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Intradural Extramedullary Lesions in the Cervical Spine in Neurofibromatosis

Authors' Contribution:
Study Design A
Data Collection B
Statistical Analysis C
Data Interpretation D
Manuscript Preparation E
Literature Search F
Funds Collection G

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Conflict of interest: None declared

Patient: Male, 30-year-old
Final Diagnosis: Neurofibromatosis
Symptoms: Spastic quadriparesis
Medication: —
Clinical Procedure: Cervical laminectomy • durotomy • excision of intradural extramedullary masses
Specialty: Neurosurgery

Objective: Unknown etiology

Background: Neurofibromatosis (NF) is categorized into 3 diseases: neurofibromatosis type 1, type 2, and schwannoma. NF2 is associated with a mutation in gene 22q11.2. It is present in about 1/25 000 to 33 000 births, and it is passed in an autosomal dominant fashion. Diagnosis is made based on clinical and radiological features. A few clinical features have been characterized and included in the Manchester criteria. A few neurofibromatosis type 2 patients have been diagnosed with over 25 cervical lesions. We report a case of an intradural extramedullary cervical lesion in a patient later diagnosed with neurofibromatosis type 2.

Case Report: The patient was 30-year-old man admitted through the emergency unit, presenting with gradual onset and progressive spastic quadriparesis of 6 months duration. An MRI spine showed intradural extramedullary masses in the right side of C4 and left side of C6. He underwent cervical intradural excision of 2 masses under general anesthesia with neuromonitoring. The tumor was sent to histopathology and reported as neurofibromatosis 2.

Conclusions: Neurofibromatosis is a common entity, but the diagnosis of a cervical mass is judicious to avoid any misfortune in neurological function. It requires a multidisciplinary approach and screening modalities.

Keywords: Cervical Cord • Neurofibromatosis 2 • Spinal Cord Neoplasms

Full-text PDF: https://www.amjcaserep.com/abstract/index/idArt/933090

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Background

Neurofibromatosis (NF) is divided into 3 diseases: neurofibromatosis type 1, type 2, and schwannoma [1]. NF2 is found about 1/25 000 to 1/33 000 births. It is associated with with a mutation in gene 22q11.2, and it is passed in an autosomal dominant fashion [2,3]. The average age of presentation is in the mid-twenties, with no sex or ethnic predilection, and an average delay in diagnosis of 7 years [4]. There has been a rise in discovery, with a prevalence of 1/100 000 in 2005 and screening by advancements in technology, which leads to earlier treatment [5].

Diagnosis is made based on clinical and radiological features. A few clinical features have been characterized and included in the Manchester criteria, which incorporate one-sided/two-sided vestibular schwannoma, family history (one-sided vestibular schwannoma or 2 NF2 lesions incorporating meningioma, glioma, neurofibroma, schwannoma, and cataract), and numerous meningiomas [5]. A comparative algorithm created through the NIH incorporates reciprocal vestibular schwannomas with numerous meningiomas, optic gliomas, cranial nerves tumors, and spinal tumors. The presence of bilateral vestibular schwannoma before the age of 30 years with first-degree family history, or unilateral vestibular schwannoma less than 30 years with 2 of meningioma, glioma, schwannoma, and posterior subcapsular cataract can confirm the diagnosis [4].

A few neurofibromatosis type 2 patients diagnosed with cervical lesions have had over 25 lesions [6-12]. The most recent case reports, in 2016 and 2019, mention the main presentation was cervical pain (C1-2 lesion) [8] and lower limb weakness (C4-5 lesion) [10]. The patients underwent decompression (suboccipital for upper cervical lesion and laminectomy for middle cervical lesion), and neither had any deficits postoperatively [8,10]. Here, we report the case of an intradural extramedullary cervical lesion in a patient later diagnosed with neurofibromatosis type 2.

Case Report

The patient was 30-year-old man admitted through the emergency unit, presenting with gradual onset and progressive course of spastic quadriparesis of 6 months duration. He started to have progressive weakness in his lower limbs until he could no longer walk and required a wheelchair, and upon further questioning, there was no history of trauma, infection, or malignancy. He had no incontinence or urine leakage, was voiding freely, and had no history of febrile UTI.

Upon admission, the Glasgow Coma Score (GCS) was 15/15, he was vitally stable, and pupils were equal and reactive. He could move his upper limbs but had limitations in fingers, with overall power 3/5. Upper limb power was elbow flexion 3/5, wrist extension 3/5, elbow extension 2/5, middle finger flexion 3/5, and little finger abduction 2/5. He had lower limb weakness with overall power 2/5 with spasticity. Lower limb power was hip flexion 2/5, hip extension 1/5, knee flexion 2/5, knee extension 2/5, plantar flexion 2/5, plantar dorsiflexion 1/5. The reflexes were +2 in triceps and +3 in biceps and brachioradialis, +1 in lower limbs (knee and ankle). There were no sensory deficits or urinary incontinence, only occasional urgency. There was +ve Hoffman sign. He was not able to walk, but was mobilized with help of another person and self-rolling in bed. Urology was consulted, and KUB and US renal results were normal.

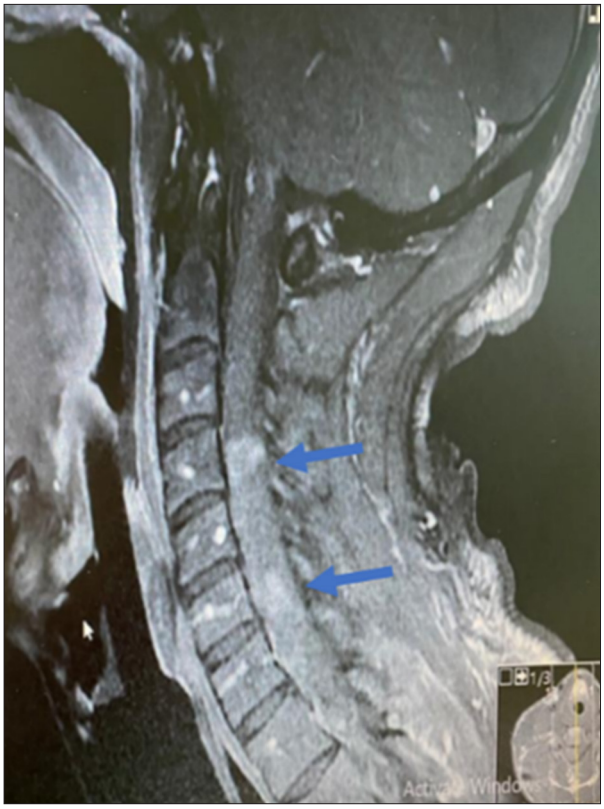

Figure 1. Preoperatively, showing 2 cervical masses in C4 and C6 (arrows).

Lab values upon admission were PT 17.3 (normal value 10-12), INR 1.48 (normal value 0.9-1.2), PTT 62.9 (normal value 25-36), Hb 15.2g/dl (normal value 13-17), RBC 5.38* 10^{12} (normal value 4.5-5.5×10^{12}), WBC 4.7×10^{9} (normal value 4-10×10^{9}), and PLT 263×10^{9}L (normal value 150-410), and his cardiac profile, RFT, and LFT were normal. On admission, he was started on prophylactic enoxaparin, pain medications (ibuprofen, paracetamol), baclofen 10 mg TID, and dexamethasone 4 mg QID with gastric protection. MRI spine showed intradural extramedullary masses in the right side of C4 and left side of C6 (**Figure 1**).

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He underwent cervical intradural excision of 2 masses under general anesthesia and was given 2 g preoperative ceftriaxone. Cervical laminectomy, durotomy, and excision were done, followed by a midline nuchal skin incision from the posterior occipital protuberance to the spinous process of D2. The patient was placed in a prone position. Subperiosteal muscle separation was made from C2 to D1, laminectomy of C3, C4, C5, C6, and C7, midline durotomy, starting intraoperative neurophysiological monitoring by the motor evoked potential, which showed no traces of any receptors in the lower limbs. We performed dissection of the right-sided mass at the level of C6 and total excision with its attached posterior ramus, as well as dissection of the left-sided mass at the level of C4 and total excision with its attached posterior ramus. Hemostasis was achieved, and postprocedural motor evoked potential confirmed good response in both triceps muscles and starting weak response in the lower limbs. Closure of anatomical layers was done, and dural suturing showed tightening of the cord, so it is closed by a layer of gel foam and a TachoSil™ fibrin sealing patch. Then, a subcutaneous drain was attached; blood loss was 300 cc, and the procedural time was 4 h. The tumor was sent to histopathology and reported as neurofibromatosis 2. He tolerated the procedure well. He was kept in the ICU for observation. The drain was 125 cc on positive suction and was removed 2 days after the operation. He was shifted to the floor 2 days after the operation, strength significantly improved in lower limbs 3+-4/5, and he received extensive physiotherapy. MRI brain was done and was normal.

Discussion

Patients usually present with deafness, hearing loss, and tinnitus if there was vestibular schwannoma, and in late stages there may be signs of increased intracranial pressure. The other presentation is compression of nerves and spinal cord, and the tumors are usually low-grade gliomas. Sometimes, patients present with reduced vision and cataracts, and more than 2/3 present with skin manifestations of plaque formation [13].

All patients should be screened with MRI brain thin cuts every 1-2 years if they are under age 20 years and every 3-5 years if older than 20 years to assess vestibular schwannoma involvement. Also, a baseline spinal MRI is needed for staging [14] and every 3 years, as spinal tumors are seen in around 60-80% of NF2 patients [15]. Studies have shown that early detection and treatment of spinal lesions can decrease morbidity [14], as 30% need surgery. In spinal tumors, intramedullary tumors are usually detected with the involvement of the syrinx, predominantly in the upper cervical spine and brainstem [16]. Visual assessment screening starting from the age of 10 years is needed for early detection of cataract, and audiological assessment should include auditory brainstem response [15].

The most common tumors are schwannomas and meningiomas, as indicated in a review by Evan in 2015. Schwannomas are detected mostly around nerves and the cerebellopontine angle, and are classified by Antoni classification. They can stain positive for S-100 and vimentin. They are usually benign; malignancy has been reported to increase 10-fold with radiation, but the risk is less in NF2 compared to NF1. Meningioma of various types (meningothelial, fibroblastic, transitional) are the second most common tumors in NF2, with a tendency to occur supratentorial in falx and lobes, except for the occipital lobe, and in the spinal cord, which can be surgically challenging to excise [17].

Treatment is usually a multidisciplinary approach involving radiotherapy, surgery, and rehabilitation. Surgery is complex and can be combined with neurosurgery along with otolaryngology. Reported surgeries include decompressive laminectomy and radiosurgery in 2 patients with pathology of ependymoma grade 3 with a mean lesion size of 6 mm (range 2-20 mm). Patients with central lesions, small lesions <1 cm, no cord signals, and homogenous lesions usually have a good prognosis [6]. Lim et al [7] reported performing a procedure beginning with a midline skin incision in the dorsal surface, followed by laminectomy, with visualization of an avascular soft grayish mass. A frozen section showed a high-grade glioma, so subtotal resection was performed, followed by confirmatory histopathology showing ependymoma, and the patient underwent total resection of the mass. In a case report in 2016, a patient presented with recurrent ependymoma located in C1-2; despite multiple decompressions and laminectomy, the patient presented with signs of increased intracranial pressure, so suboccipital craniotomy was done [8]. In a case report in 2019, the authors performed cervical laminectomy and fusion with miniplates and screws in a C4-5 cervical lesion and the patient had no postoperative deficits [10]. A retrospective series of nearly 180 patients found total gross resection was performed in nearly 2/3 of patients, with postoperative complications in 2 patients, including cerebrospinal leak and wound infection, with no mortalities [9]. Radiotherapy is a controversial treatment, but is utilized in aggressive disease with multifocal intracranial tumors of less than 3 cm, and in poor surgical candidates. However, the risk must be explained to patients, as about 70% have subsequent hearing loss [18].

Conclusions

Neurofibromatosis is a common entity, but the diagnosis of cervical mass is judicious to avoid any problems in neurological function. It needs a multidisciplinary approach and screening modalities.

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Conflict of Interest

None declared.

Declaration of Figures Authenticity

All figures submitted have been created by the authors who confirm that the images are original with no duplication and have not been previously published in whole or in part.

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