

A GUIDE FOR NF PATIENTS AND
CAREGIVERS

TRANSITIONING TO ADULT CARE

WORKING TOGETHER AS THE **NF COLLECTIVE**

Overview

This initiative of the Neurofibromatosis (NF) Collective aims to help caregivers and patients through the process of transitioning from pediatric to adult health care, which can be trickier than it sounds! Without proper planning, the health care transition process can lead to a number of avoidable issues including: insurance hassles, medical care/health complications, difficulty finding quality care, and lack of follow-up/responsibility for patients. Since neurofibromatosis occurs world-wide across all ethnic groups, we acknowledge patients' cultural and religious differences and encourage the individual right to interpretive services. Our hope is to offer you the tools that will ensure a smooth health care transition. As you begin this process, we encourage you to take the Transition Readiness Assessment Questionnaire (TRAQ). In addition to the tools we lay out here, we encourage you also to visit gottransition.org for more information on how to plan for and navigate health care transitions. Further, we welcome your feedback for future annual revisions to reflect your authentic experiences. Specific suggestions for each form of NF are located at the end of this document, along with a glossary of terms.

SUGGESTIONS FOR CHILD

Suggested Action Steps:

- Look for every opportunity to practice speaking for yourself and be actively involved in your health care.
- Learn about your condition, medications and any allergies you may have.
- If helpful, refer to the attached digital visual handout detailing NF1, NF2 & schwannomatosis manifestations.
- Access and fill out appropriate sections of the Transition Readiness Assessment Questionnaire (TRAQ). (Recommended to be completed annually)
- Develop a partnership with your parents , caregivers, or other adult mentors.
- Plan ahead for how you'll answer questions about NF, which will allow you to be a better advocate for yourself.

SUGGESTIONS FOR PARENT/CAREGIVER

Suggested Action Steps:

- Embrace a mentality to gradually shift responsibility for care to your child as he/she approaches adolescence.
- Make sure you know at what age your clinic no longer cares for young adults.
- Help your child build resilience through open communications and allow them to help guide their own treatment. Help give them healthy "scripts" for talking with others about NF. Prepare yourself for what you'll say and how you'll walk them through the inevitable, and sometimes awkward parts of adolescence.
- As your child's history of MRIs continues, consider alternative sedation forms that are age-appropriate for each individual.



11-12 YEARS OF AGE

SUGGESTIONS FOR ADOLESCENT

Suggested Action Steps:

- Continue to develop knowledge of your condition, including what to do in case of emergency.
- Learn how to fill a prescription.
- Participate in your IEP (Individualized Education Program) process, including the areas that address transition.

SUGGESTIONS FOR PARENT/CAREGIVER

Suggested Action Steps:

- Maintain a thorough medical record of your child's medical and family history.
- Help your child create an emergency wallet card or learn to use a mobile health app.
- Provide your child with an insurance card, explain its components, and how to contact the insurance company.
- Access resources on how to advocate for your child, such as during your child's IEP meeting or from your child's school counselor.
- Ensure the implementation of necessary accommodations.
- Begin conversations about financial responsibility and safety.



13-16 YEARS OF AGE

SUGGESTIONS FOR ADOLESCENT

Additional Comments:

- Explore meeting other people your age (through camps, networks, symposiums, etc.) in order to share your unique experiences together.
- Ask your doctor about your care both with/without your parent/caregiver present.
- Identify the adults you can trust to help guide you through your health care journey.

SUGGESTIONS FOR PARENT/CAREGIVER

Additional Comments:

- Empower your adolescent to begin guiding his or her own health care by asking the questions at medical appointments, etc.
- Discuss peer pressure with your adolescent, particularly as it relates to the dangers of alcohol, tobacco and drugs.
- Have an open dialogue with your adolescent about sexuality and relationships, encouraging him or her to ask questions. Explain that no question is “wrong” or “stupid”.
- Talk to your adolescent about what it means to set clear personal boundaries and to advocate for his or her own health and safety.
- This is also an ideal age to begin talking about your adolescent’s plans for life, post-high school.

13-16 YEARS OF AGE

SUGGESTIONS FOR ADOLESCENT

Suggested Action Steps:

- Learn how to schedule an appointment.
- Learn how to check your benefits with the insurance company or “sign up” for new insurance.
- Take responsibility for contacting your clinical team with a question or to obtain test results.
- Secure all medical supplies and routinely check to be sure you have an adequate quantity.
- Complete a Post Secondary/Career checklist via IEP or Guidance Counselor.
- Make decisions on living arrangements, if applicable.

SUGGESTIONS FOR PARENT/CAREGIVER

Suggested Action Steps:

- Find an adult doctor (and consider finding a social worker).
- Practice with your child the process of making doctor’s appointments, contacting the clinic, and confirming your child's medical insurance or Medicaid coverage (varies from state to state).
- **Some helpful links:**
[Healthy Adulting Toolkit](#)
- Provide your child with a copy of medical records, contact numbers, medications, and necessary providers.
- Familiarize yourself with Guardianship and Conservatorship options/issues.

17-19 YEARS OF AGE



SUGGESTIONS FOR ADOLESCENT

Additional Comments:

- Whether you are living at home, at college, or independently elsewhere, ensure you have a local medical team that you trust and who has your entire medical history on file.
- If living away from home, be sure to identify and utilize the support services offered through your post-secondary institution or local clinic.

SUGGESTIONS FOR PARENT/CAREGIVER

Additional Comments:

- Empower adolescents to manage their own medical care and underscore the importance of trusting their instincts while learning to become the primary advocate for their own personal health.

17-19 YEARS OF AGE

SUGGESTIONS FOR YOUNG ADULT

Suggested Action Steps:

- As you begin contemplating your career options, be sure you know your rights in the workplace.
- Educate yourself on how to monitor your personal medical information. Resources are readily available online and through your clinic.
- Ensure there are no gaps in your medical coverage.
- Know how to access psychosocial materials for support.

SUGGESTIONS FOR PARENT/CAREGIVER

Suggested Action Steps:

- Finalize health insurance coverage timelines.
- Check in with your child on a regular basis to be sure he/she is effectively managing his/her own health care.
- Remind your child that you are there to help him/her throughout transition.

20-21 YEARS OF AGE



SUGGESTIONS FOR YOUNG ADULT

Additional Comments:

- Identify your own personal interests, strengths and weaknesses and choose the paths that highlight these strengths and interests.
- Be aware of any health changes that may occur during this time and seek help from trusted medical professionals--who understand NF--to address these issues.
- Make sure you have an organizational system (such as a binder or folder) that includes important health documents, insurance information, prescriptions, appointment schedules and ongoing care.

SUGGESTIONS FOR PARENT/CAREGIVER

Additional Comments:

- In the ever-changing world of health insurance, teach your young adult how to shop for and examine the costs and benefits of various health insurance policies. Help him or her learn to compare various coverage options in order to select the plan that is best. Consider contacting an insurance professional or an NF organization who can recommend ways to navigate specialized care insurance coverage.
- Continue to be a guide and resource for your young adult, while encouraging his or her self-advocacy.
- Continue facilitating open dialogues with your young adult on subjects related to sexuality, relationships, genetics, birth control and substance use.

20-21 YEARS OF AGE

SUGGESTIONS FOR YOUNG ADULT

Suggested Action Steps:

- Have a plan in place to keep track of your medical appointments and maintain insurance coverage.
- Consider consulting with a genetic counselor when you begin family planning.

SUGGESTIONS FOR PARENT/CAREGIVER

Suggested Action Steps:

- Check in as needed to ensure your child is following his/her care plan and be available to provide support and guidance to your child when asked.



OVER 21 YEARS OF AGE

SUGGESTIONS FOR YOUNG ADULT

Additional Comments:

- As you consider employment and career options, evaluate all aspects of what each option would provide for you.
- Keep in mind that this is only the beginning of your life's journey as an adult! This time is about continuing to learn about yourself. Your interests may change, your career paths will likely change and, most importantly you will learn what you don't like to do. This is normal! Use this knowledge to inform ongoing decisions as they relate to your work and personal life.

SUGGESTIONS FOR PARENT/CAREGIVER

Additional Comments:

- Continue to be a source of steadfast support and encouragement for your young adult. Reserve judgment, but offer advice and remind your young adult what you've been teaching him or her since Kindergarten: we learn from our mistakes and become better-rounded individuals by turning challenges into opportunities.

OVER 21 YEARS OF AGE

NF TYPE 1

The specific manifestations of NF1 are as unique as each individual with the disorder. A myriad of complications arise at various ages, and many individuals never experience the more concerning issues that would have required treatment by the time of transition. The following categories are more typical issues that need consideration in the transition process.

Specific Suggestions for Each Form of NF

- **Cognition/Psychosocial Issues:** Learning challenges accompanied by attention deficit and hyperactivity disorder persist through the education years and employment. Social skills issues in childhood and adolescence give way to anxiety and depression in many. Quality of life issues involving pain, fatigue, parenting, employment, and social isolation require monitoring throughout adulthood.
- **Peripheral nerve sheath tumors:** Cutaneous neurofibromas emerge in late childhood through adulthood, whereas plexiform neurofibromas emerge in early childhood. Assessment of deeper peripheral nerve sheath tumors can be made after puberty and dictate ongoing surveillance for internal tumors. Malignant peripheral nerve sheath tumors tend to emerge in early to middle adulthood, and require early detection to achieve optimal management. Other, more rare sarcomas, arise at any time.
- **Central nervous system tumors:** Optic nerve pathway tumors and low-grade gliomas of the brain generally arise in early to mid-childhood, are responsive to therapy, and cease being problematic by puberty. However, high-grade astrocytomas and paraspinal issues can arise in adulthood.
- **Orthopedic manifestations:** Long-bone bowing, sphenoid wing dysplasia, dystrophic scoliosis, and short stature all arise in childhood, but treatment for those manifestations carry over into adulthood. Adolescent- and adult-onset scoliosis and osteopenia require ongoing surveillance.
- **Screening plan:** Establishing surveillance plans with an NF provider for tumor screening including MPNST, breast cancer, and brain cancer is important in transition to adulthood. Monitoring of blood pressure and potential vascular issues, including stroke, is also important in the transition to adulthood.

Any of these issues can potentially affect the process of transition, and need consideration when approaching the timing and determination of individual needs for transition. In general, a person's medical journey through adolescence will drive issues to be followed in adulthood, but there are additional issues that arise later in life that need to be addressed by healthcare providers experienced with medical issues in all adults with NF. Such providers will assemble the appropriate management teams to treat potential medical complications if they should arise.

NF TYPE 2

Similar to NF1, the specific manifestations of NF2 are as unique as each individual with the disorder. Depending on the location and size of tumor development, there are a myriad of complications that may arise at various ages, including during the time of transition. The following categories are more typical issues that need consideration in the transition process.

Specific Suggestions for Each Form of NF

- **Hearing impairment/deafness and communication:** Approximately 90 percent of individuals with NF2 develop hearing impairment or deafness during their lifetime. Changes in hearing may be gradual or, in some cases, very sudden. Since the age of onset and progression of hearing loss is often unpredictable, it is helpful to start instruction for speech reading and sign language early, prior to significant hearing impairment.
- **Surgical intervention of vestibular schwannomas:** There are different approaches to hearing preservation and surgical intervention of vestibular schwannomas. It is important to discuss these approaches with a healthcare provider who has experience in both cochlear implants (CI) and auditory brainstem implants (ABI). Young adults should participate in these discussions and be aware of surgical complications such as facial paralysis which may cause a changed facial appearance, difficulties with eye closure, and impaired oral function.
- **Central nervous system tumors:** Other than vestibular schwannomas, non-vestibular schwannomas may develop in NF2, as well as meningiomas and ependymomas. These can appear during the transition phase and monitoring of tumors with an N2 specialist is important. MRIs are a routine part of NF2 care. Young adults should be familiar with the types of symptoms that may be of concern. Some individuals consider participation in clinical trials related to NF2-related treatments and young adults should be involved in this decision-making process.
- **Other NF2-related issues:** Vertigo and balance issues in NF2 may develop over time and may interfere with different activities including lifestyle choices and physical activity. However, it is important for individuals to stay physically active so discussing safe options with an NF provider is suggested. Visual disturbance may also become a challenge in NF2 and accommodations may be needed.
- **Psychosocial issues:** The impact of hearing loss on communication can lead to social isolation affecting the education years and employment. Quality of life issues including pain, effects of the progression of tumors, and treatments give way to anxiety and depression in many. These require monitoring throughout adulthood.

Any of these issues can potentially affect the process of transition, and need consideration when approaching the timing and determination of individual needs for transition. In general, a person's medical journey through adolescence will drive issues to be followed in adulthood, but there are additional issues that arise later in life that need to be addressed by healthcare providers experienced with medical issues in all adults with NF. Such providers will assemble the appropriate management teams to treat potential medical complications if they should arise.

Schwannomatosis

Schwannomatosis is typically diagnosed in adulthood (25-30 years old). Affected people generally have multiple schwannomas, which are benign tumors of the nervous system. In schwannomatosis, these tumors can grow along any nerve in the body, although they are less common on the vestibular nerve. The signs and symptoms associated with schwannomatosis vary based on the size and location of the schwannomas. The most common symptom is chronic pain, which can develop as a growing schwannoma presses on nerves or surrounding tissues. Some people may develop a mass if the schwannomas is located just beneath the skin. Others can experience neurological symptoms such as numbness, tingling, and/or weakness in the fingers and toes. Pain management is the paramount medical consideration for an individual with schwannomatosis. Recommended discussion with physician and NF provider could entail the asking about blood tests, possible MRI (before off parent's insurance), consulting with a neurologist for clinic follow-up, and addressing any issues related to chronic pain, depression and anxiety.

Specific
Suggestions for
Each Form of NF

Additional Resources

ADDITIONAL RESOURCES

Got Transition

<https://www.gottransition.org/>

Healthy Adulthood Toolkit

<https://younginvincibles.org/wp-content/uploads/2018/10/2018-Healthy-Adulthood-Toolkit.pdf>

Adulthood 101

<https://younginvincibles.org/what-we-do/consumer-education/>

National Human Genome Research Institute

<https://www.genome.gov/Genetic-Disorders/Neurofibromatosis>

National Institute of Neurological Disorders and Stroke

<https://www.ninds.nih.gov/disorders/patient-caregiver-education/fact-sheets/neurofibromatosis-fact-sheet>

Glossary of Terms

GLOSSARY OF TERMS

Auditory Brainstem Implant (ABI) - a device that provides hearing sensations to individuals who are deaf or hard of hearing and may not benefit from a hearing aid or cochlear implant.

Cafe-au-lait spots or macules (CALs or CALM) - flat, pigmented spots on the skin associated mainly with NF1. They are commonly referred to as “birthmarks”, but are often not present at birth.

Cochlear Implant (CI) - a small electronic device that electrically stimulates the cochlear nerve (nerve for hearing).

Endocrinology - the branch of medicine concerned with the endocrine glands and hormones.

Genetics - the branch of biology that studies genes, including how they are inherited, mutated, activated, or inactivated, often including the role that genes play in health and disease.

Individualized Education Plan (IEP) - a written legal document or plan that describes instruction, support, services, and goals of a student.

Lisch nodules - raised lesions developing on the surface of the iris often seen in NF1.

Malignant peripheral nerve sheath tumor (MPNST) - a cancerous type of tumor that may occur in NF1.

Neurofibroma - a type of benign nerve tumor seen in NF1 that forms soft bumps on or under the skin. A neurofibroma can develop within a major or minor nerve anywhere in the body.

Neurology - the branch of medicine involving disorders of nerves and the nervous system.

Glossary of Terms

Neurofibromatosis (NF) - a term that refers to a complex set of separate genetic disorders known as neurofibromatosis type 1 (NF1), neurofibromatosis type 2 (NF2), and schwannomatosis, collectively called the neurofibromatoses.

Neurofibromatosis type 1 (NF1) - the most common form of neurofibromatosis affecting approximately 1 in 3,000 people. Features of NF1 include brown skin spots (café-au-lait spots), tumors (neurofibromas), eye findings (Lisch nodules or optic pathway gliomas), bone abnormalities and learning issues.

Neurofibromatosis type 2 (NF2) - a form of neurofibromatosis occurring in about 1 in 25,000 people. Findings of NF2 include tumors along the hearing/balance nerve (vestibular schwannoma, sometimes referred to as acoustic neuroma) typically leading to hearing loss or deafness and balance problems. In addition, individuals with NF2 may have visual issues (cataracts or other eye anomaly) and develop neurologic or functional issues related to different types of tumors in the brain and/or spinal cord.

Oncology - the branch of science involved in the diagnosis and treatment of tumors and cancer.

Ophthalmology - the branch of medicine concerned with the study and treatment of disorders and diseases of the eye.

Optic gliomas (also called optic nerve glioma or optic pathway glioma) - a tumor that affects the optic nerve and may be a manifestation of NF1. Optic gliomas may or may not cause vision issues and may affect other areas of the brain causing hormone dysfunction such as precocious puberty.

Orthopedics - a branch of medicine involved with the evaluation and treatment of the bones or muscles.

Primary Care Physician (PCP) - a healthcare professional who practices general medicine.

Glossary of Terms

Schwannomas - a tumor of the tissue that covers nerves, called the nerve sheath. These are typically benign and seen often in NF2 and schwannomatosis.

Schwannomatosis - the least common of NF occurring in roughly 1 in 40,000 people. The main feature of this condition is the development of schwannomas in many areas of the body which often cause pain.

Transition Readiness Assessment Questionnaire (TRAQ) - a validated tool that providers and families can use to assess the abilities of an individual during the transition to adult care.

Vestibular Schwannoma (VS) (sometimes called an acoustic neuroma) - a tumor along the hearing/balance nerve typically leading to hearing loss/deafness and balance problems. These tumors are most commonly seen in NF2, and seen only occasionally in schwannomatosis.

Magnetic resonance imaging (MRI) - a procedure looking at detailed images of organs, tissues and tumors in the body.

Vestibular nerves - the eighth pair of cranial nerves, conveying sensory impulses from the organs of hearing and balance in the inner ear to the brain.



WORKING TOGETHER AS THE NF COLLECTIVE