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What is Neurofibromatosis? (pronounced: neuro-fibroma-tosis)

Neurofibromatosis or NF, is a term for three distinct genetic disorders which cause tumours to form on the nerves anywhere on the body and produce other abnormalities such as bone defects, cardiovascular problems, deafness, disfigurement, cancer, learning disabilities, blindness, and pain. NF manifestations vary greatly but can cause serious health issues and disfigurement.

How common is NF?

NF is not a rare disorder. It is the most common single gene disorder. There are over 13,000 Canadians currently diagnosed and living with neurofibromatosis¹, with over 1,600 of those living in BC². There are more Canadians living with neurofibromatosis than the combined number of Canadians living with cystic fibrosis, hereditary muscular dystrophy, Huntington's Disease and Tay-Sachs Disease.

What are the three NF disorders?

1. Neurofibromatosis Type 1 (NF1) is the most common form and is also called von Recklinghaus Disease. One in every 3,000 Canadians has NF1³. It can affect the nervous system, skin, bones and other tissues. NF1 is a progressive disorder with a wide variety of manifestations and severity.
2. Neurofibromatosis Type 2 (NF2) is a less common form of NF and differs greatly from NF1. One in every 25,000 children born in Canada has NF2⁴. NF2 is distinguished by multiple tumours in the brain and spine which may cause deafness, severe balance problems, facial nerve paralysis, and spinal cord compression. Symptoms usually appear during adolescence or in the early 20s.
3. Schwannomatosis is the third form of NF and has recently been recognized as a distinct disorder. It is estimated that one in every 40,000 Canadians has the disorder⁵. Symptoms usually first appear in adulthood and may manifest as multiple tumours (known as schwannomas) on cranial, spinal and peripheral nerves.

How does a person get NF?

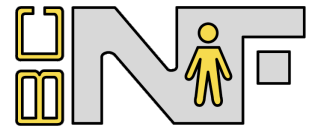
NF is not contagious. NF is a genetic disorder present at birth and can affect anyone, regardless of family history, race, gender, or ethnic background. Fifty per cent of NF cases are passed from one generation to the next, although Schwannomatosis may skip generations. The other 50 per cent of cases have no family history and the cause of the genetic mutation is unknown.

How does NF affect day to day life?

Because there is a wide range of NF symptoms and severity, each person is affected differently. Many deal with deformities caused by tumours or damage to the nerves. Individuals and families may isolate themselves to avoid public exposure rather than face ridicule and fear from others who don't understand the disorder. If there are no overriding complications from NF, life expectancy of people with NF is almost normal and they lead normal lives.

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What is the cure for NF?

Currently there is no cure for NF — only the symptoms are treated at this time.

How is NF diagnosed?

Diagnosis depends on the NF disorder but may include diagnosis based on the combination of symptoms, magnetic resonance imaging, audio testing, genetic testing and pathological examination of tumours.

How is NF treated?

Treatment strategies are still in the trial stages. Removal of tumours may be difficult and even impossible based on location on nerves and their removal may raise a host of other issues.

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¹ based on World Bank, World Development Indicators population figures, 2008 and Children's Tumor Foundation incidence rates

² based on Statistics Canada, Canadian Census 2006 and Children's Tumor Foundation incidence rates

³ Children's Tumor Foundation, May 2007

⁴ Children's Tumor Foundation, May 2007

⁵ National Institute of Neurological Disorders, Publication No. 09-2126