Neurofibromatosis

Autosomal dominant conditions resulting in the growth of nerve tumours.

Suggested Approach to Neurofibromatosis Type 1 OSCE Station

Describing lesions:

- Neurofibromas
  - "There are multiple, well circumscribed brown circular nodules and papules present across the back in a generalized distribution."
  - "These range in size from 1 to 7mm."
  - "There are no secondary features (such as crusting, scaling or erosion)."
  - "On palpation, these lesions are soft and non-tender, with some exhibiting the button-hole sign."
  - "These lesions are consistent with neurofibromas."
- Café au lait spots
  - "There are also irregularly shaped, evenly pigmented, flat brown patches"
  - "These are consistent with café au lait spots"
- Other areas
  - "I would like to look for axillary and inguinal freckling, and abdominal scars (e.g. for pheochromocytoma)"
- Conclusion
  - "Given there are 2 or more neurofibromas and 6 or more au lait spots, the likely diagnosis is neurofibromatosis type 1."
  - "In addition to examining the rest of the skin and taking a full history, I would like to measure blood pressure, do a full neurological exam to determine the presence of any spinal cord neurofibromas, examine the vision for signs of optic nerve gliomas, and use a slit lamp to look for lisch nodules in the iris."

If you are asked to ask the patient questions:

- Family history
- Eyes, ears, blood pressure
- Any nerve lesions

Differential diagnosis:

- Neurofibromatosis type 2
- Tuberous sclerosis (multi-system hamartoma formation)

Types

Type 1 (peripheral)

- NF-1 gene defect (chromosome 17) resulting in **peripheral neurofibromas** (i.e. in skin or subcutaneous tissues)
- Skin
  - Neurofibromas (2 or more): well-defined nodules (1-2mm) which are erythematous and varying size and shape
  - Café au lait spots (6 or more): flat patch of asymmetrical darkened skin (brown/black)
- Other features
  - Optic nerve glioma → non-correctable visual loss
  - Lisch nodules (iris hamartomas i.e. brown spots on iris seen with slit lamp)
  - Dorsal root spinal cord tumours
  - Plexiform neurofibromas (diffuse and invasive → bony erosion)

Type 2 (central)

- Merlin protein gene defect (chromosome 22) resulting in **central schwannomas**
- Main presenting condition is bilateral acoustic neuromas (CN8) causing hearing loss in 20's
- Other conditions include: meningiomas, gliomas, vestibular schwannomas, jeuvenile cataracts
- Other symptoms include: headache, balance loss, vertigo, facial weakness, deafness, tinnitus
- Skin symptoms uncommon

Complications

- CNS tumours (described above)
- Cardiovascular
  - Hypertension
  - Pheochromocytoma
  - Renal artery stenosis
- GI neurofibromas → bloating, pain, dyspepsia, haemorrhage, constipation
- Malignant tumours

Investigations

- Diagnosed on clinical features and MRI

Management

- Lifelong annual monitoring
    - Vision
    - Heart and blood pressure
    - Hearing
- Treat complications
- Education