

## *Glossary of Terms Often Used With NF*

**Atrophy** – Decrease in size or wasting away of a body part or tissue.

**Auditory Nerve** – Any of the eighth pair of cranial nerves connecting the inner ear with the brain and transmitting impulses concerned with hearing and balance.

**Autosomal Dominant inheritance** – Only one abnormal gene of a pair is necessary for expression of the trait or disorder. Such a gene has a 50% chance of being passed on to each son or daughter of an affected parent. Neurofibromatosis is an autosomal dominant trait.

**Benign** – Not malignant or not cancerous.

**Bilateral** – Affecting or simply “on” both sides

**Café au lait Spots** – Brown oval spots on the skin the color of coffee with milk.

**Cataract** – Clouding of the lens of the eye or of its surrounding transparent membrane that obstructs the passage of light.

**Chemotherapy** – The use of chemical agents in the treatment or control of disease.

**Chromosomes** – The basic units of heredity. The nucleus of each body cell contains 23 pairs of chromosomes.

**Computerized tomography (CT) of the brain** – (Also known as CAT or EMT scans). An automatic electronic machine which provides x-ray scans of tissue planes at a given thickness. Scans show the internal structures of the brain. Tumors, brain injury or other abnormalities can be shown. It is also useful to evaluate orbital pathology.

**Dominant** – Being the one of a pair of bodily structures that is the more effective or predominant in action.

**Dysplasia** – Abnormal development of a part of the body.

**Gene** – The basic unit of heredity. Thousands of genes, arranged in specific linear order, form a chromosome. Genes come in pairs. Each pair is located on one chromosome with the matching gene on the other chromosome of that pair.

**Genetic** – Inherited or basic, relating to information contained on genes.

**Glioblastoma** – Malignant brain tumor (also called astrocytoma Grade III, IV).

**Glioma** – A type of brain tumor

**Learning Disability** – A disorder that affects people’s ability to either interpret what they see and hear or to link information from different parts of the brain. These limitations can show up in many ways: as specific difficulties with spoken and written language, coordination, self-control or attention.

**Lisch Nodule** – Spot on the iris, the colored part of the eye. It does not affect vision. Multiple Lisch nodules are a criteria used in diagnosing NF-1.

**Meningioma** – A benign tumor of the covering of the brain.

**MRI (Magnetic Resonance Imaging)** – A diagnostic technique that uses magnetic energy to image the brain and body.

**Mutation** – A permanent change in the genetic material, usually in a single gene.

**Myelin** – The lipid substance forming an “insulating” sheath around many nerve fiber.

**Neurofibroma** – A benign tumor consisting of nerve fibers and connective tissue caused by proliferation of Schwann cells and fibroblasts.

**Neurofibromatosis (NF)** – A genetic disorder of the nervous system that causes multiple, soft tumors to grow anywhere on the body. The disorder is found in both sexes and all races. The inheritance pattern is autosomal

dominant and, to date, there is no known cure. Two major genetically distinct forms of NF have been identified, NF-1 and NF-2.

**Neurofibromatosis Type 1 (NF-1)** – The condition is characterized by multiple brown spots on the skin, neurofibromas of varying sizes on or under the skin, Lisch nodules on the iris of the eyes, and freckling in the underarm or groin area. Bone deformities, learning disabilities, and optic glioma can also be associated with NF-1. The gene for NF-1 is located on chromosome 17.

**Neurofibromatosis Type 2 (NF-2)** – The condition is characterized by bilateral vestibular schwannomas that cause balance problems, hearing loss, deafness, other tumors of the central and peripheral nervous systems, and cataracts occurring at an early age. The gene for NF-2 is located on chromosome 22.

**Neurofibromin** – A complex protein substance produced by the NF-1 gene.

**Neurofibrosarcoma** – Also known as peripheral nerve sheath tumor (MPNST); a malignant tumor that develops in the cells surrounding these peripheral nerves.

**Optic Glioma** – Tumor affecting the optic nerve.

**Orbit** – The bony cavity of the skull in which the eyeball is located.

**Peripheral** – Situated away from the center or central nervous system.

**Pheochromocytoma** – A tumor of the adrenal gland that causes severe high blood pressure.

**Plexiform Neurofibroma** – A diffuse mass of tissue that is vascular, affecting bundles of nerves.

**Pseudarthrosis** – A “false joint” within a long bone, which is a rare but serious complication of children with NF-1. Other bone-related abnormalities seen in NF-1 include bowing of the long bones of the leg and fractures of long bones that do not heal.

**Radiation Therapy** – The use of high-energy rays or particles to treat disease.

**Sarcoma** – Malignant soft tissue tumor.

**Schwann Cell** – The cell in which myelin is composed.

**Schwannoma** – A benign tumor causes the proliferation of Schwann cells.

**Scoliosis** – Curvature of the Spine.

**Segmental Neurofibromatosis** – A variant of neurofibromatosis in which manifestations are limited to a single part of the body, thought to be due to somatic mosaicism.

**Slit lamp** – Device used by ophthalmologists to examine the eyes for Lisch Nodules in individuals with NF-1.

**Spontaneous Mutation** – A change in a gene occurring with no identifiable cause.

**Tinnitus** – Ringing noise in the ear.

**Tumor** – An abnormal mass of tissue that results from excessive cell division.

**Vestibular Schwannoma** (Acoustic Neuroma) – Benign tumor of the eighth cranial nerve (balance nerve) that can cause hearing impairment, balance problems, and deafness. In NF-2, vestibular schwannomas form on both acoustic nerves; therefore, it is bilateral. Formerly known as acoustic neuroma.

**Von Recklinghausen disease** – Former term for Neurofibromatosis Type 1.