Neurofibromatosis type two with associated spinal schwannomas

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Abstract: Neurofibromatosis (NF) is a rare genetic disorder characterized by the development of benign tumors in the nervous system. This pathology is classified into neurofibromatosis type one (NF1) and neurofibromatosis type two (NF2) [1]. Schwannomatosis is a newly recognized third form of NF. It is characterized by multiple schwannomas, without vestibular schwannomas which are diagnostic of NF2. This case report describes the role of imaging and the management of a patient diagnosed as NF2 who later developed multiple spinal schwannomas [1-3]. NF2 occurs as a result of genetic defects caused by a mutation on a gene located on chromosome number 22 [1]. It is characterized by multiple schwannomas, meningiomas, and ependymomas. Multiple cranial nerve schwannomas, as well as bilateral acoustic neuromas or bilateral vestibular schwannomas, are the hallmark of the disease. Tumors of the brain and spine eventually occur in most patients with NF2.

Keywords: genetic disorder, vestibular schwannomas.

Case report
A 37 year old female was diagnosed with neurofibromatosis type two (NF2) in 2004 when she presented with bilateral acoustic neuromas. The left acoustic neuroma was surgically removed and then treated with stereotactic radiotherapy. She also underwent stereotactic radiotherapy treatment for a right acoustic neuroma. She developed left cranial nerve VII palsy and bilateral cranial nerve VIII palsy causing loss of hearing. Due to loss of hearing the patient learned to lip read.

As this patient had been diagnosed as NF2 she underwent magnetic resonance imaging scans to monitor her condition. In 2007 she presented with pain and progressive weakness of her lower limbs, back pain and she was developing spasticity. She had multiple schwannomas related to almost every cranial nerve intracranially, as well as widespread spinal schwannomas. She had slight scoliosis to the right. She also had multiple tiny schwannomas involving her entire cauda equina. She experienced pain in the left eye with no left eyelid closure.

She was referred for a magnetic resonance imaging scan of the spine. This scan revealed a large T11/12 schwannoma dumbelling out through the neural foramina. Compared to her previous scans this schwannoma had substantially increased in size and was causing displacement of the spinal cord (Figure 1). A large cervical schwannoma at C1/2 level was also thought to be associated with significant spinal cord compression but did not appear to have changed in size since the previous scan (Figure 2).

The patient was referred to a neurosurgeon for surgical removal of the large thoracic schwannoma. Surgical resection of the cervical schwannoma was contraindicated due to the risks involved regarding the extensive nerve supply in that area and since no significant size increase had occurred.

The patient underwent a T10-12 laminectomy and tumor resection. The surgery was successful and the patient was then transferred to the intensive care unit and kept overnight so that rapid intervention would be available if postoperative complications occurred. She was mobilized following physiotherapy treatments and now uses a walking frame.

The patient was discharged on day four post-surgery. She was prescribed pain management medication. The patient was scheduled...
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[3]. Classic features of NF2 are bilateral acoustic neuromas and
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presented case the patient had previously developed bilateral acoustic
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multiple schwannomas in the spine [3, 4].
Spinal schwannomas account for approximately 29% of all spinal
tumors. Schwannomas tend to occur in the 30-60 year age group but
they may occur at any age. They have no gender predilection. They can
occur at any level in the spine, typically from the posterior nerve roots.
Schwannomas are benign and slow growing but can cause
compression of the spinal cord giving rise to neurologic symptoms
(Figure 3) [5].
Although symptoms reflect the function mediated by the involved
nerve and surrounding structures, presentation is usually of a slowly
advancing spastic paraparesis with an unilateral band of pain and
sensory loss from the affected nerves. As the lesion progresses these
initial symptoms may be followed by weakness and bladder and bowel
dysfunction [5,6]. In the case presented the patient’s symptoms
included spasticity, weakness of lower limbs, back pain, pain
experienced in the lower limbs and left eye pain.
Magnetic resonance imaging with the use of gadolinium-based
contrast medium is the technique of choice for diagnosing
schwannomas; it provides the highest degree of soft tissue resolution.
The images are in multiple planes and are not encumbered by bone
artifacts from the skull base. Computed tomography is ideal for
evaluating the secondary effects of spinal schwannomas on the neural
foramen and to delineate extraspinal extension [6]. Since the
emergence of magnetic resonance imaging other imaging methods,
such as computed tomography including gas cisternography and
myelography, have limited roles in diagnosing schwannomas but
occasionally are used in patients where magnetic resonance imaging is
contraindicated [6].
Treatment for spinal schwannomas includes surgical resection of
the schwannoma, arresting schwannoma growth using radiation
therapy or careful serial observation and monitoring. Each treatment
modality has its own risks and benefits [7]. Genetic screening and
testing of blood can reveal damaging mutations of the NF2 gene.
Screening may be conducted during pregnancy to aid in early
diagnosis of individuals with a family history of the disorder thereby
allowing for early treatment [7].

Conclusion
NF2 is a rare genetic disorder characterised by multiple schwannomas
of the nervous system, brain and spine. Due to the fact that NF2 is so
rare, few studies have been done to observe the natural progression of
this disorder. As there is no cure for NF2 the main aim of treatment is
to alleviate symptoms, closely monitor its development and to intervene
when necessary.
One concern that should not be overlooked is the risk of isolation,
depression and loneliness in NF2 patients. Patients with NF2 are
anxious about future complications of their condition and often
deteriorate into a state of chronic depression brought on by
progressive symptoms of this disorder. Geneticists are working hard to
improve understanding of how mutations in these genes occur so that
they can develop gene therapy to help prevent NF2.

References
weakness of the limbs and multiple tumours of spinal nerves. The
2. Evans, D G R, Sainio, M et al. Neurofibromatosis type 2. Journal of
Medical Genetics, 37:12; 2000: 897-901.
schwannomatosis or neurofibromatosis type 2. Journal of Neurology,
and type 2 neurofibromatosis-associated meningiomas and
5. Roach, E S & Miller, V S. Neurocutaneous Disorders. London: