Fragile X Syndrome

= most common cause of sex-linked general learning disability

Prevalence ~2.13/10,000

Presentation:

Usually diagnosed before age 1

- Learning difficulties
- Delayed milestones
- High forehead
- Large testicles
- Facial asymmetry
- Large jaw
- Low voice
- Obsessiveness and anxiety can occur
- Echolalia and perseveration can occur

Investigations:

- Southern blotting and PCR on blood tests
- FMRP antibody testing via blood smear (to detect affected males)

Management:

- Genetic counselling
- Special needs education
- Behavioural therapy
- Speech therapy

Life expectancy is unaffected
• >3 weeks – prolonged/persistent neonatal jaundice

Unconjugated

□ Breast milk
□ Infection
□ Hypothyroidism though should be picked up on Guthrie heel prick

Conjugated

□ Biliary atresia all/part of biliary tree not developed – prevents bile into gut, chalky stools
□ Neonatal hepatitis syndrome

• Look out for signs of kernicterus = bilirubin in very high concentrations can cross blood brain barrier and deposit in basal ganglia and brainstem → neurological deficits, coma, death

Investigations

• Thorough history and examination
• Bloods
  □ Serum bilirubin
  □ FBC
  □ Blood film
  □ Blood group
  □ LFTs
  □ TFTs
  □ Blood cultures (TORCH screen)
  □ Coomb’s test
• Urine dipstick (for bilirubin) and culture (for sepsis)
• If prolonged or indicated by other investigations, US or other imaging of biliary tree

Management

• Correct dehydration and poor intake
• Phototherapy with 450nm wavelength blue-green light to convert unconjugated in harmless pigment
• If unsuccessful, exchange transfusion
Diabetes mellitus

The majority of paediatric diabetes is Type I diabetes mellitus.

**Type 1 diabetes**
= inability to produce insulin due to autoimmune destruction of pancreatic beta cells.
= progressive condition occurring in genetically susceptible individuals

**Signs and symptoms**
- Hyperglycaemia
- Glycosuria
- Polydipsia
- Unexplained weight loss
- Nonspecific malaise
- Ketoacidotic symptoms – nausea, abdo pain, SOB → tachycardia, hyperventilation, hypotension, ketone breath, confusion

**Diagnosis**

Capillary blood glucose
- Fasting plasma glucose 7mmol/L or
- 2 hour plasma glucose 11mmol/L during 75g OGTT or
- Random plasma glucose 11mmol/L in patient with symptoms of hyper

HbA1c (glycated haemoglobin)
- Normal non diabetic: 20-42mmols/mol
- Low: <42mmols/mol
- Ideal diabetic control: 42-53mmols/mol
- Acceptable control: 53-64mmols/mol
- Needs improving: 64-75mmols/mol
- Not acceptable: >75mmols/mol
- back (coarctation of the aorta)
- **Quality**
  - Musical/harmonic (vibratory)
  - Noisy/dissonant (non vibratory)
- **Pitch**
  - Low pitched best hear with bell, high pitched with diaphragm
- **Manoeuvres**
  - Supine → standing (decreases blood pressure, increased HR but lower stroke volume so systolic murmurs decrease in intensity. Hypertrophic cardiomyopathy increases in intensity).
  - Standing → squatting (muscle contraction → increased preload but also vascular resistance → most systolic murmurs increase in intensity. Hypertrophic cardiomyopathy decreases in intensity).
  - Valsalva (raises intrathoracic pressure, decreases venous return and therefore stroke volume → compensatory increase in HR leading to similiary findings to supine→standing).
History

**straining does not necessarily indicate constipation**

- When symptoms began
- Associated symptoms – abdominal pain, pain with defecation, systemic symptoms, soiling, enuresis
- Stools
  - Frequency
  - Amount
  - Appearance – colour, texture, blood
- Meconium (no meconium by 48hrs suggests Hirschprung’s/CF/congenital malformation)
- Failure to thrive (Hirschsprung’s, malabsorption, CF, metabolic)
- Toilet training – age
- Diet
- Family history
- Past medical history
- Developmental and growth history
- Psychosocial history

Examination

- General observation
  - Ill or well?
  - Weight, height appropriate?
- Abdominal exam
  - Sensory/motor perianal deficits
Enuresis and wetting

Enuresis = involuntary discharge of urine in the absence of organic disease at an age when expected to be dry

By 3-5 years, should have developed ability to inhibit voiding unconsciously and voluntarily.

Intervention advised at 7 years+

- Primary enuresis – never been dry
- Secondary enuresis – period of continence for >6months

- May be nocturnal or diurnal

Primary nocturnal enuresis is common.

- More common in boys
- More common in first borns
- May be due to
  - Genetic component
  - Maturational delay
  - Emotional stress
  - UTI
  - Reduced ADH
  - Diabetes or renal disease

Secondary enuresis:

- Often due to **emotional stress**
- UTI
- Diabetes
- Threadworm

Diurnal enuresis:

- UTI
- Neurogenic bladder (spina bifida, CP, tumour)
- Severe constipation
- Congenital abnormalities (e.g. ectopic ureter)
- Psychogenic
- **Sexual abuse**
- Physiological (urgency)
Epiglottitis

=inflammation of structures above insertion of glottis (usually due to bacterial infection)

- Most common between ages 2-8 but can occur at any age
- Now rare due to Hib vaccination
- Acute epiglottitis associated with upper airway obstruction → respiratory arrest → death

- Bacteria: Hib, penumococci, pseudomonas, TB
- Viruses: HSV
- Candida and aspergillus in immunosupressed

In child, epiglottis more anterior and superior than adult, and at greater angle with trachea

Bacteria penetrate mucosal barrier → invasion of bloodstream and seeding of epiglottis and surrounding tissues → acute onset inflammatory oedema → reduced airway aperture → airway obstruction/aspiration or oropharyngeal secretions/distal mucous plugging → respiratory arrest

Presentation

- Abrupt onset of severe symptoms
- Fever usually first symptom
- Rapidly followed by stridor, laboured breathing
- Hoarse voice, sore throat
- Clinical triad of drooling + dysphagia +distress

Examination

- Child appears toxic
Inherited/genetic

**Trisomy 21** – can suffer from cataracts, strabismus, blepharitis, conjunctivitis, myopia, hypermetropia

**CHARGE** – rare genetic disorder (Ch8) resulting in sensory, physical, developmental problems **Coloboma (hole in an eye structure)**, **Heart Atresia choanae Retardation GI/genitourinary Ears**

Congenital

**Cataract** – lens opacity present at birth, 1/3 isolated trait, 1/3 component of syndrome/disease, 1/3 undetermined cause

**Albinism** – lack of melanin results in non-degenerative visual impairment (due to improper crossing of optic nerve fibers, lack of melanin retinal pigment epithelium, decreased ability of iris to absorb light) and nystagmus

**Retinoblastoma (rubella, CMV)** – sporadic or familial tumour of retina, absent red reflex, visual deterioration, good prognosis

**Congenital infection (TORCH)** – particularly lead to chorioretinal scar

Antenatal/perinatal

**Retinopathy of prematurity** – 20-60% of VLBW infants, associated with arterial hyperoxia and retinal ischaemia before 32 weeks gestation so important to prevent hyperoxia in infants requiring O₂

**Hypoxic ischaemic encephalopathy** – resulting from perinatal cerebral hypoxia

**Cerebral damage** – causing cortical blindness/cortical visual impairment

**Optic nerve hypoplasia** – underdevelopment of optic nerve(s) (risks include young maternal age and primiparity) --> vision problems, nystagmus, strabismus

Post-natal

**Trauma** – to eye or head

**Infection** – measles, external eye infection

**Juvenile idiopathic arthritis**

**Vitamin A deficiency** – due to Vitamin A’s role in phototransduction and immunity, usually in developing countries/malnourished children,