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EXTENSIVE NEUROFIBROMATOSIS TYPE 1 WITH MULTIPLE CUTANEOUS NEUROFIBROMAS, LUMBOSACRAL MENINGOCELES, VERTEBRAL DYSPLASIA AND INCIDENTAL CEREBRAL FINDINGS IN A MIDDLE-AGED MALE: A RADIOLOGICAL CASE REPORT

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ABSTRACT
Neurofibromatosis type 1 (NF1) is a common autosomal dominant neurocutaneous disorder characterized by multisystem involvement, including cutaneous neurofibromas, skeletal dysplasia, and central and peripheral nervous system abnormalities. Extensive spinal meningoceles associated with vertebral dysplasia are uncommon manifestations and are frequently detected incidentally during imaging. We report a rare case of a 52-year-old male with a history of post-polio paralysis since childhood who presented with intermittent headache of two months' duration. Clinical examination revealed multiple painless cutaneous neurofibromas involving the scalp, trunk, back, gluteal region, and extremities. Magnetic resonance imaging (MRI) of the brain and whole spine demonstrated multiple subcutaneous neurofibromas, a large multiloculated lumbosacral meningocele measuring approximately 11.2 × 7.8 cm, vertebral dysplasia involving the lower lumbar and sacral vertebrae, posterior vertebral scalloping, widened sacral neural foramina, and dural ectasia. Mild cerebral atrophic changes, cervical spondylotic changes, and incidental inflammatory changes within the paranasal sinuses were also identified, without evidence of intracranial neoplasm or optic pathway glioma. Computed tomography (CT) further confirmed extensive dysplastic remodeling of the lower lumbar and sacral vertebrae, posterior vertebral scalloping, widened sacral foramina, and anterior as well as posterior sacral meningoceles, providing superior characterization of the associated osseous abnormalities. The constellation of clinical and radiological findings was diagnostic of Neurofibromatosis Type 1. This case highlights the broad radiological spectrum of adult NF1 and underscores the complementary roles of MRI in delineating neural and soft-tissue involvement and CT in characterizing skeletal abnormalities. Recognition of these characteristic imaging findings is essential for establishing the diagnosis, assessing disease burden, guiding multidisciplinary management, and ensuring appropriate long-term surveillance. This case further contributes to the limited literature describing extensive lumbosacral meningoceles with vertebral dysplasia in adults with Neurofibromatosis Type 1.

KEYWORDS: Neurofibromatosis type 1; Magnetic resonance imaging; Computed tomography; Neurofibroma; Lumbosacral meningocele; Vertebral dysplasia; Dural ectasia.

INTRODUCTION
Neurofibromatosis type 1 (NF1), also known as von Recklinghausen disease, is one of the most common inherited neurocutaneous disorders, characterized by multisystem involvement affecting the skin, peripheral nerves, central nervous system, bones, and soft tissues. It results from mutations in the NF1 gene, leading to abnormal proliferation of neural crest-derived cells and the development of multiple benign nerve sheath tumors known as neurofibromas. Although the disorder follows an autosomal dominant pattern of inheritance, a significant proportion of cases arise due to spontaneous mutations. The clinical presentation is highly variable, ranging from isolated cutaneous lesions to extensive neurological and skeletal abnormalities.

Cutaneous neurofibromas are the most frequent manifestation of NF1; however, deeper lesions involving the spinal canal, meninges, and peripheral nerves may remain clinically silent until adulthood. Skeletal abnormalities such as vertebral dysplasia, posterior vertebral scalloping, dural ectasia, enlargement of neural foramina, scoliosis, and meningoceles represent important but relatively uncommon manifestations. These abnormalities may lead to compression or displacement of adjacent neural structures, resulting in pain, neurological deficits, or incidental findings during imaging performed for unrelated complaints.

Magnetic resonance imaging (MRI) plays a pivotal role in the evaluation of patients with NF1 by accurately delineating the extent of soft tissue, neural, and skeletal involvement. MRI provides excellent visualization of intracranial lesions, spinal abnormalities, peripheral nerve tumors, and associated complications without exposure to ionizing radiation.

Early recognition of these findings is essential for appropriate surveillance, multidisciplinary management, and timely intervention in symptomatic patients.

The coexistence of extensive cutaneous neurofibromas with multiple spinal meningoceles and vertebral dysplasia is an uncommon radiological presentation. Such cases demonstrate the broad phenotypic spectrum of NF1 and emphasize the importance of comprehensive imaging in identifying associated abnormalities that may otherwise remain undetected. We present a rare case of a middle-aged male with long-standing post-polio paralysis who underwent MRI evaluation for headache, revealing extensive neurofibromatosis type 1 with multiple subcutaneous neurofibromas, lumbosacral meningoceles, vertebral dysplasia, and characteristic spinal abnormalities.

Case Presentation

A 52-year-old male presented to the Department of Radiology with complaints of intermittent headache following an oil bath for the preceding two months. There was no history of seizures, loss of consciousness, focal neurological deficits, vomiting, visual disturbances, bowel or bladder dysfunction, or back pain. He had a known history of post-polio paralysis involving the lower limb since childhood. Physical examination revealed multiple painless cutaneous nodules distributed over the scalp, trunk, back, gluteal region, and extremities, which had gradually increased in number over several years. There was no significant past medical or surgical history, and the family history was unremarkable.

Magnetic resonance imaging (MRI) of the brain with whole-spine screening was performed to evaluate the cause of headache. MRI demonstrated multiple well-defined T1-isointense and T2/FLAIR-hyperintense subcutaneous lesions involving the frontal, parietal, and occipital scalp regions, with similar lesions extending into the cervical, thoracic, lumbar, and bilateral gluteal subcutaneous tissues, consistent with multiple neurofibromas. No focal intracranial parenchymal lesion, diffusion restriction, intracranial hemorrhage, or abnormal post-contrast enhancement was identified. Mild generalized prominence of the cerebral sulci and cisternal spaces, suggestive of age-related cerebral atrophy, was noted. Mild mucosal thickening was present within the bilateral ethmoid, frontal, and left maxillary sinuses. Cervical spine screening demonstrated a posterior disc-osteophyte complex at the C5–C6 level producing mild ventral thecal sac indentation without spinal cord compression or abnormal cord signal.

Whole-spine screening revealed a large multiloculated lumbosacral meningocele measuring approximately 11.2 × 7.8 cm, containing multiple internal septations. Associated osseous abnormalities included dysplastic changes involving the L3 vertebra to the sacrum, marked posterior vertebral scalloping, dural ectasia, widening of the bilateral sacral neural foramina, and displacement with partial encasement of the cauda equina nerve roots. Additional lateral thoracic and posterior sacral meningoceles were also identified. Subsequent computed tomography (CT) of the lumbosacral spine confirmed extensive dysplastic remodeling of the lower lumbar and sacral vertebrae with anterior vertebral dysplasia, posterior vertebral scalloping, widened sacral foramina, and both anterior and posterior sacral meningoceles, providing excellent delineation of the associated bony abnormalities. The constellation of characteristic clinical and radiological findings was diagnostic of Neurofibromatosis Type 1 (NF1). The patient was referred for neurosurgical evaluation and multidisciplinary management with recommendations for long-term clinical and radiological surveillance.

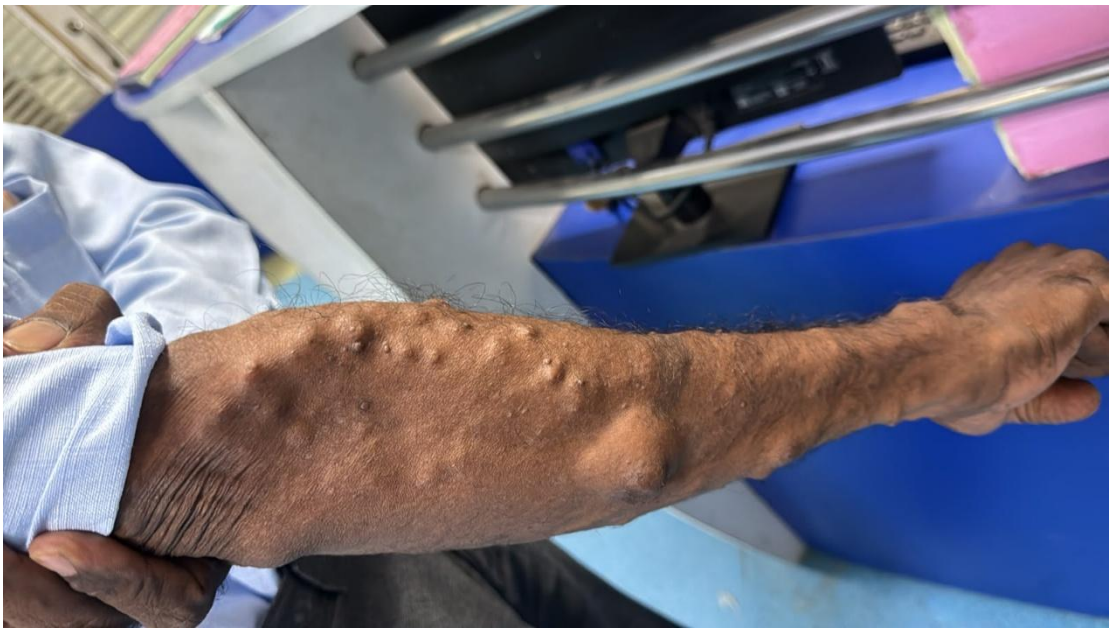

Figure 1. Multiple cutaneous neurofibromas involving the upper limb.

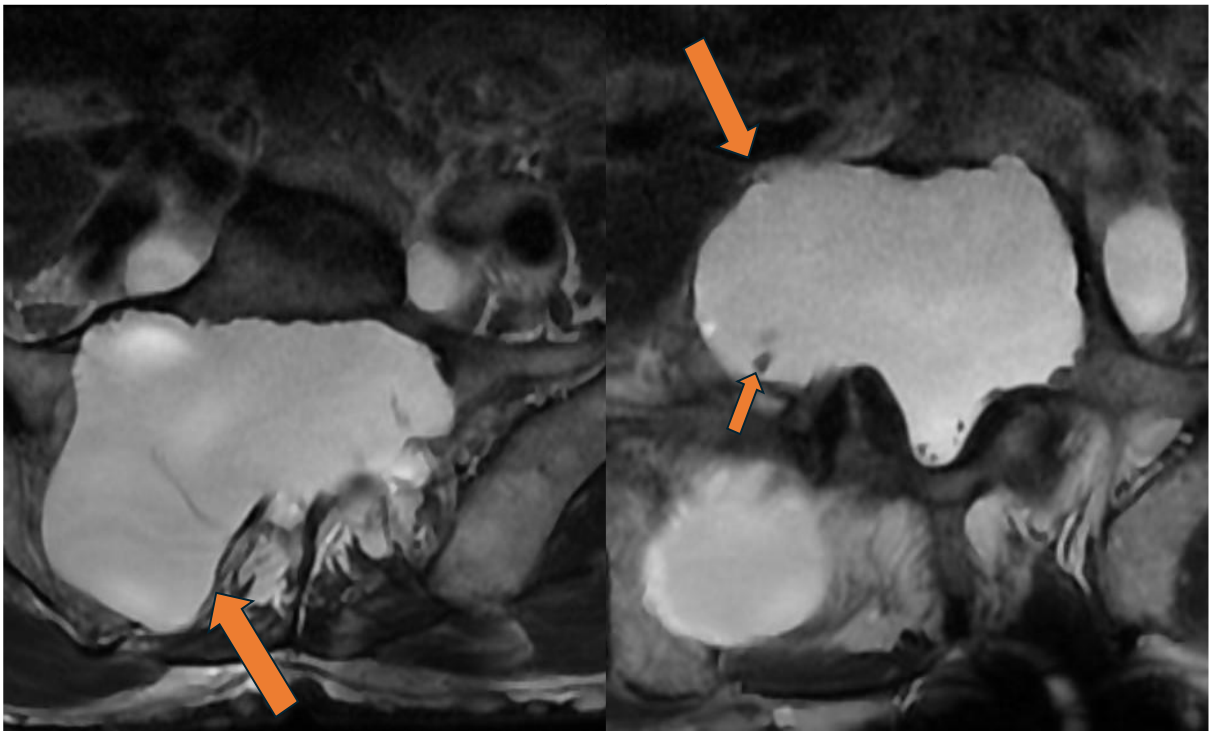

Figure 2. Axial T2-weighted MRI of the lumbosacral spine demonstrating a large multiloculated lumbosacral meningocele with internal septations and associated posterior vertebral scalloping.

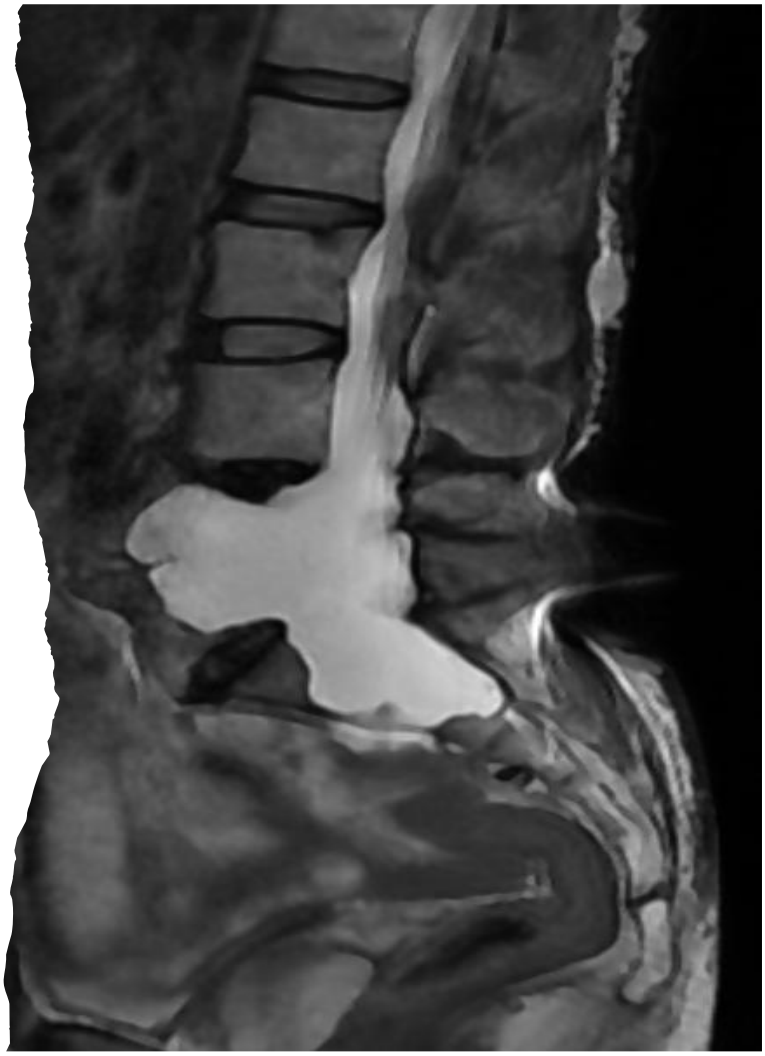

Figure 3. Sagittal T2-weighted MRI demonstrating extensive lumbosacral meningocele associated with vertebral dysplasia and dural ectasia.

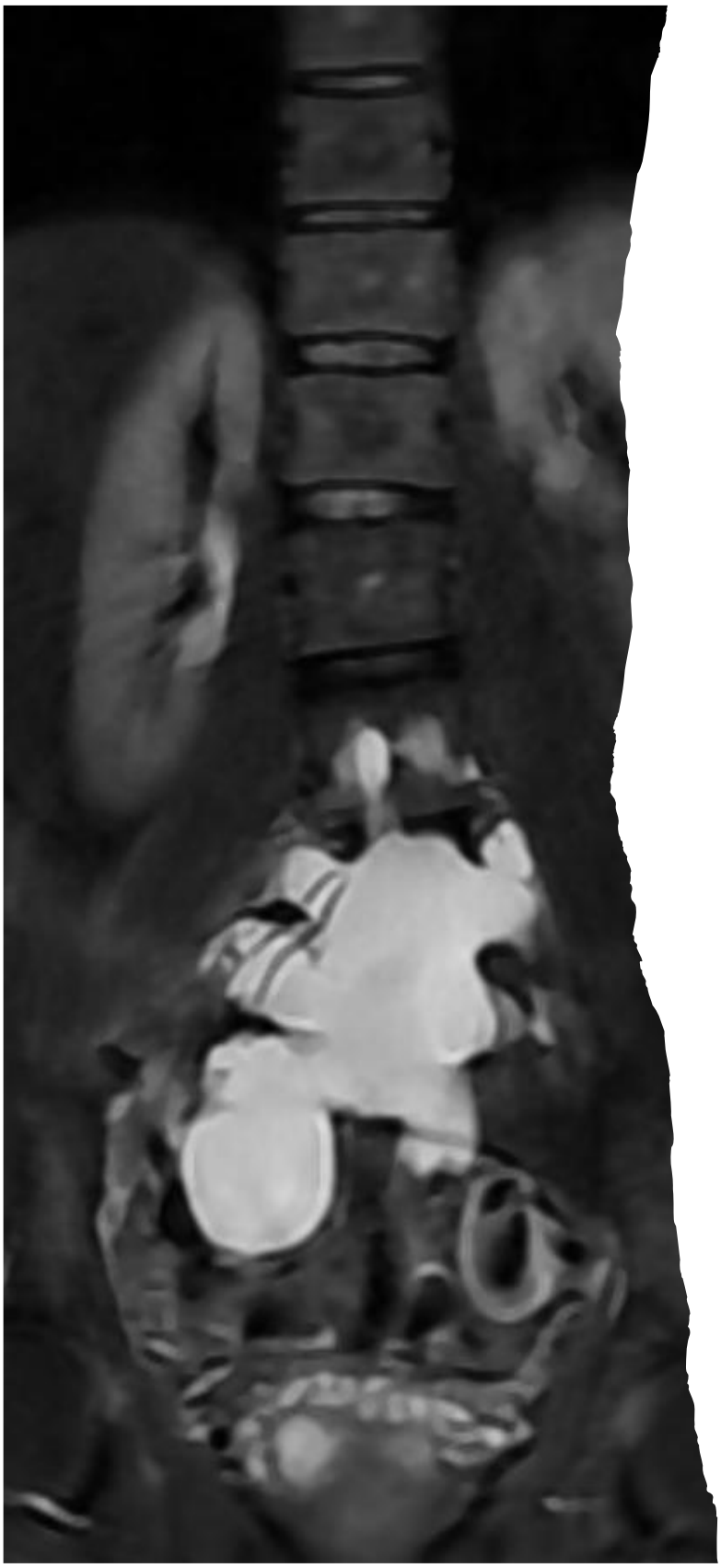

Figure 4. Coronal MRI of the lumbosacral spine demonstrating dysplastic lower lumbar and sacral vertebrae with widening of the bilateral neural foramina and associated meningocele.

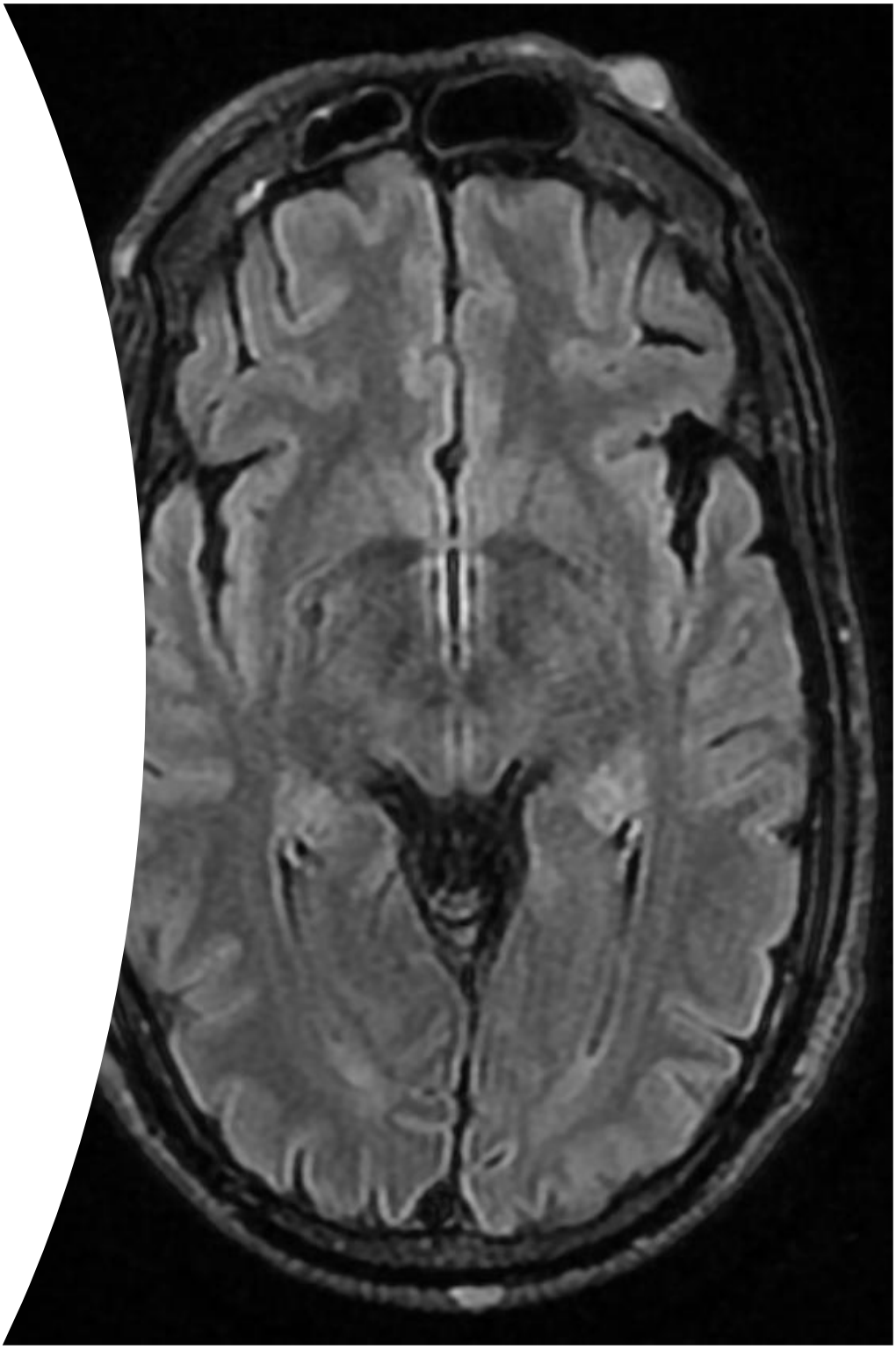

Figure 5. Axial FLAIR MRI of the brain demonstrating multiple subcutaneous scalp neurofibromas, with no focal intracranial parenchymal abnormality identified. The case report describes these lesions as T1-isointense and T2/FLAIR-hyperintense.

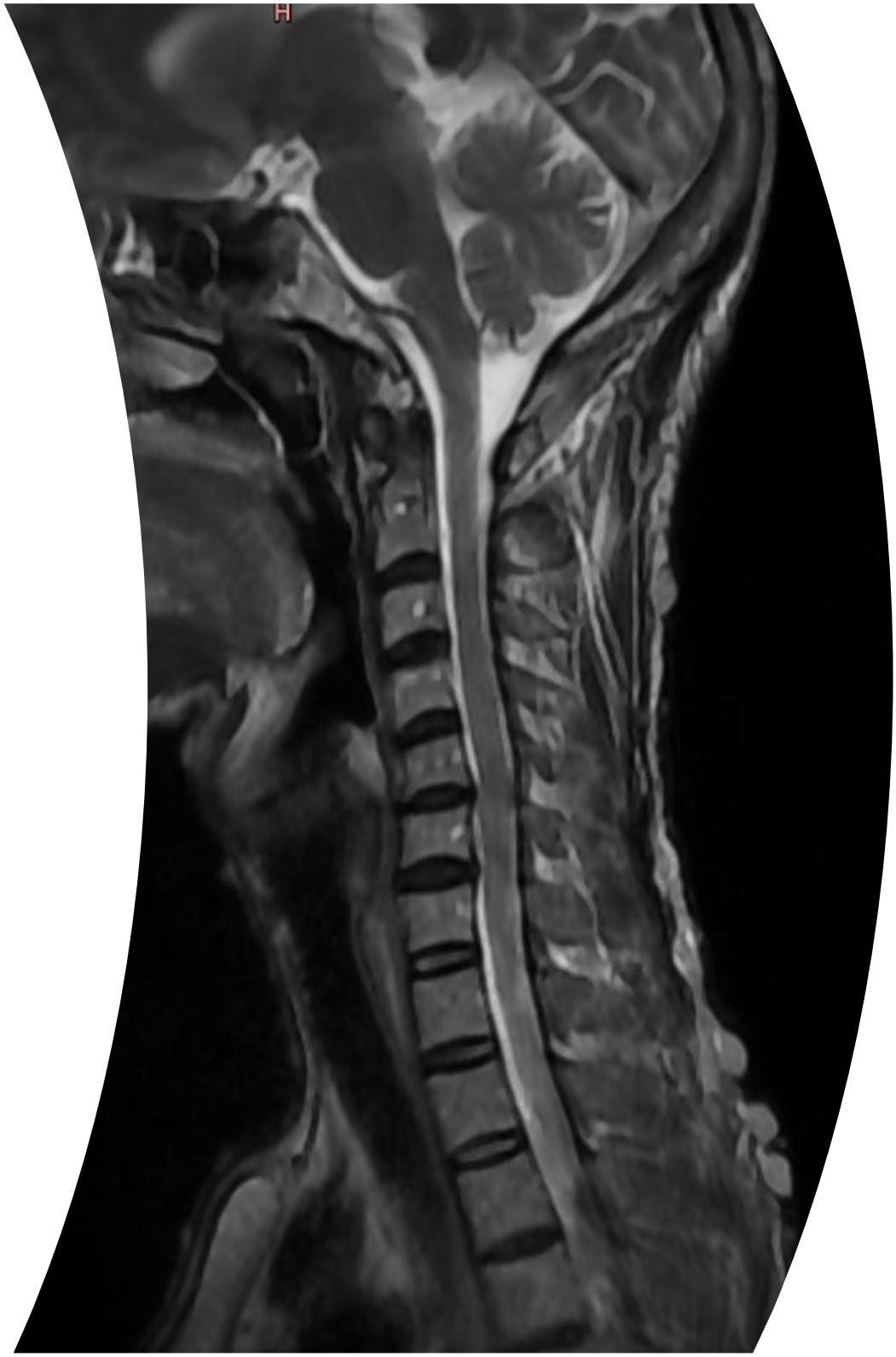

Figure 6. Sagittal T2-weighted MRI of the cervical spine demonstrating multiple subcutaneous neurofibromas and mild C5–C6 disc-osteophyte complex with ventral thecal sac indentation without spinal cord compression.

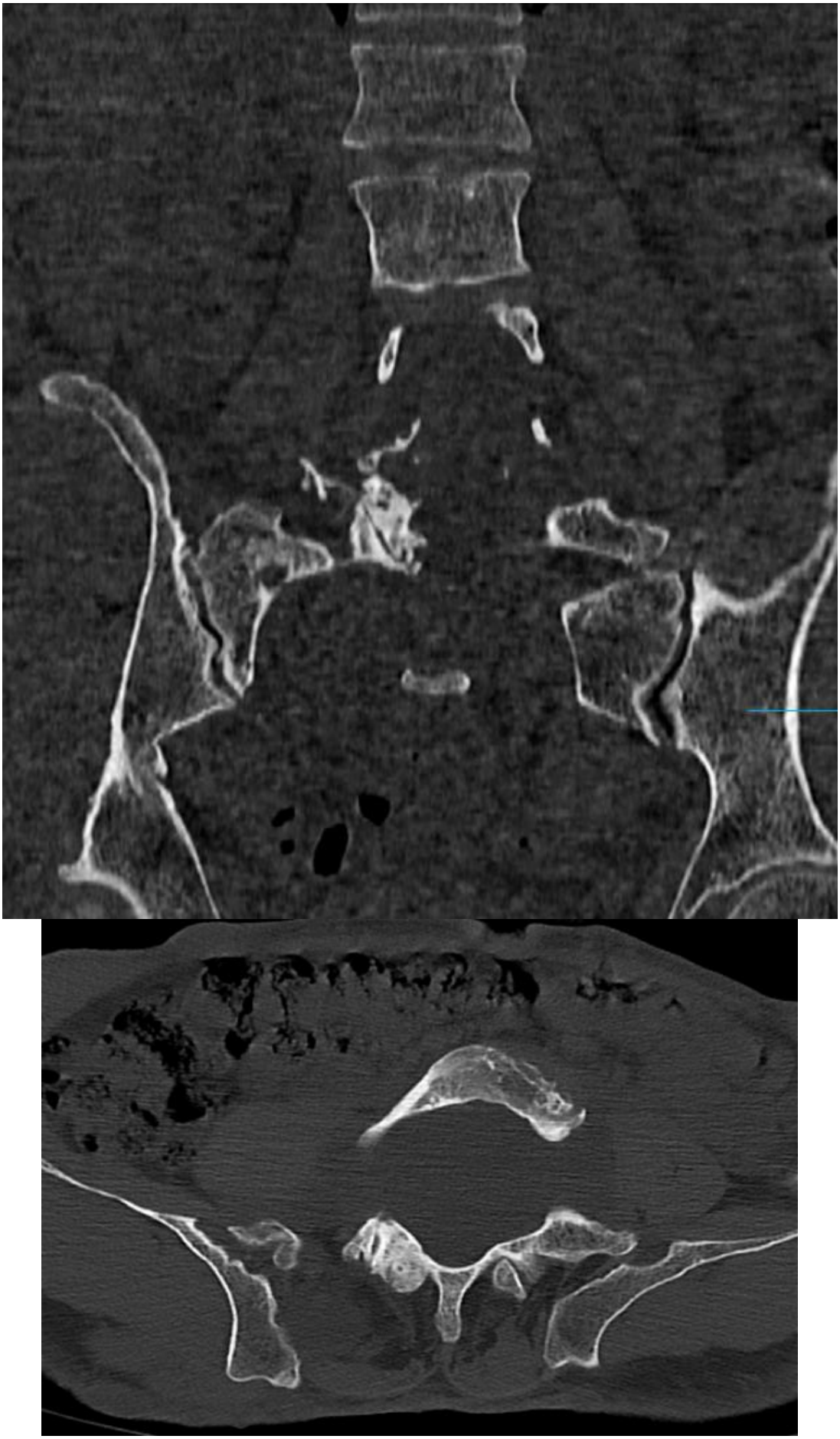

Figure 7: Coronal CT of the lumbosacral spine demonstrating extensive dysplastic remodeling of the lower lumbar and sacral vertebrae with posterior vertebral scalloping, widened sacral neural foramina, and associated meningocele.

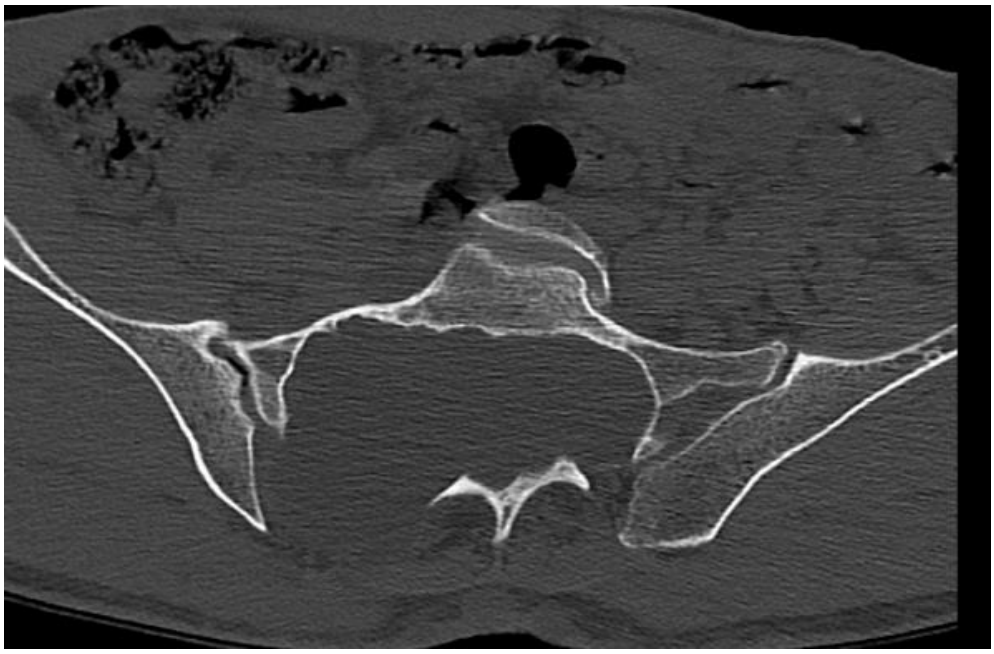

Figure 8: Axial CT images of the lumbosacral spine demonstrating vertebral dysplasia with posterior scalloping and associated anterior and posterior sacral meningoceles, with widening of the neural foramina.

DISCUSSION

Neurofibromatosis type 1 (NF1) is a common autosomal dominant neurocutaneous disorder characterized by multisystem involvement affecting the skin, peripheral nerves, central nervous system, and skeletal system. The clinical manifestations are highly variable, ranging from isolated cutaneous lesions to extensive spinal and osseous abnormalities. Magnetic resonance imaging (MRI) remains the imaging modality of choice for evaluating disease burden, delineating neural and soft-tissue involvement, identifying associated complications, and guiding long-term surveillance. Complementary computed tomography (CT) is particularly valuable for detailed assessment of osseous abnormalities, including vertebral dysplasia, neural foraminal enlargement, and bony remodeling associated with dural ectasia.[1]

The predominant imaging finding in the present case was the presence of multiple cutaneous and subcutaneous neurofibromas involving the scalp, trunk, back, gluteal region, and extremities. These lesions represent the most common benign peripheral nerve sheath tumours encountered in NF1 and typically increase in both size and number with advancing age. Nix et al. described similar MRI characteristics, demonstrating well-defined T1-isointense and T2-hyperintense lesions that allow excellent delineation of soft-tissue involvement, reinforcing MRI as the preferred modality for evaluating the overall extent of disease.[1]

A particularly striking feature of our patient was the presence of a large multiloculated lumbosacral meningocele associated with vertebral dysplasia, posterior vertebral scalloping, widened sacral neural foramina, and dural ectasia. These abnormalities result from chronic cerebrospinal fluid pulsations acting on congenitally weakened dura, producing progressive osseous remodeling and herniation of the meninges through dysplastic vertebral defects. MRI accurately demonstrated the relationship of the meningoceles to the neural elements, whereas CT clearly depicted the associated vertebral dysplasia, posterior scalloping, widened sacral foramina, and anterior as well as posterior sacral meningoceles, highlighting the complementary role of both imaging modalities. Similar radiological findings were reported by Hsieh et al., who emphasized the indispensable role of MRI in preoperative anatomical assessment of giant meningoceles in NF1.[2]

Likewise, Mousavi et al. reported that giant meningoceles associated with NF1 may remain clinically silent for years and are often detected incidentally during imaging performed for unrelated complaints. Conservative management may be appropriate in asymptomatic patients, whereas surgical intervention is generally reserved for neurological deficits, progressive enlargement, or compression of adjacent structures.[3] Our patient similarly had extensive meningoceles identified during imaging performed for headache rather than symptoms directly attributable to the spinal lesions.

The skeletal abnormalities observed in this patient, including vertebral dysplasia, posterior vertebral scalloping, dural ectasia, and widening of the sacral neural foramina, represent characteristic osseous manifestations of NF1. Chronic dural ectasia produces gradual remodeling of the vertebral bodies, resulting in the classical imaging appearance demonstrated in this case. CT was particularly useful in confirming these osseous changes and complementing the MRI findings. Comparable radiological features have been described by Giannopoulos et al., who highlighted the importance of recognising these characteristic imaging findings to avoid diagnostic confusion with other cystic thoracic or spinal lesions.[4]

Although our patient did not demonstrate intracranial tumours or optic pathway glioma, MRI revealed multiple scalp neurofibromas together with mild cerebral atrophic changes and incidental inflammatory changes within the paranasal sinuses. Adult patients with NF1 frequently exhibit peripheral nerve tumours and skeletal manifestations rather than

intracranial neoplasms. Matsumoto et al. similarly demonstrated that neurological manifestations in adults may arise secondary to large meningoceles and their complications rather than primary intracranial disease, further supporting the value of comprehensive craniospinal imaging.[5]

An additional noteworthy feature of the present case was the coexistence of post-polio paralysis with extensive NF1. Although no direct pathogenic relationship has been established between these conditions, underlying neuromuscular disability may delay recognition of progressive spinal abnormalities or neurological deficits related to meningoceles. Comprehensive clinical examination together with whole-spine MRI and targeted CT evaluation is therefore essential to identify clinically occult but significant abnormalities.

Current clinical recommendations advocate lifelong multidisciplinary follow-up for adults with NF1 because of the risk of progressive skeletal deformities, neurological complications, malignant peripheral nerve sheath tumours, and other systemic manifestations. Miller et al. highlighted the importance of regular surveillance for early detection of disease progression, while Stewart et al. recommended coordinated multidisciplinary management involving neurology, orthopaedics, radiology, neurosurgery, and clinical genetics to optimise long-term outcomes.[6,7]

CONCLUSION

Neurofibromatosis type 1 is a multisystem genetic disorder with diverse clinical and radiological manifestations. This case highlights a rare presentation of extensive cutaneous neurofibromas associated with giant multiloculated lumbosacral meningoceles, vertebral dysplasia, posterior vertebral scalloping, widened sacral neural foramina, and dural ectasia in a middle-aged male with coexisting post-polio paralysis. Although the patient presented with headache, imaging revealed widespread spinal and soft-tissue abnormalities that were largely incidental but clinically significant. MRI was invaluable for delineating neural, meningeal, and soft-tissue involvement, whereas CT complemented these findings by accurately characterising the associated osseous abnormalities. Recognition of these characteristic imaging features is essential for establishing the diagnosis, assessing disease burden, planning multidisciplinary management, and ensuring appropriate long-term surveillance. This case further expands the limited literature describing extensive lumbosacral meningoceles with vertebral dysplasia in adults with Neurofibromatosis Type 1.

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