

Fact Sheet: Schwannomatosis



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Schwannomatosis

Schwannomatosis is characterized by the growth of multiple schwannomas anywhere except on the vestibular nerve – which is a defining characterization of NF2. This disorder appears to occur as often as NF2 – approximately 1 in 40,000 births. Schwannomatosis is a rare form of NF that has only recently been recognized. People with schwannomatosis develop multiple schwannomas on cranial, spinal and peripheral nerves – but they do not develop vestibular tumours and do not go deaf.

Schwannomas are tumours that come from the cells that form a protective sheath around the body's nerve fibers. They are usually benign and surgically removed when possible. They usually appear as a single tumour. Rarely will they develop into multiple tumours. Affected individuals usually have much greater problems with pain than with neurological disability, although as with all forms of NF, schwannomatosis may vary greatly between patients. They also do not develop any other kinds of tumours (for example, meningiomas, ependymomas or astrocytomas) and do not have learning disabilities.

For reasons not yet understood, people with schwannomatosis have problems with chronic pain that often exceeds their neurological problems. The first symptom of schwannomatosis is almost always pain, which can occur in any part of the body. Many patients with schwannomatosis go several years before the source of their pain is realized because they have few or no neurological symptoms. Once a tumour is found, it is important to determine its pathological type by examining a piece of it under a microscope. Patients with schwannomatosis are always found to have schwannomas, not neurofibromas. For reasons not yet understood, about 1/3 of patients with schwannomatosis have tumours limited to a single part of the body, such as an arm, a leg or a segment of the spine. Once a person is found to have multiple schwannomas, the possibility of NF2 must be excluded before a diagnosis of schwannomatosis is given. In an older person with no hearing loss, NF2 is unlikely. In a younger person, or in any person with hearing loss and multiple schwannomas, it is imperative that a high quality MRI scan of the base of the brain be done to exclude the possibility of vestibular tumours and NF2.

At the current time there is no “blood test” to determine if a patient has schwannomatosis. Surgical management of schwannomatosis is often quite effective. When tumours are completely removed, pain often subsides – although it may recur if new tumours form. When surgery is not feasible, management in a multidisciplinary pain clinic is advisable. There is no currently accepted medical treatment or drug for schwannomatosis. Because schwannomatosis has been a relatively recent “discovery,” there are no set schedules of tests that all patients should undergo. The tumours of schwannomatosis are relatively slow growing, and probably only need to be imaged when symptoms change.

Schwannomatosis is a genetic condition, but may often skip generations so that more distant family members with unexplained neurological symptoms and/or unexplained pain should be evaluated for the possibility that they are also affected.

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Schwannomas

Both schwannomas and neurofibromas originate in the insulating covering of peripheral nerves called the nerve sheath. Schwannomas are very homogenous tumours consisting **only** of nerve sheath cells or Schwann cells. They stay on the outside of the nerve, but may push it aside or against a bony structure causing damage. Neurofibromas are very heterogeneous tumours which incorporate all sorts of cells and structural elements in addition to the Schwann cells. They infiltrate the nerve and splay apart the individual nerve fibers. Neurofibromas sometimes degenerate into cancer, but for reasons that we do not understand, schwannomas never do.