CLINICAL MANIFESTATION OF CERVICAL NEUROFIBROMA TYPE 1 IN PATIENTS OF A TERTIARY CARE HOSPITAL

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ABSTRACT

Objective: *To determine the clinical presentation of cervical neurofibroma type 1 (NF 1) in patients of a tertiary care hospital.*

Methodology: *This descriptive study was conducted at Neurosurgery department, Lady Reading Hospital Peshawar from February 2001 to July 2011. A total 31 patients with symptomatic cervical spine neurofibromas who underwent surgical decompression and tumor resection were included in this study. Both gender (male and female) and patients in the age range of 20-70 years were included in this study. The patients' demographic details and clinical manifestation were entered into a semi structured proforma. Data was analyzed through statistical program SPSS version 11.*

Results: *Out of 31 patients, there were 17(54.8%) males and 14(45.2%) females. The age of patients ranged from 20 to 70 years. In this study the overall mean age was 32.38 years. Majority of patients fifteen (48.38%) were in the age range of 31-40 years. Most common clinical presentation of patient was quadrapresis in seventeen (54.8%) patients.*

Conclusions: *Cervical neurofibroma type 1 commonly occurred in the third decade of life. quadriparesis was the common clinical presentation of cervical cord neurofibroma type 1.*

Key Words: *Cervical neurofibroma type 1, Surgical decompression, Tumor resection*

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INTRODUCTION

Nerve sheeth tumors together with meningioma accounts for ninety percent intradural extra medullary tumors[1]. Neurofibromatosis (NF) is a multisystem genetic disorder that commonly is associated with cutaneous, neurologic, and orthopedic manifestations. It is the most frequent of the so-called hamartoses[1,2]. In many surgical series spinal neurofibromas and schwannomas have been grouped together under headings such as neurinoma or neurofibroma. It has recently been shown that neurofibromatosis 1 (NF1) is associated with a defect in the neurofibromin gene on chromosome 17 and NF2 with a defect in the merlin/ schwannomin gene on chromosome 22. The spinal nerve root tumors in NF1 are typically neurofibromas, those in NF2 are schwannomas[3,4].

The estimated incidence of NF1 is 1 in 3000. Neurofibroma shows no sex predilection. They are in contrast to meningiomas ,are evenly distributed along the spinal axis. NF type 1 (NF1) is differentiated from central NF or NF type 2 in which patients demonstrate a relative paucity of cutaneous findings but have a high incidence of meningiomas and acoustic neuromas. NF1 has a better prognosis with a lower incidence of CNS tumors than NF2. However, morbidity and mortality rates in NF1 are not negligible[5,6]. As the tumor arise from the nerve root, chronic radicular pain is the most common presentation. NF 1 arising from conus madularis may present with bowel and bladder dysfunction. There can be

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sexual dysfunctions for months to years. Change in clinical presentation may be observed especially NF 1 in the cauda aquina region. These are called mobile neurofibromatosis. Some of the more severe complications are visual loss secondary to optic nerve gliomas, spinal cord tumors, scoliosis, vascular lesions, and long-bone abnormalities, which sometimes necessitate amputation[7,8].

The purpose of our study was to determine the clinical presentation of cervical neurofibroma type 1.

METHODOLOGY

This was a retrospective review of 31 cases involving NF-1 patients with symptomatic cervical spine neurofibromas who underwent surgical decompression and tumor resection from February 2001 to July 2011 at Lady Reading Hospital Peshawar.

Clinical diagnosis required the presence of at least 2 of 7 criteria to confirm the presence of neurofibromatosis, type 1. The 7 clinical criteria used to diagnose NF1 are six or more café-au-lait spots, Axillary or inguinal freckles, two or more typical neurofibromas or one plexiform neurofibroma, Optic nerve glioma, two or more iris hamartomas (Lisch nodules), Sphenoid dysplasia or typical long-bone abnormalities such as pseudarthrosis and first-degree relative with NF1 patients.

The patients' demographic details and clinical manifestation were entered into a semi structured proforma.

Data was analyzed through statistical program SPSS version 11. All the qualitative variables like gender, quadrapresis, paraparesis, neck pain and urinary incontinence were analyzed for percentages and frequencies. Mean $\pm$ standard deviation was calculated for quantitative variable like age. The results were presented through tables.

RESULTS

Out of 31 patients, there were 17(54.8%) males and 14(45.2%) females. The age of patients ranged from 20 to 70 years. In this study the mean age was 32.38$\pm$3.73 years. Fifteen (48.4%)patients were in the age range of 31-40 years (Table 1).

Most common clinical presentation of patient was quadrapresis in seventeen (54.8%) patients fallowed by paraparesis and radicular pain. Detailed clinical features are shown in Table 2.

DISCUSSION

Spinal neurofibroma is pathologically benign but neurologically a devastating tumour. The insidious nature of growth of these tumors and varied clinical presentation result in a delay in diagnosis[5,7,9]. As ignorance and poverty prevails in our country, these patients initially visit quacks, which result in further delay in diagnosis. Sometimes the neurological deficit may be irreversible, when they reach a neurosurgical department.

In our study age range was from 20 to 70 years. The mean age of presentation was 32.38 years. Majority of patients 48.4% were presented

Table 1: Age distribution

S. No	Age group	Number of patients	Percentages
1	20-30	7	22.6
2	31-40	15	48.4
3	41-50	5	16.2
4	51-60	3	9.7
5	61-70	1	3.3

Table 2: Clinical presentation

S. No	Clinical presentation	Number of patients	Percentages
1	Quadriparesis	17	54.8
2	Paraparesis	5	16.2
3	Neck pain radiating to upper limb	4	12.9
4	Urinary incontinence	3	9.7
5	Neck pain	2	6.4

in age group of 31 to 40 years. A study was conducted by Taleb FS et al showed mean age of 42.5 years[10]. Ma J et al conducted study in which mean age was 19 years[11]. Leonard JR et al showed the mean age of 29.9 years[12].

In this study majority of male 17 (54.8%) presented with cervical neurofibroma type 1 compared to the females 14(45.2%) with a male predominance. Taleb FS et al[10] showed male predominance. Ma J et al also reported male predominance in his study[11]. Scheithauer BW et al also documented male predominance[13].

It is observed that in various international studies more women are diagnosed with neurofibromas. Seppälä MT et al conducted study in which 13 were women and 9 were men[14]. Another group of patient showed that the patient was 69.2% female and 30.7% male[12]. Most probable reasons for this difference is that our females are shy and hesitant to take the medical consultation for their health related problems so lesser number of patient presenting to doctor for treatment. There might be some genetic differences responsible for this gender distribution.

Clinical presentation of the patients in our study were quadrapresis in seventeen (54.8%) patients, paraparesis in five (16.2%) patients, neck pain radiating to upper limb in four (12.9%) patients, urinary incontinence in three (9.7%) patients and neck pain in two (6.4%) patients. This means that the common clinical presentation was quadrapresis. Two of our female patients with neurofibroma-1 died suddenly. The cause of death in both of them could not be ascertained. Leonard et al also documented the same results of presenting symptoms like progressive quadriparesis (seven patients), paraparesis involving the lower extremities (two patients) or upper extremities (one patient), incontinence (one patient) and neck pain (three patients). One patient presented with a sensory level at C2 and one individual experienced tingling without neurological deficit[12]. Seppälä MT et al reported that quadriparesis is the common clinical presentation of cervical neurofibroma type 1[14]. The study of Taleb FS et al showed the same result[10].

CONCLUSION

Cervical neurofibroma type 1 commonly occurred in the third decade of life and quadriparesis was the common clinical presentation of cervical cord neurofibroma type 1.

REFERENCES

1. Ng ES, Ong CK, Wilder-Smith E. Neurological picture. Traumatic upper limb weakness in a man with type 1 neurofibromatosis. J Neurol Neurosurg Psychiatry 2011;82:883-4.
2. Higa G, Pacanowski JP , Jeck DT, Goshima KR, León LR .Vertebral artery aneurysms and cervical arteriovenous fistulae in patients with neurofibromatosis 1. Vascular 2010;18:166-77.
3. Buckley M, Kuan S, Barton D. An unusual cause of collapse and neck pain. Emerg Med J 2008;25:857-8.
4. Tsuji M, Harada S, Ueno Y, Osaka N. Neurological pictures. Familial spinal neurofibromatosis: three generations of identical level symptomatic tumours. J Neurol Neurosurg Psychiatry 2010;81:1382.
5. Cheong JH, Kim CH, Kim JM, Oh YH. Transformation of intracranial anaplastic astrocytoma associated with neurofibromatosis type I into gliosarcoma: case report. Clin Neurol Neurosurg 2010;112:701-6.
6. Charfeddine I, Mnejja M, Hammami B, Hasnaoui M, Hadj KA, Frikha I, et al. Neurofibromatosis type 1 revealed by malignant peripheral nerve sheath tumor. Rev Laryngol Otol Rhinol (Bord) 2009;130:327-30.
7. Sarica FB, Cekinmez M, Tufan K, Erdoğan B, Sen O, Altinörs MN. A rare case of massive NF1 with invasion of entire spinal axis by neurofibromas: case report. Turk Neurosurg 2008;18:99-106.
8. Kelleher MO, Quraishi NA, Tan G, Guha A, Massicotte EM. Intermittent atlantoaxial subluxation caused by a prolapsing neurofibroma. Case report. J Neurosurg Spine 2008;8:288-91.
9. Tonogai I, Sakai T, Katoh S, Higashino K, Sairyo K, Hirohashi N, et al. Myelopathy in a 6-year-old girl caused by neurofibromatosis Type 1: a case report. Spine J 2008;8:836-40.
10. Taleb FS, Guha A, Arnold PM, Fehlings MG, Massicotte EM. Surgical management of cervical spine manifestations of neurofibromatosis Type 1: long-term clinical and radiological follow-up in 22 cases. J Neurosurg Spine 2011;14:356-66.
11. Ma J, Wu Z, Yang X, Xiao J. Surgical treatment of severe cervical dystrophic kyphosis due to neurofibromatosis Type 1: a review of 8 cases. J Neurosurg Spine 2011;14:93-8.
12. Leonard JR, Ferner RE, Thomas N, Gutmann DH. Cervical cord compression from plexiform neurofibromas in neurofibromatosis 1. J Neurol Neurosurg Psychiatry 2007;78:1404-6.

13. Scheithauer BW, Erdogan S, Rodriguez FJ, Burger PC, Woodruff JM, Kros JM et al. Malignant peripheral nerve sheath tumors of cranial nerves and intracranial contents: a clinicopathologic study of 17 cases. Am J Surg Pathol 2009;33:325-38.

14. Seppälä MT, Haltia MJ, Sankila RJ, Jääskeläinen JE, Heiskanen O. Long-term outcome after removal of spinal neurofibroma. J Neurosurg 1995 ;82:572-7.

CONTRIBUTORS

MS conceived the idea and planned the study. KK, RUR, MIH, MS & A did the data collection, analyzed the study and helped in writing the manuscript. All the authors contributed significantly to the research that resulted in the submitted manuscript.