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A Rare Case Of C1-2 Unilateral Dumbbell Ganglioneuroma In A Patient With Neurofibromatosis Type-1

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Abstract

Background

The autonomic nervous system's ganglion cells give rise to an uncommon and benign tumour known as a ganglioneuroma (GN). The most frequently affected locations are the retroperitoneal space, the abdominal cavity, and the posterior mediastinum. When it occurs in the cervical area, the spinal cord is rarely squeezed. About 1 in 3000 people have neurofibromatosis type 1 (NF-1), an autosomal dominant genetic condition.

Case Summary

We present a fairly unusual instance of unilateral dumbbell GN of the cervical spine with NF-1. A 25-year-old NF-1 patient was examined who had been suffering right lower limb weakness that had been getting worse over the previous six months. The spinal cord was compressed by a left-sided dumbbell lesion at C1-C2 levels, according to magnetic resonance imaging. We completely removed the lesion, and the excised mass's postoperative histological diagnosis was GN. Following surgery, the preoperative symptoms progressively subsided without any negative effects. This is the eighth report of unilateral dumbbell GNs that we are aware of.

Conclusion

Dumbbell GNs may occasionally be linked to NF-1. Before surgery, it is impossible to make a precise diagnosis; the current gold standard is the outcome of the histopathology. The most effective method is surgical resection, and disease outcomes are typically positive following treatment.

Core Tip:

The primary mature form of neuroblastic tumours, known as ganglioneuroma, primarily affects the posterior mediastinum and the retroperitoneal area. We describe a rare instance of type 1 neurofibromatosis in conjunction with a unilateral dumbbell ganglioneuroma. Only seven cases have been documented in the literature thus far.

Prior to postoperative pathology, a definitive diagnosis is difficult due to the non-specificity of symptoms, signs, and radiological imaging. This tumour has a positive prognosis. The most effective course of action is complete resection. Furthermore, if tumour residuals are less than 2 cm, partial resection does not raise the risk of progression.

Keywords: NIL

Introduction

60% of ganglioneuromas (GN) in children and young adults are rare, slow-growing, fully- differentiated tumors that arise from sympathetic ganglia or adrenal medulla neural crest cells [1-3]. This benign tumor may develop spontaneously, as a result of

neuroblastoma maturation (spontaneous remission phenomenon), or as a result of chemotherapy or radiation treatment [4]. It consists of mature Schwann cells, ganglion cells, @ibrous tissue, and nerve @ibers histologically [5].

The most frequent anatomical sites are the posterior mediastinum (41.5%), abdominal cavity with adrenal glands (21%), and retroperitoneal space (37.5%) [6]. The trigeminal GN, as described by Deng et al. [7], as well as the external auditory canal and middle ear GN, as described by Almofada et al. [8], are examples of sympathetic nerve cells that can be found anywhere.

On rare occasions, it develops into the spinal canal and severely compresses the spinal column. GN is typically unidirectional and lonesome. Here, we describe an incredibly unusual instance of neuro@ibromatosis type 1 and left-sided dumbbell GN of the cervical spine (NF-1). Von Recklinghausen illness, commonly known as NF-1, typically manifests during the first ten years of life..

The National Institute of Health developed the NF-1 diagnostic standards[9]. Neuro@ibroma is the most prevalent benign tumour in NF-1[10,11]. As far as we are aware, there have only been seven cases of dumbbell GNs of the cervical spine published in the English literature, including two that were linked to NF-1[1,3,12,13]. The objective of this report is to present a second case and assess its clinical features, diagnostic methods, surgical alternatives, and prognosis.

Case Presentation

A 25-year-old lady who had been experiencing progressively worsening right lower limb paralysis for six months was referred to the department of neurosurgery. Her parents had noticed her walking erratically and spinning around a month earlier. Several coffee spots, cutaneous, and subcutaneous neurofibromas were discovered during a physical examination. No one in his family had neurofibromatosis.

A neurologic evaluation gave the right lower limb's muscular strength a grade of 4. Spasticity of the right lower limb muscles and hyperactivity of the deep reflexes were both present.

We discovered a sizable dumbbell tumour at the C1-C2 level on cervical magnetic resonance imaging (MRI), which showed low signal intensity on T1-weighted pictures, somewhat high signal intensity on T2-weighted images, and significant contrast enhancement. Compression caused the spinal cord signal intensity to rise at the C2 level (Figure 1).

The patient underwent surgical decompression of the spinal cord in the prone position through posterior midline incision. Muscles we're separated sub periosteally on left side of spine and lamina of C1 and C2. Tumor was extending from intra spinal region to extra spinal region between C1 and C2. The tumor was @irm and avascular. The tumor was then dissected all around, end was found to be arising from left C2 root, angulated. The tumor involved nerve root was excised along with the tumor and the cut ends of the root were anastomosed end to end. Complete tumor resection was done. The dura was found to be intact and there was no evidence of intra dural extension.

Under microscopy, section shows an un encapsulated lesion composed of scattered mature ganglion cells with compact eosinophilic granular cytoplasm with distinct cell border and single eccentric nucleus. Few of them show @ine granular golden brown pigment. Background shows intersecting fascicles of Schwann cells separated by myxoid stroma. No atypia/mitosis or necrosis noted in the sections studied. Spindle-shaped and essentially the same size, tumour cells. Fusiform nuclei and ganglion cells were dispersed throughout the tumour tissue.

S-100 (+), vimentin (+), NF (Scattered+), NeuN (Scattered+), and the Ki67 labelling index of 3% were all detected by immunohistochemistry (Figure 1,2). The mass was identified as GN based on the aforementioned measurements.

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FIG 1: IHC S-100 +

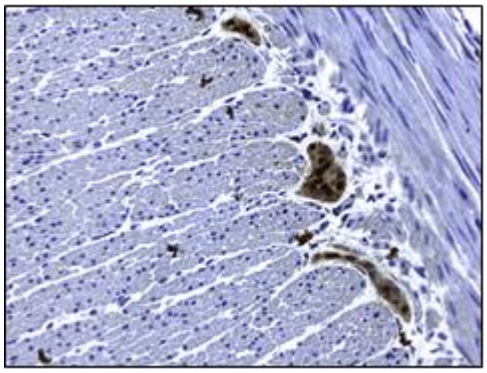

FIG 2: IHC VIMENTIN +

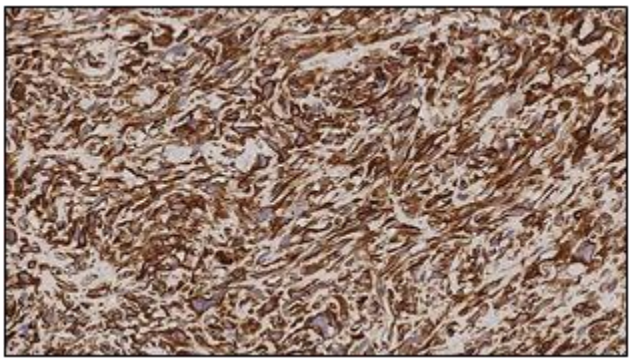

Table 1 Summary of previously reported cases with cervical bilateral dumbbell ganglioneuromas

Ref.	Age/Sex	Extension	Site	Treatment	NF-1	Presentation	Prognosis	Follow-up imaging
Hioki *et al*[12]	72/male	Intradural/extradural	C1-2	PR	-	Tetraparesis; sensory dysfunction	Symptom remission	No recurrence or increase after 2 yr
Kyoshima *et al*[3]	35/male	Intradural/extradural	C2-3	PR	+	Tetraparesis; respiratory dysfunction	Hypesthesia	None
Miyakoshi *et al*[2]	15/male	Extradural	C1-2; C3-4	PR	+	Tetraparesis; sensory dysfunction	Symptom remission	No recurrence or increase after 2 yr
Ugarriza *et al*[1]	53/male	Extradural	C2	TR	-	Tetraparesis; respiratory dysfunction; sensory dysfunction	Tetraparesis	None
Ando Hioki *et al*[13]	20/male	Intradural/extradural	C1-2; C2-3; C3-4	PR	-	Tetraparesis; sensory dysfunction	Symptom remission	No recurrence or increase after 2 yr

PR: Partial resection; TR: Total resection.

Table
Summary of previously published case reports with cervical ganglioneuroma producing spinal cord compression

Authors	Age/sex	Site of origin	Dumbbell shape	Intraspinal extension	Multiplicity	NF-1
Shephard and Sutton [7]	35/M	Unilateral C2–C7	+	Intradural	+	+
Sinclair and Yang [8]	44/F	Unilateral C2–C5	+	Intradural	+	+
Strang and Nordenstam [9]	63/F	Unilateral C2–C4	+	Intradural	–	–
Maggi et al. [10]	1.5/F	Unilateral C2–C6	+	Extradural	–	–
Ugarriza et al. [11]	53/M	Bilateral C2 (symmetric)	+	Extradural	+	–
Kyoshima et al. [5]	35/M	Bilateral C2 and C3 (symmetric)	+	Intradural	+	+
Bhand [2]	22/F	Unilateral C3	+	Extradural	–	–
Radulovi et al. [12]	39/M	Unilateral C5	+	Extradural	–	–
Tei et al. [13]	51/F	Unilateral C1	–	Intradural	–	–
Present case	15/M	Bilateral C2 (symmetric), unilateral C4	+	Extradural	+	+

M, male; F, female; NF-1, neurofibromatosis type 1.

Final Diagnosis

Left sided dumbbell GN of the cervical spine associated with NF-1.

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Treatment

In the prone position, the patient underwent posterior midline incision spinal cord decompression surgery.

Outcome And Follow-Up

After surgery, there were no complications with the nervous system. Within two weeks, the patient's preoperative symptoms started to subside. A month later, the patient visited our hospital with a postoperative MRI scan revealing complete removal of both tumours without any indication of recurrence (Figure 3).

Discussion

GN was initially described in 1870[14]. Since then, an increasing number of pertinent case reports with in-depth details have been released. Although research suggested that it might account for 0.1 to 0.5% of central nervous system tumors[15], GN incidence is still not completely understood. In the cervical region, GN rarely compresses the spinal cord. However, cervical dumbbell GNs with NF-1 are much less prevalent.

There have only been seven examples reported in the most recent English literature (Table 1). These examples included the current one, which also involved the extradural space, and three that demonstrated intra dural extension and two that demonstrated extradural growth. However, extradural extension from the intervertebral foramen is more typical in the majority of cases, whether multiple or single, given that these tumors arise from sensory root ganglions. The cases summarized here were all male, notwithstanding Stout et al's[16] claim that GN has a roughly 2/3 male to female ratio, and NF-1 is not gender- or ethnicity- speci@ic[17]. We don't know if there are gender differences in GN. According to Weiss, GN is most usually diagnosed in patients between the ages of 10 and 29 years old[18].

In addition, NF-1 is typically present within the @irst decade of life, and patients with NF-1 have malignancies up to four to six times more frequently than people without NF-1[9]. As a result, it appears that GN connected to NF-1 will be diagnosed sooner. Depending on where it is, the tumor might either be hardly symptomatic or completely asymptomatic. Only 10% of GNs may involve the spinal canal, according to prior reports[19]. A number of compressive symptoms could appear as a result of tumor growth in the spinal canal. The most prevalent symptom in the six cases was tetra paresis, which frequently manifested earlier and was brought on by bilateral tumors pressing against the spinal cord. Additionally, sensory impairment might be present. GN is still dif@icult to diagnose before surgery due to its non-speci@ic features.

MRI is a relatively precise diagnostic method for preoperative identification. The most common characteristics of this tumour include a clearly defined boundary, encapsulation, homogenous low or intermediate signal intensity in T1-weighted imaging, and heterogeneous intermediate or high signal intensity in T2-weighted images. It also exhibits a slight, uneven contrast enhancement. The reporting of MRI signals in various GN papers varies, nevertheless. Diffusion-weighted MRI may assist discriminate between benign and malignant paraspinal peripheral nerve sheath tumours, according to recent research. The apparent diffusion coefficient value of schwannomas is higher than that of neuro@ibromas because schwannomas are more cellular malignancies [22].

GN is classified as a category of neuroblastic tumours by the International Neuroblastoma Pathology Classi@ication[23], which can range from immature, undifferentiated to mature, differentiated tumours. Despite the lesion's benign origin, it has progressed, late relapsed, and even turned malignant been recorded [24,25]. Therefore, ongoing radiological monitoring is crucial. The most straightforward and ef@icient course of action for such lesions is presently resection. All seven of the recorded cases underwent surgery; of these, two, including the current one, achieved full excision. No patients experienced problems. In every case, the postoperative quality of life was superior to the prior one. Large residual tumors that are advancing locally may include immature components.

Therefore, it seems particularly crucial to figure out how to guarantee that immature components are eliminated during the initial operation. Recently, biopsy has been used to diagnose GN, however it is not always accurate and can even be deceptive before surgery. It would be worthwhile to research whether

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it could improve a patient's prognosis following surgery.

Conclusion

To summarize, cervical dumbbell GNs that compress the spinal cord and result in symptoms are incredibly uncommon. Such malignancies may occasionally be linked to NF-1. The pathological outcome is the current gold standard because the precise diagnosis cannot be determined prior to surgery. The most effective method is surgical resection, and disease outcomes are typically positive following treatment.

In our situation, the patient got a complete resection. Our research shows that this tumour has an excellent prognosis following surgery.

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