NF and Self-Advocacy Glossary

A plain-language reference for NF, care, research, insurance, workplace, education, and advocacy terms

How to use this guide	Look up a term before an appointment, appeal, school meeting, workplace conversation, or research discussion. Definitions are starting points; a term may be used differently in a particular setting.

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Important: This guide provides general educational information. It is not medical or legal advice and does not determine diagnosis, urgency, eligibility, coverage, or legal rights. Ask the appropriate clinician, health plan, school, employer, attorney, agency, or support organization how a term applies to your situation.

NF conditions and genetics

8 plain-language definitions

Neurofibromatosis (NF)

An umbrella term often used for NF1, NF2-related schwannomatosis, and other forms of schwannomatosis. These are distinct conditions with different features and care needs.

NF1

A genetic condition associated with features that can include cafe-au-lait macules, freckling, neurofibromas, learning differences, bone changes, and certain tumors. Features vary widely.

NF2-related schwannomatosis

The current name for the condition previously called NF2. It is associated especially with vestibular schwannomas and may also involve other tumors of the nervous system.

Schwannomatosis

A group of genetic conditions associated with schwannomas. Symptoms, tumor locations, genetic causes, and care needs differ among people.

Pathogenic variant

A change in a gene that evidence indicates can cause or contribute to a genetic condition. A laboratory report may use this term instead of mutation.

Variant of uncertain significance (VUS)

A genetic change for which available evidence is not sufficient to classify it as disease-causing or benign. A VUS generally should not be treated as a confirmed diagnosis by itself.

Mosaicism

A situation in which a genetic variant is present in some of a person's cells but not all of them. This can affect symptoms and whether a variant is detected in a blood test.

Genetic counseling

A process that helps people understand genetic test options and results, inheritance, family implications, and possible next steps.

Tumors, symptoms, and care

11 plain-language definitions

Neurofibroma

A usually benign tumor that develops from cells associated with nerves. Neurofibromas can occur in different forms and locations.

Cutaneous neurofibroma

A neurofibroma arising in or on the skin. The number, size, and effect on comfort or daily life vary.

Plexiform neurofibroma

A neurofibroma that grows along multiple branches of a nerve and may involve nearby tissue. It is associated with NF1 and may require monitoring or treatment.

Schwannoma

A usually benign tumor arising from Schwann cells, which help insulate peripheral nerves. Symptoms depend in part on the nerve and location involved.

Vestibular schwannoma

A schwannoma involving a nerve responsible for hearing and balance. It may affect hearing, balance, or other nearby functions.

Meningioma

A tumor arising from the membranes surrounding the brain or spinal cord. Many are benign, but effects and treatment needs depend on size, location, growth, and symptoms.

Malignant peripheral nerve sheath tumor (MPNST)

A cancer arising from cells associated with peripheral nerves. People should discuss concerning new or changing symptoms with an appropriate clinician; this glossary cannot determine urgency.

Multidisciplinary care

Care coordinated across more than one specialty, such as genetics, neurology, oncology, surgery, hearing, pain management, psychology, or rehabilitation.

Surveillance

Planned monitoring intended to identify changes over time. The tests and schedule depend on the condition, age, symptoms, history, and clinical guidance.

Baseline

An initial measurement or description used for comparison with later findings, such as hearing, imaging, pain, function, or tumor size.

Patient-reported outcome

Information reported directly by a patient about symptoms, function, quality of life, or treatment effects, without interpretation by another person.

Research and treatment

18 plain-language definitions

Children’s Tumor Foundation (CTF)

A nonprofit organization that supports NF research, care resources, education, advocacy, and community programs.

National Institutes of Health (NIH)

The United States medical research agency. This hub uses NIH sources, including PubMed, ClinicalTrials.gov, and NIH RePORTER, to help visitors locate original research information.

Food and Drug Administration (FDA)

The United States agency that regulates medicines and other health products. FDA approval applies to specific uses described in a product’s approved labeling.

PubMed

A free database maintained by the National Library of Medicine that provides citations and abstracts for biomedical literature. A PubMed listing does not by itself establish research quality or clinical relevance.

ClinicalTrials.gov

A public registry and results database for clinical studies. Registration is not proof that an intervention works.

Magnetic resonance imaging (MRI)

An imaging method that uses a magnetic field and radio waves. The body area, technique, contrast use, and schedule depend on the clinical question and professional guidance.

MEK inhibitor

A medicine that blocks MEK signaling within the RAS–RAF–MEK–ERK pathway. Some MEK inhibitors are approved for specific NF1-related uses, while other uses remain investigational or off label.

Clinical trial

A research study involving people that follows a planned protocol. Registration or participation does not by itself show that an intervention is effective or appropriate for a particular person.

Preclinical research

Research performed before testing an intervention in people, often using laboratory methods, cells, computer models, or animals.

Study protocol

The written plan for a research study, including its purpose, design, procedures, eligibility rules, outcomes, and analysis.

Eligibility criteria

The inclusion and exclusion rules used to decide who may participate in a study. Not meeting criteria is not a judgment about a person's needs or worth.

Informed consent

A process for learning a study's purpose, procedures, possible risks and benefits, alternatives, privacy practices, and voluntary nature before deciding whether to participate.

Placebo

An inactive comparison used in some studies. Not every trial uses a placebo, and study staff should explain whether and how one is used.

Randomization

Assignment to study groups by chance. It is used to reduce bias when comparing interventions.

Endpoint

A specific outcome a study is designed to measure, such as tumor response, hearing, pain, function, safety, or quality of life.

Adverse event

An unfavorable medical occurrence during a study or treatment. It is not automatically proof that the study intervention caused the event.

Off-label use

Use of an approved medicine in a way not included in its FDA-approved labeling, such as for a different condition, age group, dose, or route.

Shared decision-making

A process in which a patient and clinician discuss available options, evidence, uncertainties, risks, benefits, and the patient's goals and preferences.

Health insurance and access

10 plain-language definitions

Medical necessity

An insurance or benefit-plan standard used to decide whether requested care meets coverage criteria. Definitions and evidence requirements depend on the plan.

Prior authorization

A plan requirement to obtain approval before certain care, tests, or medicines are covered. Authorization is not always a guarantee of final payment.

Formulary

A health plan's list of covered prescription medicines, often organized into tiers with different coverage rules or costs.

Step therapy

A coverage rule that may require trying one treatment before the plan covers another treatment. Exceptions or appeals may be available depending on the plan and circumstances.

Peer-to-peer review

A discussion, often between a treating clinician and a health-plan reviewer, about an authorization request or coverage decision. Availability and procedures vary.

Internal appeal

A request for a health plan to reconsider its own coverage decision. The denial notice should explain applicable procedures and deadlines.

External review

When available and applicable, review of an eligible health-plan dispute by an independent reviewer outside the plan.

Expedited appeal

A faster appeal process that may be available when waiting through the standard timeline could seriously affect health or recovery. Standards vary.

In network / out of network

Terms describing whether a provider has a contract with a health plan. Coverage, cost sharing, and authorization rules may differ.

Case manager

A person who may help coordinate services, benefits, or care. The role differs across clinics, insurers, employers, and community programs.

Workplace and education

9 plain-language definitions

Americans with Disabilities Act (ADA)

A United States civil rights law that prohibits disability discrimination in covered settings. Which title, requirements, and remedies apply depends on the situation.

Individuals with Disabilities Education Act (IDEA)

A United States education law governing special education and related services for eligible children. Eligibility and procedures differ from Section 504 protections.

Reasonable accommodation

A change to a job or work environment that may help a qualified person with a disability apply for, perform, or access work. What is reasonable depends on the situation and applicable law.

Interactive process

A workplace discussion used to understand an accommodation request and identify possible effective options.

Essential job functions

The fundamental duties of a position. They are relevant when considering whether a person is qualified for a job and what accommodations may be effective.

Section 504 plan

A plan describing disability-related accommodations or services for an eligible student under Section 504. Eligibility and procedures differ from special education under IDEA.

Individualized Education Program (IEP)

A written education program for an eligible student who receives special education and related services under IDEA.

Disability services office

A college, university, or training-program office that coordinates requests for disability-related accommodations and documentation.

Functional limitation

A description of how a condition or symptom affects an activity, task, or access need. It can be more useful than a diagnosis alone when requesting support.

Self-advocacy and coordination

8 plain-language definitions

Self-advocacy

Communicating about your needs, questions, goals, rights, or preferences and taking part in decisions that affect you.

Care coordination

Organizing information, responsibilities, referrals, and communication across providers, services, settings, or caregivers.

Health care transition

The gradual process of preparing for, transferring to, and becoming established in adult-centered care.

Second opinion

An evaluation by another qualified clinician. It may confirm a plan, offer another option, or clarify uncertainty.

Patient navigator

A person who helps patients understand and move through care, referrals, appointments, services, or practical barriers. Duties vary by program.

Release of information

Written permission that allows specified health information to be shared with named people or organizations for a stated purpose and time.

After-visit summary

A document summarizing an appointment, which may include diagnoses discussed, medications, instructions, tests, referrals, and follow-up plans.

Escalation

Moving an unresolved concern to another person or level, such as a supervisor, patient relations office, plan appeal, or agency process.

A useful follow-up question	What does this term mean in my situation, and what action - if any - should I take next?