A 42-year-old man presents with a two-year history of increasing right facial numbness. He has a history of intermittent unsteadiness, mild hearing loss and vertigo but has otherwise been well. Cranial magnetic resonance imagine (MRI) (T1 weighted following gadolinium contrast) is shown below.

The most likely diagnosis is:
A. multiple sclerosis
B. neurofibromatosis type 2
C. cerebellar haemangioblastoma
D. meningioma
E. pontine glioma

A. multiple sclerosis
Neurofibromatosis, a common neurocutaneous disorder, includes numerous different forms. Neurofibromatosis type 1, also known as von Recklinghausen's NF or NF1 is the most common type, and accounts for at least 85 percent of patients.

Diagnostic criteria — The hallmarks of NF1 are the multiple café-au-lait spots (CALS) and associated cutaneous neurofibromas. The diagnostic criteria developed by the NIH Consensus Conference in 1987 and updated in 1997 are based upon specific clinical features of NF1 (show table 1) [16-18]. According to these criteria, at least two of the following clinical features must be present to make the diagnosis of NF1: Six or more café-au-lait macules of greatest diameter >5 mm in prepubertal and >15 mm in postpubertal individuals Two or more neurofibromas of any type or one plexiform neurofibroma Freckling in the axillary or inguinal regions Optic glioma Two or more Lisch nodules (iris hamartomas) A distinctive bony lesion such as sphenoid dysplasia or thinning of the long bone cortex with or without pseudoarthrosis A first-degree relative (parent, sibling, or offspring) with NF1 based upon the above criteria

Neurofibromatosis type 2 (NF2) is less frequent than NF1 and has a birth incidence of 1 in 30 000-40 000. It is caused by a dominant mutation in the NF2 gene which is located on chromosome 22. The NF2 gene codes for the protein Merlin/Schwannomin. The hallmarks of NF2 are the bilateral vestibular schwannomas (formerly called “acoustic neuromas”). There are also clinical diagnostic criteria available to make the diagnosis:

1. Bilateral vestibular schwannomas (histology or MRI)

2. A parent, sibling or child with NF2 and either
   - Unilateral vestibular schwannoma
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- Any two of the following: meningioma, glioma, schwannoma, posterior subcapsular lenticular opacities, cerebral calcification

3. Unilateral vestibular schwannoma and two or more of the following: meningioma, glioma, schwannoma, posterior subcapsular lenticular opacities, cerebral calcification

4. Multiple meningiomas (two or more) and one or more of the following: glioma, schwannoma, posterior subcapsular lenticular opacities, cerebral calcification

Patients with NF2 sometimes have a few café-au-lait spots but not more than 5. They can have tumors of the peripheral nerves (schwannomas). These schwannomas are sometimes mistaken for neurofibromas. Symptoms of bilateral vestibular schwannomas usually begin in early adult life with decreased hearing, tinnitus or vertigo. Symptomatic spinal tumors are frequently observed. The actuarial survival after diagnosis is on average 15 years.

**NF type 2 — Spinal tumors** are found in up to 90 percent of patients with NF-2. The tumors are often multiple, consisting primarily of schwannomas, but including some meningiomas [7]. About 2 percent of all spinal schwannomas are seen in patients with NF-2, while the rest are sporadic [8]. (See "Risk factors for brain tumors" section on Genetic factors).

**DIAGNOSIS —** Contrast-enhanced magnetic resonance imaging (MRI) is the most sensitive and specific imaging modality to evaluate possible spinal column lesions. The majority of NSTs enhance diffusely with contrast, but heterogeneous enhancement can be seen when intratumoral cysts, hemorrhage or necrosis are present.

Cerbellar haemangioblastoma
Gadolinium-enhanced T1-weighted axial magnetic resonance image reveals a large right-sided cerebellar tumor.

Meningioma

Signs in CT or MRI images will vary according to the cellular expression of the tumour. It is usually well circumscribed, but can become locally invasive or may rarely undergo metaplasia. Typically on CT, the tumour is extra-axial, of greater density than adjacent normal brain, may contain calcification and enhances densely, after intravenous contrast. Cystic change has been recorded, but is rare (1-2 percent). MRI scans of meningioma almost invariably show enhancement with IV Gadolinium contrast.

E. pontine glioma

Gliomas arising in the brainstem (midbrain, pons, and medulla oblongata) account for 10 to 20 percent of all central nervous system (CNS) tumors in children. They are more common in children than adults [1-4]. In the United States, for example, there are approximately 300 pediatric cases and 100 adult cases reported each year. In children, the median age at diagnosis is five to nine years of age, and the incidence is approximately equal between males and females.
On CT, these tumors infiltrate and enlarge the brain stem and secondarily displace the fourth ventricle. They commonly are hypodense but occasionally are of increased density compared to normal brain as seen on Film .3 in a different child. Calcification and cyst formation are infrequent (12%), and approximately half demonstrate enhancement after contrast (Film .4). When present, enhancement patterns are variable, with diffuse, nodular, or ring enhancement seen. A small number of these tumors may demonstrate exophytic growth into the posterior fossa cisterns, particularly the cerebellopontine angle cistern. These tumors tend to enhance with contrast administration.

Because of Hounsfield artifact in the posterior fossa, a small percentage of brain stem gliomas will be overlooked on initial CT scans. The superior imaging capability of MRI for brain stem and posterior fossa pathology allows recognition and characterization of lesions that are poorly delineated by CT, such as isodense or nonenhancing tumors.