Case Study

DURAL ECTASIA WITH BONE SCALLOPING AND CERVICAL MENINGOCELE: A PREDICTIVE SIGN OF NEUROFIBROMATOSIS TYPE 1

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ABSTRACT

Neurofibromatosis (NF) is a group of rare genetic disorders characterized by the formation of benign tumors, predominantly neurofibromas, along the peripheral and central nervous system, as well as in other mesodermal and ectodermal tissues. The main clinical forms of NF include NF type 1 (NF1), NF type 2 (NF2), and schwannomatosis, each with specific genetic and phenotypic characteristics. NF1, the most common form, has an incidence of 1 in 3,000 live births and is associated with mutations in the NF1 gene on chromosome 17, responsible for the production of neurofibromin. This protein regulates the RAS-MAPK signaling pathway. Aberrant activation of this pathway leads to uncontrolled cellular proliferation, with clinical manifestations such as café-au-lait spots, neurofibromas, optic gliomas, and skeletal dysplasias.

KEYWORDS: *neurofibromatosis, neurofibromas, NF, schwannomatosis, mutation, chromosome*

INTRODUCTION

Neurofibromatosis 2 (NF2) has an incidence of approximately 1 in 25,000 individuals and is caused by mutations in the NF2 gene on chromosome 22, responsible for merlin, a tumor suppressor protein. Loss of merlin function is associated with schwannomas and meningiomas, with a 90% prevalence of bilateral vestibular schwannomas, leading to an increased risk of deafness and postural instability (1-13). Schwannomatosis is the rarest form of the disease, characterized by the presence of schwannomas along peripheral nerves, which can cause neuropathic symptoms such as chronic pain, often associated with mutations in the SMARCB1 and LZTR1 genes (3, 11, 12).

Early diagnosis of neurofibromatosis (NF) is of fundamental importance for the management of the disease and for the mitigation of the risk of associated complications. Imaging plays a crucial role, allowing not only the monitoring of tumor growth but also the timely identification of any complications (14). When analyzing localized portions of the body, different imaging modalities should be used. Magnetic resonance imaging (MRI) is recommended for the characterization of intracranial or spinal lesions, while computed tomography (CT) represents a secondary choice. For the preliminary evaluation of skeletal lesions, the use of radiography (XR) is indicated. Furthermore, ultrasonography is

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a useful option for evaluating superficial lesions and for characterizing peripheral and intra-abdominal nerve tumors (14, 15).

In situations where it is necessary to analyze the entire body, methods such as whole-body MRI are used, which allows a global evaluation of the growth pattern and extension of the peripheral nerve sheath tumor (PNST). Positron emission tomography with fluorodeoxyglucose (FDG PET) or CT is used as a tool for differential diagnosis, proving particularly useful in the evaluation of potential malignant transformation, as in the case of malignant peripheral nerve sheath tumor (MPNST) (14, 15).

In recent years, innovative therapeutic approaches, such as MEK kinase inhibitors, have demonstrated efficacy in reducing tumor volume in patients with NF1 (16). Despite these advances, the management of NF remains complex, due to phenotypic variability and unpredictability of disease progression.

Currently, there are no standardized protocols for imaging NF. Diagnostic strategies may vary depending on the type of NF and the presence of clinical symptoms (15-17). This variability represents a significant obstacle to progress in research and technology, making NF a particularly complex disease to diagnose. It is, therefore, imperative to follow standardized protocols based on empirical evidence derived from clinical practice to improve patient management and therapeutic outcomes. The present study aims to illustrate a clinical case of neurofibromatosis type I (NF1) in which the efficacy of the diagnostic sequences employed led to an accurate diagnosis, highlighting the importance of a systematic and targeted diagnostic approach.

CASE STUDY

SD, male, born October 2005, presents to clinical attention due to neck pain; this was the reason why the doctor requested a cervical MRI, which revealed the presence of ectasia of the dura mater, that is, an enlargement of the dural sac, such as to determine posterior vertebral bone scalloping associated with herniation of the nerve root sheaths with the formation of cervical meningoceles at the C5-C7 passage on the left hemiside, (Fig 1A-C, Fig. 2A, B), this picture enters into the differential diagnosis with Marfan syndrome and Ehlers-Danlos syndrome.

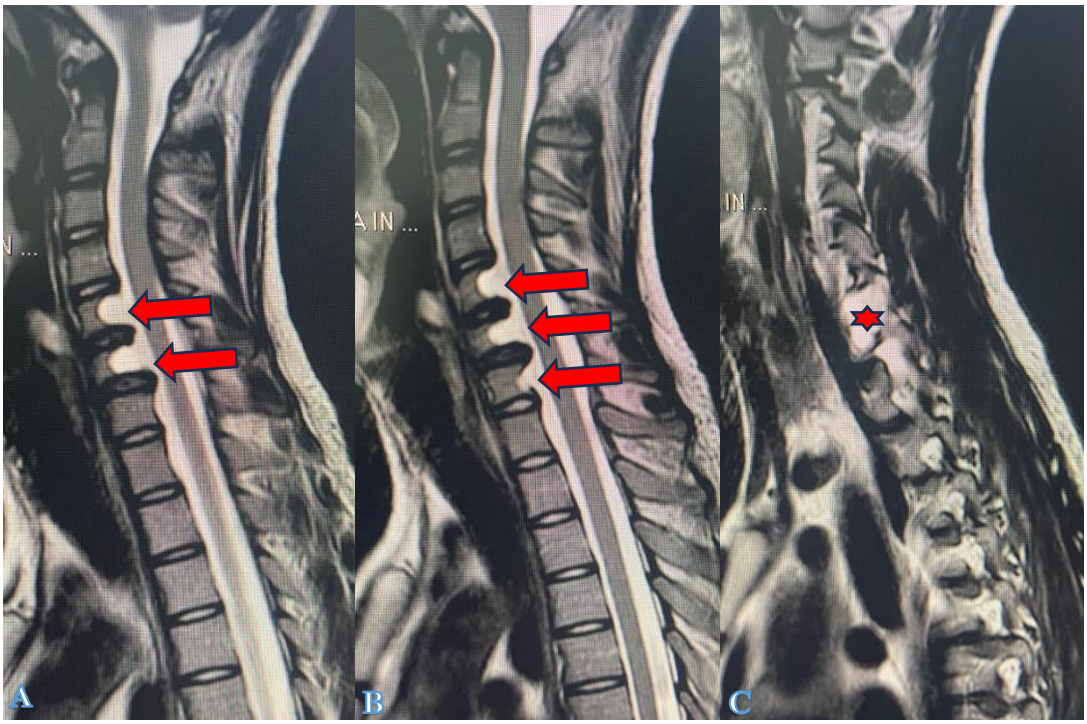

Fig. 1. ***A-C****): Sagittal MRI T2; posterior vertebral bone scalloping of C5, C6, C7 (****arrows****) and left meningocele at C6-C7 (*).*

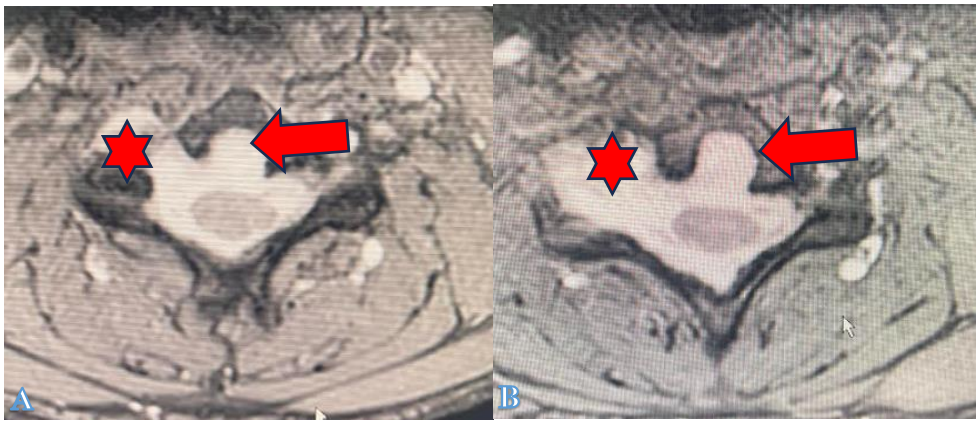

Fig. 2. ***A, B****): Axial T2 MRI; posterior vertebral bone scalloping of C5, C6, C7 (****arrows****) and left meningoceles at C5-C6 and C6-C7 (*).*

In light of these findings, to reach a diagnosis of the nature of the disease, the investigations are completed with an MRI of the brain and an ultrasound of the subcutaneous soft tissues of the lateral cervix.
Brain MRI allows us to appreciate the presence of four focal areas of altered signal intensity at the subtentorial level with cerebellar localization, compatible with UBO (unidentified bright objects). These areas appear characterized by signal hyperintensity in T2 and Flair sequences that do not exert mass effect on adjacent structures (Fig. 3A, B) and do not show enhancement after intravenous administration of contrast medium (Fig. 4).

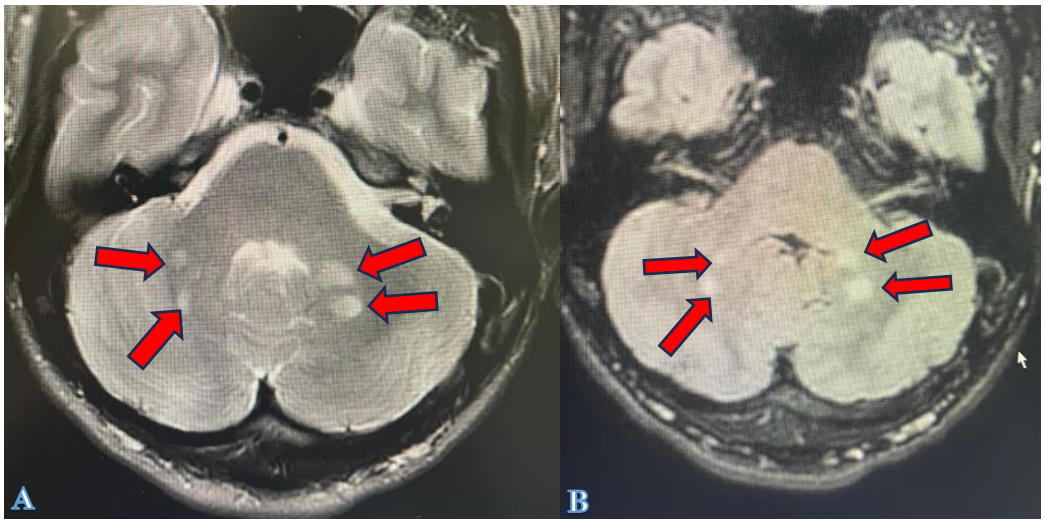

Fig. 3. ***A****): Axial T2 MRI;* ***B****): Flair; focal areas of altered signal intensity at the infratentorial level with cerebellar localization, compatible with UBO (****arrows****).*

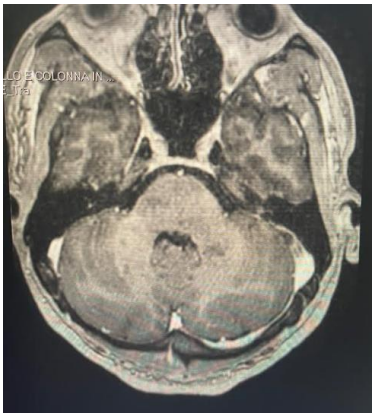

Fig. 4. *Axial MRI after intravenous contrast medium administration: there is no enhancement of the lesions.*

Therefore, by adding the results of the cervical and encephalic MRI investigations, we tend towards a possible diagnosis of NF1, which is also confirmed by the support of the ultrasound investigation of the subcutaneous soft tissues of the lateral carpi, which allows us to appreciate the presence of some small subcutaneous neurofibromas.

DISCUSSION

NF1 is a well-documented genetic condition, but its clinical presentation can vary significantly, making it possible to make late or incorrect diagnoses. In the case presented, neck pain was the only initial clinical manifestation, a symptom that is not always directly and immediately associated with NF1. This situation highlights the importance of accurate diagnosis, considering that initial symptoms may be atypical and overlap with those of other musculoskeletal or neurological pathologies. Targeted MRI revealed significant findings, such as dural ectasia and the presence of meningoceles, information that directed the diagnosis toward a more complex condition.

The finding of infratentorial UBOs in correspondence with the cerebellar hemispheres in brain MRI further supported the diagnosis of NF1, as this finding is well-known and frequently associated with this pathology. Finally, the presence of subcutaneous neurofibromas, documented by ultrasound, completed the clinical picture, highlighting the importance of performing multimodal investigations for an accurate diagnosis. The systematic approach that led to the diagnosis is in line with current recommendations for managing NF1, in which early identification of lesions and associated complications is crucial for timely therapeutic intervention.

Furthermore, the phenotypic variability in patients with NF1 requires continuous monitoring and adaptation of diagnostic and therapeutic strategies. The implementation of standardized protocols based on consolidated evidence can improve not only diagnostic accuracy but also long-term clinical outcomes.

CONCLUSIONS

The clinical case analyzed demonstrates the importance of an accurate diagnostic evaluation of NF1, highlighting how common symptoms, such as neck pain, can mask more serious conditions. The combined use of imaging techniques, such as MRI and ultrasound, has proven to be essential for the timely and accurate identification of the pathology.

This study highlights the need to develop standardized and appropriate imaging protocols that can guide clinicians and radiologists in the early diagnosis of NF1 and in managing associated complications. Early identification of visceral alterations and predisposition to develop malignancies require regular surveillance and timely therapeutic intervention.

In conclusion, a systematic and targeted diagnostic approach is essential not only to confirm the diagnosis of neurofibromatosis but also to improve the overall management of patients and to optimize long-term therapeutic outcomes. Future research should focus on the definition of practical guidelines for monitoring and treatment of NF1 based on scientific evidence and consolidated clinical experience.

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