34 year-old female with genetic disorder

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Question 1:
What is the genetic disorder this patient likely has?
Answer 1: Neurofibromatosis
Question 2

Dx?
Answer 2: NF1 with malignant transformation of neurofibroma to malignant peripheral nerve sheath tumor
Bilateral neurofibromas (orange arrows)
Neurofibromas of cervical spine, seen in NF1 (orange arrows)

Incidentally noted is ferromagnetic susceptibility artifact from skeletal hardware (yellow arrows).
Neurofibromas of cervical spine, in NF1 (orange arrows)
Interval enlargement of neurofibroma (orange arrows)
Interval enlargement of neurofibroma (orange arrows), which now deforms cord (yellow arrow)
Heterogenous enhancement, suggesting central necrosis (yellow arrow)
Heterogenous enhancement, suggesting central necrosis (yellow arrow)
“Tongue-like” extension into spinal cord (yellow arrow)
Heterogenous enhancement, suggesting central necrosis (yellow arrow)
Neurofibromatosis 1

• Autosomal dominant neurocutaneous disorder (phakomatosis) in which NF1 gene (tumor suppressor gene) is turned off via nonsense mutation.
• Gene locus = chromosome 17q11.2
• Imaging manifestations include multiple neurofibromas, plexiform neurofibromas, visual pathway gliomas, optic hamartomas, sphenoid wing dysplasia, and thinning of long bones.
Malignant transformation of neurofibroma to malignant peripheral nerve sheath tumor in NF1

Malignant peripheral nerve sheath tumor (MPNST)
- Rare soft tissue sarcoma of peripheral nerve sheath.
- Peripheral nerve sheath tumors include schwannomas, neurofibromas, & plexiform neurofibromas.
- 5-13% of patients with NF1 develop MPNST
- May or may not cause clinical symptoms when transformation occurs including pain, muscle weakness in nerve territory.
Malignant peripheral nerve sheath tumor (Imaging)

- Imaging cannot reliably distinguish benign vs. malignant, but having at least 2 of these features suggests malignancy (specificity 90%; sensitivity 61%) & these lesions need biopsy:
  - Increased size
  - Peripheral enhancement pattern
  - Perilesional edema-like zone
  - Intratumoral cystic lesion
- Often can see tapered transition to normal contiguous nerve at margin of mass.

Malignant transformation of neurofibroma to malignant peripheral nerve sheath tumor in NF1
References


2. Statdx.com