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WHAT'S YOUR DIAGNOSIS?

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FIGURE 1.



FIGURE 2.

HISTORY

A three-year-old male had a four-month history of reduced movement of the right upper limb. The clinical examination revealed skin hyperpigmentation distributed all over the body, and concentrated on the back and the arms. Hyperpigmentation of the sclera was also noted. The neurological examination showed signs of lower motor neuron lesion in the right upper limb, but was otherwise unremarkable. The father and the sister showed similar skin lesions. There was first-degree consanguinity between the parents.

MRI of the cervical spine was performed, including T1 sagittal and axial images, post-gadolinium administration.

1. What do these images show?
2. What is the radiographic diagnosis?
3. What other area in the central nervous system might be examined to further support the diagnosis?

ANSWER TO WHAT'S YOUR DIAGNOSIS? (PREVIOUS PAGE)

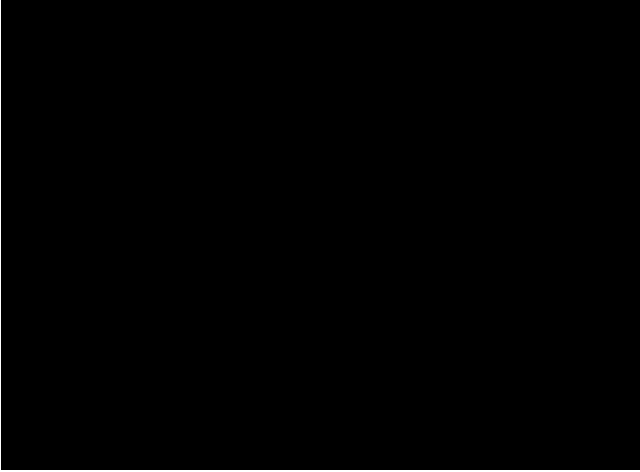


FIGURE 1. Sagittal T1 with contrast, showing an enhanced tumor mass extending from C4 to C7.

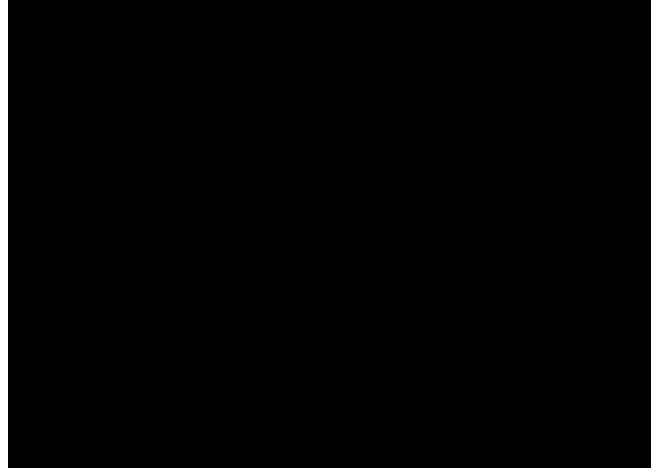


FIGURE 2. Axial T1 with contrast, demonstrating clearly the extradural extension of the tumor through the vertebral foramen, which appears widened (arrow), and the intradural component displacing the cord to the other side. Note also that the other side is also affected by the same process.

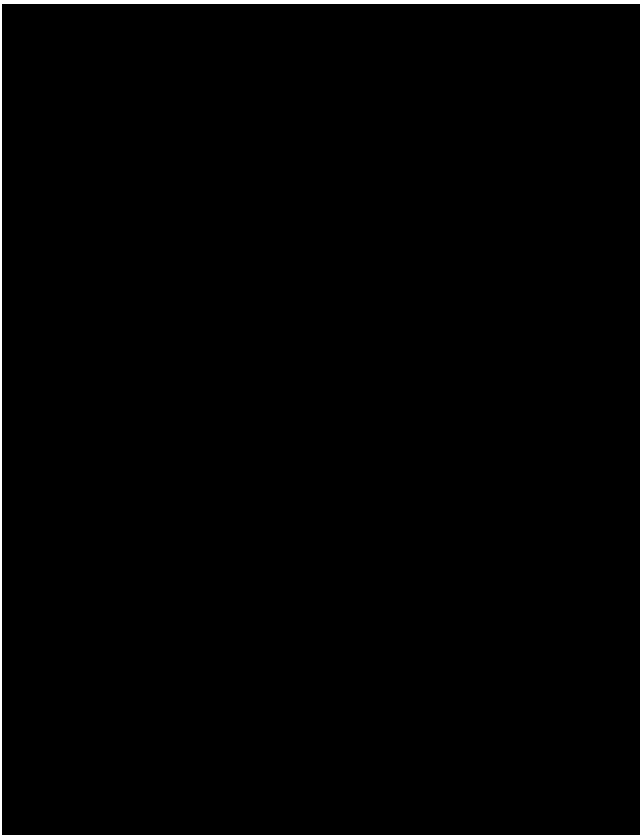


FIGURE 3A. Axial T2 of the brain showing the hyperintense foci in the pons, thalami, and the left globus pallidus.

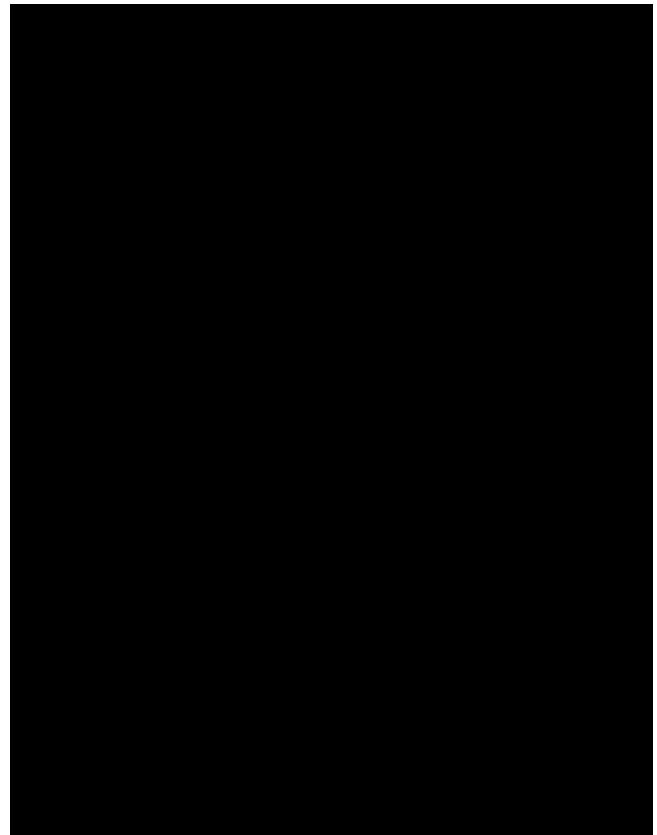


FIGURE 3B. Axial T2 of the brain showing the hyperintense foci in the pons, thalami, and the left globus pallidus.

Radiographic Findings: The T1 sagittal images of the cervical spine (Figure 1) show markedly enhanced lobulated masses within the spinal canal, extending from the level of C4 through C7. The axial images (Figure 2) reveal the masses to have both an intradural component displacing the cord, and an extradural component within the intervertebral foramen, with the typical appearance of a dumbbell-shaped tumor.

Radiographic Diagnosis: The appearance is typical of plexiform dumbbell-shaped schwannoma or neurofibroma. However, with the above given history of skin hyperpigmentation, which was in fact typical of café-au-lait spots, the diagnosis of neurofibromatosis was confirmed. In view of this, MRI of the brain was carried out (Figures 3A and B) and T2 axial image through the pons and basal ganglion revealed hyperintense scattered foci in the pons, left putamen, and the thalami, with no mass effect or contrast enhancement, in keeping with multiple hamartomas.

Discussion: Phacomatosis is a heterogeneous group of disorders which have central nervous system manifestations and cutaneous manifestations. Although eight variants have been proposed by the NIH consensus conference, only two major categories are defined: NF1 (Von Recklinghausen disease) and NF2 (bilateral acoustic neuroma). Both are autosomal dominant. The diagnostic criteria of NF1 are shown in Table 1.

CNS manifestations occur in 15% to 20% of all patients with NF1. They involve basal ganglia, mainly globus pallidus, optic radiation, brainstem, and cerebral peduncles. Pathologically, these lesions represent foci of dysplastic glial proliferation. They will appear as high-signal intensity in T2WI, typically with no mass effect or contrast enhancement.

Spinal neurofibroma are rare tumors of the cord or nerve roots. They comprise 3% of all spinal tumors. They occur sporadically or in association with neurofibromatosis, and the cervical region is the most common site. The plain film may show erosion of the adjacent bones, typically involving the pedicles with increased interpediculate distance and enlargement of the neural foramina, with scalloping of the vertebral bodies. CT scan will show these abnormalities as well as the extension of the tumor into the neural foramina or the paravertebral soft

tissues. Dumbbell-shaped tumors refer to those tumors extending into the extradural space through one or more neural foramina. They are typical of neurofibromatosis, but are not pathognomonic. They may also be seen in sarcoma, neuroblastoma, metastasis and hemangioblastoma. MRI provides an accurate topographical definition of the tumor and will differentiate between lateral meningocele and neurofibroma in the clinical context of neurofibromatosis. Spinal neurofibroma appear as isointense to the spinal cord in T1W1 and as high signal in T2W1. They enhance homogeneously after intravenous gadolinium. The use of contrast enhancement provides tumor contrast and more clearly delineates compartmentalization of the lesion into intramedullary, intradural, extramedullary, or extradural. MRI is also useful in assessing spinal cord compression and associated syringomyelic cavity.

Although it may not be possible to differentiate between schwannoma and neurofibroma with preoperative imaging, the histologic diagnosis does have clinical importance. A study suggested that a patient with an intraspinal nerve sheath neurofibroma is very likely to have NF1, while patients with schwannoma are likely to have sporadic conditions or NF2.

TABLE 1. *Diagnostic features of NF1.*

1.	Six or more café-au-lait spots
2.	Two or more neurofibroma or plexiform neurofibroma
3.	Axillary or inguinal freckling
4.	Optic glioma
5.	Two or more iris hamartoma (Lisch nodules)
6.	Characteristic bone lesions (sphenoid wing dysplasia, thinning of the bones with or without pseudarthrosis)
7.	First degree relative with NF1

References

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