyes), Dennie-Morgan (infra-orbital skin) folds, mouth breathing. On nasal inspection, pale hypertrophied inferior nasal turbinates. If polyps are seen, think of cystic fibrosis or aspirin-sensitive asthma in the older child.

PATHOPHYSIOLOGY Allergen on the nasal mucosa/eyes binds to IgE on mast cells and leads to release of inflammatory mediators (histamine/leukotriene). Mucosal cellular infiltration leads to the late phase response and chronic exposure leads to hypertrophy of the nasal turbinates and increased mucous production.

INVESTIGATIONS Skin prick tests/specific IgE for seasonal (grass/tree pollen and moulds) and perennial (house dust mite, animal dander) inhalant allergens.

MANAGEMENT Assess severity and impact on quality of life/sleep. Check compliance with medication and method of delivering intranasal steroids. Ask about asthma symptoms; treating AR will improve asthma control. Avoid irritants (e.g. cigarette smoke).

Allergen avoidance: *Pollen:* wear glasses during and shower after being outside, close windows (especially early morning and evening), don't dry clothes outside, fit pollen filter in car. *House dust mite prevention strategies:* ventilate room, bedding barrier covers, remove soft furnishings/cuddly toys, HEPA filter vacuums, acaricidal sprays.

Non-sedating antihistamines: Relieves itch, sneeze and rhinorrhoea but less effective for nasal congestion. Recommended for mild intermittent AR as a single therapy.

Decongestants: Modifies nasal obstruction. Recommended for short-term use only due to risk of rhinitis medicamentosa (rebound congestion).

Topical nasal steroids: Most effective single therapy. Relieves and prevents itch, sneeze, rhinorrhoea and congestion. Best when used regularly. S/E nasal bleed mainly if administered incorrectly. Newer formulations have minimal (<0.5%) systemic steroid absorption.

Leukotriene receptor antagonists: Second-line treatment. Synergistic effect when combined with non-sedating antihistamines but not as effective as combining antihistamines and nasal steroids.

Immunotherapy: Available in sublingual and subcutaneous preparation. Treatment shows sustained reduction in symptoms and medication scores, prevents new sensitisation to inhalant allergens and decreases the risk of developing asthma.

COMPLICATIONS Decreased quality of life: poor sleep, impairment of daily activities, problems at school (particularly during exam term).

PROGNOSIS Seasonal allergic rhinitis tends to diminish with age. If symptoms start in early childhood, the likelihood of improvement is greater than if the onset is in adulthood.

DEFINITION Pancytopenia (deficiency of all blood cell elements) associated with bone marrow hypoplasia.

AETIOLOGY

Idiopathic (>40%): Possibly 2° to immunological suppression of multipotent myeloid stem cells by cytotoxic T cells. Phenotypically normal but often have genetic markers of congenital marrow failure syndromes.

Acquired:

1. Viral infection (parvovirus, CMV, HIV, hepatitis, measles).
2. Drugs (chloramphenicol, alkylating agents, methotrexate).
3. Chemicals (DDT, benzene).
4. Radiation.

Inherited:

1. Fanconi anaemia, FA (autosomal recessive, error of DNA repair).
2. Dyskeratosis congenital (rare sex-linked disorder with skin and nail atrophy).
3. Shwachmann syndrome (pancytopenia in 25%).

ASSOCIATIONS/RELATED

FA: Growth retardation, forearm bones abnormalities, heart and renal tract defects (horseshoe or pelvic kidney) and skin pigmentation.

EPIDEMIOLOGY 2–5/1,000,000/yr. Any age. M > F.

HISTORY May present with either slow (months) or rapid (days) onset:

1. ↓ **RBC:** Tiredness, lethargy and dyspnoea
2. ↓ **Platelets:** Easy bruising, bleeding gums, epistaxis
3. ↓ **WCC:** Increased frequency and severity of infections (immunodeficiency).

EXAMINATION Signs of anaemia (pallor), petechiae, bruises, bacterial or fungal infections. No hepatomegaly, splenomegaly or lymphadenopathy.

PATHOPHYSIOLOGY

Macro: Pale or white bone marrow.

Micro: Hypocellular bone marrow composed of empty marrow spaces, fat cells, fibrous stroma and isolated foci of lymphocytes and plasma cells. Classic ‘chicken wire’ appearance.

INVESTIGATIONS

Blood: ↓Hb, ↓platelets, ↓neutrophils, normal MCV, low or absent reticulocytes.

Blood film: Leukaemia exclusion.

Bone marrow trephine biopsy: For diagnosis and exclusion of other causes (bone marrow infiltration: lymphoma, leukaemia, malignancies).

Chromosomal abnormalities: ↑ Random breaks in peripheral lymphocytes in Fanconi anaemia. Also prognostic guide; certain mutations associated with earlier onset of leukaemia/haematological abnormalities.

Ham test: For paroxysmal nocturnal haemoglobinuria: measures sensitivity of affected red blood cells to lysis by complement following activation in acidified serum.

MANAGEMENT

Treat the underlying cause: Medication review and underlying infection treatment.

Supportive: Blood and platelet transfusions, antibiotics (therapeutic/prophylactic).

Medical: Immunosuppression; corticosteroids, cyclosporin A, antithymocyte globulin (ATG), androgen (oxymetholone for FA), and antilymphocyte globulin (ALG). Pts who relapse or are unresponsive to an immunosuppressive regimen may benefit from a repeat or alternative course.

Surgical: CVC/Portacath placement for repeated phlebotomy/transfusions. Splenectomy with heavily transfused and allosensitised pts refractory to medical therapies and with no HSCT donor.

Haematopoietic stem cell transplantation (HSCT): Definitive treatment in severe aplastic anaemia; from an HLA-identical family member. Cure rate approaching 90%.

COMPLICATIONS

Complication of disease process: Bleeding, infections (bacterial and fungal), sepsis and ↑d risk of developing myelodysplastic syndromes or leukaemia if the duration of illness is prolonged.

Complication of HSCT: Graft rejection, graft versus host disease, infection (new or reactivated).

PROGNOSIS Poor prognostic features include: platelets $<10 \times 10^9/l$, neutrophils $<0.5 \times 10^9/l$, reticulocytes $10 \times 10^9/l$. >50% of patients with all these features lasting more than 3 weeks will die.

DEFINITION Premature erythrocyte breakdown (haemolysis) causing ↓d erythrocyte lifespan (<120 days) and anaemia (2° to bone marrow activity unable to compensate).

AETIOLOGY Intravascular or extravascular (reticuloendothelial system; by splenic macrophages). 2° to either hereditary or acquired factors.

Hereditary:

Membrane defects: Spherocytosis (abnormal spectrin: structural membrane protein which alters deformability of RBCs), elliptocytosis (elliptical RBCs), hereditary pyropoikilocytosis.

Metabolic defects: G6PD deficiency, pyruvate kinase deficiencies.

Haemoglobinopathies: Sickle cell disease, thalassaemia.

Acquired (immune):

Autoimmune: Warm or cold antibodies attach to RBCs → activation of complement → intravascular haemolysis and extravascular haemolysis in hypersplenism.

Warm antibodies: Idiopathic or associated with SLE, lymphomas or methylidopa.

Cold antibodies: Idiopathic/associated with infections (*Mycoplasma*, EBV) or lymphoma.

Isoimmune: Transfusion reaction, haemolytic disease of the newborn.

Acquired (non-immune):

Trauma: RBC fragmentation in abnormal microcirculation; thrombotic thrombocytopenia purpura, haemolytic uraemic syndrome, disseminated intravascular coagulopathy, malignant hypertension, pre-eclampsia, artificial heart valves.

Paroxysmal nocturnal haemoglobinuria (PNH): Acute-onset haemoglobinuria; idiopathic or 2° to cold due to complement-mediated lysis.

Infection: Malaria.

ASSOCIATIONS/RELATED Parvovirus B19.

EPIDEMIOLOGY

Hereditary causes: Prevalent in African, Mediterranean and Middle Eastern populations.

Hereditary spherocytosis (HS): Most common inherited haemolytic anaemia in northern Europe.

HISTORY AND EXAMINATION

Depends on age: Jaundice and anaemia most common symptoms. Neonatal jaundice may require exchange transfusion. Older children may present with chronic anaemia. Hepatosplenomegaly and specific signs of underlying pathogenesis may also be present.

PATHOPHYSIOLOGY

Blood film (haemolytic anaemia): Leucoerythroblastic picture, microspherocytosis, macrocytosis, nucleated RBCs/reticulocytes, polychromasia.

Blood film (underlying cause): Spherocytes, elliptocytes, sickle cells, fragmented RBCs (DIC), malarial parasites, RBC Heinz bodies (G6PD deficiency).

INVESTIGATIONS

Bloods: ↓Hb, ↑MCV due to reticulocytes, ↑unconjugated bilirubin, ↑LDH, ↓haptoglobin, ↓red cell G6PD and pyruvate kinase assays, Hb electrophoresis (identifies variants).

Urine: ↑Urobilinogen 2° to excess unconjugated bilirubin, haemoglobinuria.

Direct Coombs' test: Identifies RBCs coated with antibodies using antihuman globulin.

Warm antibodies: IgG, agglutinate RBCs at 37 °C. *Cold antibodies:* IgM, agglutinate RBCs at room temperature.

Osmotic fragility test: Detects membrane abnormalities (spherocytosis).

Ham's test: PNH.

Bone marrow biopsy (rarely required): Erythroid hyperplasia; may be hypoplastic in PNH.

MANAGEMENT

Contributing factor avoidance: Cold exposure (cold antibodies, PNH), drugs (G6PD deficiency), transfusions, folate deficiency (HS), splenectomy (postpone until after childhood).

Autoimmune (warm): Prednisolone, splenectomy, azathioprine/cyclophosphamide.

PNH: Blood transfusions (leucocyte-depleted), anticoagulants for thrombotic episodes, bone marrow transplantation is successful in a small number of patients.

COMPLICATIONS Renal failure may develop in all cases due to accumulation of RBC breakdown products in the renal tubules. *PNH:* Can transform into aplastic anaemia or leukaemia. *HS:* Gallstones, aplastic anaemia in parvovirus infection, megaloblastic and haemolytic crises (\downarrow folate due to hyperactive bone marrow), leg ulcers and corneal opacities.

PROGNOSIS Depends on the cause.

DEFINITION ↓Hb with low mean cell volume (MCV <80 fl) and depleted iron stores.

AETIOLOGY

WHO definition: Hb <11 g/dl (110 g/l) aged 1–2 years and <11.2 g/dl (112 g/l) aged 3–5 years.

General: Three stages in the pathogenesis: Fe^{2+} depletion → Fe^{2+} -deficient erythropoiesis → Fe^{2+} deficiency anaemia.

↓d Fe^{2+} stores at birth: Prematurity, multiple pregnancy, perinatal bleeding, early umbilical cord clamping, and maternal Fe^{2+} deficiency.

Nutritional (inadequate Fe^{2+} supply): Exclusive breastfeeding >6/12 (insufficient Fe^{2+} in breast milk); early cow's milk introduction (↓bioavailability of iron than breast milk and formula milk is fortified with 6 mg iron/l); excessive reliance on milk in the second year of life; ↑d infant fruit juice intake; strict vegetarian diet; behavioral (food refusal, grazing, dieting, eating disorders); Crohn's disease, coeliac disease (↓d absorption).

↑d Fe^{2+} loss: Acute haemorrhage (Meckel's diverticulum, intestinal duplication cyst, peptic ulcer); chronic haemorrhage (cow's milk protein intolerance, intestinal duplication, IBD, telangiectasia, intestinal polyp); parasites (hookworm – *Ancylostoma duodenale* in developing countries); menstruation.

↑d Fe^{2+} demand: Growth; prematurity; IUGR; post malnutrition.

ASSOCIATIONS/RELATED Inner city centres (UK) and Asian communities.

EPIDEMIOLOGY Most common nutritional deficiency disorder worldwide. *Incidence:* 5–40% depending on community. *Peak ages:* 6 months to 3 years and adolescent girls. Uncommon in non-premature infants <6/12 (fetally acquired iron reserves).

HISTORY

General: Gradual onset; failure to thrive, poor exercise tolerance, global developmental delay, headaches, and irritability.

Behavioural: Anorexia, pica (ingestion of odd materials with ↑d Fe^{2+}), irritability, impaired concentration, impaired progress at school.

EXAMINATION

General: Signs of anaemia (pallor of skin and mucous membranes, tachycardia) and systolic flow murmurs.

Rarely: Brittle nails and hair (↓epithelial cell iron), spoon-shaped nails (koilonychia), glossitis (atrophy of tongue papillae), angular stomatitis, mild hepatosplenomegaly and lymphadenopathy.

PATHOPHYSIOLOGY

Blood film: Microcytic, hypochromic (central pallor), anisocytosis (variable sizes), poikilocytosis (variable shapes).

INVESTIGATIONS

Bloods: ↓Hb, ↓serum ferritin, ↓serum Fe^{2+} , ↑TIBC, ↓haematocrit, ↓MCV.

Hb electrophoresis: Exclude β -thalassaemia trait or Sickle cell disease.

Bone marrow (only in complicated cases): Erythroid hyperplasia and ↓ bone marrow Fe^{2+} or total absence of iron.

MANAGEMENT

Preterm: Fortify breast milk with Fe^{2+} . Use Fe^{2+} -fortified milk formula.

Infants: ↑ Highly absorbable haem iron sources (meat, fish) and sources of non-haem iron (such as grains) in vegetarian families. Enhance non-haem iron absorption by eating vitamin C-rich foods at the same meal.

Oral FeSO_4 : Maximum rise of Hb 0.25–0.4 g/d/day.

Blood transfusion: Indicated with severe anaemia leading to CHF and cardiovascular compromise. Packed RBCs should be given slowly or partial exchange transfusion.

COMPLICATIONS Possible impaired mental and psychomotor development. High output cardiac failure in severe cases.

PROGNOSIS Good outcome if cause is nutritional or due to \uparrow d demand and prompt action is taken. If underlying GI cause, outcome dependent on underlying cause.

DEFINITION Normocytic, normochromic, hyporegenerative anaemia in a preterm infant associated with a low serum erythropoietin (EPO) level.

AETIOLOGY

Inadequate RBC production 2° to low EPO: EPO initially produced by the fetal liver then the kidneys nearer to term. Liver EPO production stimulated with ↓d degree of anaemia and hypoxia than for the fetal kidney. With the premature neonate RBC production ↓d as liver still 1° source of EPO production.

Shortened fetal RBC lifespan: Fetal RBCs have 50–66% of the lifespan of an adult RBC. 2° to ↓d intracellular ATP, carnitine and enzyme activity combined with ↑d susceptibility to lipid peroxidation and fragmentation. At birth fetal haemoglobin represents 60–90% of haemoglobin. Adult levels of <5% by 3–6/12.

Blood loss: Intrapartum and aggravated by repeated blood sampling. A blood sampling can account for 10–15% of the total circulating volume.

Low iron stores: Combined with nutritional deficiencies of iron, vitamin E, vitamin B12 and folate may exaggerate the degree of anaemia.

Rarer pathological causes of anaemia in preterm infants

1. Haemolysis: 2° to ABO/Rh blood group incompatibility or haemoglobinopathies.
2. Bone marrow suppression: 2° to infection or renal failure.
3. Bone marrow failure: aplastic anaemia or malignancy.

ASSOCIATIONS/RELATED Low birthweight, FHx. No association with sex/race.

EPIDEMIOLOGY Frequency of anaemia of prematurity is inversely related to the gestational age and/or birthweight of the population. 50% of infants <32 weeks will develop symptoms secondary to this condition. Up to 80% of low-birthweight (<2.5 kg) infants require transfusions and 95% of extremely low birthweight (<1.25 kg).

HISTORY AND EXAMINATION Symptoms and signs of anaemia in a preterm infant:

1. ↓d activity which is improved by transfusion.
2. Poor weight gain despite adequate calorie intake.
3. Tachypnoea, tachycardia, pallor and flow murmurs.
4. If severe, will result in respiratory depression; episodes of apnoea.

PATHOPHYSIOLOGY See Aetiology.

INVESTIGATIONS

Bloods: Hb <10 g/dl, normochromic, normocytic; normal platelet count and white cell count.

Blood film: ↓Reticulocyte count (2° ↓ EPO), abnormal RBC forms (sickle cells, target cells in thalassaemia), red cell fragmentation (haemolysis).

Blood typing: ABO/Rh blood group incompatibility of neonate and mother.

MANAGEMENT

Indications for packed RBC transfusion:

1. Hb <8 g/dl.
2. Failure to thrive.
3. Cardiovascular/respiratory compromise.
4. Co-existing pathologies that may be exacerbated by anaemia.

Iron supplementation: May ↓ need for transfusion.

Recombinant EPO: Not advised (Cochrane review).

COMPLICATIONS Transfusion-acquired infection, transfusion-associated fluid overload, electrolyte imbalances or haemolysis.

PROGNOSIS Preterm infants are usually started on iron therapy for 2–3/12. Anaemia usually resolves spontaneously by 3–6/12, as adult haemoglobin is produced and intrinsic RBC/EPO production ↑s.

DEFINITION Wide spectrum of congenital disorders affecting the distal anus and rectum as well as the urinary and genital tracts.

AETIOLOGY

General: Wide spectrum of defects. Exact aetiology unknown but close genetic association.

All have absence of an anus in the normal position. *Mild forms:* bowel outlet via fistula in the perineal region separately from the normal sphincter complex. *Severe forms:* bowel outlet ectopically opens in the urogenital tract (males) and genital tract (females).

Classification (Wingspread): Traditional system. Relationship of the pouch to the levator muscle complex: either low, intermediate and high.

Classification (Krickenbeck): Recent international consensus. *Major group:* presence of fistula (perineal, rectourethral, prostatic, bulbar, rectovesical, vestibular) or no fistula and cloaca or anal stenosis. *Minor group:* pouch colon, rectal atresia/stenosis, rectovaginal fistula, H-fistula, others.

ASSOCIATIONS/RELATED Associated anomalies >60%. Multianomaly syndromes: VACTERL, CHARGE, MURCS, OEIS. Trisomy 13, 18, 22. Hirschsprung's disease. Vertebral and sacral anomalies.

EPIDEMIOLOGY 1 in 5000 live births. M > F.

HISTORY AND EXAMINATION

General: Clinical examination by an experienced surgeon will reveal the type in >90%. The presence and position of fistula. Associated anomalies.

Specific (low lesions): Prominent midline skin bridge ('bucket-handle'), subepithelial midline raphe fistula ('chain of meconium pearls'), rectovestibular fistula, anal membrane. A flat perineum (absence of midline gluteal fold and anal dimple) may indicate absence of perineum muscle and therefore a high ARM.

PATHOPHYSIOLOGY See Aetiology.

INVESTIGATIONS

General: Screening for associated anomalies: ECHO, CXR (NGT position/heart), RUSS, serum karyotype, USS for hydrometrocolpos/spinal anomalies, sacral XR, MRI, urinalysis (?urinary tract association).

Specific: Prone cross-table lateral XR with the pelvis elevated and marker on the perineum (air column to marker <1 cm = treat as low lesion, >1 cm = colostomy), high-pressure distal colostography (distal stomal contrast to delineate distal rectum and urinary connection).

MANAGEMENT

Medical (neonatal): Resuscitation as appropriate. Associated anomalies screening, NGT, NBM, IVI, antibiotics, transfer to paediatric surgical centre. Often have 24 hours to stabilise as low obstruction. Clinically observation vital ?passage of meconium via fistula/urinary tract.

Surgical (neonatal): Ascertain provisional diagnosis of ARM level with surgical planning. *Severe:* primary diverting colostomy. *Low:* cutback anoplasty/limited PSARP. Large rectovestibular fistulas may be dilated.

Surgical (definite): Posterior sagittal anorectoplasty (PSARP) allows excellent visualisation. Colostomy closed separately. Laparoscopic-assisted approach also possible with mobilisation of the rectal pouch/ligation of fistula and delivery of the pouch to the perineum through a minimal posterior incision.

Postsurgical incontinence: Effective bowel management programme including enemas, laxatives and dietary manipulations.

COMPLICATIONS Most common functional disorder post surgery is constipation. Possible urinary and faecal incontinence even with excellent anatomical repair 2° to poorly developed sacrum, spinal cord anomalies and deficient nerve supply.

PROGNOSIS Generally good. Low ARM has fewer complications than high. Dependent on associated anomalies.

DEFINITION Acute inflammation of the vermiform appendix.

AETIOLOGY Obstruction of the appendiceal lumen, causing a cycle of progressive inflammation and bacterial overgrowth.

ASSOCIATIONS/RELATED Poor dietary fibre intake: ↑s faecal viscosity, bowel transit time, and the formation of faecaliths.

EPIDEMIOLOGY Most common cause of an acute abdomen in children. *Incidence:* 4/1000 children. Any age, most common >5 years of age, uncommon <2 years.

HISTORY Large variation in clinical picture:

- Classically colicky pain starts periumbilically then localises to the right iliac fossa. Constant with peritoneal inflammation and ↑d with movement.
- Anorexia (vague abdominal pain and won't eat their favourite food).
- Vomiting (young children).
- Constipation or diarrhoea (less common).
- Low-grade pyrexia.

EXAMINATION

General: Tachycardia, pyrexia, reluctance to move.

Abdominal examination: Percussion tenderness signifies inflammation of the peritoneum.

Guarding may be present in RIF (McBurney's point). Rovsing's sign (RIF pain reproduced with palpation in the LIF). There may also be pain on expansion and recession of the abdomen. Cough may exacerbate pain. Peritoneal irritation signs may be absent with a retrocaecal appendicitis.

Rectal examination: Should be performed by the most senior doctor only when diagnosis is in doubt. There is marked tenderness against anterior rectal wall, especially with a retrocaecal appendix.

PATHOPHYSIOLOGY Obstruction of the lumen by impacted faeces or faecalith leads to mucosal inflammation. Inflammation extends into the submucosa to involve the muscular and serosal layers. Fibrinopurulent exudates from the serosal surface extend to the peritoneal surface causing localised peritonitis. The lumen subsequently becomes distended with pus and thrombosis of end-arteries leads to gangrene and perforation. Ineffective lymphatic and venous drainage allows bacterial invasion of the appendiceal wall.

INVESTIGATIONS

General: Appendicitis is a clinical diagnosis; investigations may aid diagnosis in difficult cases.

Bloods: ↑WCC (normal WCC doesn't exclude appendicitis), ↑CRP, U&Es (especially if vomiting), clotting (raised neutrophil count is the most sensitive serological investigation for appendicitis).

Urine: MC&S to exclude UTI, leucocytes may be present with an inflamed appendix against bladder wall (nitrite -ve).

Radiology: Plain AXR not indicated; if performed, may show dilated loops of bowel and a fluid level in the RIF. USS may show the inflamed appendix as a non-compressible tubular structure, presence of free fluid or appendiceal mass.

MANAGEMENT

Surgical: Once diagnosis confirmed clinically. May be performed via the traditional open approach (Lanz incision) or laparoscopically. Washout essential with complicated appendicitis.

Conservative: With a confirmed appendiceal abscess that responds to intravenous antibiotics. If this management fails then surgical intervention (+/- drain insertion) is indicated. If conservative management succeeds then the patient is offered an interval appendicectomy.

COMPLICATIONS

Perforation: <3 years old = 80–100%; >10 years old = 10–20%. Complicated appendicitis (perforated/presence of pus); wound infection/intra-abdominal abscess formation. ↓d fertility in girls after complicated appendicitis (ovarian/fallopian tube involvement), small bowel obstruction, adhesions.

PROGNOSIS Usually excellent.

DEFINITION Chronic inflammatory airways disease characterised by variable reversible airway obstruction, airway hyper-responsiveness and bronchial inflammation.

AETIOLOGY

Genetic factors: Positive family history of asthma or atopy.

Environmental triggers: Passive or active smoking, URTIs, exercise, cold weather, inhalant allergies (house dust mite/pollens/moulds/pets) and food allergens.

ASSOCIATIONS/RELATED Eczema, allergic rhinitis, previous CLD of prematurity.

'Hygiene' hypothesis: Exposure to microbial products in infancy leads to switching off Th2 predisposition of T cells and increasing regulatory T cells to prevent an allergic predisposition.

DD in <2 years: Aspiration, pneumonia, tracheomalacia, CF, tracheo-oesophageal fistula (H-type), bronchiolitis.

EPIDEMIOLOGY

Prevalence: 10–15%. **Age:** 80% of asthmatic children are symptomatic by the age of 5. M:F, 2:1; equalises in adulthood. **Distribution:** Viral-associated wheeze/recurrent wheezy bronchitis ↑ in urban areas and in children of low socio-economic status families.

HISTORY

Age-related symptoms:

- **<1 year:** Persistent or recurrent nocturnal cough, wheezing with URTIs.
- **2–3 years:** Nocturnal cough, wheezing during exercise with URTIs.
- **<5 years:** Non-productive cough may be the only symptom, often worse at night and in the morning.

Assess severity: Frequency of attacks (mild: <1 attack in 2 months; moderate: >1 attack in 2 months; severe: persistent symptoms, ↓exercise tolerance), effect on school attendance, hospital attendances and admissions to PICU.

EXAMINATION

Respiratory: End-expiratory wheeze, recession, use of accessory muscles, tachypnoea, hyper-resonant percussion note, diminished air entry, hyperexpansion, Harrison sulcus (anterolateral depression of thorax at insertion of diaphragm).

Peak flow: Useful in >5 years of age; use as baseline (predicted best) and as determinant for efficacy of treatment.

BTS guidelines for assessment of acute asthma attack

Severe asthma	Life-threatening asthma
Too breathless to speak or feed	Silent chest
Tachycardia:	Cyanosis
>120 bpm in 2–5 years	Poor respiratory effort
>130 bpm in <2 years	Hypotension
Tachypnoea:	Exhaustion
>30 breaths/min in 2–5 years	Confusion
>50 breaths/min in <2 years	Coma
Peak flow: <50% predicted >5 years	Peak flow: <33% predicted in >5 years

PATHOPHYSIOLOGY

Acute phase (within minutes): Contact with exacerbating factor (cigarette smoke, inhalant or food allergen or viral infection) leads to ↑ airway receptor hyper-responsiveness → narrowing of airways.

Late phase (onset after 2–4 hours, effect may last up to 3–6 months): Persistent bronchoconstriction 2° to vicious cycle of inflammation, oedema and excess mucous production.

INVESTIGATION

CXR: In acute severe cases to exclude pneumothorax or first presentation to exclude congenital anomaly.

Lung function (spirometry): Can be performed in >5 years. Obstructive airways disease: FEV1 <80%, FVC normal or reduced, FEV1/FVC <70%. Assess reversibility after 400 mg salbutamol inhalation.

MANAGEMENT

BTS guidelines 2008 for the management of acute asthma attack

- High-flow oxygen via reservoir bag.
- Salbutamol and ipratropium bromide via volumatic spacer or nebulised.
- Oral prednisolone 20 mg (2–5 years), 30–40 mg (>5 years) or IV hydrocortisone if unable to retain oral medication.
- Commence IV salbutamol (bolus then infusion) or aminophylline infusion.
- Magnesium sulphate (40 mg/kg) IV.
- If not responding (<92% O₂ saturations) or any life-threatening features present, discuss with PICU for ventilatory support.

Discharge criteria: Patients can be discharged when stable on 3–4-hourly inhaled bronchodilators. Peak flow 75% of predicted best, and O₂ saturations >94%.

Education: On adherence to medication, recognition of acute attacks, emergency protocol, maintaining normal activities.

BTS stepwise management of chronic asthma

Key principles:

1. Avoid obvious precipitants, e.g. passive smoking, allergen avoidance.
2. Ensure good inhaler technique +/- volumatic spacer.
3. Check compliance.
4. Review treatment every 3–6 months.
5. 'Rescue' prednisolone in acute deterioration.
6. If obese advise weight reduction.

When to start preventer inhaler:

1. Symptomatic/use β_2 -agonist inhalers ≥ 3 times/week.
2. Waking one night/week.
3. Frequent exacerbations.

Children <5 years

Step 1, mild intermittent asthma: Short-acting β_2 -agonist inhalers (e.g. salbutamol) as necessary.

Step 2, regular preventer control: Add low-dose inhaled steroid (200–400 mg/day budesonide equivalent) or leukotriene receptor antagonist if steroid cannot be used.

Step 3, add-on therapy: Trial of leukotriene receptor antagonist.

Step 4, persistent poor control: Refer to respiratory paediatrician.

Children 5–12 years

Step 1, mild intermittent asthma: Short-acting β_2 -agonist inhalers as necessary.

Step 2, regular preventer control: Add low-dose inhaled steroid (200–400 mg/day).

Step 3, add-on therapy: Add LABA, e.g. salmeterol.

1. Good response: continue LABA.
2. Benefit from LABA but control still inadequate: \uparrow dose of inhaled steroids to 400 mg/day.

3. No response to LABA: stop LABA, ↑ dose of inhaled steroids to 400 mg/day, and add trial of oral theophylline (monitor plasma levels) or leukotriene receptor antagonist.

Step 4, persistent poor control: ↑ Dose of inhaled steroids to 800 mg/day.

Step 5, continuous or frequent use of oral steroids: Maintain ↑ dose of inhaled steroids.

Add oral prednisolone at lowest dose to provide adequate control.

Refer to respiratory paediatrician.

COMPLICATIONS Decreased linear growth rate due to poorly controlled asthma more usual than from overprescription of inhaled steroids, chest wall deformity, recurrent infections, status asthmaticus can be fatal. One-third of deaths occur under the age of 5 years.

PROGNOSIS Asthma often remits during puberty and many children are symptom free as adults, especially those who have mild asthma and are asymptomatic between attacks, or who develop asthma at >6 years. Rates of admission and mortality in asthma have ↓ since the early 1990s.

DEFINITION Chronic inflammatory itchy skin condition. Also known as atopic dermatitis.

AETIOLOGY Parental atopy. Dysfunction in the epidermal barrier protein filaggrin (due to genetic loss of function mutations) has recently been shown to be a major predisposing factor.

ASSOCIATIONS/RELATED Eczema can be triggered by environmental factors including irritants (soaps and detergents), infections, contact with food or inhalant allergens.

EPIDEMIOLOGY Affects 15–20% of UK children; 2–3-fold increase in the last three decades.

HISTORY The majority of eczema begins in the first year of life. Intense itchy skin and chronic relapsing inflammation of the skin are cardinal features.

EXAMINATION Infantile eczema affects the face and extensor surfaces and spares the nappy area. Flexural involvement predominates in older children. In eczema herpeticum there are small uniform circular ‘punched-out erosions’.

PATHOPHYSIOLOGY Acute phase is characterised by intercellular epidermal oedema (spongiosis) and leukocyte infiltration. In the chronic phase there is thickening of the epidermis, stratum corneum and dysfunction of keratinisation.

INVESTIGATIONS Infants with moderate to severe eczema with a history of immediate reaction to food warrant skin prick testing to common food allergens. Patch testing of foods in eczema produces conflicting data but in contact dermatitis it is well established.

MANAGEMENT Assess eczema severity and quality of life, including everyday activities and sleep, and psychosocial well-being. Identify and manage any trigger factors. Give advice on prompt recognition and treatment of infection (in particular eczema herpeticum) and provide a written eczema management plan.

Emollients: ‘Total emollient care’ includes liberal application of creams and ointments, using a soap substitute and bath oil.

Topical corticosteroids: Use a stepwise approach to topical steroids depending on the severity of eczema. Use mild steroids for the face. In children with recurrent flares, use a topical steroid for 2 consecutive days/week.

Topical calcineurin inhibitors: Can be used as second-line treatment of moderate to severe atopic eczema in children ≥ 2 years that is not controlled by topical corticosteroids, or where there are adverse effects to topical steroids.

Wet wraps: May be particularly effective for troublesome areas (feet/hands). Not to be used over infected skin, topical potent steroids or calcineurin inhibitors.

Dietary manipulation: In bottle-fed infants <6 months with moderate to severe eczema not controlled by optimal treatment, 6–8-week trial of an extensively hydrolysed or amino acid formula in place of cow’s milk formula.

Antihistamine: If children suffer from severe itching, trial a non-sedating antihistamine.

COMPLICATIONS Skin infections leading to cellulitis. Residual pigmentation, lichenification or skin atrophy related to long-term potent steroid use. Poor quality of life for the child and parent. Psychosocial issues.

PROGNOSIS Sixty to 70% of UK children with eczema at 7 years clear their eczema as teenagers, but may go on to develop asthma or allergic rhinitis (the ‘allergic march’).

DEFINITION Acyanotic congenital heart condition characterised by malformation in the atrial or atrioventricular septum.

AETIOLOGY

Secundum ASD: Patent foramen ovale (abnormal resorption of the septum primum during formation of foramen secundum).

Primum ASD/Partial AVSD: Defect in lower atrial septum, 2° to incomplete fusion of septum primum with the endocardial cushion. Immediately adjacent to the atrioventricular valves.

Sinus venosus ASD: Defect high in the atrial septum near the entry of the SVC (abnormal fusion between the embryological sinus venosus and the atrium).

Coronary sinus defect: Unroofed coronary sinus and persistent left SVC that drains into the left atrium.

Complete AVSD: Large central defect due to ASD and VSD with single large atrioventricular valve.

ASSOCIATIONS/RELATED Wide variety of chromosomal and genetic disorders and syndromes. Trisomy 21 ~50%.

EPIDEMIOLOGY 4/100,000 live births.

HISTORY Depends on defect size and degree of mitral regurgitation. May take decades to manifest symptoms (PHT, atrial tachyarrhythmias, mitral valve disease). Childhood diagnosis often after routine examination reveals a murmur. Large defects: recurrent respiratory infections, symptoms of congestive heart failure, dyspnoea, palpitations (older children), failure to thrive.

EXAMINATION

ASD: Left to right shunt (pink +/-breathless): Murmurs are due to ↑d flow across valves, not the ASD itself. Ejection systolic murmur (pulmonary valves). Mid-diastolic murmur (tricuspid valve). Fixed splitting of the second heart sound 2° to ↑d volume causing prolonged contraction time of the RV.

ASVD: Mixed shunt (blue and breathless): Atrial and ventricular components therefore PHT present as with a large VSD. May present with cyanosis at birth, a murmur in the first few weeks or signs of CHD at 1–2/12.

PATHOPHYSIOLOGY See Aetiology.

INVESTIGATIONS

CXR: RAH, RVH, prominent PA and ↑d pulmonary vascular markings.

ECG: Right axis deviation, first-degree heart block, right bundle branch block, RAH and RVH.

Doppler + Echo: For diagnostic, assessment of size and congestive heart failure features.

MANAGEMENT

Supportive management: Antibiotic prophylaxis for dental surgery and other minor procedures. Medical treatment of associated CHF.

Surgical management: Closed to prevent RVH and arrhythmias. Repair of associated anomalies (e.g. PDA) under same GA. Elective repair at 2–5 years unless significant MR indicates earlier closure.

ASD/partial AVSD: Direct closure of the defect via an open median sternotomy with extracorporeal support. Use of autologous pericardium or synthetic patches made of polyester polymer (Dacron) or polytetrafluoroethylene (PTFE). Percutaneous transcatheter closure via femoral venous approach is also possible.

Complete AVSD: 3–5/12; requires treatment of pulmonary vascular resistance if present at birth.

COMPLICATIONS Infective endocarditis, congestive heart failure, atrial fibrillation, pulmonary hypertension.

PROGNOSIS 95% remain open, 5% close spontaneously. Only large defects cause significant morbidity due to complications.

DEFINITION Disorder characterised by attention deficit, hyperactivity and impulsiveness for ≥ 6 months which causes moderate psychological, social and/or educational impairment in ≥ 2 important settings (home, school, peers).

AETIOLOGY

Genetic factors: Twin studies 81–67% concordance; first-degree relatives 50% concordance.

Environmental factors: Intrauterine complications, maternal smoking, alcohol and drug abuse during pregnancy.

ASSOCIATIONS/RELATED Depression, anxiety, addictive, obsessional and behavioural disorders (conduct disorder), learning difficulties including receptive and expressive language problems, tic disorders and Tourette syndrome.

EPIDEMIOLOGY $\sim 9\%$ school-age children affected (DSM-IV). M:F = 4:1. Onset < 7 years.

HISTORY AND EXAMINATION

Attention deficit: Inability to sustain mental effort, listen, organise or finish tasks.

Hyperactivity: Fidgeting, running, climbing or talking excessively.

Impulsiveness: Inability to wait their turn, intruding or interrupting others. Some children are predominantly hyperactive and impulsive, while others are principally inattentive.

PATHOPHYSIOLOGY PET scan studies suggest abnormality is of ascending projections of catecholaminergic and serotonergic neurons into the frontal cortex.

INVESTIGATION It is important to evaluate each child comprehensively and take a full developmental, educational and behavioural history. Rating scales such as the Conners Rating Scales and the Strengths and Difficulties Questionnaire (parent, teacher, child) are valuable adjuncts. Exclude medical conditions such as impaired vision and hearing or, less commonly, epilepsy (absence seizures) or hypothyroidism. ECG is warranted prior to starting central nerve stimulants if there is past medical or family history of serious cardiac disease/sudden death or abnormal findings on cardiac examination.

MANAGEMENT

Involvement of health and education: GPs, community paediatricians, educational psychologists, SENCO (special educational needs co-ordinator), social workers, CAMHS.

Child (individual) therapy: Behaviour modification programmes (home, school), CBT, training in social skills, psychotherapy to improve self-esteem.

Parent training/education programme: Parental positive reinforcement of desired behaviour and appropriate negative feedback for unacceptable behaviour.

Drug therapy

Drug treatment should always form part of a comprehensive treatment plan that includes psychological, behavioural and educational advice and interventions.

- *Methylphenidate, dexamfetamine (>6 years):* CNS stimulant which increases arousal in areas of inactivation, thereby improving attention span and reducing impulsivity and hyperactivity. S/E: insomnia, nervousness, headache, \downarrow d appetite, abdominal pain, exacerbation of tics, cardiovascular effects such as tachycardia, palpitations and minor increases in BP. Weight, height and BP should be monitored every 6 months.
- *Atomoxetine (>6 years):* Selective noradrenaline reuptake inhibitor; second-line therapy. S/E: abdominal pain, \downarrow appetite, nausea and vomiting, liver damage (rare), early morning awakening, irritability, mood swings, suicidal ideation, CVS effects, dysmenorrhoea.

COMPLICATIONS ADHD often negatively affects a person's educational achievements. This can contribute to economic, social and life adjustment problems throughout a person's life and lead to substance abuse and crime.

PROGNOSIS Hyperactivity symptoms may improve with maturation and development of self-control. Children with appropriate educational input, support and good compliance with treatment have the best prognosis.

DEFINITION Developmental disorder affecting social interaction, social communication, rigidity of thinking and difficulties with social imagination. Autistic spectrum disorder (ASD) is an umbrella term that ranges from classic autism (severe childhood onset) to Asperger syndrome (milder social impairment with preservation of language development).

AETIOLOGY Genetic factors: 80% concordance in monozygotic twins. RF: maternal rubella infection, ↑paternal age. A link with the MMR vaccine has been proven to not exist.

ASSOCIATIONS/RELATED TS, fragile X syndrome. Epilepsy may be present in 25%.

EPIDEMIOLOGY National Autistic Society (NAS) UK prevalence: 535,000 (2006). ASD is increasing in prevalence.

HISTORY AND EXAMINATION

Motor: Stereotypical and repetitive motor mannerisms (hand flapping, body twirling), clumsy and unco-ordinated movements.

Behavioural: Repetitive activities, ritualistic behaviour, the disruption of which → violent temper tantrums, particular interests/obsessions.

Social: Indifference to others, avoiding eye contact, preferring to be alone, limited facial expressions and understanding of others' gestures, lack of understanding of social rules (e. g. turn taking), attachments to unusual objects, no danger awareness.

Speech: Delayed speech and language development, echolalia, poor comprehension and expression, abnormalities in vocal pitch or rhythm, socially inappropriate comments.

Learning disability: Varies with severity of autistic spectrum.

Other: Sensory abnormalities, difficulty with sleeping, eating, toileting, fears/phobias.

PATHOPHYSIOLOGY Normal children learn social habits without being consciously aware of them. It is these instinctive relations that are disturbed in autistic children. 'They have to learn everything via the intellect' (Hans Asperger).

INVESTIGATIONS

Diagnostic assessments:

1. Strength and Difficulties Questionnaires: parent/teacher and child.
2. School reports/observation in class.

Bloods: Chromosomes for fragile X, FBC, ferritin, thyroid function, lead.

Others: Developmental assessment, EEG (if presenting with fits/funny turns).

Multidisciplinary approach: Should be initiated as early as possible. *Behavioural interventions:* NAS EarlyBird programmes employ the SPELL (Structure, Positive (approaches and expectations), Empathy, Low arousal (calm environment), Links (child, parent and teachers)) and TEACCH (Treatment and Education of Autistic and related Communication handicapped Children) approaches. Other private interventions include Applied Behavioural Analysis and the Son-Rise programme. *Communication support:* SALT work on listening and attention skills, play, social skill and understanding; they may use picture exchange communication system (PECS) or Makaton signing in children with severe language impairment. *Educational support:* portage (preschool children) at home. School-age children may access mainstream school with support (liaison with special educational needs co-ordinator (SENCO)) or special needs school (statement of special educational needs). *Parental support:* charities (NAS, Aspergers UK, Autism UK, Contact A Family), parent partnership services (educational advice), Disability Living Allowance (DLA).

COMPLICATIONS

Physical abuse: Frustrated parents or annoyed social contacts.

Psychological/emotional: May result in aggressive or self-injurious behaviour.

PROGNOSIS Varies with degree of speech development, learning disability and severity of ASD. Adults with severe autism usually live with their parents or require care in special communities. However, many can attend higher education, be employed successfully and lead independent lives.