Case 3

- Hepatoblastoma, combined fetal and embryonal epithelial subtype
Hepatoblastoma

- Wholly epithelial
  - Pure fetal
  - Mixed fetal / embryonal
    - Small cell undifferentiated (may show rhabdoid features and be INI1-negative)

- Mixed epithelial / mesenchymal
  - Without teratoid features (fibrous/osteoid/cartilage)
  - With teratoid features (diverse heterologous components incl. endodermal/neuroectodermal (melanin, glial, neuronal)/skeletal muscle)
Case 14

- 6 week old boy, jaundice, abnormal LFTs
Case 14

- Extrahepatic biliary obstruction (extrahepatic biliary atresia) with cirrhosis
Extrahepatic biliary atresia

- 2 forms
  1. Embryonic/fetal (10-35%, presents at birth)
  2. Perinatal (65-90%, jaundice free period of several weeks)

- Aetiology unclear
  - ? Viral
  - ? Autoimmune
  - ? Cilia defect
  - ? Multifactorial

- Diagnosed on liver biopsy (core vs open at time of surgery)
Extrahepatic biliary atresia

- **Histology:**
  1. Bile ductular proliferation
  2. Inflammation (lymphocytes +/- neutrophils)
  3. Fibrosis $\rightarrow$ cirrhosis
  4. Hepatocellular effects: cholestasis, feathery degeneration, focal giant cell change

- NB Similar changes in choledochal cyst, alpha-1-antitrypsin defic., TPN cholestasis

- Correlate with clinical, imaging findings
Case 19

- 8 week old boy, abnormal LFTs
Case 19

- Neonatal (giant cell) hepatitis
Neonatal giant cell hepatitis

- Cholestatic liver disease
- May be associated with wide range of disorders:
  - Infectious
    - Viral, bacterial, toxoplasma
  - Metabolic
    - Glycogen storage disease
    - Lipid storage disorders
    - Alpha-1-antitrypsin deficiency
  - Toxic
    - TPN
  - Anatomic
    - Extrahepatic biliary atresia
    - Sclerosing cholangitis
- Idiopathic form most common (25-40%, diagnosis of exclusion)
  - Sporadic (85-90%, good prognosis)
  - Familial (10-15%)
Neonatal giant cell hepatitis

- Cholestasis (esp zone 3)
- Giant cell transformation
- Ballooning
- Acidophilic necrosis
- Pseudoglandular/acinar formation
- Inflammation usually sparse
- Bile ducts normal
- Usually not much fibrosis
Case 19

- Giant cell hepatitis, alpha-1-antitrypsin deficiency
Case 4

- Neonate, stoma reversal
- Perinatal volvulus
Case 4

- Inspissated secretions, consistent with cystic fibrosis
Cystic fibrosis (CF)

- Manifestations of CF in routine paediatric surgical pathology specimens:
  1. Nasal polyps
  2. Intestinal biopsies/resections (esp if history of meconium ileus, neonatal volvulus, intestinal atresia)

- Always look for inspissated brightly eosinophilic secretions in such specimens when patient is a child
- Chase newborn CF screening results
Case 8

- Ex 25 week prem, now 29 weeks. Acute abdomen.
Case 8

- Necrotising enterocolitis
Necrotising enterocolitis (NEC)

- Seen in premature babies, low birthweight infants
- Mortality 20-40%
- Difficult to diagnose early (signs non-specific)
- May present in fulminant fashion with extensive bowel involvement, perforation, shock
- Sequelae: strictures, short gut syndrome (if extensive resection required)

Pathology is that of ischaemic enteritis/colitis
- Mucosal ischaemia (lamina propria haemorrhage, surface necrosis, inflammation)
- Mural necrosis
- Transmural infarction
- Pneumatosis (but not always)
Case 11

- 4 day old boy, abdominal distention, failure to pass meconium
This gives away the diagnosis!
Case 11

- Hirschsprung’s disease
Hirschsprung’s disease (HSCRD)

- Developmental disorder
- Failure of neural crest cells to migrate/proliferate/differentiate during development of enteric nervous system
- Absence of ganglion cells in distal rectum and variable length of proximal contiguous bowel

- 2 clinical presentations:
  - Failure to pass meconium in a neonate (may be a surgical emergency)
  - Chronic constipation in older child (or even adult, rarely)
Hirschsprung’s disease

- Diagnosis involves 3 modalities:
  1. Serial H&E sections on an adequate rectal biopsy looking for submucosal ganglion cells
  2. Acetylcholinesterase staining on frozen section (positive reaction is abnormal)
  3. Calretinin immunohistochemistry (negative reaction is abnormal)

- Neonates: suction rectal biopsy
- Older children/adults: strip rectal biopsy
### Hirschsprung’s disease

<table>
<thead>
<tr>
<th></th>
<th>Rectal biopsy histology</th>
<th>Acetylcholinesterase (Frozen section)</th>
<th>Calretinin (IHC)</th>
</tr>
</thead>
<tbody>
<tr>
<td><strong>Normal</strong></td>
<td>Submucosal GCs present</td>
<td>Negative</td>
<td>Positive</td>
</tr>
<tr>
<td><strong>HSCRD</strong></td>
<td>No submucosal GCs *</td>
<td>Positive</td>
<td>Negative</td>
</tr>
</tbody>
</table>

*Minimum of 60 consecutive serial sections (NOT levels) in an adequate biopsy (ie all must have adequate submucosa)*
Acetylcholinesterase
Calretinin
Case 13

- 3 year old girl, rectal bleeding, ?juvenile polyposis
Case 13

- Intestinal ganglioneuromatosis
Intestinal ganglioneuromas

- Single/multiple polyps (usually sporadic, indolent)

- Diffuse, murally extending – high association with MEN2B, NF1

- Correlate with clinical features, endoscopy, imaging to define extent

- Genetic evaluation
6 year old boy, recurrent lip / gingival swelling, and oral mucosal tags. Intraoral bx.