Neurofibromatosis

National Institute of Neurological Disorders and Stroke (NINDS)

Source

National Institute of Neurological Disorders and Stroke (NINDS). Neurofibromatosis Information Page.

The neurofibromatoses are genetic disorders that cause tumors to grow in the nervous system. The tumors begin in the supporting cells that make up the nerves and the myelin sheath--the thin membrane that envelops and protects the nerves. These disorders cause tumors to grow on nerves and, less frequently, in the brain and spinal cord, and produce other abnormalities such as skin changes and bone deformities. Although many affected persons inherit the disorder, between 30 and 50 percent of new cases arise spontaneously through mutation (change) in an individual's genes. Once this change has taken place, the mutant gene can be passed on to succeeding generations. There are three forms of neurofibromatosis (NF): NF1 is the more common type of the disorder. Symptoms of NF1, which may be evident at birth and nearly always by the time the child is 10 years old, may include light brown spots on the skin ("cafe-au-lait" spots), two or more growths on the iris of the eye, a tumor on the optic nerve, a larger than normal head circumference, and abnormal development of the spine, a skull bone, or the tibia. NF2 is less common and is characterized by slow-growing tumors on the vestibular branch of the right and left eighth cranial nerves, which are called vestibular schwannomas or acoustic neuromas.. The tumors press on and damage neighboring nerves and reduce hearing. The distinctive feature of schwannomatosis is the development of multiple schwannomas (tumors made up of certain cells) everywhere in the body except on the vestibular branch of the 8th cranial nerve. The dominant symptom is pain, which develops as a schwannoma enlarges or compresses nerves or adjacent tissue. Some people may develop numbness, tingling, or weakness in the fingers and toes.