Saudi Journal of Medicine and Public Health

https://saudijmph.com/index.php/pub

https://doi.org/10.64483/202631693

Jugular Foramen Meningocele in a 37-Year-Old Saudi Woman with Neurofibromatosis Type I: A Clinical Case Report

Fahad Alshehri[(1)]

(1) Medical Imaging Department, College of Medicine, Medical Collage and Medical City, Qassim University, Saudi Arabia, Saudi Arabia

Abstract

Summary

A 37-year-old woman presented with a five-year history of progressively worsening hearing loss in the right ear accompanied by pulsatile tinnitus. She also reported recurrent headaches described as dull and aching, typically lasting several hours and mainly affecting the right side of the head. There was no history of head trauma or previous surgical procedures, and the symptoms appeared without any identifiable precipitating factor. The patient denied experiencing visual disturbances, vertigo, dizziness, ear pain, nausea, or recurrent upper respiratory tract infections. General physical and neurological examinations were unremarkable, and no abnormalities were detected on systemic evaluation. Ophthalmological and otorhinolaryngological assessments also revealed no significant findings. Routine laboratory investigations were within normal limits and did not contribute to the diagnosis. Imaging evaluation revealed skeletal abnormalities including spinal meningocele and sphenoid wing dysplasia, findings consistent with Neurofibromatosis type I (NF1). Based on these radiological features, NF1 was diagnosed in this patient, an autosomal dominant neurocutaneous disorder known to be associated with various skeletal and neurological manifestations.

Keywords: Jugular foramen meningocele, 37 year female, neurofibromatosis type I, case report.

Introduction

Neurofibromatosis type I (NF1) is a hereditary neurocutaneous disorder characterized by multisystem involvement and autosomal dominant inheritance. The condition results from mutations in the NF1 gene located on chromosome 17, which encodes the protein neurofibromin [1]. Loss or dysfunction of neurofibromin disrupts normal regulation of cellular growth and signaling pathways, predisposing affected individuals to a range of cutaneous, neurological, and skeletal manifestations [2]. The diagnostic criteria for NF1 were established during the National Institutes of Health (NIH) Consensus Development Conference and emphasize characteristic findings involving the skin, bones, and nervous system, including café-au-lait macules, neurofibromas, skeletal dysplasia, and specific nervous system abnormalities [3]. Although NF1 is primarily recognized for its cutaneous and peripheral nerve manifestations, skeletal abnormalities are also well documented. These may include spinal deformities, sphenoid wing dysplasia, and other osseous defects [4]. Cranial abnormalities, particularly involving the skull base, are relatively uncommon but can have important clinical implications. Among these, defects of the sphenoid bone and calvarial abnormalities have been reported in association with NF1 [5]. Meningoceles, defined as herniations of the meninges through a bony defect, may occur in various locations but are rarely described in the region of the jugular foramen [6]. The presence of a jugular foramen meningocele associated with NF1 is an exceptionally rare finding, with only limited cases reported in the literature. Recognition of such anomalies is important because they may present with nonspecific symptoms and are often detected incidentally on imaging studies [6,7]. In this report, we describe a case of a 37-year-old female diagnosed with Neurofibromatosis type I who presented with progressive right-sided hearing loss, pulsatile tinnitus, and recurrent headaches. Imaging studies revealed skeletal abnormalities consistent with NF1, including sphenoid wing dysplasia and spinal meningocele, as well as a rare jugular foramen meningocele. This case highlights the role of imaging in identifying unusual skull base manifestations of NF1 and contributes to the limited literature describing this rare association.

Case Report

A 37-year-old woman from central region of Saudi Arabia was presented in this case, with a diagnosis of Neurofibromatosis type I presented with a long-standing history of progressive right-sided hearing loss accompanied by pulsatile tinnitus. She also reported recurrent episodes of dull, aching headaches that typically lasted several hours and were

*Corresponding author e-mail: Fshehri@qumc.edu.sa (Fahad Alshehri).

Receive Date: 5 April 2026, Revise Date: 13 May 2026, Accept Date: 13 May 2026

predominantly localized to the right side of the head. There was no history of trauma, previous surgical procedures, nausea, vomiting, vertigo, dizziness, or visual disturbance. On clinical examination, the patient was neurologically intact and no focal neurological deficits or cranial nerve palsies were identified. Routine laboratory investigations were unremarkable. Given the persistence of her symptoms, radiological evaluation of the brain and skull base was performed.

Magnetic resonance imaging (MRI) of the brain demonstrated a cerebrospinal fluid (CSF)–containing pouch arising from the anteroinferior surface of the cerebellum and extending into the right jugular fossa, consistent with a meningocele. The lesion appeared hyperintense on T2-weighted images and measured approximately 3 × 2.5 cm. Associated herniation of gliotic cerebellar tissue was also observed. These findings were demonstrated on sagittal, axial, and coronal T2-weighted sequences (Figure 1A, 1B, and 1D). Post-contrast imaging showed no abnormal enhancement of the lesion (Figure 1C). In addition, the lesion produced a mild mass effect, resulting in compression of the right internal jugular vein along its posterior margin.

Complementary computed tomography (CT) imaging of the skull base revealed widening of the right jugular foramen compared with the contralateral side, indicating expansion of the bony margins associated with the lesion (Figure 2A and 2B). The imaging findings were consistent with a jugular foramen meningocele, an uncommon manifestation that may occur in association with NF1.

Based on the clinical presentation and imaging findings, a diagnosis of right jugular foramen meningocele associated with Neurofibromatosis type I was established. The patient was advised periodic clinical and radiological follow-up to monitor for potential progression of the lesion.

Discussion

NF1 is a relatively common inherited neurocutaneous disorder with an estimated prevalence of approximately 1 in 3,000–4,000 live births. The condition follows an autosomal dominant inheritance pattern and results from mutations in the NF1 gene on chromosome 17, which encodes the tumor suppressor protein neurofibromin [8]. Dysfunction of this protein leads to dysregulation of cellular growth pathways and contributes to the diverse clinical manifestations associated with the disorder. The disease is primarily recognized for its dermatological features, including café-au-lait macules and axillary freckling, as well as the development of benign peripheral nerve sheath tumors known as neurofibromas [8,9]. In addition to cutaneous and neural manifestations, NF1 is also associated with a range of skeletal abnormalities. These include spinal deformities, vertebral defects, and characteristic cranial abnormalities such as sphenoid wing dysplasia. Skeletal involvement in NF1 reflects abnormal bone development and remodeling associated with the underlying genetic defect [10]. Spinal meningoceles have also been described in patients with NF1, particularly within the thoracic region [11]. However, the presence of meningoceles involving the skull base remains an uncommon finding.

Cephaloceles represent herniation of intracranial structures through defects in the skull and are broadly categorized as congenital (primary) or acquired (secondary). Congenital forms arise from developmental defects of the skull during embryogenesis, whereas secondary cephaloceles may occur as a consequence of trauma, surgical procedures, or other structural disruptions [12]. Cephaloceles most frequently occur in the occipital region, while basal variants are considerably less common. Basal meningoencephaloceles are estimated to account for only 1–10% of all encephaloceles and may remain clinically silent until adulthood depending on their size and anatomical location [12]. The jugular foramen is a complex skull base structure that transmits several important neurovascular structures, including the internal jugular vein and cranial nerves IX, X, and XI. Lesions involving this region are most commonly neoplastic, such as paragangliomas, schwannomas, or meningiomas [13]. In contrast, meningocele formation within the jugular foramen is extremely rare and is not typically included within the conventional classification systems of cephaloceles. Only a limited number of cases describing jugular foramen meningocele in association with NF1 have been reported in the literature [6-8]. Previous report by Siddiqui et al. described similar findings detected incidentally during imaging evaluation in adult patients with NF1 [7]. Imaging plays a crucial role in identifying and characterizing these uncommon lesions. Magnetic resonance imaging (MRI) is particularly valuable for demonstrating the CSF signal characteristics of meningoceles and for assessing associated brain tissue herniation. In the present case, MRI revealed a CSF-filled pouch extending from the anteroinferior cerebellar surface into the right jugular fossa (Figure 1A, 1B, and 1D) without post-contrast enhancement (Figure 1C), consistent with a meningocele. Complementary CT imaging provided important information regarding the bony anatomy of the skull base and demonstrated widening of the right jugular foramen (Figure 2A and 2B), supporting the diagnosis. Although many meningoceles may remain asymptomatic, their clinical significance lies in the potential for mass effect on adjacent neurovascular structures. In the present case, the lesion resulted in compression of the right internal jugular vein and was associated with symptoms including pulsatile tinnitus and progressive hearing impairment.

Recognition of such lesions is therefore important for accurate diagnosis and appropriate follow-up.

In short, jugular foramen meningocele represents a very rare skull base manifestation of NF1. The present case contributes to the limited body of literature describing this unusual association and highlights the importance of multimodality imaging in identifying rare structural abnormalities in patients with NF1. Continued documentation of similar cases may help improve understanding of the spectrum of skeletal and skull base abnormalities associated with this condition.

Conclusion

This case highlights an unusual skull base manifestation of Neurofibromatosis type I (NF1) in the form of a jugular foramen meningocele. Although NF1 is commonly associated with cutaneous findings and peripheral nerve tumors, skeletal abnormalities involving the skull base are much less frequently encountered. The present report demonstrates that meningoceles may rarely arise in the region of the jugular foramen and may present with nonspecific clinical symptoms such as hearing impairment, pulsatile tinnitus, or headache. Advanced imaging techniques play a crucial role in recognizing such abnormalities. In this case, magnetic resonance imaging clearly demonstrated a CSF-filled herniation extending into the jugular fossa, while computed tomography confirmed the associated widening of the jugular foramen. The combination of these imaging modalities allowed accurate characterization of the lesion and its relationship with adjacent neurovascular structures. Awareness of this rare association is important for clinicians and radiologists evaluating patients with NF1, particularly when symptoms related to the skull base are present. Careful imaging assessment and periodic follow-up are recommended to monitor potential progression and to guide appropriate clinical management.

Authors' Contributions:

Both authors were involved in all stages of the study, including collection and evaluation of the clinical data, review of relevant literature, preparation of the manuscript, and approval of the final version submitted for publication. Each author takes full responsibility for the integrity and accuracy of the work.

Declaration of Patient Consent:

The patient provided written informed consent for the publication of this case report and the related images. All reasonable measures have been taken to ensure the patient's privacy and anonymity.

Conflicts of Interest:

The authors declare that they have no competing interests related to this work.

Availability of Data and Materials:

The datasets generated and/or analyzed during this study can be obtained from the corresponding author upon reasonable request.

References

1. O'Donohue AK, Ginn SL, Burgio G, Berman Y, Dabscheck G, Schindeler A. The evolving landscape of NF gene therapy: Hurdles and opportunities. Mol Ther Nucleic Acids. 2025 Feb 3;36(1):102475. doi: 10.1016/j.omtn.2025.102475. PMID: 40034205; PMCID: PMC11872496.
2. Báez-Flores J, Rodríguez-Martín M, Lacal J. The therapeutic potential of neurofibromin signaling pathways and binding partners. Commun Biol. 2023 Apr 20;6(1):436. doi: 10.1038/s42003-023-04815-0. PMID: 37081086; PMCID: PMC10119308.
3. Legius E, Messiaen L, Wolkenstein P, Pancza P, Avery RA, Berman Y, Blakeley J, Babovic-Vuksanovic D, Cunha KS, Ferner R, Fisher MJ, Friedman JM, Gutmann DH, Kehrer-Sawatzki H, Korf BR, Mautner VF, Peltonen S, Rauen KA, Riccardi V, Schorry E, Stemmer-Rachamimov A, Stevenson DA, Tadini G, Ullrich NJ, Viskochil D, Wimmer K, Yohay K; International Consensus Group on Neurofibromatosis Diagnostic Criteria (I-NF-DC); Huson SM, Evans DG, Plotkin SR. Revised diagnostic criteria for neurofibromatosis type 1 and Legius syndrome: an international consensus recommendation. Genet Med. 2021 Aug;23(8):1506-1513. doi: 10.1038/s41436-021-01170-5. Epub 2021 May 19. PMID: 34012067; PMCID: PMC8354850.
4. Ozarslan B, Russo T, Argenziano G, Santoro C, Piccolo V. Cutaneous Findings in Neurofibromatosis Type 1. Cancers (Basel). 2021 Jan 26;13(3):463. doi: 10.3390/cancers13030463. PMID: 33530415; PMCID: PMC7865571.
5. Arrington DK, Danehy AR, Peleggi A, Proctor MR, Irons MB, Ullrich NJ. Calvarial defects and skeletal dysplasia in patients with neurofibromatosis Type 1. J Neurosurg Pediatr. 2013 Apr;11(4):410-6. doi: 10.3171/2013.1.PEDS12409. Epub 2013 Feb 15. PMID: 23414129.
6. Serindere M, Tasar M, Hamcan S, Bozlar U. Imaging Findings of Jugular Foramen Meningocele in a Neurofibromatosis Type 1 Patient. Case Rep Radiol. 2017;2017:7047696. doi: 10.1155/2017/7047696. Epub 2017 Dec 24. PMID: 29435380; PMCID: PMC5757098.
7. Siddiqui A, Connor S, Gleeson M. Jugular foramen meningocoele in a patient with neurofibromatosis type 1. J Laryngol Otol. 2008 Feb;122(2):213-6. doi: 10.1017/S0022215107007244. Epub 2007 Apr 16. PMID: 17437652.
8. Liao M, Lee HL, Liu YL, Wong TT. Neurofibromatosis Type 1 : A General Review. J Korean Neurosurg Soc. 2025 Nov;68(6):644-651. doi: 10.3340/jkns.2025.0073. Epub 2025 May 20. PMID: 40393479; PMCID: PMC12582889.
9. Almuqbil MA, Ammari MA, Al Dhayyan NS, Kaithathara ST, Al Balwi M. Clinical characteristics and healthcare burden of neurofibromatosis type 1 in Saudi Arabia: a single centre experience. Hered Cancer Clin Pract. 2025 Nov 26;23(1):27. doi:

10.1186/s13053-025-00325-8. PMID: 41299544; PMCID: PMC12659344.
10. Russo C, Russo C, Cascone D, Mazio F, Santoro C, Covelli EM, Cinalli G. Non-Oncological Neuroradiological Manifestations in NF1 and Their Clinical Implications. Cancers (Basel). 2021 Apr 12;13(8):1831. doi: 10.3390/cancers13081831. PMID: 33921292; PMCID: PMC8070534.
11. Chen N, Li W, Min L, Huang Q, Bian J. Neurofibromatosis type 1 with huge intrathoracic meningoceles misdiagnosed as pleural effusion: A case report and literature review. J Cardiothorac Surg. 2024 May 30;19(1):303. doi: 10.1186/s13019-024-02819-3. PMID: 38816890; PMCID: PMC11138057.
12. David DJ, Proudman TW. Cephaloceles: classification, pathology, and management. World J Surg. 1989 Jul-Aug;13(4):349-57. doi: 10.1007/BF01660747. PMID: 2773494.
13. Constanzo F, Coelho Neto M, Nogueira GF, Ramina R. Microsurgical Anatomy of the Jugular Foramen Applied to Surgery of Glomus Jugulare via Craniocervical Approach. Front Surg. 2020 May 15;7:27. doi: 10.3389/fsurg.2020.00027. PMID: 32500078; PMCID: PMC7243180.

Figures with Legends

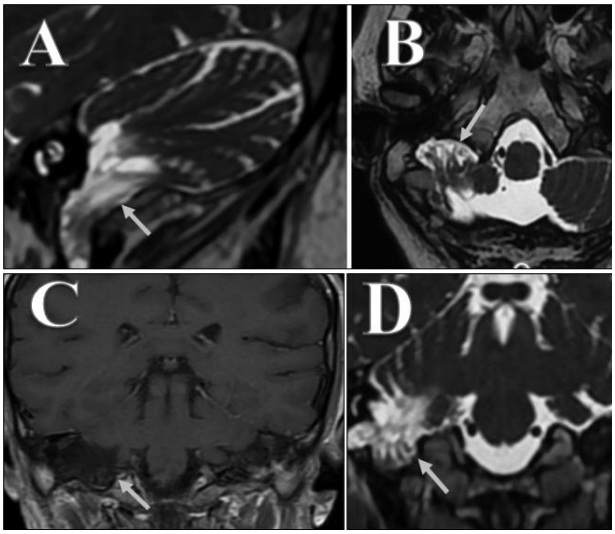

Figure 1. Sagittal (A), axial (B), and coronal (D) T2-weighted MR images demonstrate a cerebrospinal fluid (CSF)–filled pouch protruding from the anteroinferior surface of the cerebellum into the right jugular fossa (shown by arrows), with associated herniation of gliotic cerebellar tissue. The lesion measures approximately 3 × 2.5 cm. Post-contrast imaging (C) shows no abnormal enhancement. The right internal jugular vein is displaced and compressed along the posterior aspect of the lesion

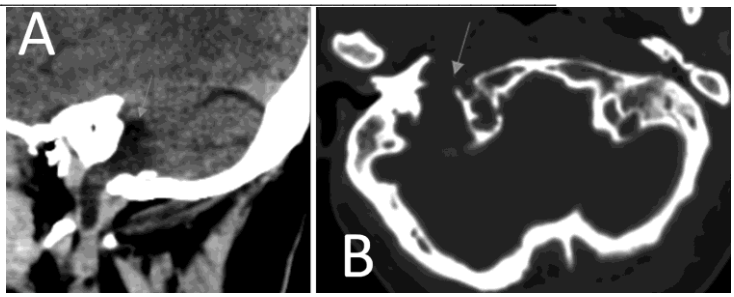

Figure 2. Complementary CT images. **(A)** Sagittal and **(B)** axial CT views demonstrate widening of the right jugular foramen compared with the contralateral side (shown by arrows), indicating bony expansion associated with the lesion.