Differential Diagnosis in Paediatrics

Global developmental delay
Genetic - Downs syndrome, fragile X, Prader-Willi, Williams.
Metabolic - hypothyroidism, PKU.
Perinatal insult - hypoxia, intracranial haemorrhage, teratogens
Neurological - infection (e.g. meningitis, encephalitis), head trauma, hypoglycaemia.
40% chromosomal abnormality, 10% developmental malformations, 4% metabolic cause.

History:
Birth history – for anoxia, prematurity.
Family history – of learning disability.
Developmental history – milestones.
Social history – risk factors e.g. deprivation.
Distinguish delay from regression.

Examination:
Developmental assessment.
Appearance e.g. dysmorphic features of syndromes, skin lesions of neurocutaneous syndromes.
Head circumference.

Delayed speech
Normal variation, deafness, global developmental delay, lack of stimulus, autism, problem with articulation (e.g. cleft palate, CP).

History:
Development – babbling, gurgling, words. Other areas.
PMH – things affecting hearing, cleft palate.
Family history.
Distinguish delay and mechanical problems e.g. cleft palate, CP.

Examination:
Development.
Hearing.

Delayed walking
Normal variation (esp bottom shuffler and commando crawler), cerebral palsy, muscular dystrophy, hip dysplasia, lack of stimulus.

History:
Development: gross motor. Any locomotion. Other areas.
Hip problems.

Examination:
Hips – signs of dislocation e.g. waddling gait, leg length uneven, limited abduction.
Tone, power and reflexes. Gower’s sign for muscular dystrophy.
Locomotion.

Investigations:
Imaging of hips and spine.
CKase for Duchenne muscular dystrophy.
Fever
Exercise or emotion.
Minor infection – URTI, non-specific viral, gastro-enteritis without dehydration.
Major – meningitis, pneumonia, UTI, septicaemia.]
Rarely – neoplasia, thyrotoxicosis, drug reaction, heart failure.

History:
Duration - >1wk suggests TB, malaria, typhoid, autoimmune disorders.
Localising symptoms – cough and coryza (URTI), D and V (GI tract, vomiting alone is less specific), painful limb, lower abdo pain (UTI, lower lobe pneumonia), meningism (headache, photophobia, neck pain). Probably none in infants.
Recent foreign travel.

Examination:
Systemically unwell – if well unlikely to be septic.
Local signs of infection – ENT, bulging fontanelle.
Rash.

Investigations and management:
Markers of inflammation e.g. WCC, CRP.
Septic screen – blood cultures, urine, throat swab, CSF, CXR.
In infants or those looking ill, usually start antibiotics before results. Antipyretics prevent against febrile convulsions.

Rash
Infection – viral, toxin related, streptococcal, meningococcal, scabies.
Dermatitis – eczema, vasculitis.
Allergy – drug related, urticaria.

Scalp – seborrhoeic dermatitis, eczema, psoriasis, fungal.
Flexor – eczema.
Extensor – psoriasis, Henoch-Schonlein purpura.
Web spaces – scabies.
Trunk – viral xanthems, molluscum.
Mucous membranes – measles, Kawasaki disease, SJ syndrome, herpes.

Maculopapular (viruses and drugs) – measles (prodrome, Koplik's spots in mouth), rubella (pink on face, lymphadenopathy), Kawasaki disease (protracted fever, red lips, conjunctivitis, lymphadenopathy), scarlet fever (face, strawberry tongue).
Vesicular – chicken pox, eczema herpeticum.
Haemorrhagic – meningococcal septicaemia (unwell, petechial), leukaemia (pallor, hepatosplenomegaly), ITP (looks well, bruising, epistaxis), Henoch-Schonlein purpura (legs and buttocks, arthralgia, abdo pain), NAI.

History:
HPC – duration, site, spread, persistent or comes and goes (urticaria), itch (eczema, scabies), drug ingestion or provocative agents, other family members affected (viral, infestations), associated symptoms, history of skin conditions.

Examination:
Other features – fever, mucous membranes, lymphadenopathy, splenomegaly, arthropathy.
Describe – morphology, arrangement, distribution
**Short stature**

Constitutional, endocrine pathology (GH deficiency, hypopituitarism, hypothyroidism, Cushing’s), bone dysplasia (achondroplasia), systemic illness (CHD, CF, CP, chronic renal failure), psychosocial, genetic syndromes (Turner’s, Silver-Russel, Down’s, Noonan’s), nutritional.

**History:**

Early childhood illness.
Parental heights – midparental height is average of parent’s heights +7 for boys, -7 for girls.
Target centile range is 10cm either side in boys, 8.5cm either side in girls.

**Examination:**

Height and growth velocity.
Dysmorphic features – Turner’s (neck webbing, wide spaced nipples), Prader-Willi (obesity, hypotonia, small testicles).
Visual fields (pituitary tumour).
Stage of puberty.

**Investigations:**

Bone age (early puberty with premature bone fusion or delayed with delayed growth spurt).
Karytype (Turner’s).
Skeletal survey (dysplasias).
Endocrine investigations – thyroid function, growth hormone provocation test.
Skull Xray (craniopharyngoma).

**Abdominal pain**

Acute: medical (abdominal and systemic) – colic, constipation, mesenteric adenitis, gastroenteritis, hepatitis, UTI, DKA, sickle cell, lower lobe pneumonia, HSP. Surgical – appendicitis (migrate from centre to RIF), intussusception (episodic), ovarian or testicular torsion (see on examination), strangulated inguinal hernia (groin mass), volvulus.

Recurrent – usually functional. Rarely, UTI, obstructive uropathy, food intolerance IBD, ulcer, malrotation, pancreatitis, celiac disease, CF, porphyria, lead poisoning.

**History:**

Babies – drawing up legs, screaming.
Duration - >4hrs usually significant.
Location – further from umbilicus is usually significant.
Nature – intermittent, constant.
Other symptoms – vomiting (gastroenteritis, obstruction), bloody stools (intussusception in infant, IBD in older child), dysuria (UTI), cough (pneumonia).

Functional recurrent abdo pain – periumbilical, worse on waking, short-lived, no appetite loss, family history of functional illnesses, healthy thriving child.

**Examination:**

Check for fever, jaundice (hepatitis), rash (HSP), respiratory tract (RLL pneumonia), hernial orifices, genitalia.

**Investigations:**

FBC and WCC (bacterial infection), sickling test, U&Es (electrolyte disturbances before surgery if vomiting), glucose.
Urinalysis – glucose, ketones, infection.
AXR (constipation, renal calculi, obstruction).
USS (obstructive uropathy, appendix mass, intussusception).
**Intestinal obstruction**
Neonate – congenital malformation of gut e.g. atresia, malrotation, volvulus, exomphalos, gastrochisis, Hirschprung’s, imperforate anus, CF, NEC.
Young child – intussusception, volvulus, strangulated hernia.
Older child – strangulated hernia.

**Constipation**
Baby – inadequate fluid intake, overstrength formula, change to cow’s milk.
Infants – normal, simple, lack of fibre or water, painful anal fissure, Hirschprung’s, CP, hypothyroidism, hypercalcaemia, renal tubular disorders.

**History:**
Frequency and consistency.
Pain or blood.
Soiling (overflow, simple constipation).
History of delay in passing meconium (Hirschprung’s).

**Examination:**
Failure to thrive and dehydration.
Abdominal distension (Hirschprung’s) or palpable colon (simple).
Anal fissure.
Rectal loading.

**Diarrhoea**
Chronic - toddler’s diarrhoea, malabsorption (steatorrhoea), lactose intolerance, coeliac disease, cystic fibrosis, IBD, parasitic infection, constipation with overflow.
Acute – viral gastroenteritis (rotavirus, SRSV, adenovirus), bacterial gastroenteritis, suggested by high fever (Ecoli, campylobacter, salmonella, shigella), protozoo (giardia, entamoeba, cryptosporidium).
Bloody – infective e.g. campylobacter, shigella, amoeba, intussusception, haemolytic uraemic syndrome, UC.

**Examination:**
Signs of malabsorption – anaemia, think, wasting, abdominal distension.

**Investigations:**
Stool microscopy and culture, test for reducing substances, tests for malabsorption (e.g. Hb), jejunal biopsy (celiac disease), sweat test (CF).

**Vomiting**
Persistent in a baby - pyloric stenosis, GORD, overfeeding, systemic illness, lactose intolerance.

Neonate – regurgitation (normal), systemic infection, inborn errors of metabolism, bowel obstruction (duodenal atresia, volvulus, strangulated hernia, meconium ileus in CF, Hirschprungs’s), tracheo-oesophageal fistula, NEC, CAH.
Infant – reflux, gastroenteritis, RTI, UTI, meningitis, pyloric stenosis, intussusception.
Older children – infections, appendicitis, raised ICP, migraine, eating disorders.

Haematemesis in a neonate – swallowed maternal blood (predelivery or cracked nipple), trauma from a feeding tube, haemorrhagic disease of the new born.
Haematemesis in children – liver disease, oesophagitis, gastritis.
**History:**
Bile or blood – need to investigate urgently.
Duration.
Associated symptoms – fever, abdo pain, constipation, diarrhoea.
Last wet nappy. Feeding.

**Examination:**
Dehydration.
Fever, abdominal distension, hernial orifices.

**Painful joints**
Monoarthritis - reactive arthritis, juvenile chronic arthritis (chronic pain and swelling), septic arthritis (fever, immobile), haemophilia, trauma.
Polyarthritis – JIA, SLE, HSP, viral, rheumatic fever.

**History:**
Acute or insidious.
One or multiple joints.

**Limp**
‘Abnormality of gait’

**History:**
Presence of pain.
Duration – chronic pain is unlikely to be infection.
Prodromal illness.

**Examination:**
Observe walking if possible.
Fever (infection), rashes, range of movement, point tenderness, unequal leg length, spinal abnormality, neurological signs (tone, power, reflexes).

**Investigations:**
Xray, USS (hips), isotope bone scans, acute phase reactants, blood cultures.

**Painful limb**
Growing pains, osteomyelitis, fracture or trauma, any febrile illness, sickle cell crisis, haemophilia, malignant deposits (worse at night), rickets.
May present as pseudoparalysis in infants.

**History:**
Onset – acute with trauma, osteomyelitis, sickle crisis.
**Bleeding disorder**
Can cause bleeding into skin, epistaxis, haemarthrosis, haematuria.

Vascular defects – heriditary haemorrhagic telaniectasia, Ehler Danlos syndrome, HSP (most common, purpura on buttocks and lower limbs), scurvy, meningococcal septicaemia. Platelet defects – ITP (bruising in well child), DIC, HUS, marrow failure, drug induced e.g. aspirin. Coagulation defects – haemophilia (male), von Willebrand disease, vit K defiency (haemorrhagic disease of the newborn, malabsorption, liver disease), anticoagulant drugs.

**History:**
Inherited or acquired, family history.
Age of onset.
Previous haemostatic challenges e.g. operation.
Site and type of bleeding.

**Investigations:**
Blood film, liver and renal function, platelet count, coagulation screen.

**Anaemia**
Decreased red cell production, increased red cell breakdown, blood loss.
Also pallor due to vasoconstriction.

**History:**
Chronic disease esp renal.
GI symptoms.
Dietary history (insuffient iron is most common).
Family history e.g. of sickle cell, thalassaemia, spherocytosis.

**Examination:**
Ethnicity – Afro-Caribbean (sickle cell), Mediterranean and Asian (thalassaemia). Jaundice (haemolysis), bruising (marrow failure), splenomegaly (haemolysis).

**Hypoglycaemia**
Metabolic – liver disease, inborn errors of metabolism e.g. glycogen storage diseases, ketotic (fasting).
Hormonal – hyperinsulinaemia (treated DM, islet cell adenoma), Addison disease, CAH, panhypopituitarism, GH deficiency.

Changes in EEG at <2.6mmol/l, always send for lab glucose as well as capillary prick test.

**Management** – sugary drink, if unconscious 10% dextrose.
Jaundice
Neonatal in first 24 hours – excess haemolysis (rhesus or ABO incompatibility, G6PD, pyruvate kinase deficiency, spherocytosis), congenital infections.
Neonatal 2d-2wks – physiological, breast milk jaundice, Gilbert’s syndrome (common, mild hepatic enzyme defect), infection, haemolytic disease, resolving haematoma.

Child – infectious hepatitis esp hep A (also EBV, malaria, leptospirosis), injury due to drugs (e.g. halothane), Gilbert syndrome (hepatic enzyme defect), haemolysis (G6PD deficiency, spherocytosis, HUS, malaria), biliary obstruction (e.g. CF).

Investigations: for causes. Bilirubin levels, conjugated or unconjugated, liver enzymes, FBC, urine culture, viral titres, liver USS, liver biopsy.
For neonatal conjugated – screen for infection (e.g. hepatitis) and genetic causes (e.g. A1AT and galactosaemia. USS of bile ducts, if dilated then cholangiogram (choledocal cyst), if normal or not seen then radionucleide scan. If get excretion then patent biliary tree (need liver biopsy), if no excretion then suspect biliary atresia.

Hepatosplenomegaly
Hepatomegaly with jaundice: biliary atresia, infective hepatitis.
Hepatomegaly without jaundice: cardiac failure, hyperinflation of lungs.
Hepatosplenomegaly: advanced liver disease, leukaemia, thalassaemia, rare storage disorders.
Splenomegaly – neonates and thin children (tip palpable), acute infection (e.g. CMV, EBV, septicaemia), malignancy (lymphoma, leukaemia), haemolytic anaemia (spherocytosis, G6PD deficiency), haemoglobinopathy, ITP, collagen diseases (JCA, SLE). Also consider non-spleen e.g. Wilms tumour, hydronephrosis, neuroblastoma.

Neck swelling
Lateral – acute lymphadenitis (short history, inflammation), TB, malignant nodes (non-tender, rubbery, other signs), branchial cyst (middle third of SCM, transilluminates), haemangiomia (bruit, may get cardiac failure or platelet consumption), lymphangioma (transilluminable, rapid enlargement, soft), sternomastoid tumour (neonate, swelling in muscle, no inflammation, torticollis), salivary gland (pain with eating).

Midline – lymph nodes (regional infection), dermoid cyst, thryroglossal cyst, ectopic thyroid, goitre.

Lymphadenopathy
Generalised – infection (EBV, TP, CMV, HIV), haemotological malignancy, immunological (JCA, sarcoidosis, Kawasaki).

History:
Duration - <4wks usually infection, >1yr unlikely to be neoplastic.
Constitutional symptoms e.g. weight loss, fever, rash.
Pets (cat scratch fever, TP).

Examination:
All nodal sites.
Size, mobility, tenderness (bacterial adenitis), drainage region, abdomen.

Worrying lymph nodes – rapid growth, skin ulceration, fixation, >3cm, persist.

Investigations:
If general – FBC, EBV serology, CXR, abd USS, bone marrow aspiration, biopsy.
**Collapse or coma**
Infective – septicaemia, meningitis.
Metabolic – hypoglycaemia, DKA, inborn errors of metabolism, liver or renal failure.
Poisoning or trauma.
Neurological - head injury, post-ictal, intracranial tumour or haemorrhage.
Congenital heart disease.

**Management:**
ABC, temperature, blood glucose, AVPU/GCS, signs of injury, fever or rash, focal neurological signs.
Signs of coning – hypertension, bradycardia, irregular respiration (Cushing’s response).
Don’t do lumbar puncture as raised ICP likely.

**Seizures**
Firstly, distinguish true seizures from others. In infants – jitteriness (doesn’t have fast and slow component), benign myoclonus (jerks when asleep), apnoea, reflux. Toddlers - breath holding attacks, reflex anoxic seizures, rigors. Children – syncope, tics, migraine, tantrums, heart block, arrhythmia.

Seizures - epileptic fit (one off or recurrent), febrile convulsion, hypoglycaemia, meningitis, encephalitis, head injury, hyponatraemia, cerebral tumour.
In the neonate – encephalopathy (hypoxic, bilirubin), intracranial haemorrhage, infection, congenital abnormality, hypoglycaemia.

**History:**
Provoking factors – when and where.
Episode – loss of consciousness, abnormal movements, altered tone, pallor or cyanosis, eye movements, duration of episode.
Previous history – birth, developmental delay, head injury, family history.

**Examination:**
Usually normal in epilepsy.
Skin – neurocutaneous syndromes e.g. tuberous sclerosis, neurofibromatosis (look with Wood’s light).
Optic fundi – fundal changes with congenital infections or neurodegenerative diseases.
If actively convulsing – ABC, fever, fontanelle (raised ICP), meningism, optic fundi, focal neurological signs, level of consciousness.

**Investigations:**
EEG – identify a particular epilepsy syndrome or underlying lesion. A single interictal EEG will be normal in 50% children with epilepsy and can be abnormal in normal children. Can get more from ambulatory monitoring or recordings after sleep deprivation.
Neuroimaging – for partial seizures, intractable seizures, focal neurological deficit, evidence of neurocutaneous syndrome.
Investigation of underlying neurometabolic disorder.
Epilepsy is diagnosed from a history from a witness rather than investigations.
**Recurrent headaches**
Tension headache, migraine, sinusitis, refractive errors, hypertension, raised ICP, brain tumour.

**History:**
Site, intensity, duration and frequency, provoking factors, associated symptoms (e.g. weakness, paraesthesia, nausea).

**Examination:**
Blood pressure and radiofemoral delay (coarctation), visual acuity (refractive error), papilloedema (raised ICP), focal neurological deficit (tumour).

**Bed wetting**
Primary nocturnal enuresis, nocturnal epilepsy, UTI, neuropathic bladder, psychological.
May be due to polyuria – diabetes, UTI, excessive drinking, chronic renal failure.

**History:**
Frequency, time of night.
Daytime symptoms – urgency or wetting (suggests underlying cause).
Emotional stresses.

**Examination:**
Any evidence of neurological problem e.g. hairy patch.
Enlarged bladder
Urinalysis.

**Daytime enuresis**
Psychological (acute onset, previously continent), infection, neurological (continual since birth), ectopic ureter (continuous dribbling since birth), urethral obstruction, sphincter damage.
If secondary likely to be psychological or due to UTI or polyuria e.g. diabetes.

**School absence**
Illness (90%).
Truancy – mostly lower socio-economic class boys with poor academic records.
Parental refusal.
School-refusal – pre-pubertal children, normal intelligence, shy.
Separation anxiety.
School phobia.

**Hypertension**
Primary – becomes more common the older the child (about 50% in teenagers).
Renal (most) – chronic pyelonephritis, hydronephrosis, tumours, chronic glomerulonephritis, renal artery stenosis.
Other – coarctation of the aorta, Cushings, CAH, phaeochromocytoma, neuroblastoma, thyrotoxicosis.

**History:**
Symptoms of renal or endocrine disease.
Symptoms of malignant hypertension.

**Examination:**
Malignant hypertension – retinal haemorrhage, papilloedema, heart failure.
Renal masses, renal artery bruit.
Goitre.
Femoral pulses.
**Investigations:**
Renal – urinalysis, ultrasound kidneys, IVP, kidney function tests.
Endocrine – serum thyroxine, electrolytes, urine VMA and cortisol.

**Proteinuria**
Mostly benign – fever, exercise, orthostatic.
Nephrotic syndrome, UTI, glomerular nephritis.

**Examination:**
Nephrotic syndrome suggested by oedema, ascites, pleural effusions.

**Investigations:**
Early morning protein:creatinine ratio, renal function, plasma albumin, MSU, throat swab.

**Haematuria**
Either frank (naked eye) or on strip. Confirm by microscopy (as strips positive in myoglobinuria and dyes may discolour urine).

Glomerular (also white cells and protein) - acute glomerular nephritis: post-streptococcal, HSP, IgA nephropathy, Alport syndrome.
Non-glomerular - UTI, Wilm's tumour, schistosomiasis, trauma, stones, bleeding disorders, transient benign haematuria (diagnosis of exclusion).

**History:**
Duration and recurrence.
Urinary symptoms (suggest UTI).
Loin pain (pyelonephritis).
Foreign travel (schistosomiasis).
Sore throat (post-streptococcal).
Family history (Alport syndrome).

**Examination:**
Fever (UTI, pyelonephritis), oedema (nephrotic syndrome), hypertension (nephritis, scarring), rash and joint swelling (HSP), bruises (ITP), abdo mass (Wilm's tumour).

**Respiratory difficulties in neonate**
Airway – choanal atresia, macroglossia, micrognathia, goitre, subglottic stenosis, cord paralysis, laryngomalacia, tracheoesophageal fistula.
Lung problems – meconium aspiration, RDS, pneumothorax, transient tachypnoea of newborn, pneumonia.
Malformations – diaphragmatic hernia, pulmonary hypoplasia, CCAM.
Non-pulmonary – excessive air swallowing, septicaemia, anaemia, CHD, persistent fetal circulation

**Wheeze**
Viral LRTI e.g. bronchiolitis (infants), asthma (children over 2), reflux, foreign body, pneumonia (fever), congestive cardiac failure, CF (other problems), GO reflux (vomiting), central airways disease (also stridor), postviral wheeze.

**History:**
Pattern of symptoms.
Family and personal history of atopy.
Trigger factors.
Estimation of severity – e.g. exercise tolerance, sleep disturbance, school absense.
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Cough
Chronic – CF (productive, other symptoms), TB, post-viral cough receptor sensitivity, asthma (most common, usually at night and also wheeze), whooping cough, inhaled foreign body, primary ciliary dyskinesia (productive), reflux.
Acute – usually URTI (coryzal symptoms), bronchiolitis (<1y, wheeze), pneumonia (fever, dyspnoea), foreign body (acute onset). Be aware of barking cough of croup and paroxysmal prolonged bouts of coughing in pertussis.

History:
Duration - >3wks is chronic.
Type of cough – dry, productive (may suggest CF).
Presence of wheeze (asthma).
Trigger factors e.g. smoking, animals.

Stridor
Acute - croup, epiglottitis, inhaled foreign body, bacterial tracheitis, angioedema.
Persistent – laryngomalacia, anatomical e.g. vascular ring lord of the ring-Aragorn son of Arathorn.

Breathlessness
Pneumonia, bronchiolitis, asthma.

Microcephaly
Normal child, severe mental retardation, anencephaly, craniosynostosis.

Big head
Normal child, gigantism, hydrocephalus, megalencephaly (e.g. storage diseases), subdural haematoma, intracranial tumour.

Inconsolable crying infant
Colic, otitis media, incarcerated hernia, UTI, anal fissure, intussuception.

Swollen testes
Testicular torsion, hydrocoe, torsion of testicular appendix, epididymo-orchitis, idiopathic scrotal oedema, trauma “kick in the balls”.

Important Note
These notes were written by Liz Tatman, as a fourth year medical student in 2006. They are presented in good faith and every effort has been taken to ensure their accuracy. Nevertheless, medical practice changes over time and it is always important to check the information with your clinical teachers and with other reliable sources.
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