Schwannomatosis shares many features of the neurofibromatoses, and may be viewed as another form of neurofibromatosis. Much less is known about schwannomatosis than about NF1 and NF2, however. Current evidence suggests that schwannomatosis is associated with alteration of a gene that is distinct from the NF1 and NF2 genes.

Schwannomatosis is a disorder that causes tumors called schwannomas to grow on nerves. By definition, individuals with schwanomatosis do not develop the vestibular tumors characteristic of individuals with NF2. It is unclear how many people suffer from this disorder or how many have been misdiagnosed because of similarities to NF2 or other nerve tumor disorders. There are published reports that indicate a prevalence of schwannomatosis that is similar to that of NF2, i.e., about 1 in 40,000 births.

Schwannomatosis is a genetic condition, but there are many unexplained observations about its occurrence. For example, well over half of patients with schwannomatosis have no discernable affected relatives. When schwannomatosis is familial, it may often skip generations, unlike either NF1 or NF2. The Children's Tumor Foundation is supporting two research groups who are looking for the genetic locus causing schwannomatosis.

Affected individuals usually have much greater problems with pain than with neurological disability, although schwannomatosis severity varies greatly between patients. Most comonly,schwannomatosis patients experience their first symptoms as adults, but many experience a frustrating lag between onset of symptoms and achieving a proper diagnosis.

Once a person is found to have multiple schwannomas, the diagnosis of NF2 must be excluded before a diagnosis of schwannomatosis is established. 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 skull be done to exclude the possibility of vestibular tumors and NF2. At the current time there is no blood test to determine if a patient has schwannomatosis.

In October 2003, the Children's Tumor Foundation sponsored an International Consensus Conference of schwannomatosis researchers to develop standards for diagnosis and evaluation of patients with schwannomatosis. Their formal suggestions for diagnostic criteria are shown in Table 1. Apart from skull base imaging, there are no set schedules of tests that all patients should undergo. The tumors of schwannomatosis are relatively slow growing, and probably need to be imaged only when symptoms change. Patients with schwannomatosis do not develop neurofibromas or the malignant peripheral nerve sheath tumors associated with NF, and do not have learning disabilities. Approximately 1/3 of patients with schwannomatosis have tumors limited to a single part of the body, such as an arm, leg or a segment of the spine. Surgical management of schwannomatosis is often quite effective at the beginning of the course of schwannomatosis. When tumors are completely removed, pain often subsides, although it may recur if new tumors form. When surgery is not feasible, or during later stages of the disease when surgery does not help, management in a multidisciplinary pain clinic is advisable. There is no currently accepted medical treatment or drug for schwanmatosis.

If you think that you might have schwannomatosis or any other form of NF and would like a referral to an NF clinic, visit: http://www.ctf.org/patientinfo/clinics/.

Clinical Criteria for Schwannomatosis
Table 1

DEFINITE
Age over 30 years and no evidence of vestibular tumor on high quality MRI scan, no known constitutional NF2 mutation and two or more non-intradermal (within or between layers of the skin) schwannomas, at least 1 with histologic confirmation
or
One pathologically confirmed schwannoma plus a first degree relative who meets above criteria

POSSIBLE
Age under 30 years and otherwise meeting criteria for definite schwannomatosis
or
Radiographic evidence of a schwannoma and first degree relative meeting criteria for definite schwannomatosis

SEGMENTAL
Meets criteria for either definite or possible schwannomatosis but limited to one limb or five or fewer contiguous segments of the spine

Glossary of Terms

Chromosome - String of genetic information carried by the cells of the body. Each person receives 22 non-sex chromosomes and 1 sex chromosomes from each parent at conception. As the body grows, a complete set of these 46 chromosomes is replicated and placed in every cell of the body.

Intradermal- Within or between the layers of the skin.

Locus - Position a given gene occupies on a chromosome.

Magnetic Resonance Imaging (MRI) - Method of visualizing the internal structures of the body by exposing the tissues to magnetic fields and measuring their response. MRI produces highly detailed pictures of the anatomy of the body and is especially useful for visualizing the brain. MRI produces no functional information on the area scanned.

Neurofibroma - Neurofibromas originate in the insulating covering of peripheral nerves called the nerve sheath. Neurofibromas are composed of many types of cells. Neurofibromas grow within the center of a nerve, causing the nerve to expand.

NF2 - Neurofibromatosis Type 2 (NF2) is a rare genetic disorder that causes affected individuals to develop tumors on both nerves to the ears and other tumors of the nervous system.

Schwann cell - The cell of which myelin, the insulation of peripheral nerves, is composed.

Schwannoma - Schwannomas are tumors that consist only of nerve sheath tumors or Schwann cells. They form solid, encapsulated tumor masses, growing out from the parent nerve and compressing rather than invading them. They stay on the outside of the nerve but may push it aside or against a bony structure causing damage.

Sheath - The outer covering of the nerve.

Tumor - An abnormal growth of cells. Tumors may be malignant in which case they are called cancers. Non-malignant tumors (benign) do not aggressively invade surrounding tissues or spread to other parts of the body, but they may cause significant symptoms and even death depending on their location.

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Ending Neurofibromatosis Through Research

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Schwannomatosis

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