# 34 year-old female with genetic disorder

Ryan Joyce, MD
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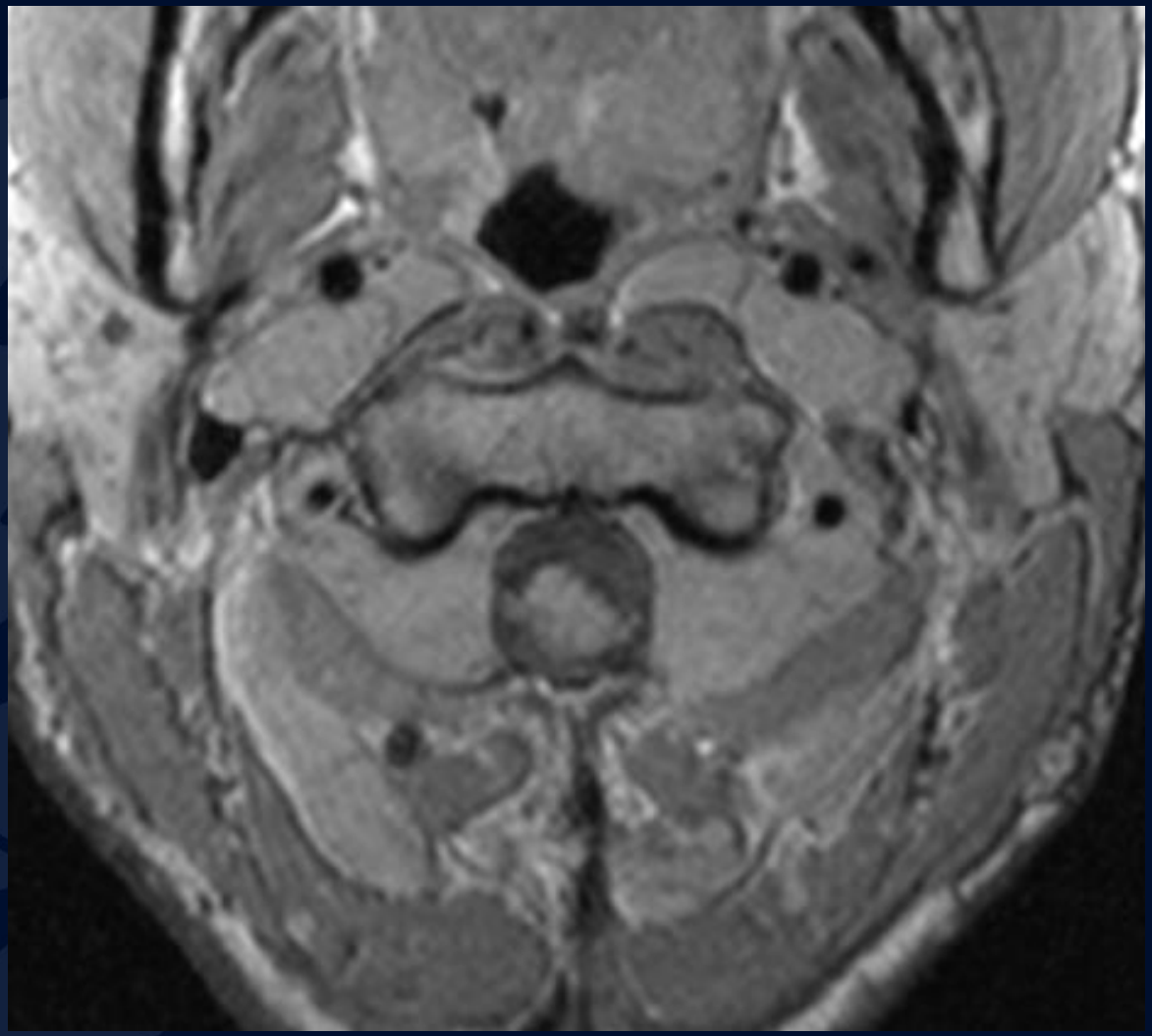

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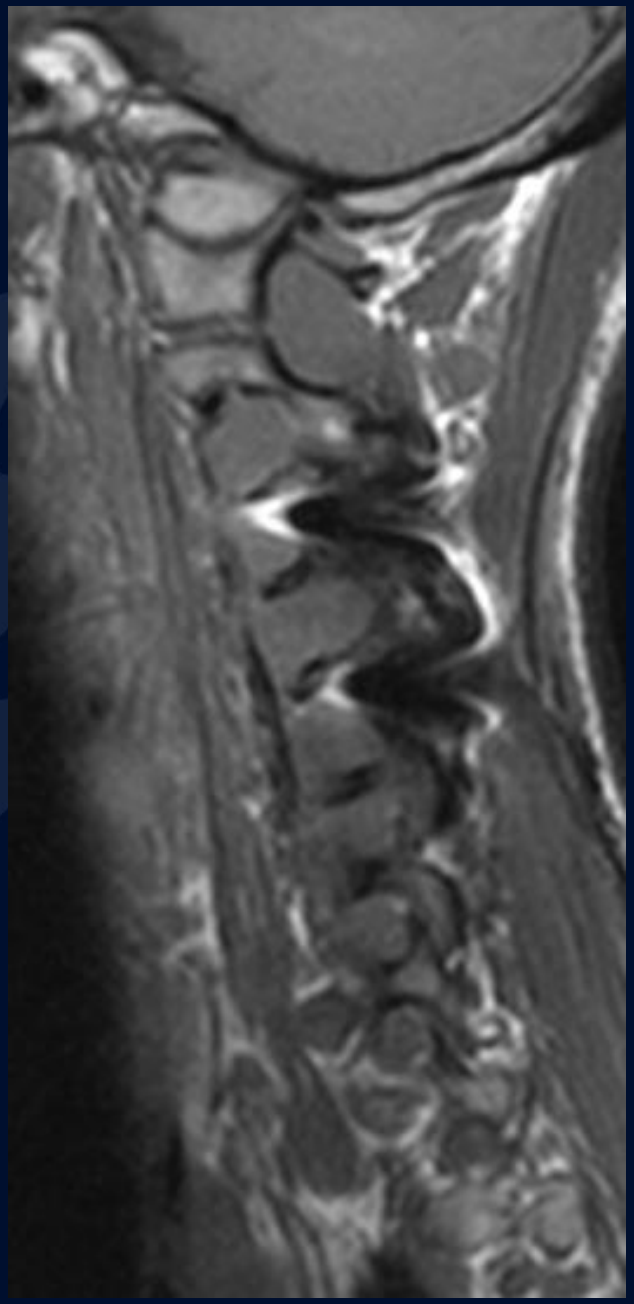

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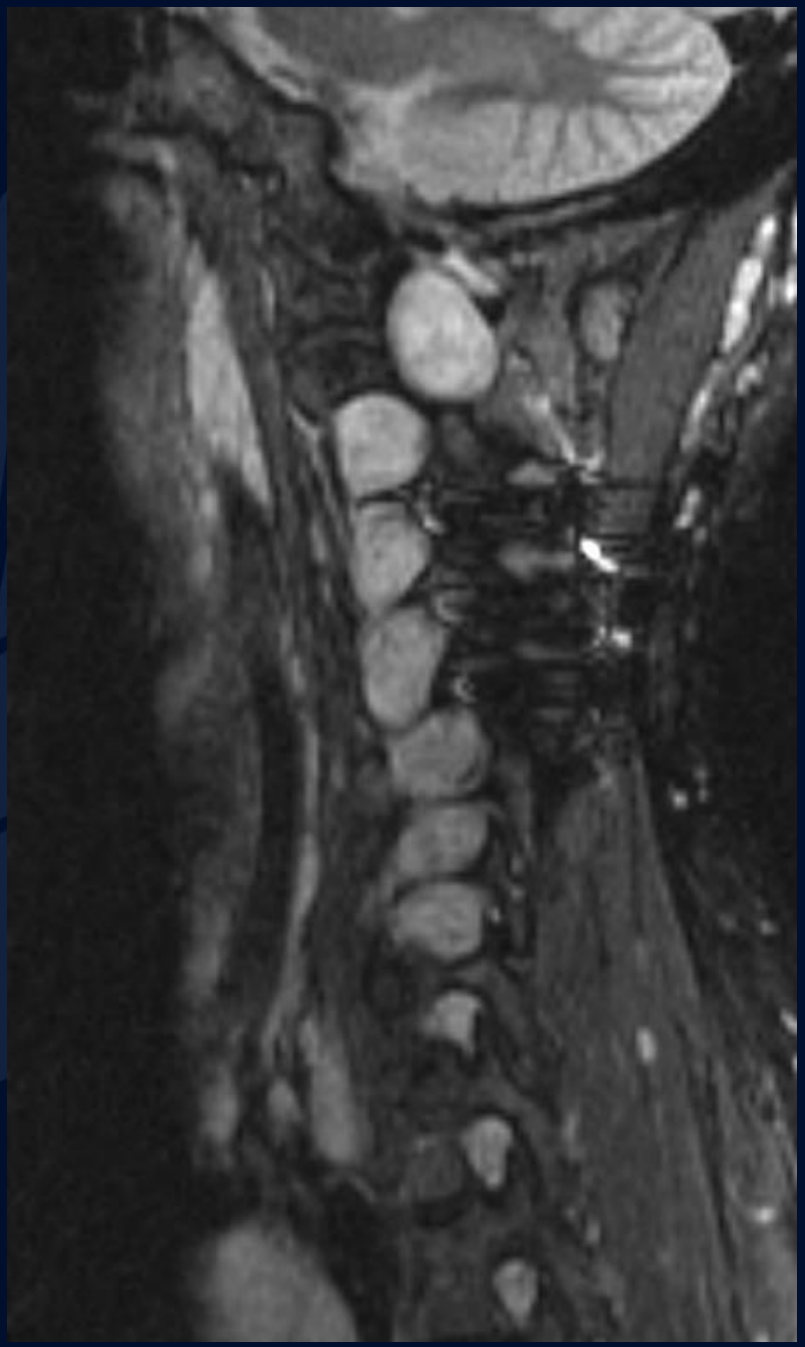

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# Question 1:
# What is the genetic disorder this patient likely has?

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# Answer 1:
# Neurofibromatosis

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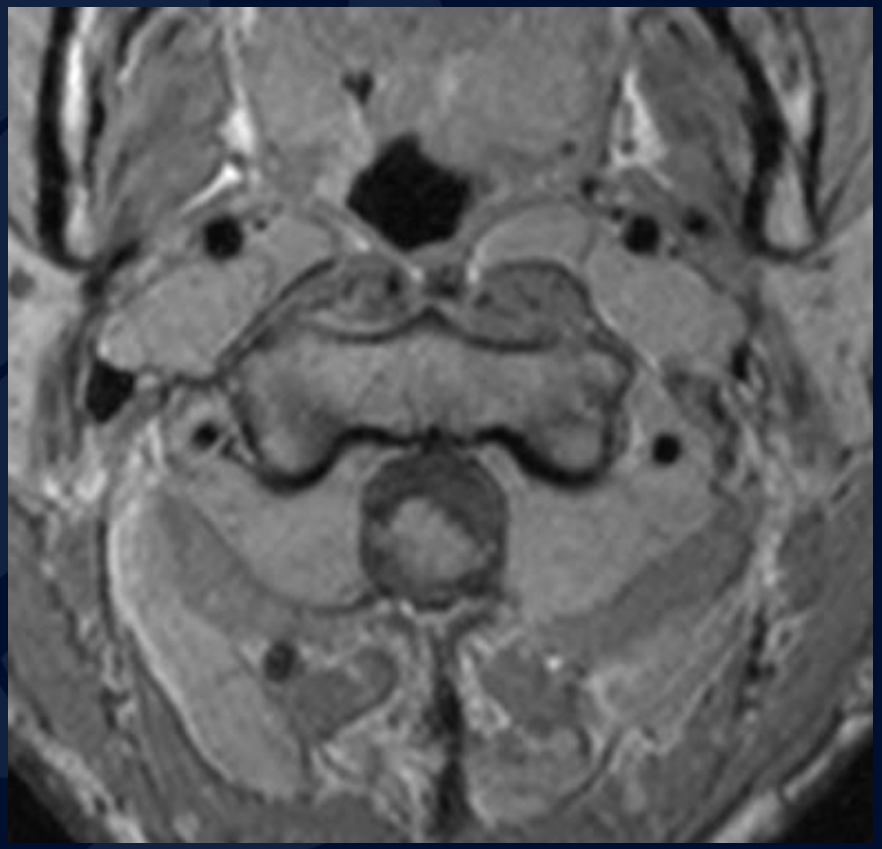

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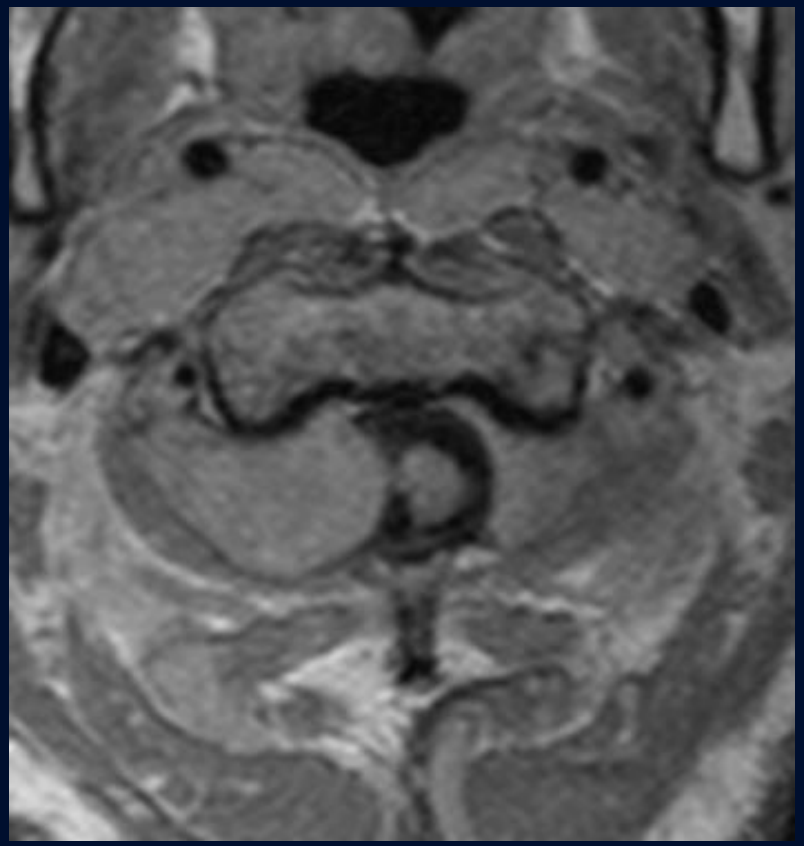

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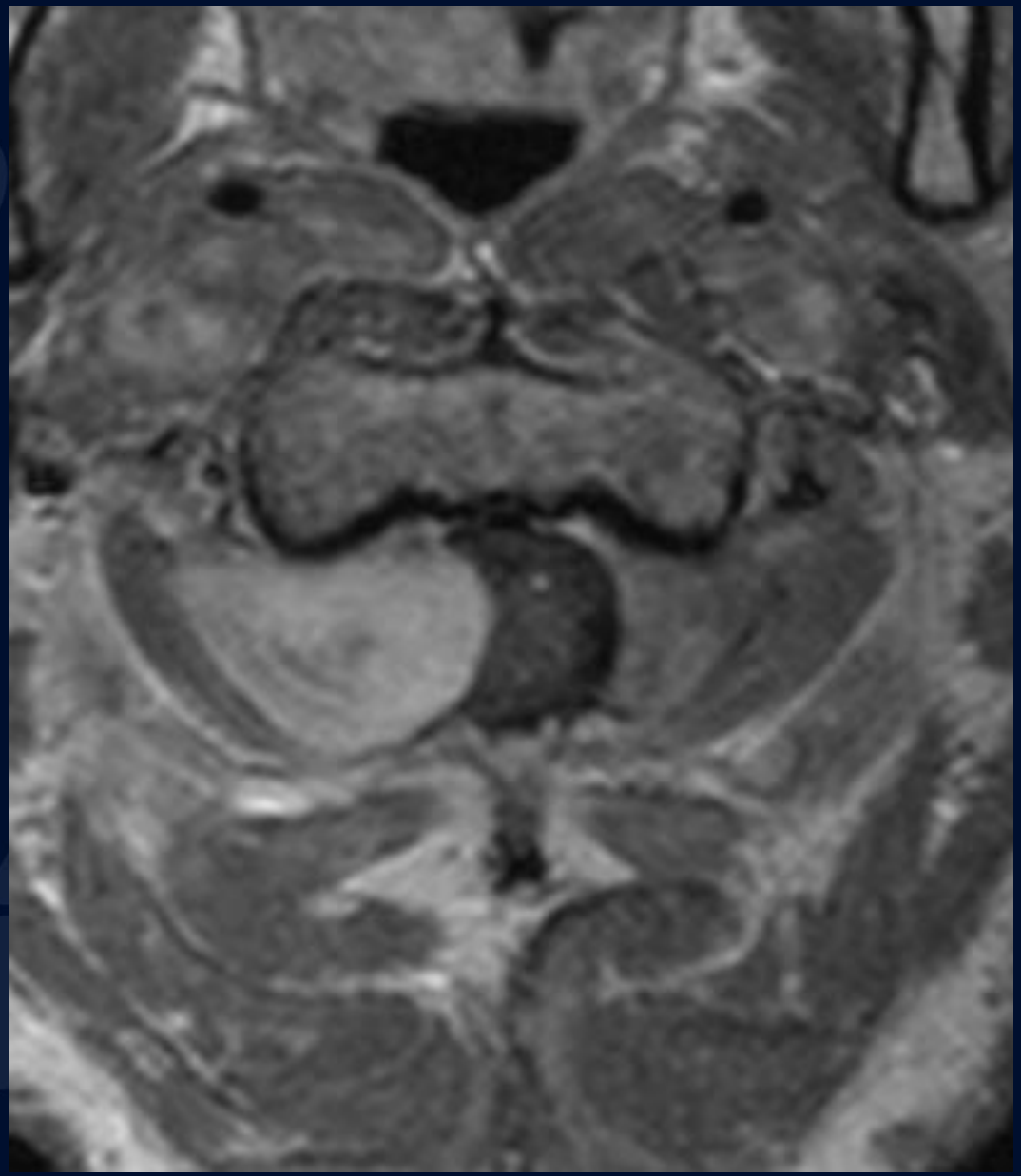

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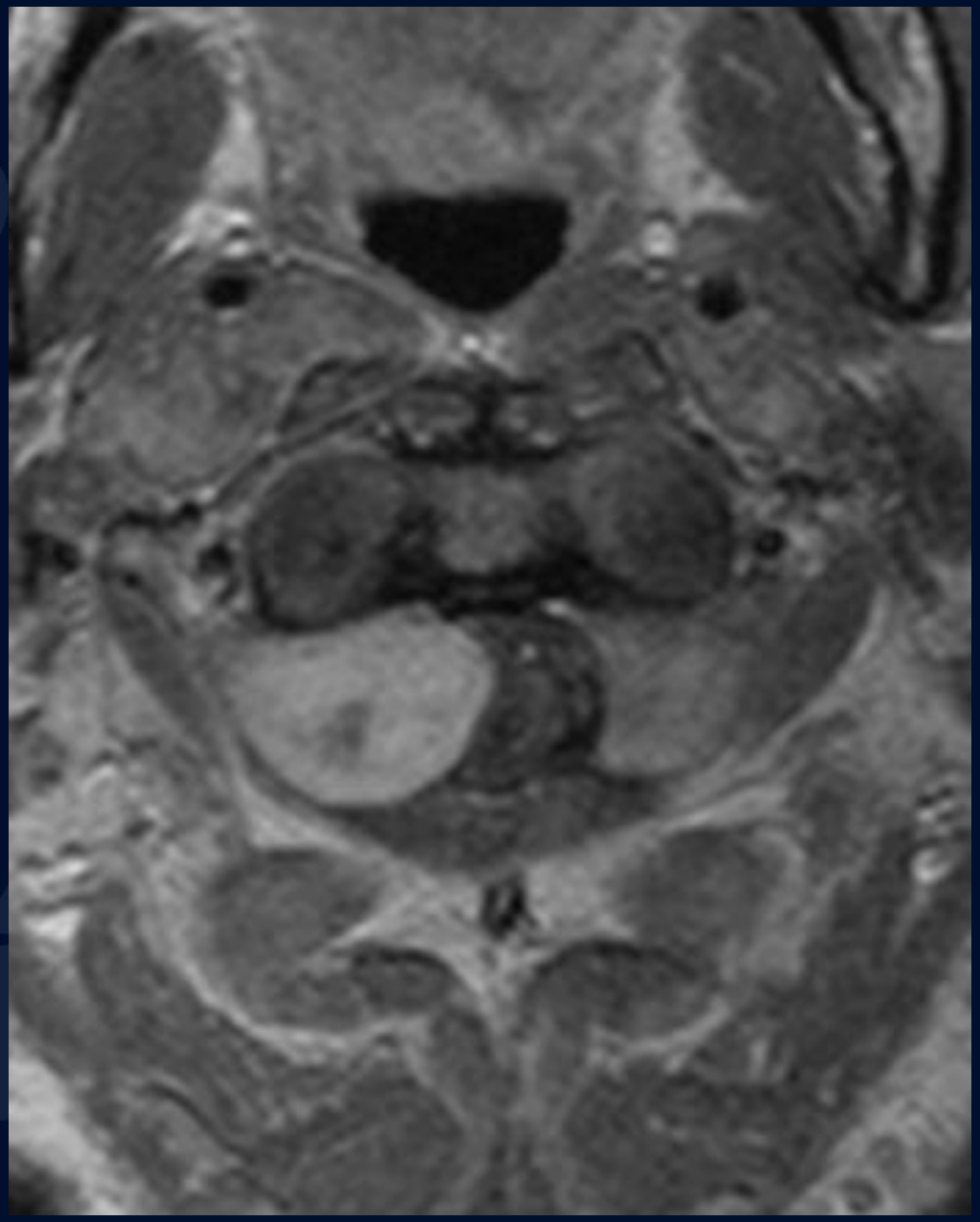

2017

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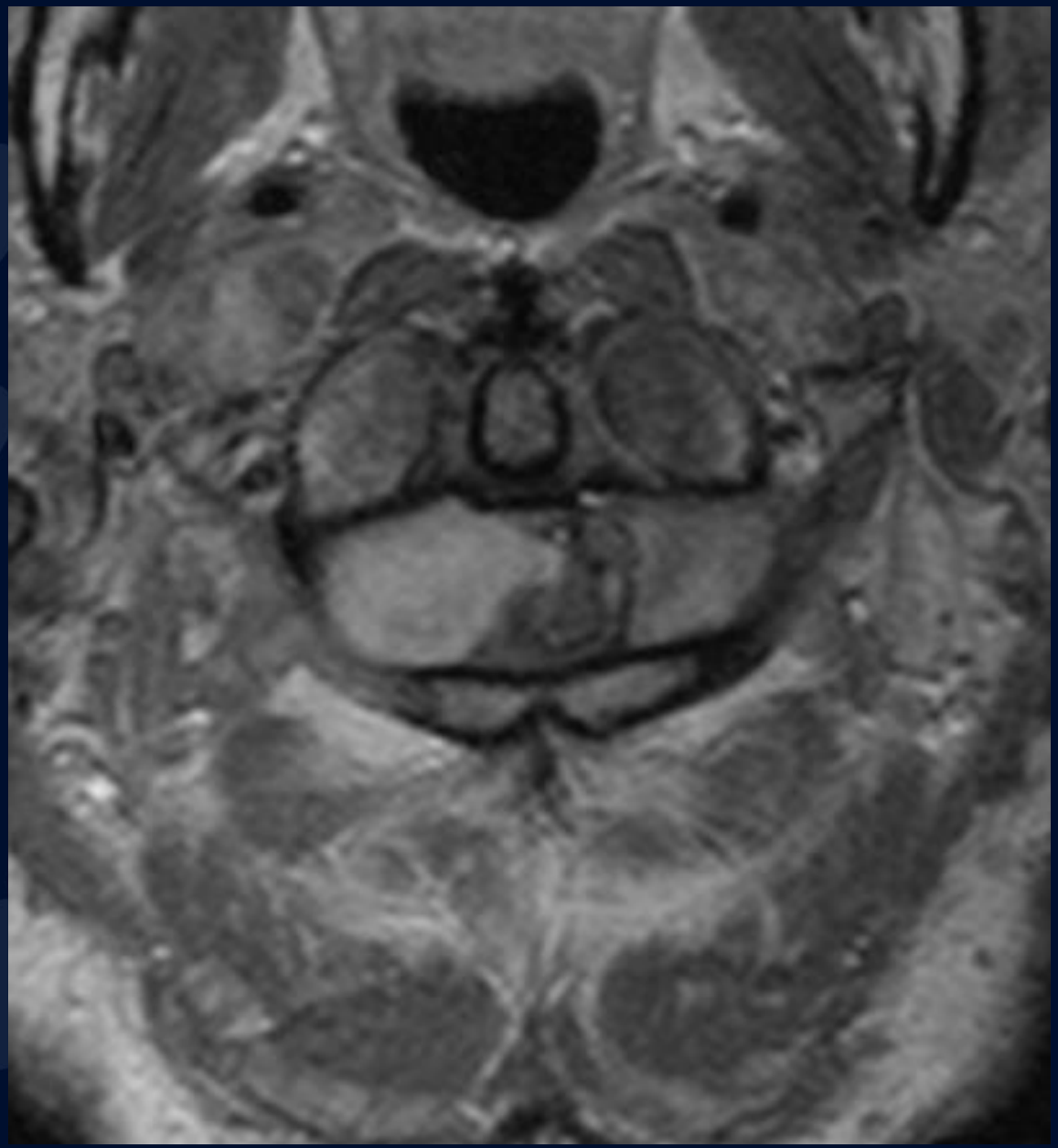

2017

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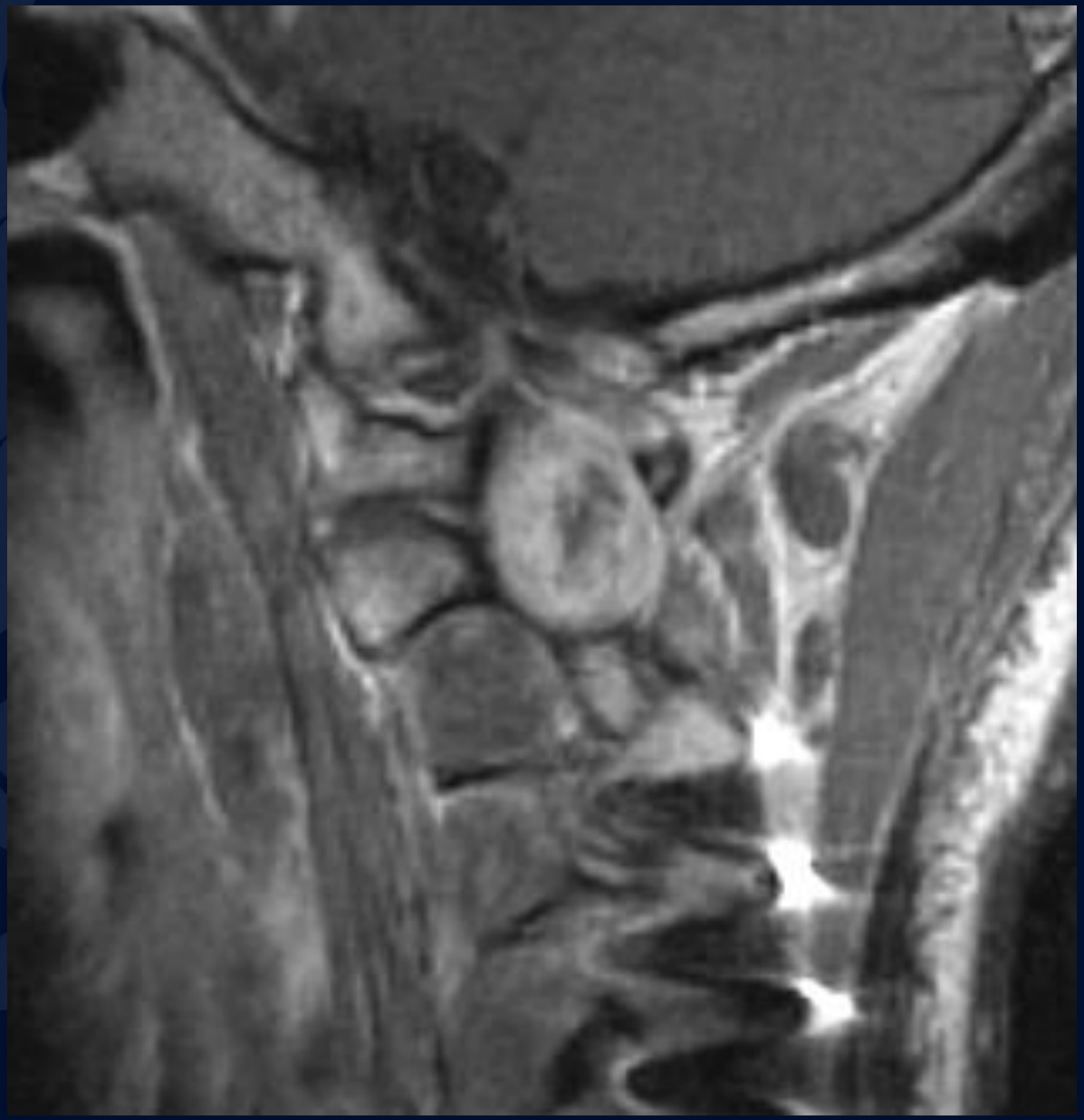

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# Question 2

# Dx?

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# Answer 2:
# NF1 with malignant transformation of neurofibroma to malignant peripheral nerve sheath tumor

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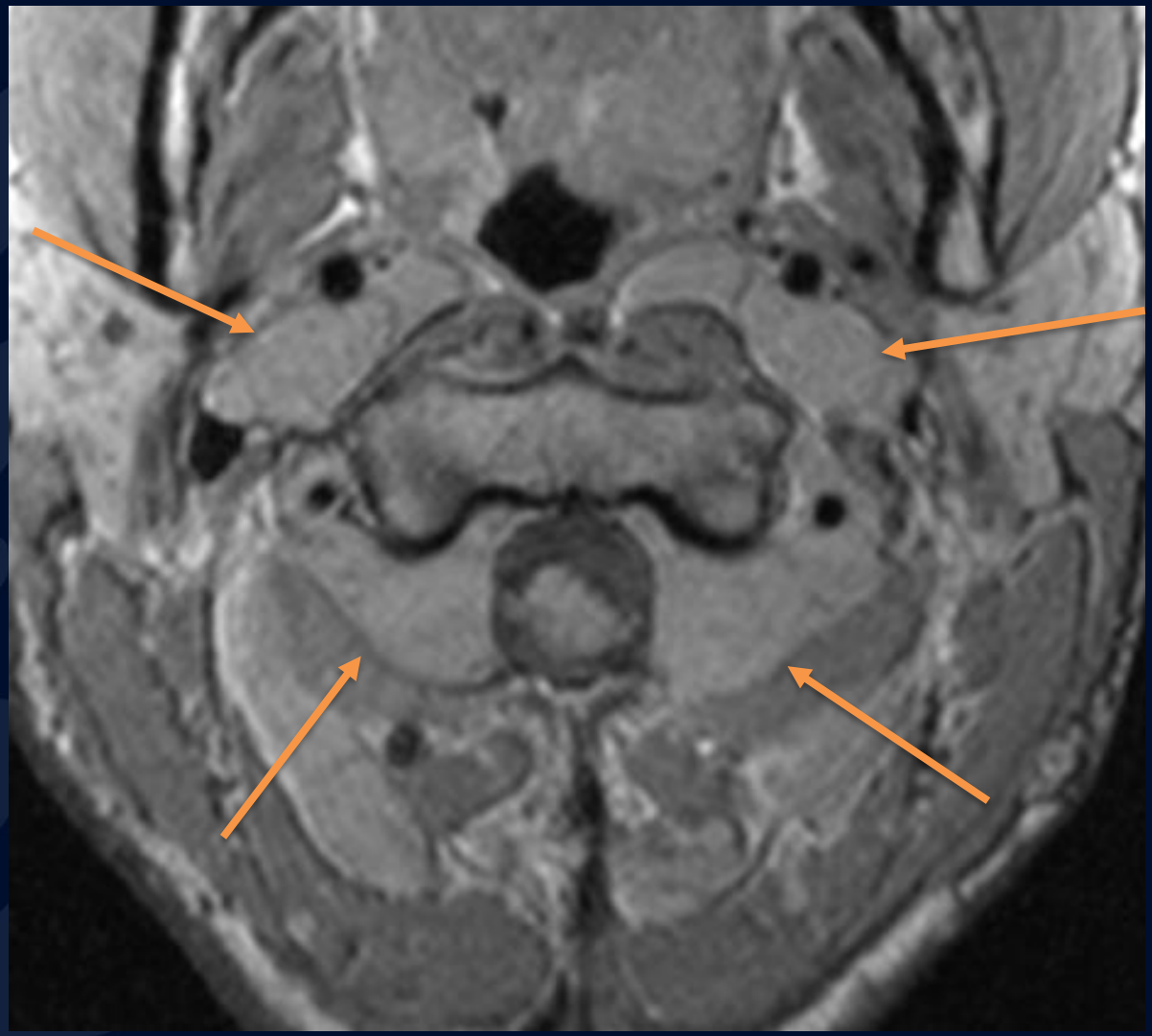

T1
2008

Bilateral neurofibromas (orange arrows)

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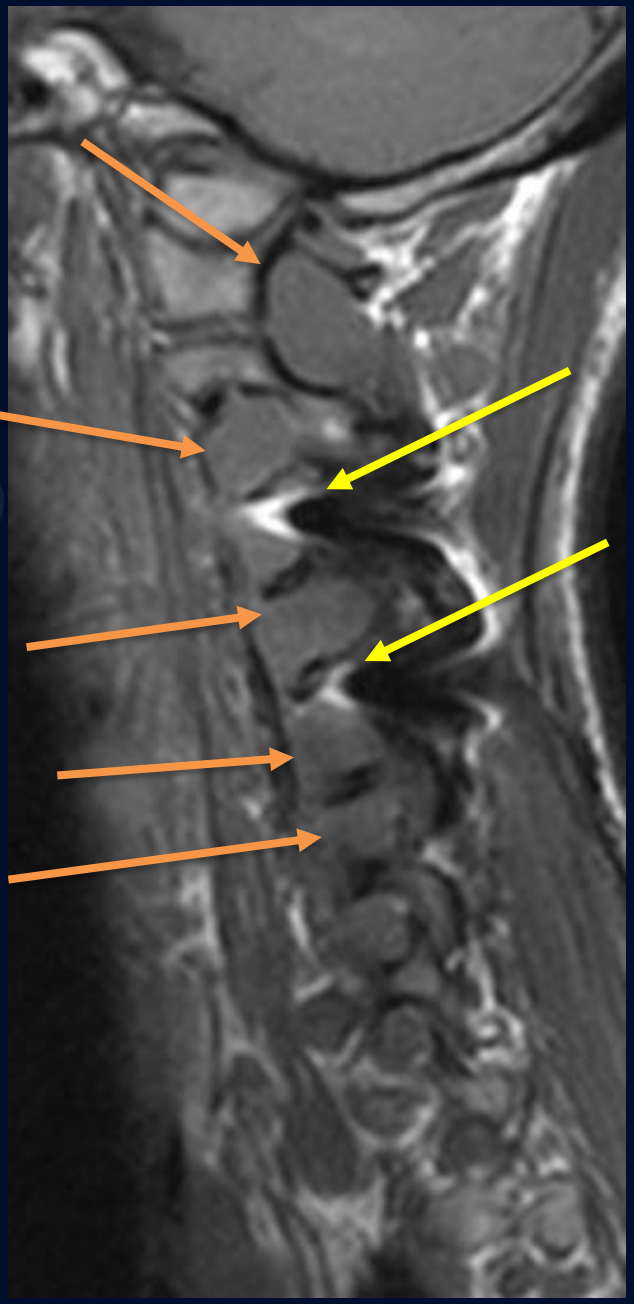
Neurofibromas of cervical spine, seen in NF1 (orange arrows)

Incidentally noted is ferromagnetic susceptibility artifact from skeletal hardware (yellow arrows).



T1
2008

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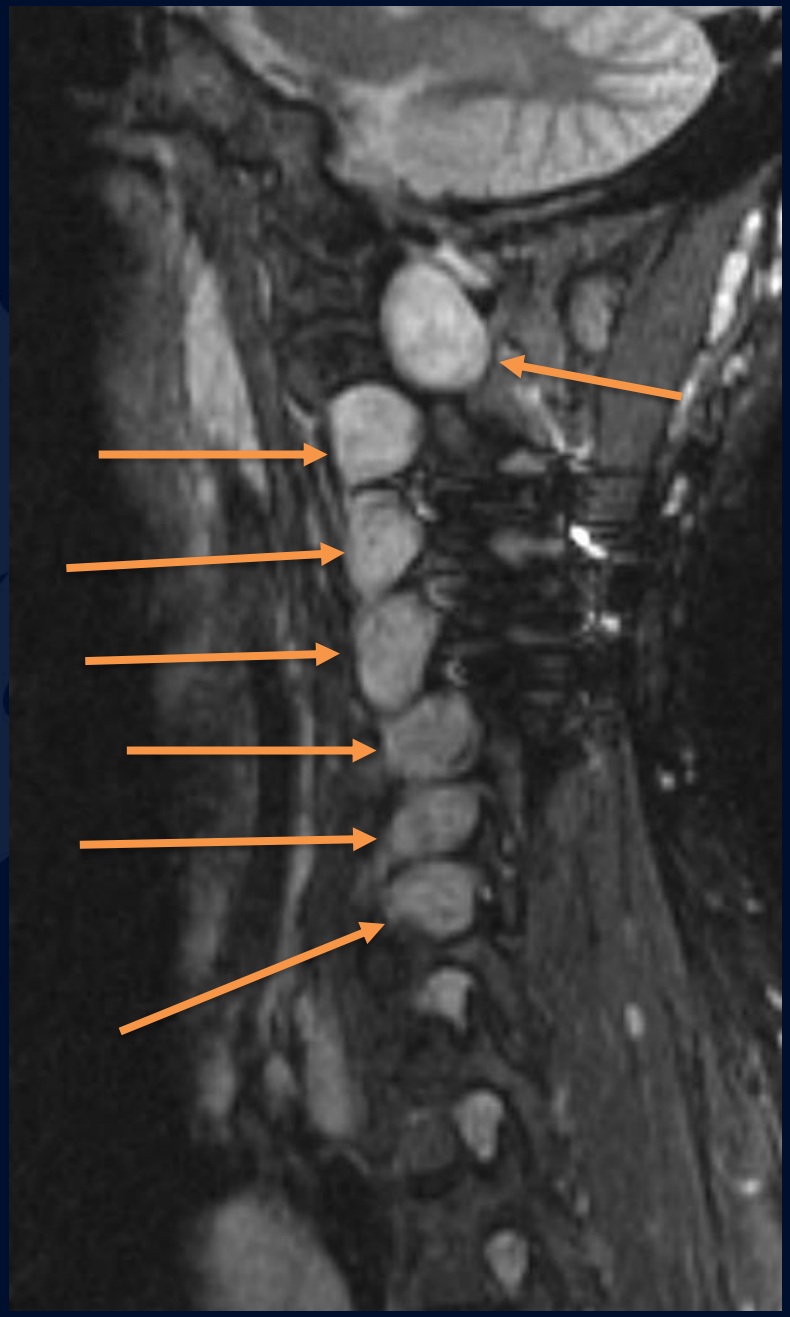
Neurofibromas of cervical spine, in NF1 (orange arrows)



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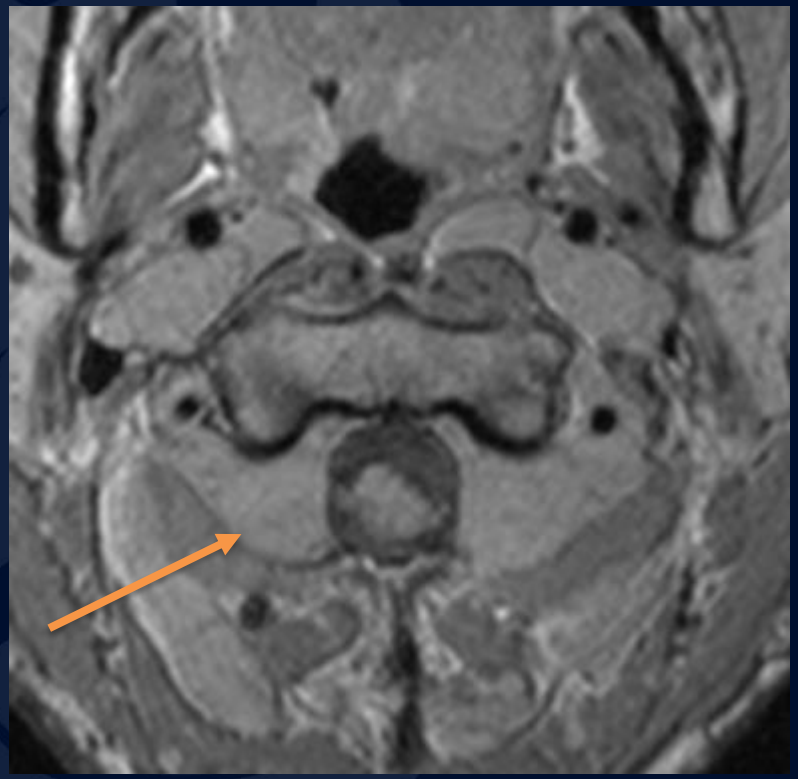

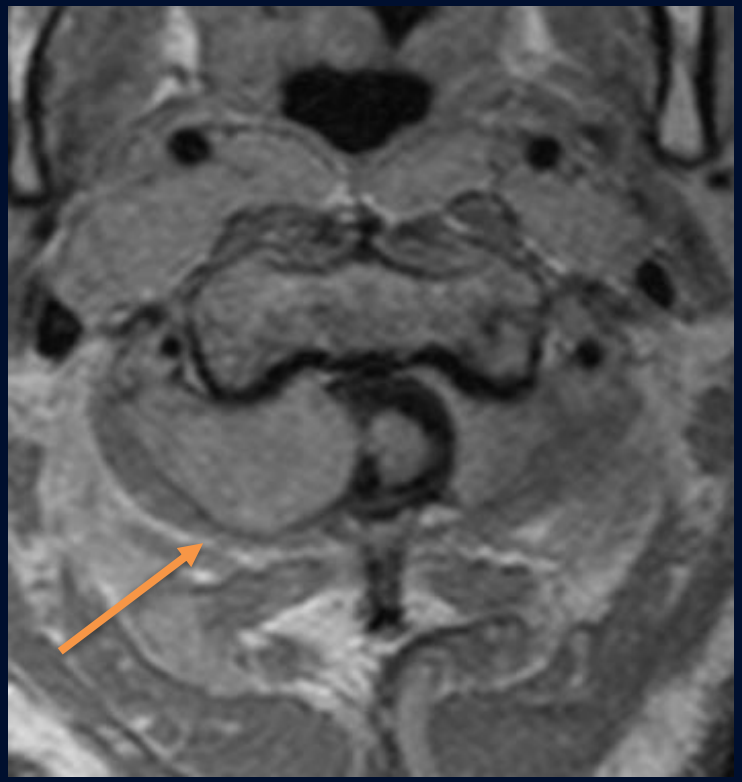

Interval enlargement of neurofibroma (orange arrows)

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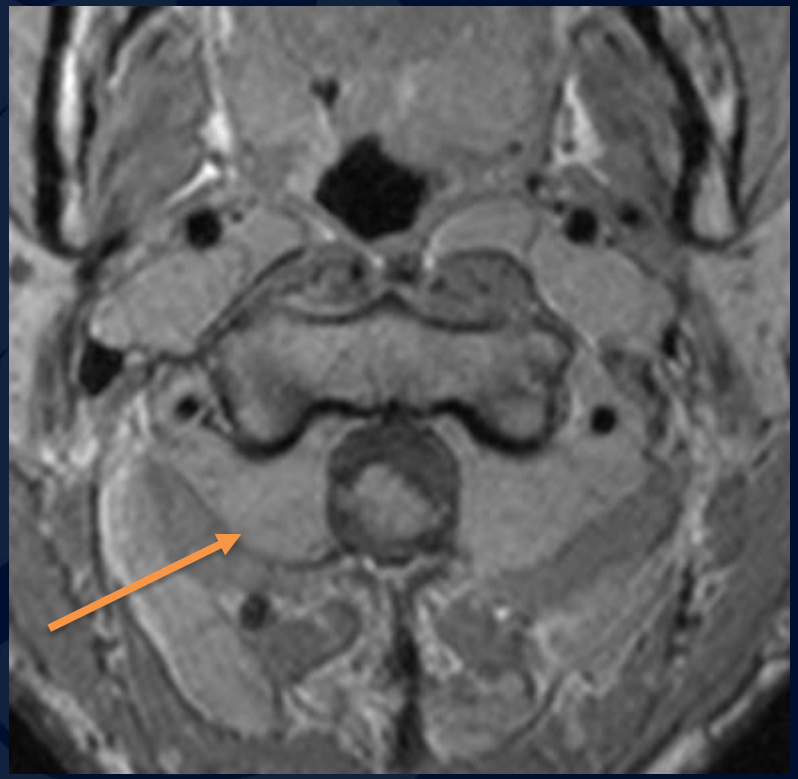

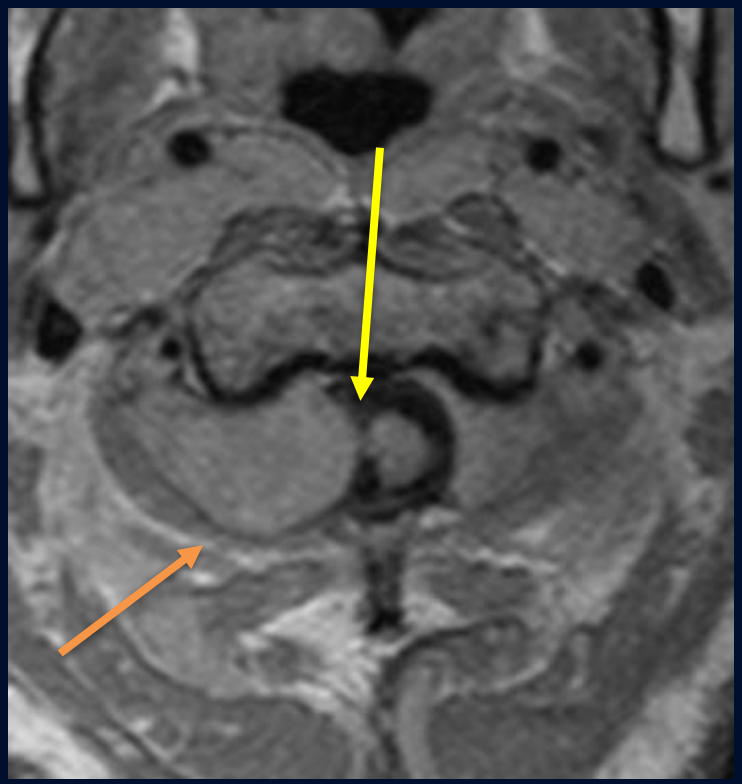

Interval enlargement of neurofibroma (orange arrows), which now deforms cord (yellow arrow)

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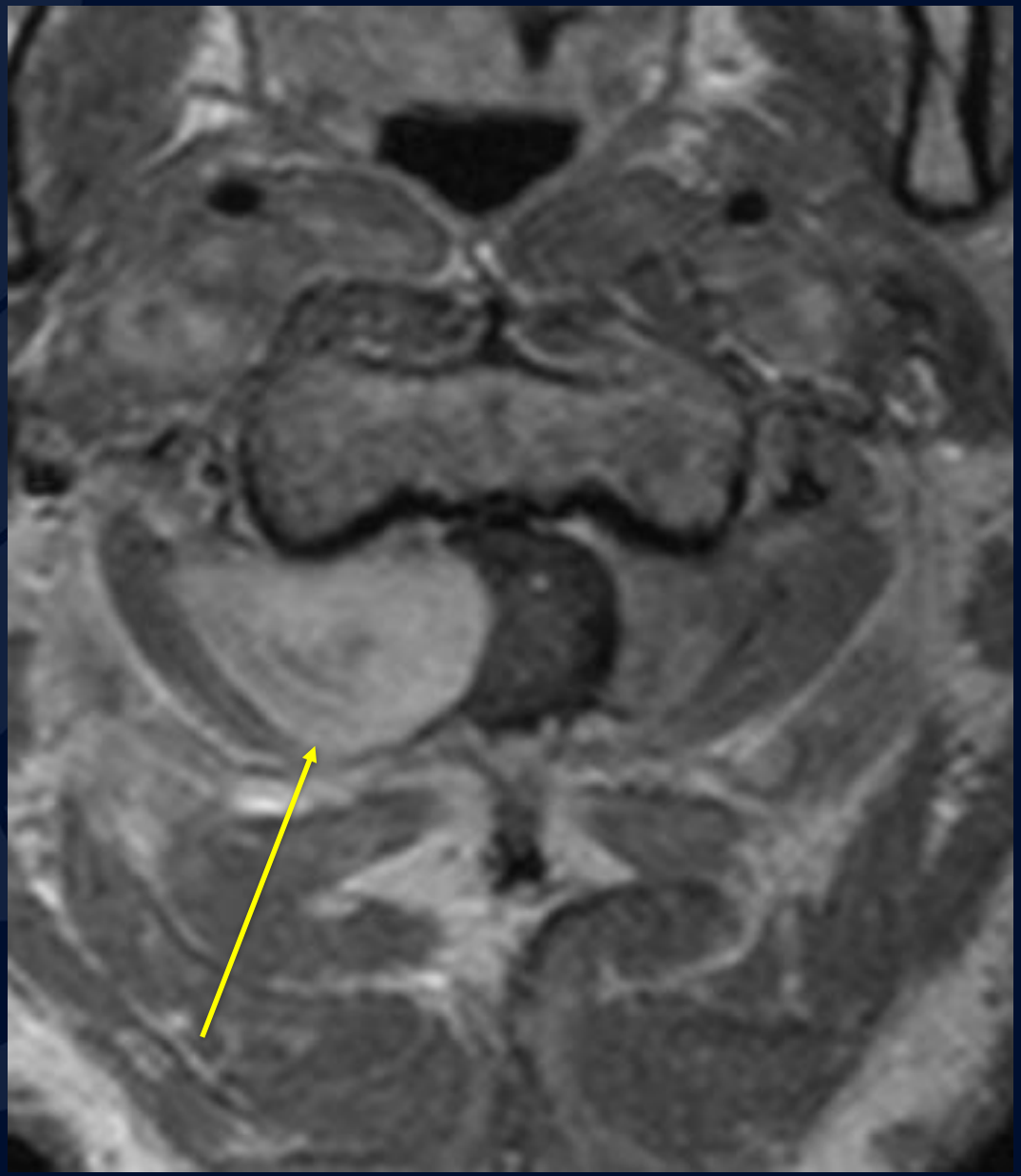

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Heterogenous enhancement, suggesting central necrosis (yellow arrow)

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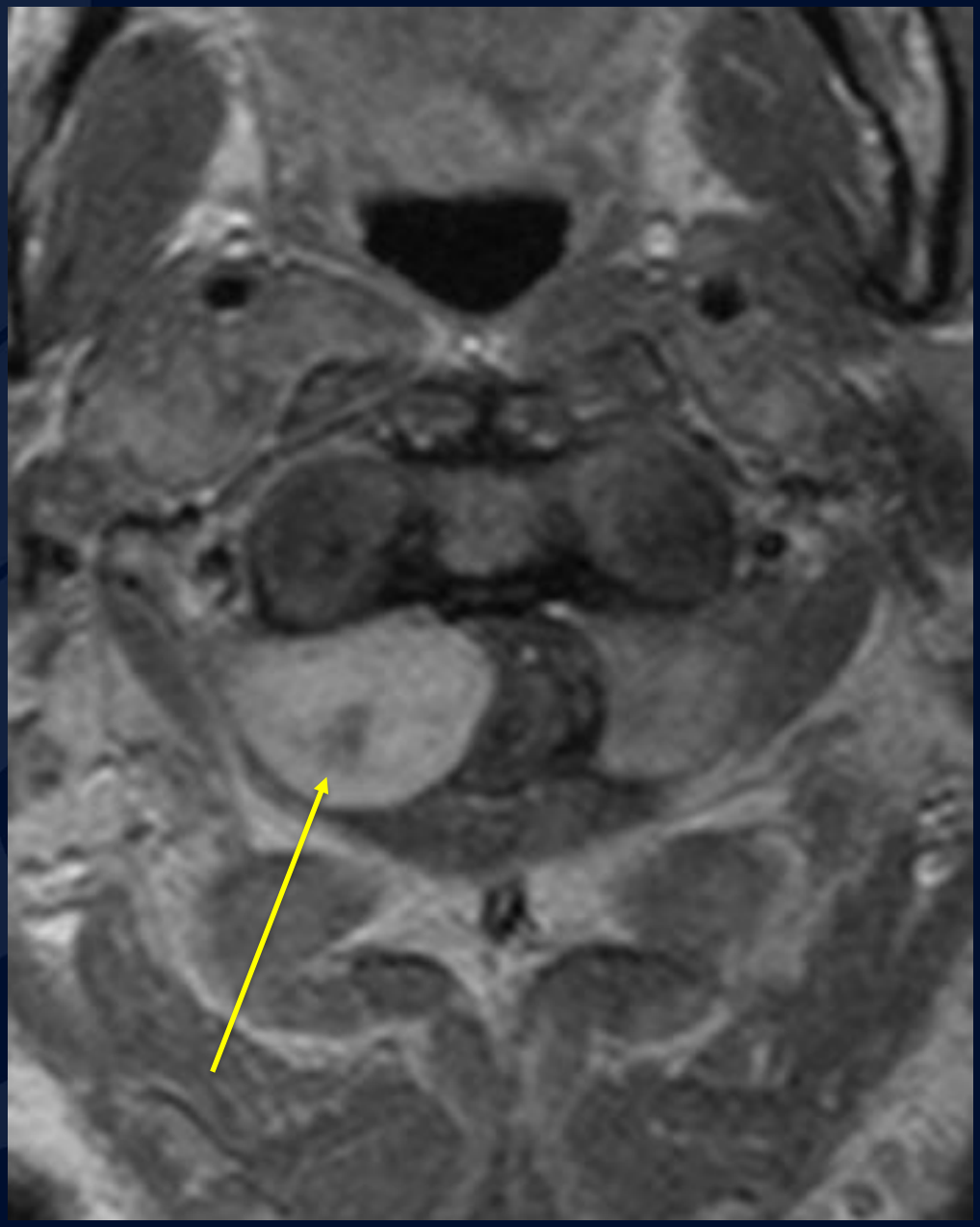

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Heterogenous enhancement, suggesting central necrosis (yellow arrow)

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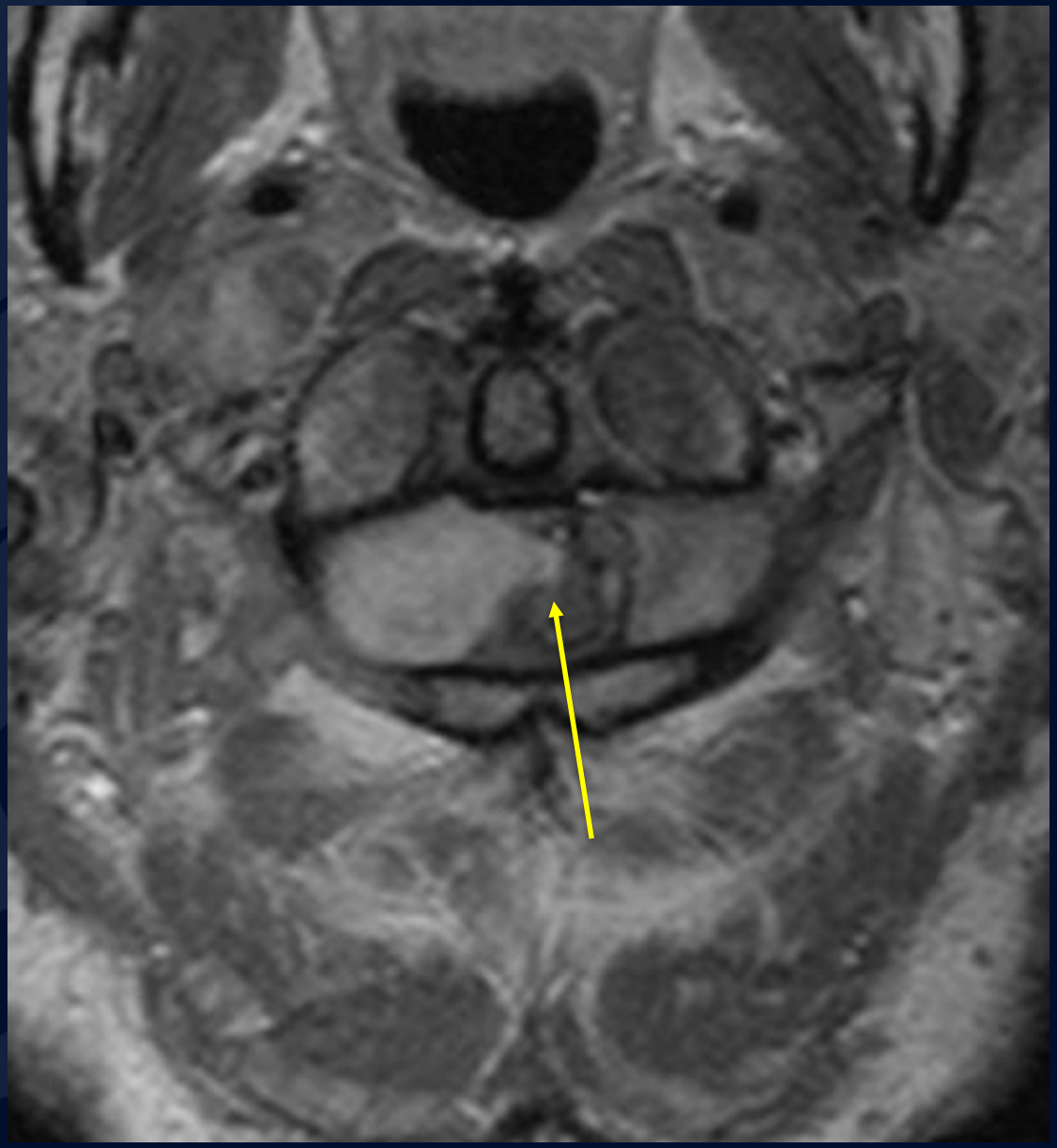

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“Tongue-like” extension into spinal cord (yellow arrow)

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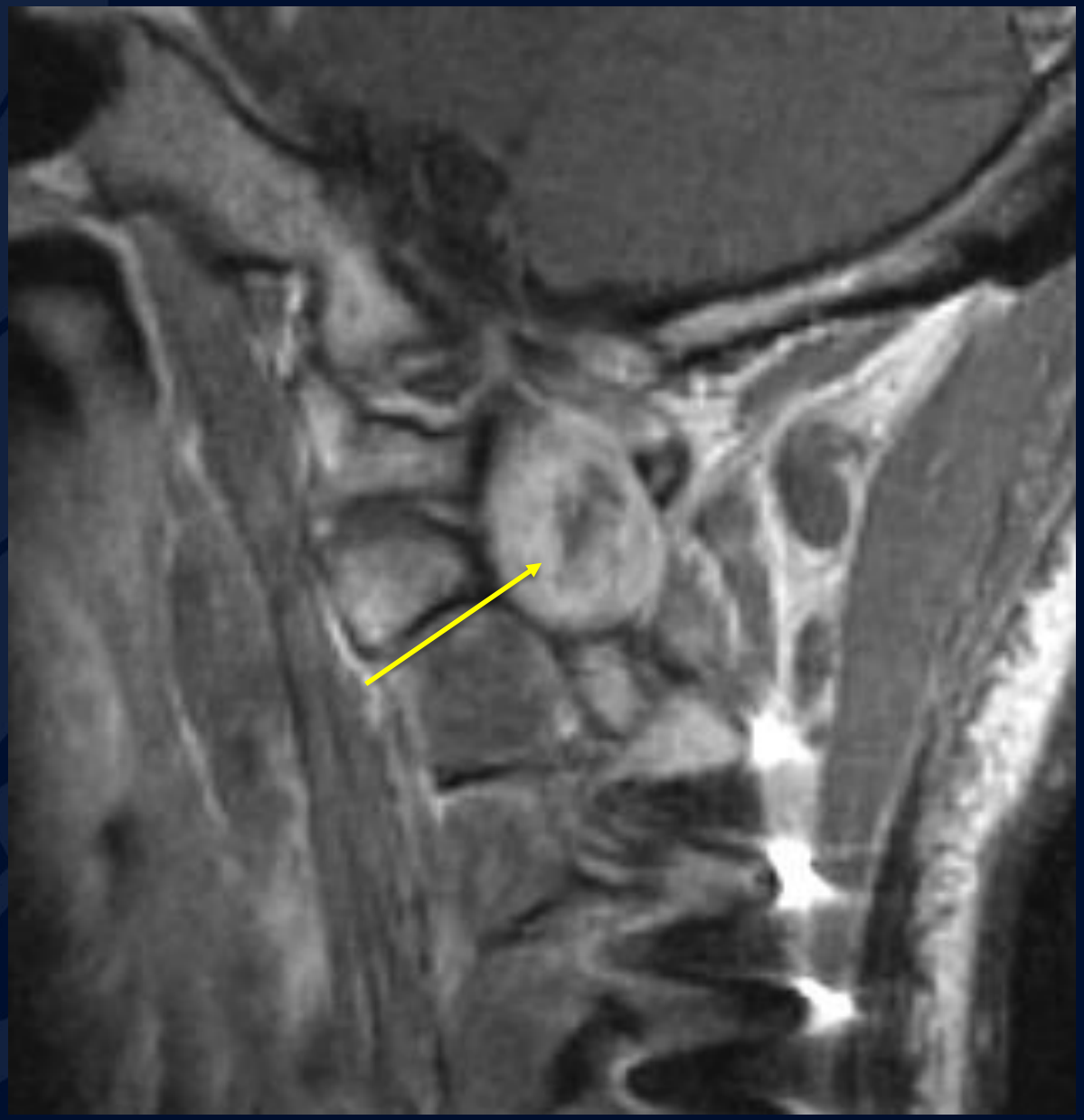

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Heterogenous enhancement, suggesting central necrosis (yellow arrow)

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# 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.

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# 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.

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

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.

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# References


1. MRI Features in the Differentiation of Malignant Peripheral Nerve Sheath Tumors and Neurofibromas. J Wasa, Y Nishida, S Tsukushi, et al. American Journal of Roentgenology. 2010 194:6, 1568-1574.
2. Statdx.com


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