Case 16

- Orofacial granulomatosis
Orofacial granulomatosi

- Presents with swelling of the lips, adjacent cheeks
- May be associated with facial nerve palsy, fissured tongue (Melkersson-Rosenthal syndrome)
- **May be associated with Crohn’s disease, or may pre-date Crohn’s**

- Histology:
  - Oedema
  - Perivascular mixed inflammatory cell infiltrate
  - Poorly formed granulomas
  - Fibrosis
Case 20

- 2 year old boy, diarrhea, lung mass, colonic biopsy
Case 20

- Granulomatous colitis
- Secondary to chronic granulomatous disease
Chronic granulomatous disease

- Genetically heterogeneous (X-linked, auto recessive) immunodeficiency disorder of neutrophil function
- Defect in NADPH oxidase (can’t generate bactericidal oxidative metabolites in neutrophils)
- Suspect in:
  - Young children with history of recurrent infections (bacterial, fungal)
  - Esp when the granulomas are “too good to be true”

Initial diagnostic screening test:

Treatment:
- Interferon
- Bone marrow transplant
Chronic Granulomatous Disease
NADPH Oxidase Deficiency

- X-linked CGD: 60% of cases
- Autosomal recessive CGD: < 5%

From Elsori, ResearchGate
Case 15

- 9 year old boy, posterior fossa mass
Case 15

- Medulloblastoma, classic subtype, WHO Grade IV
Brain tumours in children

- Vast majority are **primary** CNS tumours
- Metastases very rare
- Before assessing, need to know:
  - Age (infantile vs older)
  - Location (supratentorial vs infratentorial; cortical/dural based vs deep)
  - Imaging (this is your macro)
  - Associated conditions (eg tuberous sclerosis, neurofibromatosis)

- Histological classification AND genetic classification
Brain tumours in children

- Pilocytic astrocytoma
- Medulloblastoma
- Diffuse midline glioma
- Glioneuronal tumours
- Diffuse astrocytoma
- Supratentorial embryonal tumours
- Ependymoma
- Meningioma
- Craniopharyngioma
- Teratoma
Medulloblastoma

- 4 Histologic subtypes:
  1. Classic
  2. Desmoplastic/nodular
  3. Medulloblastoma with extensive nodularity
  4. Large cell anaplastic

- 3 Main molecular subtypes:
  1. WNT-activated
  2. SHH-activated
  3. Non-WNT/non-SHH (Group 3, Group 4)
RETICULIN
Case 9

- 11 year old boy, intraventricular brain tumour.
Case 9

- Subependymal giant cell astrocytoma, WHO Grade I
Subependymal giant cell astrocytoma (SEGA)

- WHO grade I
- Lateral walls of lateral ventricles
- Strong association with tuberous sclerosis
- Looks very scary on frozen section (esp if not given hx)

Histology:
- Solid, calcified
- Large cells (gemistocytic-like, ganglion cell-like)
- Multinucleated cells
- Mitoses/necrosis/endothelial proliferation may be seen
Case 5

- 15 year old boy, expansile lesion right maxilla
Case 5

- Fibrous dysplasia
Fibrous dysplasia

- Proportion of fibrous and osseous tissue may vary
- **Submit plenty of blocks**
- Osseous component: irregular, curvilinear, trabecular. *May see surrounding osteoblasts*
- May see nodules of cartilage
- **Look at radiology carefully (esp in long bone fracture with history of minimal trauma)**
  - Compare with other comparable bones, radiological findings can be very subtle (ground class appearance to medulla)
Case 18

- 11 year old boy, scapula mass
Case 18

- Ewing sarcoma
Ewing sarcoma

- Prototypical small round blue cell tumour
- Usually completely undifferentiated cells BUT
- Can see subtle morphological variations
  - Larger cells with nucleoli
  - Neuroectodermal differentiation (rosette-like structures)

- Genetics:
  - Demonstration of rearrangement of EWSR1 gene confirmatory
  - 85% t(11;22)(q24;q12) EWSR1-FLI1
  - Remainder have different partner genes (ERG, ETV1, ETV4 etc)
Case 26

- 12 year old boy, painful lesion middle phalanx, index finger
Case 26

- Osteoid osteoma
Osteoid osteoma

- Small size (but painful – relieved by NSAIDs)
- Any bone
- Children/adolescents
- Radiology: dense sclerosis surrounding radiolucent nidus
- Microtrabecular osteoid with intervening vascular connective tissue incorporating osteoblasts
- Excellent prognosis
Case 17

- 11 year old boy, orbital mass
Case 17

- Embryonal rhabdomyosarcoma
Embryonal rhabdomyosarcoma

- Most common subtype of rhabdomyosarcoma in children
- Head and neck / periorbital
- Genitourinary (bladder/paratesticular/periprostatic)
- <10% in extremities
- “Botryoid” subtype (polypoid ERMS) occur in relation to epithelial surfaces
  - Bladder, nasopharynx, orbit

Histopathology
- Very primitive
- Small round blue cells, stellate cells, loosely arranged/sheets
- Rhabdomyoblasts (tadpole cells, strap cells) – look hard for these!
- “Cambium layer” in botryoid subtype
  - BEWARE THE NASAL/BLADDER POLYP
- IHC: myogenin patchy/weak
Embryonal rhabdomyosarcoma

Genetics:
- Multiple numerical chromosomal abnormalities
- No specific translocation / fusions
17 year old girl, forearm nodule