NF1 Glossary of Terms

Acoustic neuroma – Benign tumor of the eighth cranial nerve (acoustic or hearing nerve) which causes hearing impairment (also called acoustic neurinoma or acoustic schwannoma).

Allele – Specific form of a gene. Each individual has two copies of every gene, one inherited from each parent. The particular form of the gene inherited from each parent is referred to as an allele. Neurofibromatosis is due to an alteration of one of the two alleles at the neurofibromatosis gene locus.

Amniocentesis – Method of prenatal diagnosis in which a small amount of amniotic fluid which bathes the fetus is removed for genetic study. Usually performed at 16 to 20 weeks of gestation.

Astrocytoma – A brain tumor containing cells derived from the glial cells (astrocytes or "star-shaped" cells) of the nervous system, which may be benign or malignant.

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 – A single, abnormal gene on one of the autosomal (non-sex determining) chromosomes from either parent can cause certain diseases. One of the parents will usually have the disease (since it is dominant) in this mode of inheritance. Only one parent must have an abnormal gene in order for the child to inherit the disease. For an autosomal dominant disorder - If one parent has an abnormal gene and the other parent a normal gene, there is a 50% chance each child will inherit the abnormal gene, and therefore the dominant trait. Neurofibromatosis is an autosomal dominant trait.

Axilla – The hollow area under the arm where it joins the body. Skin-fold freckles related to NF1 often develop in this area.

Benign – Not malignant or not cancerous.

Bilateral – Affecting or simply “on” both sides

Café au lait – 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.

Cerebrospinal fluid (CSF) – Fluid which bathes the brain and spinal cord.

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

Chorionic villus sampling – Method of prenatal diagnosis in which a small piece of fetal placenta is removed for genetic study. Can be performed in the first third of pregnancy.

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.

Congenital – Present at birth.

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

DNA – The chemical substance that makes genes.

Dysplasia – Abnormal development of a part of the body.

EEG – Electroencephalogram, used to examine electrical activity of the brain for diagnosis of seizures and other problems of brain function.

Expressivity – Degree to which a genetic trait is manifested.

Fibroblast– Cell type that is found in connective tissue throughout the body and involved in the development of neurofibromas.

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.

Genotype – A tiny biochemical structure in cells that transmits characteristics from one generation to the next.

Glaucoma – Increased pressure within the eyeball, which may occur in individuals with neurofibromatosis type 1 having a plexiform neurofibroma of the orbit.

Glioblastoma – A type of malignant brain tumor.

Glioma – A tumor composed of connective tissue of the nervous system and affecting the brain or spinal cord. Sometimes found on the optic nerve of people with NF1.

Heterozygote – Individual who possesses different alleles at a gene locus on each of a pair of homologous chromosomes.

Homozygote – Individual who possesses the same allele at a gene locus on each of a pair of homologous chromosomes.

Hormone – Chemical substances secreted by glands in the body which serve particular roles.

Hydrocephalus – Presence of increased spinal fluid pressure within the ventricles of the brain.

Hypertrophy – Increase in the size of a part of the body.

Hypothalamus – Part of brain responsible for control of hormone secretion, appetite, and other "automatic" functions

Kyphoscoliosis – Spinal deformity combining sideways curvature and hunching forward of the upper part of the spine.

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.

Leukemia – Malignancy involving white blood cells, found rarely in association with neurofibromatosis type 1.

Linkage analysis – Method of mapping genes in relation to one another along the chromosome.

Lisch Nodule – Small masses of pigment on the iris of the eye that do not affect vision. They are often seen in the eyes of individuals with Neurofibromatosis type 1 (NF-1), and used as a diagnostic criteria for NF-1.

Macrocephaly – Enlargement of the head.

Mast cell – Type of cell that is widely distributed throughout the body and particularly prevalent in neurofibroma tissue. Mast cells secrete substances that may affect the behavior of other cells nearby, including those involved in the development of neurofibromas.

Megalencephaly – Enlargement of the head.

Meiosis – Process of cell division which occurs exclusively in germ cells and functions to reduce the chromosome number from 46 to 23 in this cell lineage.

Melanosome – Structure inside melanocytes which contains pigment. Giant melanosomes are seen within pigment cells in a café-au-lait spot.

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

Mitosis – Process of cell duplication.

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

MPNST - A malignant peripheral nerve sheath tumor (also known as "Malignant schwannoma" "Neurofibrosarcoma," and "Neurosarcoma") is a form of cancer of the connective tissue surrounding nerves.

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.

Neural crest – An embryonic structure which contains many of the cell types that tend to grow abnormally in neurofibromatosis.

Neurilemma – The thin membrane which spirally enwraps the myelin layers of myelinated nerves.

Neuro – Denotes relationship to a nerve or nerves, or to the nervous system.

Neurofibroma – A benign tumor 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, freckling in the underarm or groin area, learning disabilities, and optic glioma. 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; a malignant tumor that develops in the cells surrounding these peripheral nerves.

Neurons – Electrically active cells of the nervous system responsible for controlling behavior and body functions.

Noonan syndrome – Dominantly inherited disorder characterized by short stature, congenital heart disease, and distinctive appearance. Features of Noonan syndrome have been found in a small proportion of individuals with neurofibromatosis type 1.

Optic Glioma – Tumor affecting the optic nerve.

Optic nerve – The nerve that transmits visual information from the eye to the brain.

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

Orbital dysplasia – A deformity in the orbit, the bony wall behind the eye, and/or the bone behind it, which may result in displacement of the eye.

Penetrance – Proportion of individuals possessing a genetic trait who manifest specific features of the trait.

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

PET scan (positron emission tomography) – A specialized test that measures a region of the body's metabolic activity. The test may be helpful in distinguishing benign neurofibromas from other types of tumors

Phenotype – Expression of a genetic trait.

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

Pituitary gland – Gland located at the base of the brain which secretes hormones involved in growth, sexual function, and control of other endocrine glands. **Plexiform Neurofibroma** – A diffuse mass of tissue that is vascular, affecting bundles of nerves.

Polymorphism – Genetic trait for which multiple alleles exist with appreciable frequency in the population.

Precocious puberty – Attainment of puberty abnormally early.

Prenatal diagnosis – The testing for the presence of a medical condition in an embryo or fetus.

Proteus syndrome – Recently described disorder that causes overgrowth of parts of the body, as well as abnormalities of skin and bone. It has been suggested that John Merrick, the "Elephant Man," actually had Proteus syndrome rather than neurofibromatosis.

Pseudarthrosis – A "false joint" within a long bone, which is a rare but serious complication of NF-1 children. 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.

Recombination – Process whereby homologous chromosomes exchange segments during meiosis.

Recessive – Both members of a pair of genes need to be altered in order to have expression of the trait or disorder.

Rhabdomyosarcoma – Malignant tumor of muscle cells, found rarely in association with neurofibromatosis type 1

Sarcoma – Malignant soft tissue tumor.

Schwann Cell – The cell in which myelin is composed.

Schwannoma – A benign tumor cause the proliferation of Schwann cells.

Scoliosis – Curvature of the Spine.

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

Seizure – Abnormal electrical discharge of brain tissue, often resulting in abnormal body movements or behaviors.

Skin-fold freckles – Freckles on areas of the skin not exposed to the sun. Often seen in people with NF1.

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.

Schwannomatosis – The condition is characterized by multiple schwannomas (nerve sheath tumors) without evidence of vestibular schwannoma.There is pain associated with the schwannomas and symptoms possibly limited to one area of the body.

Sphenoid – Skull bone that forms a portion of the back of the orbit. Sometimes the sphenoid is abnormally formed in people with NF1, resulting in deformity of the orbit.

Spinal fusion – A means of surgical treatment of scoliosis.

Spontaneous mutation – A mutation occurring without apparent external influence.

Sporadic trait – Occurring in a single individual in a family, with no prior family history.

Tibia – The long bone in front of lower part of leg (shinbone), sometimes bowed in people with NF1.

Tibial dysplasia – A deformity involving excessive bowing or curvature of the tibia, also known as the shin bone.

Tinnitus – Ringing noise in the ear.

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

Ventricles – Cavities within the brain which are filled with cerebrospinal fluid.

Vertebra – Bone of the spinal column.

Vestibular nerve – Cranial nerve that conveys information about sense of balance to the brain. This nerve may be affected by complications of NF2 known as vestibular schawannomas.

von Recklinghausen neurofibromatosis – Another name for neurofibromatosis type 1, the peripheral form of neurofibromatosis.

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.