Schwannomatosis is a genetic disorder that affects approximately 1 in 40,000 individuals.

It causes tumors to grow on peripheral nerves anywhere in the body. These tumors are frequently associated with debilitating pain, the primary concern of persons with Schwannomatosis and a complication that is often difficult to treat.

About the Foundation
The Children’s Tumor Foundation is focused on finding treatments for the three types of neurofibromatosis (NF): NF1, NF2 and Schwannomatosis. For over 25 years we have funded innovative research and clinical initiatives, serving as a catalyst to accelerate progress in NF research.

The Children’s Tumor Foundation has committed over $30 million to NF research programs. In 2008, we will commit $500,000 specifically to Schwannomatosis research.

Founded in 1978, the Children’s Tumor Foundation is a national, not-for-profit health organization dedicated to meeting the unique needs of individuals with neurofibromatosis (NF) and their families.

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What are the features of Schwannomatosis?
Schwannomatosis causes the development of nerve tumors called schwannomas, which can appear anywhere in the peripheral nerves of the body. Schwannomas are benign (non-cancerous) tumors. Schwannomatosis patients typically do not develop malignancies, although these can develop in NF1 and NF2.

Is Schwannomatosis inherited?
Unlike NF1 and NF2, in which 50% of new cases are inherited and there is a 50% chance of passing on the disorder to a child, only 10-20% of Schwannomatosis cases are inherited with 80-90% being sporadic (non-inherited). The genetics are still not well understood however, and it is thought that at least some forms of Schwannomatosis could arise from a combination of mutations in both the NF2 and INI1 genes.

When is Schwannomatosis typically diagnosed?
Schwannomatosis patients most commonly experience their first symptoms in adulthood. Because diagnostic criteria were only recently established, many patients had a frustrating lag between developing symptoms and obtaining a diagnosis.

How is Schwannomatosis diagnosed?
At this time there is no definitive genetic test that can be done on blood or tumor tissue to diagnose Schwannomatosis. Tests are available to identify mutations in one of the genes involved in Schwannomatosis, INI1. However, INI1 is believed to be only one of the genes involved in Schwannomatosis, and mutations in additional genes not yet identified may play a role. For this reason, an INI1 test would not give a definitive diagnosis of Schwannomatosis. Therefore, Schwannomatosis is currently diagnosed based on clinical criteria which were developed by the Children’s Tumor Foundation.

Schwannomatosis shares some clinical features with NF2, and patients must be evaluated for potential diagnosis of either disorder.
The severity of Schwannomatosis varies between individuals. Approximately one-third of patients have “mosaic” Schwannomatosis, with tumors limited to a single part of the body, such as an arm or leg.

What are the clinical management options for Schwannomatosis?
Pain is the principal concern of Schwannomatosis patients. Surgical management is the most common option; early intervention surgery to remove schwannomas can provide some relief and alleviate pain, though this can recur if new tumors form. In cases where Schwannomatosis is advanced or surgery is not feasible, management in a multidisciplinary pain clinic is advisable. However the underlying biological mechanisms of Schwannomatosis-related pain are poorly understood. As a result, it is difficult for physicians to know which drugs will provide adequate relief. Drugs may work for some patients but not others, or may work for a time then lose efficacy in a patient.

What causes Schwannomatosis?
Although genes for NF1 and NF2 were identified in the early 1990’s, a candidate gene for Schwannomatosis — called INI1 — was only recently identified, in early 2007. INI1 is a tumor suppressor and lies on Chromosome 22. Researchers are now studying this gene to better understand its role in Schwannomatosis. In addition to INI1, it is believed that mutations in additional genes may be required for Schwannomatosis to develop in an individual. Notably, it is thought that at least some forms of Schwannomatosis could arise from a combination of mutations in both the NF2 and INI1 genes.

What about drug treatments for Schwannomatosis?
Currently, there is no drug treatment for Schwannomatosis. However, the recent landmark identification of INI1 has opened up the molecular and biological study of Schwannomatosis in an unprecedented way. Research progress is accelerating toward unraveling the genetic basis of Schwannomatosis — a portal to understanding the disorder and identifying effective drug therapies for Schwannomatosis tumor growth and pain.

Currently, there is no effective treatment for Schwannomatosis.
The Children’s Tumor Foundation (CTF) is working to change this by funding innovative Schwannomatosis research programs and facilitating collaborations between researchers and physicians. In 2007-2008, CTF is committing $500,000 to Schwannomatosis research. These funds will be used to advance our knowledge of Schwannomatosis genetics and biology and to develop screening tools such as Schwannomatosis cell lines and mouse models that can be used to test and identify treatments for the disorder.