Neurofibromatosis Type 1

Most often this is presented as a “spot diagnosis” case.

On inspection of the skin: look for
- Multiple papules and nodules, sometimes pedunculated
- Truncal café au lait patches (flat and darker than surrounding skin)
- Axillary freckling (pigmented macules in axilla; present in 80% of patients)
- Larger subcutaneous swellings (plexiform neurofibromatosis)

Palpation
Palpate any larger subcutaneous swellings to see if they are in the distribution of specific nerves; check power and sensation in lower motor distribution if directed to do so or if evidence of muscle wasting.

Questions:

What is the difference between a papule and a nodule?
A: Both are raised, unlike macules which are flat.
A nodule is 5mm or more in diameter and a papule is less than 5mm in diameter

How many café au lait patches are significant? What is a patch?
A: More than 5 said to be significant.
A patch is a flat lesion 5mm or more in diameter. Macules are flat lesions less than 5mm in diameter. Sometimes small lesions are called CALM (café au lait macules).

Is there anything else you’d like to do? What associated conditions are there?
A:
- Inspect the back for an associated kyphoscoliosis and note short stature and bowing of legs if present
- Check the blood pressure (phaeochromocytoma may be associated)
- Check for an acoustic neuroma (corneal reflex, facial and auditory nerve: the cranial nerves in the cerebello-pontine angle; loss of corneal sensation is said to be the earliest sign) These are more often associated with type 2 NF but may occur in type 1 NF too.
- Check visual acuity and fundi (optic glioma rarely associated)
- Arrange slit lamp examination (hamartoma of iris- Lisch nodules)
- Malignant transformation of peripheral nerve lesions is very rare
- Learning difficulties sometimes associated

What is the cause?
Autosomal dominant condition affecting neural crest cells: melanocytes affected in skin (café au lait patches) and Schwann cells in nerves: hence it is a “neuroectodermal syndrome”.
Type 1 due to an abnormality on chromosome 17- with abnormality of the tumour suppressor gene that makes neurofibromin.

What would a CT scan of the spine show?
Typical appearance of benign tumours of the nerve roots at their exit from the vertebral column: “dumb-bell tumours” with part of tumour inside and part outside neural foramina. May lead to lower motor neurone syndromes in a nerve root distribution.
Summary of examination scheme

- Inspect skin from in front, looking for café au lait patches as well as the more obvious papules and nodules; ask patient to raise their arms (so you can see the axillae, looking for freckling)
- Inspect skin from behind (noting any kyphoscoliosis)
- Palpate skin (commenting on any larger subcutaneous swellings)
- Extras: ask for or measure BP, inspect abdomen for scars (phaeo), check fundi and corneal reflex

Suggested spiel:

“I would also like to check the blood pressure, examine the eyes and check the nerves of the cerebello-pontine angle”

“These findings are consistent with type 1 neurofibromatosis, an autosomal dominant condition due to a mutation on chromosome 17, leading to proliferation of neuroectodermal cells mainly in the skin and peripheral nerves”.

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<th>Autosomal dominant Variable expression</th>
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Important Note
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