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CASE REPORT Open Access

Giant sacral schwannoma in a neurofibromatosis type 2 patient

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Abstract

Background Neurofibromatosis type 2 is an autosomal dominant disorder, mainly characterized by multiple neurological lesions, such as schwannomas, meningiomas, neurofibromas and intramedullary ependymomas. Schwannomas are usually small circumscribed lesion. Sacral location of a schwannoma with cystic change is a very rare finding. We are presenting one such case with giant cystic schwannoma with fluid–fluid levels in sacral region.

Case presentation We present a case of 13-year-old female patient, presenting with pelvic pain and gradually progressive bilateral lower limb weakness. On MRI, giant cystic schwannoma with internal fluid–fluid levels was noted in sacral region, extending anteriorly into the presacral region, causing mass effects on pelvic organs, which explained the cause of her symptoms. She also showed the presence of bilateral vestibular schwannoma and multiple small cerebral lesions, leading to the diagnosis of neurofibromatosis type 2.

Conclusions Our current case of neurofibromatosis type 2, diagnosed by presence of bilateral vestibular schwannoma, shows atypically large sacral cystic schwannomas and cerebral subcortical lesions, probably representing glial microhamartomas. Sacral schwannomas can be of giant size with cystic changes and fluid–fluid levels, mimicking aneurismal bone cyst, as in current case.

Keywords Neurofibromatosis, Cystic schwannoma, Glial microhamartomas, Meningioangiomatosis

Background

Neurofibromatosis type 2 is a rare, autosomal dominant neurocutaneous disorder, which is mainly characterized by neurological, ocular and cutaneous manifestations. Classic neurological manifestations include the development of intracranial schwannomas, meningiomas and intramedullary ependymomas [1]. Other CNS lesions include microhamartomas, meningioangiomatosis along with intracranial calcified deposits [2]. Neurofibromatosis type 2 usually presents in early adulthood with sensorineural hearing loss from a vestibular schwannoma, which can be unilateral or bilateral. Other presenting complaints are visual disturbances, skin tumors and peripheral neuropathy [2].

Among symptomatic spinal cord lesions, most common are ependymomas and schwannomas. Most of spinal schwannomas are small, well-circumscribed tumors, which can frequently show degenerative changes such as hemorrhage, calcification and fibrosis; however, cystic changes are rather uncommon [3]. We are presenting a case of neurofibromatosis type 2 in a young female patient, showing the presence of a giant sacral schwannoma with cystic changes and multiple fluid–fluid levels mimicking the aneurysmal bone cyst.

Case presentation

A 13-year-old female patient presented in neurology department with pelvic pain and bilateral lower limb weakness for several months, which was gradually progressive. She had no other relevant history of such as trauma or constitutional symptoms. Patient did not have

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any skin lesions which were retrospectively confirmed after getting the MRI scans. There was no any similar history in her family. Patient had no history of any previous surgical interventions.

Her neurological examination revealed hyporeflexia, flaccid paraparesis and asymmetric weakness as well as reduced perineal sensations.

Her ultrasound scan of pelvis showed presence of a large multiseptated cystic lesion in left presacral region, displacing the uterus and adnexa toward right side. No other remarkable finding was present in ultrasound report.

Magnetic resonance imaging of lumbosacral spine showed a large multiseptated expansile cystic lesion of approximate size 6.1×5.0×9.4 cm (AP×TR×CC) in left presacral region arising from left S1-S2 neural foramen (Fig. 1a, b). Left S1 spinal nerve was noted to be continuous with the lesion (Fig. 1c). Lesion caused marked bone scalloping and widening in left S1-S2 neural foramen and central canal at L5, S1 and S2 vertebral body level. The lesion was T1 hypointense and T2 hyperintense, and showed multiple fluid–fluid levels.

Another smaller multiseptated cystic lesion of approximate size 22×20×31 mm (AP×TR×CC) was also noted in left L5 paravertebral region, continuous with left L4 spinal nerve (Fig. 1d). These findings were characteristic of cystic schwannomas.

We proceeded with MRI brain screening, which was proved to be diagnostic in this case. FLAIR image showed presence of two small hyperintense heterogeneous lesions at bilateral cerebellopontine angles (Fig. 2) with small intracanalicular components, which were characteristic of vestibular schwannomas. This led to diagnosis of neurofibromatosis type 2.

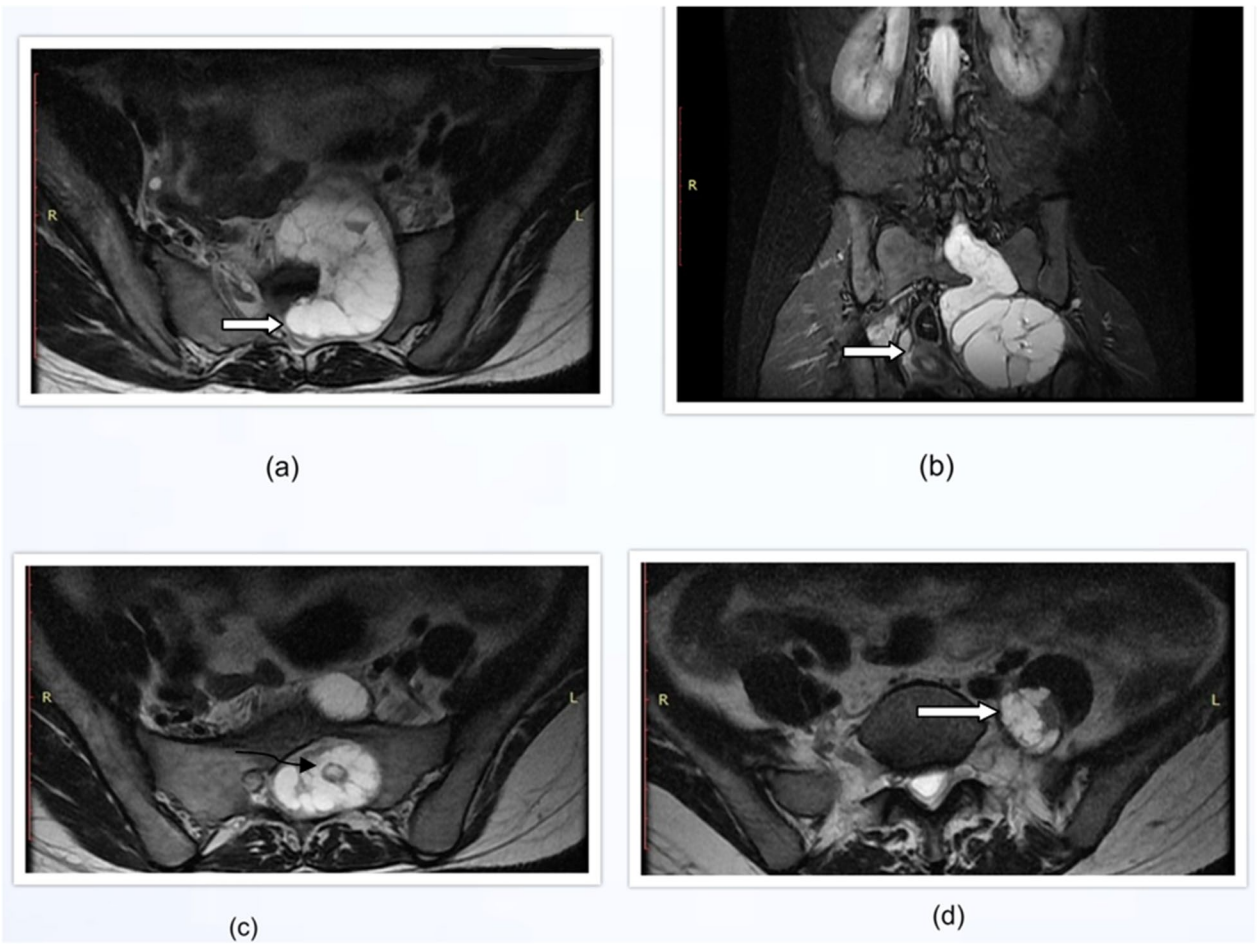

Fig. 1 Pelvis MRI of 13-year-old female patient, presenting with pelvic pain-**a** axial T2 image at S1 level shows a large multiseptated cystic lesion originating from left S1–S2 neural foramen with its scalloping. **b** Coronal STIR image shows pelvic location of the lesion, displacing the uterus toward right side. **c** Axial T2 image showing left S1 exiting nerve root within the cystic lesion, marked by curved arrow. **d** Axial T2 image at L5 level shows another cystic lesion with similar characteristics, along the course of left L5 spinal nerve

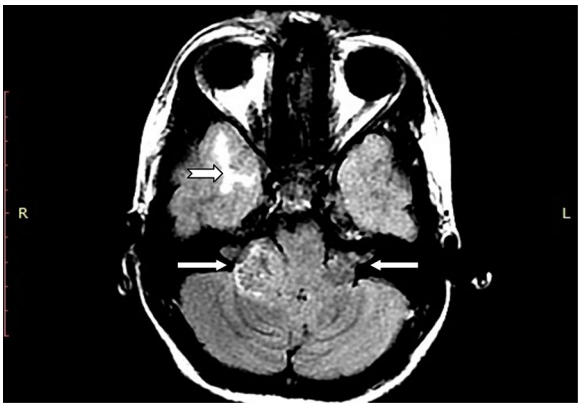

Fig. 2 T2/FLAIR sequence of brain showing heterogeneous lesions at bilateral cerebellopontine angles, with typical ice cream cone sign (marked by block arrows) indicating the intracanalicular component. These features are characteristics of vestibular schwannoma. Also note the hyperintensity in right temporal lobe (notched arrow) representing hamartomatous lesion

Multiple ill-defined T2/FLAIR hyperintense lesions were also noted in cortical and subcortical white matter of bilateral cerebral hemisphere, which may represent the glial hamartia, seen in neurofibromatosis type 2 cases. These lesions were similar to the focal area of signal intensity (FASI) seen in neurofibromatosis type 1 (Fig. 3).

Discussion

Neurofibromatosis type 2 is multiple neoplasia syndrome, resulting from mutations in NF2 tumor suppressor gene on long arm of chromosome 22. It generally presents in second and third decades of life with hearing loss, due to vestibular schwannoma [1]. Multiple diagnostic criteria have been developed for neurofibromatosis type 2, like Manchester criteria, the children's tumor foundation criteria and the national institute of health criteria and all of them indicate bilateral vestibular schwannoma as hallmark of neurofibromatosis type 2. Manchester diagnostic criteria are most widely used and are summarized in Table 1 [2].

Table 1 The Manchester diagnostic criteria for neurofibromatosis type 2 [2]

Key finding	Additional findings needed for diagnosis
Bilateral vestibular schwannomas	None
Family history	Unilateral vestibular schwannoma or two NF2-associated lesions like schwannoma, glioma, meningioma, or neurofibroma
Unilateral vestibular schwannoma	Two NF2-associated lesions
Multiple meningiomas	Unilateral vestibular schwannoma or two other NF2-associated lesions

Thus, the case we presented above is a typical case of neurofibromatosis type 2, having bilateral vestibular schwannoma. Other findings we got in the case were two other extramedullary schwannomas in sacral region as well as multiple cerebral hamartoma like lesions. Later two lesions were quite unusual in this case as sacral schwannoma are usually small, solid, well-circumscribed lesions [3]. And the cerebral intraparenchymal lesions were similar to focal area of signal intensity (FASI) seen in neurofibromatosis type 1 and glial hamartia seen in neurofibromatosis type 2; however, we did not proceed with histopathological examination to find the exact nature of lesion and relied on follow-up scans.

Sacral schwannoma in itself is a very rare entity and according to Cagli et al. [4] only about 50 cases have been reported in literature worldwide. They comprise of less than 1 to 5 percent of all spinal schwannomas [4].

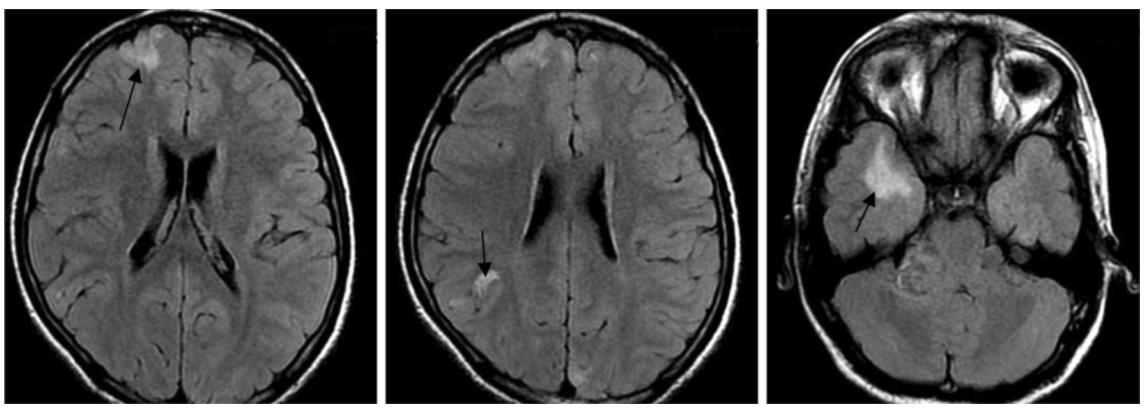

Fig. 3 T2/FLAIR sequence of brain shows multiple Ill defined hyperintense lesions (arrows) in right frontal, parietal and temporal regions—representing hamartomatous lesions

Typically spinal schwannoma is a well encapsulated and solid when small, however, may undergo necrotic changes to present like cystic mass, when it is large. Mechanism of this phenomenon is vascular insufficiency with the enlarging size of the tumor. According to Hughes et al. [3], presacral cystic schwannoma can be homogenous or heterogeneous with well-defined margins, thus may show iso- to high-intensity on T2-weighted MRI images. In our case, both sacral and lumbar lesions showed both types of signal intensity with fluid–fluid levels within the lesion. Fluid–fluid signal within the lesion was similar to the aneurysmal bone cysts; however, it was easily excluded after we noticed the multiplicity of lesion and their continuity with the spinal nerve roots. Similar cases of cystic giant sacral schwannoma have also been reported earlier in literature by Cho et al. [5] and Lee et al. [6]. Further, the finding of bilateral vestibular schwannoma, made the diagnosis almost certain.

Sridhar et al. [7] classified spinal schwannomas in five types, which is summarized in Table 2. Our case is type 5 tumor according to the classification.

Apart from vestibular schwannoma and meningioma, other intracranial findings which can be seen in neurofibromatosis type 2 cases are—(1) Benign intracranial calcified deposits within cerebral parenchyma, cerebellum and choroid plexus, (2) Microhamartomas, also called as glial hamartia—small dysplastic glial foci in the cerebral cortex, basal ganglia, cerebellum and spinal cord. Their presence in post-mortem examination is essentially pathognomonic of NF2; however, non-specific imaging features reduce its utility in radiological diagnosis criterion [6]. They may appear as T2/FLAIR hyperintense, frequently indistinguishable from focal cortical dysplasia (FCD) and may even show 'transmantle sign'. These lesions are not known to be associated with seizures or cognitive dysfunction [8]. (3) Meningioangiomatosis-Meningovascular hamartomatous plaque like lesions in cortex that may extend to overlying leptomeninges hence it is frequently both intra- and extra-axial lesions [9]. It can present as solitary lesion, or as a diffuse or multifocal lesions. Apart from being associated with NF2, it can also be sporadic. Sporadic lesions are most commonly seen in frontal lobe, whereas NF2-associated lesions are commonly seen in temporal lobe, followed by parietal lobe [9]. They may be associated with meningiomas, and in such cases, often differentiation between the two lesions cannot be made clearly. On CT, they are hypodense with heterogeneous contrast enhancement, and approximately 90% cases show calcification. On MRI, they are hypo-isointense and T2/FLAIR hyperintense with heterogeneous contrast enhancement. They frequently show gyriform pattern, because of leptomeningeal pattern of spread [10, 11].

Table 2 Sridhar classification of spinal schwannomas [7]

	Classification
Type 1	
Type 1a	Intradural intraspinal tumor, < 2 vertebral segments in length
Type 1b	Extradural intraspinal tumor, < 2 vertebral segments in length
Type 2	Intraspinal tumor > 2 vertebral segments in length—giant tumor
Type 3	intraspinal tumor with extension into nerve root foramina
Type 4	
Type 4a	Intraspinal tumor with extraspinal extension of < 2.5 cm
Type 4b	Intraspinal tumor with extraspinal extension of > 2.5 cm—giant tumor
Type 5	Tumor with erosion into the vertebral bodies (giant invasive tumor) as well as lateral and posterior extensions into myofascial planes

In our case, multiple T2/FLAIR hyperintense foci noted at cortex and subcortical regions of both cerebral hemisphere. Involved cortex was mildly thickened with dysplastic appearance and, hence, was closer to the glial hamartia. Follow-up was advised to assess the nature and stability of the lesions.

Take home messages for radiologists from our current case are—(1) For a large cystic lesion of sacral origin, schwannoma can be one of differential diagnosis. (2) Large schwannomas can present with multiple fluid–fluid levels, mimicking aneurysmal bone cyst. (3) Brain screening is necessary in suspected cases of spinal schwannoma, to assess the presence of other schwannomas, particularly at cerebellopontine angles. (4) Neurofibromatosis type 2 patients can show cerebral focal area of signal intensities, for which glial hamartia is a differential diagnosis.

Limitations of this study include—(1) Absence of genetic test of the patient to confirm the diagnosis of neurofibromatosis type 2. (2) Absence of histopathological studies of the lesions to ascertain their exact nature. (3) Follow-up and treatment history of the patient is not available.

Conclusions

Our current case of neurofibromatosis type 2, diagnosed by presence of bilateral vestibular schwannoma according to Manchester's criterion, shows atypically large sacral cystic schwannoma, mimicking aneurysmal bone cysts. Other findings were presence of multiple T2/FLAIR hyperintense predominately subcortical lesions, probably representing glial microhamartomas.

Abbreviations

NF	Neurofibromatosis
FLAIR	Fluid attenuated inversion recovery
MRI	Magnetic resonance imaging
AP	Anteroposterior
TR	Transverse
CC	Craniocaudal

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Not applicable.

Author contributions
Patient selection and follow-up were done by NS and SV. The manuscript was written by NS, SV and DV. All the authors have made necessary comments and contributions to the manuscript. All authors have read and approved the manuscript.

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Availability of data and materials
The data and images in the current study are available from the corresponding author, on reasonable request.

Declarations

Ethics approval and consent to participate
The procedure described was in accordance with the institutional ethical guidelines and conform to the WMA Declaration of Helsinki—ethical principles for medical research involving human subjects.

Consent for publication
Written informed consent was obtained from the patient and patient relatives for publication of the article.

Competing interests
The authors declare that they have no competing interests.

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