

# Neurofibromatosis Type I (NF1): Addressing the Transition from Pediatric to Adult Care

Heather B Radtke <sup>1,2,\*</sup>, Angela Berger<sup>3</sup>, Tammi Skelton<sup>4</sup>, Allison Goetsch Weisman<sup>3,5,\*</sup>

<sup>1</sup>Medical College of Wisconsin, Milwaukee, WI, USA; <sup>2</sup>Children's Tumor Foundation, New York, NY, USA; <sup>3</sup>Ann & Robert H. Lurie Children's Hospital of Chicago, Chicago, IL, USA; <sup>4</sup>UAB Heersink School of Medicine, Birmingham, AL, USA; <sup>5</sup>Northwestern University, Chicago, IL, USA

\*These authors contributed equally to this work

Correspondence: Heather B Radtke, Email [hradtke@ctf.org](mailto:hradtke@ctf.org)

**Abstract:** Health care transition, or HCT, is the process of adolescents and young adults moving from a child/family-centered model of health care to an adult/patient-centered model of health care. Healthcare providers have an essential role in this process which can be especially challenging for individuals with medical or special healthcare needs. Neurofibromatosis type 1 (NF1) is a complex multisystem disorder requiring lifelong medical surveillance, education, and psychosocial support. This review highlights the transition needs of NF1 patients and provides resources for both clinicians and families to facilitate HCT in this population. The authors propose a framework for the development of an effective NF1 transition program by using the Six Core Elements model of the Got Transition program, reviewing existing literature, and incorporating author experiences in the care and transition of NF1 patients.

**Keywords:** adolescents, young adults, neurofibromatosis, transition, healthcare

## Introduction

### Healthcare Transition

Healthcare transition (HCT) is defined as the purposeful, planned movement of adolescents and young adults (AYA) from child-centered care to an adult-oriented care system.<sup>1</sup>

This process often, but not always, involves a physical transition to a new healthcare system and/or medical provider(s) and may be complicated by the differences between pediatric and adult healthcare models of care (Table 1). Successful HCT strategies are designed to build healthcare independence<sup>2</sup> and can be characterized by individual and health-related outcomes, such as improved quality of life, decreased hospitalization, and decreased disease morbidity.<sup>3,4</sup> It is widely accepted that HCT for AYA with special healthcare needs, including those with genetic conditions, is more complex than generally healthy peers.<sup>1</sup> AYA with special healthcare needs encounter a variety of challenges, some due to the normal developmental stage that they are in and some from the stresses of their chronic medical conditions on the transition to adult healthcare and life.

Research has shown that the HCT process among AYA with special healthcare needs has been disorganized and at times results in a lack of appropriate insurance coverage and decreased access to adequate healthcare.<sup>6–11</sup> Further, underdeveloped health-related self-management skills have been observed in association with a decline in health, as well as poorer outcomes compared to the general population regarding education, employment, and socioeconomic status.<sup>11</sup> Improvements in HCT for AYA with special healthcare needs are needed to eliminate gaps in medical care and improve health outcomes, as well as general quality of life.<sup>12–15</sup>

Similar to other conditions with special healthcare needs, effective HCT in neurofibromatosis type 1 (NF1) is often deficient because of poor access to care and limited disease knowledge, and may be further complicated by psychiatric diagnoses, cognitive impairment, and executive function deficits.<sup>16,17</sup> This review focuses on the important aspects of HCT in NF1 and provides information about resources available to clinicians and families to promote successful HCT.

**Table 1** Comparison of Pediatric and Adult Health Care Models

Health Care System Characteristic	Pediatric Care Model	Adult Care Model
Practice Approach	<ul style="list-style-type: none"> <li>• Family-centered</li> <li>• Shared decision-making with parents</li> </ul>	<ul style="list-style-type: none"> <li>• Patient-centered</li> <li>• Shared decision-making with patient</li> </ul>
Clinic Location or Affiliation	<ul style="list-style-type: none"> <li>• Frequently located in children's hospitals</li> </ul>	<ul style="list-style-type: none"> <li>• Located in academic hospitals or private office-based settings</li> <li>• NF care may be provided by general healthcare provider</li> </ul>
Multidisciplinary Staffing	<ul style="list-style-type: none"> <li>• Multiple subspecialists often available for multidisciplinary team approach to care - "one-stop shop"</li> </ul>	<ul style="list-style-type: none"> <li>• Few or no "one-stop shop" multidisciplinary clinics. Separate referrals and appointments for each specialist needed.</li> </ul>
Care Coordination and Access to Support Services	<ul style="list-style-type: none"> <li>• Coordination of care often provided by clinic coordinator or nurse navigator</li> <li>• Social work and case management support often available</li> </ul>	<ul style="list-style-type: none"> <li>• Care coordination services often not provided</li> <li>• Few public care coordination programs are available for adults with NFI</li> <li>• Less social work and case management staffing and support available</li> </ul>
Length of Appointment	<ul style="list-style-type: none"> <li>• Longer</li> </ul>	<ul style="list-style-type: none"> <li>• Shorter</li> </ul>
Patient Time Alone with Healthcare Provider	<ul style="list-style-type: none"> <li>• Sometimes, part of visit</li> </ul>	<ul style="list-style-type: none"> <li>• Entire visit, with need for patient permission for others to be present</li> </ul>
Patient Role as Self Advocate	<ul style="list-style-type: none"> <li>• Less, given parental support/presence during visit</li> </ul>	<ul style="list-style-type: none"> <li>• Essential</li> </ul>
Scheduling Appointments and Medication Refills	<ul style="list-style-type: none"> <li>• Parent/guardian</li> </ul>	<ul style="list-style-type: none"> <li>• Patient</li> </ul>
Adherence to Care	<ul style="list-style-type: none"> <li>• More frequent reminders and support from healthcare system</li> <li>• Healthcare provider has the duty to report non-adherence that rises to the level of medical neglect</li> </ul>	<ul style="list-style-type: none"> <li>• Expectation of self adherence - often no appointment reminders or follow-up from missed appointments</li> <li>• Healthcare provider has no reporting duty of non-adherence unless patient has a court-appointed guardian and is being neglected per provider opinion</li> </ul>

**Note:** The table was modified from Got Transition® website.<sup>5</sup>

## Neurofibromatosis Type 1 and Healthcare Transition

### Neurofibromatosis Type 1

Neurofibromatosis type 1 (NF1) is a relatively common genetic condition that affects approximately 1 in 2500–3000 people.<sup>18,19</sup> NF1 is caused by a heterozygous pathogenic variant in the NF1 gene (*NF1*). NF1 is highly variable within and between families and has complete penetrance. It is characterized by the presence of multiple café-au-lait macules (CALM), intertriginous freckling, cutaneous and subcutaneous neurofibromas, plexiform neurofibromas, bone abnormalities, Lisch nodules, and optic pathway gliomas. Additional complications may include malignancies, other central nervous system neoplasms, chronic pain, vasculopathy, hypertension, delayed or early puberty, learning disabilities, attention deficit with or without hyperactivity, and psychological disorders. Psychological concerns include impaired socialization, low self-esteem, poor interpretation of social cues, anxiety, and depression.<sup>20</sup> Complications of NF1 may cause serious disease morbidity, and in some cases, mortality. While pediatric and adult medical care guidelines and a genetic counseling practice resource exist for the care of individuals with NF1,<sup>21–23</sup> limited information has been published to date about the deficiencies, needs, and strategies specific to provide HCT for AYA with NF1.

## Healthcare Transition Readiness and Perceptions of Adolescents and Young Adults with NF1

Few studies have addressed transition preparation and perspectives in the NF1 population. A quantitative HCT study utilizing the validated Transition Readiness Assessment Questionnaire (TRAQ) in the NF1 population was published by Goetsch Weisman et al.<sup>24</sup> Transition readiness was found to be significantly lower for AYA with NF1 than in healthy peers, consistent with the HCT literature assessing transition readiness for AYA with special healthcare needs. In addition, the authors used the TRAQ as a model to develop an NF1-specific readiness questionnaire (NF1-TRAQ) and demonstrated a high correlation between TRAQ scores and NF1-TRAQ scores, indicating that increased transition readiness correlated with greater self-reported knowledge of NF1 and associated NF1 specific HCT skills. Overall, participants reported adequate knowledge of NF1 and comfort talking to medical providers, but discomfort with appointment keeping, insurance-related tasks, addressing NF1 medical emergencies, and discussing their diagnosis with other (non-NF1) medical providers and peers. The study also found that both TRAQ and NF1-TRAQ scores were lower in individuals who reported that their NF1 diagnosis had an impact on their education, career, and relationships, suggesting that decreased transition readiness is associated with a negative impact on young adult life. The results of this study emphasize the crucial need for education and mentoring throughout adolescence.

A qualitative study by Davidson<sup>25</sup> evaluated parent perspectives of adolescents with NF1. Consistent with HCT research for AYA with special healthcare needs, parents of adolescents with NF1 had considerable concern about adolescent ability to take responsibility for medical needs and independently manage their healthcare. Importantly, the study found a positive correlation between parent perspectives of a young adult's knowledge of NF1, involvement with healthcare management, and independence with self-management tasks. Similarly, Rietman et al<sup>26</sup> completed a qualitative assessment of the worries and needs of young adults and parents of young adults with NF1. Findings indicated that young adults with NF1 desire more information about their diagnosis and medical needs, access to and improved communication with NF1 expert clinicians, daily living support, mental health services, and socioeconomic participation. Parents also stressed the concern that they often remain in a coordination role for their adult child due to their child's lack of understanding and health-related self-management skills. Parents reported significant stress and concerns about the time they spend assisting their child with factors of young adult life, including medical management. Results of these studies emphasize the importance of parent participation in a patient's HCT, and that this partnership may include working together to make medical and life decisions.

The importance of continued follow-up and education after the transition period was emphasized in a study by Oates et al<sup>16</sup> in Australia, which revealed that many young adults with NF1 have poor access to healthcare, limited disease knowledge, and high NF1 complication rates. The authors found that for many young adults, the lack of ongoing healthcare and complication surveillance might explain the large number of new medical complications they identified, and suggests that lack of knowledge of associated risks and complications, cognitive deficits, and no identified NF1 care provider likely contributes to poor health surveillance and management.

## Quality of Life and Mental Health Issues in Adolescents and Young Adults with NF1

It is well established that NF1 can have a profound effect on quality of life (QoL) across all domains, including general physical health, bodily pain, mental health, activities of daily living, independence, and social functioning and relationships.<sup>27–33</sup> An important goal of HCT is to manage factors that may decrease QoL by increasing knowledge and building independence, as well as ensuring AYA have appropriate support in all facets of their lives.

Medical worries expressed by young adults with NF1 often relate to the unpredictable nature of the disease.<sup>26,34</sup> This includes fear of potential loss of function, future surgeries, aesthetic problems, malignancy risk, pain, and the visibility of their disease (Table 2).<sup>26</sup> Studies have consistently shown that visibility of disease is a major predictor of poor QoL and poor mental health among adults with NF1.<sup>27,30,35–37</sup>

Children and adolescents with NF1 experience more psychosocial problems and mental health issues compared to their healthy peers, such as anxiety, depression, inattention and impulsivity, internalization and externalization of problems, difficulties socializing, and poor adaptive behavior.<sup>38,39</sup> Similarly, adults with NF1 have high levels of

**Table 2** Potential Health Risks in Adults with Neurofibromatosis Type I

System	NF1 Manifestation
Malignancy/Tumors	<ul style="list-style-type: none"> <li>• Cutaneous neurofibromas</li> <li>• Plexiform neurofibromas</li> <li>• Malignant peripheral nerve sheath tumors (MPNST)</li> <li>• Breast cancer*</li> <li>• Pheochromocytoma</li> <li>• Gastrointestinal stromal tumors (GIST)</li> <li>• Non-optic pathway and low-grade gliomas</li> </ul>
Cardiovascular	<ul style="list-style-type: none"> <li>• Hypertension</li> <li>• Vasculopathy</li> </ul>
Skeletal	<ul style="list-style-type: none"> <li>• Scoliosis</li> <li>• Osteopenia/Osteoporosis</li> <li>• Vitamin D deficiency</li> </ul>
Pain	<ul style="list-style-type: none"> <li>• Headache/migraine</li> <li>• Tumor burden</li> <li>• MPNST</li> <li>• Glomus tumors</li> </ul>
Neurologic	<ul style="list-style-type: none"> <li>• Seizures/epilepsy</li> <li>• Neuropathy</li> <li>• Sleep disturbance</li> </ul>
Neurocognitive/Psychiatric	<ul style="list-style-type: none"> <li>• Depression/Anxiety</li> <li>• Stress</li> <li>• Reduced self-confidence</li> <li>• Learning disabilities</li> <li>• Executive functioning deficits</li> <li>• Attention deficit hyperactivity disorder</li> <li>• Cognitive impairment</li> <li>• Autism spectrum disorder</li> </ul>
Family Planning	<ul style="list-style-type: none"> <li>• Reproductive risks and options</li> <li>• Increased risk for antenatal and peripartum complications**</li> </ul>

**Notes:** Adults with Neurofibromatosis type I (NF1) are at increased risk to have numerous medical complications requiring patient education, screening, and/or surveillance.<sup>22</sup> \*Women with NF1 have an increased risk for early-onset breast cancer. The National Comprehensive Cancer Network provides recommendations for early breast screening.<sup>79</sup> \*\*Women with NF1 may be at risk for pregnancy and delivery complications.<sup>80</sup>

depression, anxiety, and stress.<sup>30,33,34,40,41</sup> It has been theorized that adults with NF1 may experience these mental health concerns due to social withdrawal and isolation,<sup>36</sup> as well as a lack of coping skills necessary to avoid sustained heightened emotional distress.<sup>33</sup> Psychosocial interventions to address distress have been designed for individuals with NF1, and preliminary results show improvements in both QoL and pain interference.<sup>42–44</sup>

NF1 can significantly impact social roles and relationships. Rietman et al<sup>26</sup> surveyed young adults with NF1 and found that participants commonly reported loneliness and trouble initiating and maintaining social and romantic relationships. Adults with NF1, particularly women, report greater insecurity and lower self-esteem and body image than their peers.<sup>26,45</sup> Kodra et al<sup>28</sup> found that approximately 40% of participants with NF1 in their study reported feeling embarrassed about their skin and Smith et al<sup>46</sup> found that 67% of women with NF1 in their study expressed an NF1-related appearance concern. Additionally, women with NF1 often report feeling less attractive and confident, more self-conscious about their bodies, and more dissatisfied sexually secondary to these feelings.<sup>45–47</sup> These concerns should be considered by healthcare providers when caring for adolescents and adults with NF1. The University of Alabama created

a resource for providers and patients regarding intimacy in NF1<sup>48</sup> which may be useful for AYA as they struggle through these insecurities.

Achieving independence with regard to education, employment, and social interactions can be a significant challenge for some young adults with NF1, particularly those with learning disabilities and/or cognitive dysfunction.<sup>26,27,36,49–51</sup> The AYA's perceptions about themselves, their familial and community support, and their access to mental health services may contribute to these challenges. In a review of data from the National Longitudinal Study of Adolescent Health, individuals with chronic illness were more likely to be unemployed, receive public benefits, and have a generally lower income than their peers without chronic illness.<sup>52</sup> This was similarly seen by Gregory et al<sup>51</sup> in an assessment of adults with NF1 in the Pacific Northwest region of the United States, in which participants were less likely than age-matched healthy peers to have private insurance, be employed, and have children. Thus, individuals with NF1 may require more support at school and in the workplace. Connecting AYA with NF1 to assistance programs, including disability service offices and vocational rehabilitation, can allow individuals to receive appropriate accommodations in educational and work environments. This may potentially increase labor force participation and decrease barriers to successful employment.<sup>50</sup>

NF1 is an autosomal dominant condition with a 50% risk of transition to offspring which can significantly impact family planning decisions. While most individuals with NF1 report being interested in having biological children without NF1, young adults generally want more information and education about genetics and reproduction, including alternative family planning options.<sup>24,26</sup> A practice resource by the National Society of Genetic Counselors provides guidance to clinicians regarding family planning discussions, as well as general psychosocial topics that may affect QoL in individuals with NF1.<sup>23</sup>

Although there are few studies linking the two, the potential negative impact of NF1 on QoL and mental health among AYA highlights the importance of a structured HCT program, with multidisciplinary expert care before, during, and after the formal HCT.<sup>26</sup> HCT strategies can be designed to address these concerns, including facilitation of the development of health and non-health related self-management skills, psychosocial support and referrals, connection with resources to obtain accommodations at school and work, and access to genetic counseling for family planning.

## Components of a Healthcare Transition Program and Associated Challenges

### Healthcare Transition Models

Best practices for transition have been established, and several HCT models exist.<sup>53–55</sup> Regardless of the model utilized to implement a formal HCT program, a structured transition intervention results in more positive outcomes.<sup>56,57</sup>

The most widely used model is available through Got Transition (GT),<sup>5</sup> a federally funded resource center with an aim to improve the transition from pediatric to adult healthcare. GT provides several tools and resources for providers, patients, and families. The Six Core Elements of Transition<sup>58</sup> were created for clinicians to guide transition programs and interventions. These Core Elements align with approaches used by numerous professional organizations including the American Academy of Pediatrics (AAP), with the endorsement of the American Academy of Family Physicians (AAFP) and the American College of Physicians (ACP).<sup>55</sup>

Often, healthcare providers consider transition as a discrete event, such as the actual transfer of care. The concept of the Six Core Elements presents transition as a process, starting in early adolescence and continuing into young adulthood. There are three separate guides provided by GT that are specific to the type of practice facilitating the transition, including whether the AYA changes clinicians, continues to follow with the same provider but transitions to an adult model of care, or integrates into adult health care. The elements of transition highlighted below focus on transitioning AYA to a new adult healthcare provider and provides a framework to apply these strategies to NF1 patients.

The first Core Element of Got Transition's Six Core Elements is *Transition and Care Policy/Guide*. If a transition guide is not already established, the policy ideally should be crafted with input from patients and families, and be written at an appropriate literacy level. Policies often provide patients and families with helpful anticipatory guidance and indicate how the practitioner will support the patient and their family during the transition process. A transition policy

often includes age ranges that can allow an early introduction of the concept of transition to patients and their families, ideally around 12–14 years of age. Once developed, the transition policy can be communicated to families in different ways such as: during a clinic visit, a mailing, or posted on a program's website. The policy can be reviewed yearly with patients and families. The policy should also be clearly communicated to program staff. Several examples of transition guides are available on the GT website and can be easily modified for NF1 and the specific clinic setting.

The second Core Element is *Tracking and Monitoring*. This involves defining criteria to identify youth requiring HCT assistance and tracking the receipt of the HCT program. This is also applicable to AYA with NF1 with tracking and receipt of a structured HCT program to support the best outcomes and patient/family support and communication. It is not unusual to have multiple specialties involved in caring for NF1 patients. A shared HCT tracking system allows each clinician to monitor the patient as they progress through transition. Electronic Medical Record (EMR) systems often have customizable ways to track and monitor patient history, and can be applied to HCT. Other databases can also be helpful in tracking and monitoring transition progress.

The third Core Element is *Transition Readiness*. This can be evaluated by regular assessments using the validated TRAQ or other readiness measures such as the NF1-TRAQ, which is specific to NF1 transition readiness.<sup>11,24</sup> The tools should be used at regular intervals to monitor transition readiness over time,<sup>2</sup> as the information can guide the clinician's approach to the education, anticipatory guidance, and counseling of AYA with NF1. This is an important component of the HCT process leading to positive perceptions of readiness and future success.<sup>9,59</sup> Lemke et al<sup>60</sup> found that AYA with chronic illnesses who received care coordination support through an HCT program were more than two times as likely to report receiving the care they thought they needed and to speak to their providers about future care than those who did not receive care coordination support.

The fourth Core Element is *Transition Planning*. This includes readying the patient and family for the eventual shift to adult models of care, including education about the differences in the culture between pediatric care and adult care, timing of the transfer of care, identification of adult providers, and discussion of a patient's goals and priorities. During this planning stage, clinicians should develop a medical history summary and emergency care plan for the patient. Additionally, healthcare providers might provide a release of medical information, legal documents for guardianship if applicable, and insurance options and resources. Individuals with NF1 should be well informed of these important aspects of transition planning.

The fifth Core Element is *Transfer of Care* to an adult counterpart. Introducing the concept of HCT early prepares patients and families for their eventual transition to an adult provider or model of health care. It is important to develop relationships with adult providers and be willing to serve as a continued resource should an adult colleague need consultation. A "warm handoff" between providers either by phone, electronic message, or joint telehealth visit, are all ways to facilitate this transfer. Assistance locating an NF provider is available on NF organization websites,<sup>61,62</sup> however, there may not be an identified clinic in the area that provides comprehensive care for adults with NF1. In this case, it is advisable for the pediatric provider to identify and educate a local provider or subspecialist, such as a neurologist or neuro-oncologist, to ensure appropriate care for adults with NF1. This often includes forwarding existing NF1 resources, guidelines and practice resources.<sup>22,23</sup>

The sixth and final Core Element is *Transfer Completion*. This involves communication with both the patient/caregiver and adult providers, as well as between the pediatric and adult providers, to be sure the transfer has been successfully executed. Although this step can be time intensive, it is especially important for NF1 patients who may not feel comfortable with navigating a new healthcare model and either request to return to the pediatric clinic, or are lost to follow-up and therefore not receiving the comprehensive medical care and support needed.

In addition to NF1-specific HCT, the transition program needs to consider and address other life transitions common in the AYA period, including the transition to higher education and career, social and romantic relationships, and family planning. Transition readiness and anticipatory guidance are critical to consider as individuals are guided to prepare to take care of themselves and live independently.

There are several additional considerations of the transition process including recognition of barriers to successful HCT, identification of high-risk individuals, patient and provider education of HCT and NF1, and billing and reimbursement for transition services.

## Barriers to Successful NF1 Healthcare Transition

Many of the challenges of an effective NF1 HCT are the same as barriers in the general transition process. In order to start and maintain a successful HCT program, identification and acknowledgment of potential barriers are necessary. Several barriers to effective HCT have been identified (Table 3) and include limited or lack of insurance coverage, income/financial concerns, lack of service coordination, lack of support from ancillary staff (eg nursing, social work, and case management), healthcare culture differences, work involved in transferring and accessing medical records, difficulty obtaining reimbursement for extended clinic appointments, lack of provider knowledge and comfort caring for individuals with special healthcare needs, poor communication between the healthcare provider and family, lack of communication and anticipatory guidance about the HCT process, and family stress and comfort with medical complexities.<sup>2,3,26,63,64</sup>

Notably, Gray et al<sup>65</sup> found that a prominent barrier to HCT reported by patients and families is relationships. Patients are often reluctant to leave their pediatric care team and have difficulty accessing knowledgeable adult providers whom they trust with their concerns. Further, HCT requires committed parental involvement. In a review of parent experiences with HCT, Ellison et al<sup>66</sup> found that barriers to parental facilitation of HCT include concerns surrounding relationship loss, loss of parental role, lack of knowledge and/or skills, and concerns related to the healthcare system in general. Similarly, Rietman et al<sup>26</sup> found that parents of young adults with NF1 reported significant stress regarding their now adult child's medical care. Participants reported that providers do not acknowledge the worries of parents, expressed confusion about their parental role, and felt a decrease in the quality of their parent-child relationship.

**Table 3** Barriers to Effective Healthcare Transition

<b>Patient and Family Barriers</b>
<ul style="list-style-type: none"> <li>• Lack of knowledge of condition and importance of medical care</li> <li>• Difficulty finding healthcare provider with knowledge of condition</li> <li>• Stress or emotional burden of condition or medical complications</li> <li>• Limitations in decision-making abilities or self-advocacy</li> <li>• Financial concerns (income, insurance coverage)</li> <li>• Feeling of loss of parental/caretaker role</li> <li>• Discomfort with parental/caretaker transfer of responsibility</li> <li>• Psychosocial and developmental transitions common to all adolescents and young adults</li> </ul>
<b>Shared Patient/Provider Barriers</b>
<ul style="list-style-type: none"> <li>• Poor communication</li> <li>• Lack of trust</li> <li>• Difficulty in establishing new relationships</li> <li>• Cultural differences</li> </ul>
<b>Provider and Healthcare System Barriers</b>
<ul style="list-style-type: none"> <li>• Lack of knowledge or skills in patient transition needs</li> <li>• Insufficient information about the condition, management, and treatment</li> <li>• Poor communication about the transition process</li> <li>• Lack of available services and coordination of care and follow up</li> <li>• Shortage of extended clinic slots</li> <li>• Poor access to support staff (nursing, care coordinators, social workers)</li> <li>• Consultation and communication shortcomings between pediatric and adult providers</li> <li>• Low reimbursement rates and lack of knowledge in transition services billing</li> <li>• Process of transferring medical records</li> <li>• Differences in pediatric vs adult care models</li> </ul>

**Note:** Several potential barriers may interfere with the transition process.<sup>2,3,26,54,63-67</sup>

Of additional importance, for individuals with NF1 and an intellectual disability, there may be limitations in autonomy and self-management. This typically requires an assessment of decision-making abilities to determine the capacity for self-advocacy.<sup>67</sup> In these situations, parents or guardians play a significant role in the transition process and may continue as the primary caregiver.

## Identification of High-Risk Patients

Individuals at increased risk to have challenges with the HCT process may need additional support and services before, during, and after the transition to adult healthcare. The National Survey of Children's Health data details that AYA with special healthcare needs are more likely to live in poverty, be non-Hispanic Black, and have public insurance than their generally healthy peers.<sup>68,69</sup> And, while it has generally been shown that AYA with special healthcare needs are not receiving HCT counseling at high rates, it is worse for minority patients, with reports of Hispanic children, younger teens, and non-Hispanic Black children being counseled at lower rates.<sup>70</sup> In addition to receiving less counseling, there are significant disparities with regard to race, age, and disability and the availability of transition services.<sup>71</sup> Although providers may self-report no bias against people with disabilities, an overwhelming majority had implicit biases in a study by VanPuymbrouck et al.<sup>72</sup> Low health literacy has also been linked to increased emergency department (ED) utilization and lower levels of transition readiness on the TRAQ.<sup>73</sup> Especially concerning for individuals with NF1, patients who receive care based solely during ED visits typically do not receive the comprehensive and knowledgeable care that an experienced clinician in a typical healthcare setting can provide. This can lead to medical mismanagement, lack of a consistent medical plan with poor adherence to health maintenance screenings, deficiencies in patient education and counseling, and higher healthcare costs. HCT for those with low health literacy and NF1 may be further complicated by associated diagnoses of associated psychiatric diagnoses, cognitive impairment, and executive function deficits.<sup>16,17,38,39</sup> Autism spectrum disorder in particular has been found to be associated with the lowest levels of transition readiness compared to other chronic conditions,<sup>74</sup> an important consideration given the increased incidence of autism among individuals with NF1.

Lausdahl et al<sup>17</sup> from Denmark surveyed individuals with NF1 and found that those who self-reported the most difficulty with the HCT process had psychiatric diagnoses, executive function deficits, and/or special educational needs, emphasizing the importance of multidisciplinary collaboration and support to facilitate the effective transition for these individuals. Specialists involved in the HCT process for AYA with NF1 may need to include medical providers, neuropsychologists, mental health providers, social workers, therapists, and education liaisons.

Access to medical insurance that will ensure continued care is yet another challenge that AYA with special healthcare needs experience. Medicaid expansion and the Affordable Care Act have greatly benefitted youth with disabilities,<sup>75,76</sup> including those with NF1, however, concerns remain that there may be delayed care or forgoing of care completely because of changes in the usual provision of healthcare leading to poor health outcomes into adulthood.<sup>77</sup> Disabled youth with chronic medical conditions were also shown to be at increased risk of losing health insurance, both public and private, than their non-disabled peers.<sup>78</sup> Thus, as part of the HCT program, providers should identify those NF1 patients who may be at greater risk to have challenges with the HCT process, including lack of appropriate insurance coverage, and need for additional support and guidance.

## NF1 Patient Education

Healthcare transition programs should address the changing medical needs through adulthood and provide applicable resources to the family. In a study assessing worries and needs of adults and parents of adults with NF1, Rietman et al<sup>26</sup> found a common false expectation among participants that care needs decrease in adulthood. However, NF1 is a progressive condition with significant risk for disease morbidity in adulthood (Table 2). Education about adult health risks and the importance of medical care management and screenings is a vital component of an NF1 HCT plan, with particular attention to cancer risk and surveillance, cardiovascular complications, bone health, pain management, neurologic complications, neurocognitive functioning, mental health, and reproductive risks and options.<sup>22,23,79,80</sup> Table 4 and Table 5 provide several patient resources about the HCT process and information about NF1 for providers to share with their patients for ongoing education and support. Additional priorities of patient education should address medication management, appointment keeping, tracking health issues, identifying medical emergencies, understanding the insurance system, and talking with providers; all of which can be assessed using the validated TRAQ.<sup>11</sup>

**Table 4** General Transition Tools and Resources

Tool	Access	Audience
<b>Got Transition</b> Federally funded national resource center on HCT Patient and provider information and tools available	<a href="http://www.gottransition.org">www.gottransition.org</a>	Patient and Provider English and Spanish
<b>Payment for Transition Services</b> <i>Got Transition</i> Payment approaches to implement recommended HCT services, including fee-for-service coding and reimbursement options and value-based payment options	<a href="https://www.gottransition.org/six-core-elements/payment.cfm">https://www.gottransition.org/six-core-elements/payment.cfm</a>	Provider English
<b>Transition Readiness Assessment Questionnaire (TRAQ)</b> <i>Wood, Sawicki, Reiss, Livingood &amp; Kraemer (2014)</i> <i>East Tennessee State University</i> Validated tool to assess transition readiness	<a href="https://www.etsu.edu/com/pediatrics/traq/">https://www.etsu.edu/com/pediatrics/traq/</a>	Patient/Caregiver English, Spanish, and 8 other languages
<b>STARx Questionnaire</b> <i>University of North Carolina School of Medicine</i> Validated tools to assess transition readiness Three versions for pediatric patients, young adult patients, and parents	<a href="https://www.med.unc.edu/transition/transition-tools/trxansion-scale/">https://www.med.unc.edu/transition/transition-tools/trxansion-scale/</a>	Patient/Caregiver English and Spanish (only English for parent version)
<b>Healthcare Transition Research Consortium (HCTRC)</b> Organization supporting evidence-based HCT as a standard component of health services	<a href="https://sites.google.com/site/healthcaretransition/">https://sites.google.com/site/healthcaretransition/</a>	Provider English

**Note:** Several transition tools and resources exist for patients, families, educators, and providers.

Information about the genetic etiology of NF1 and risks to offspring is another important educational component of NF1 HCT. While most individuals with NF1 report being interested in having biological children without NF1, young adults generally want more information and education about genetics and reproduction, including alternative family planning options.<sup>23,24,26</sup>

## Provider Knowledge of the Healthcare Transition Process and NFI

Healthcare providers are tasked with helping patients and families navigate and address barriers of the HCT process.<sup>55,81</sup> However, multiple studies have demonstrated that the majority of practicing providers do not feel prepared to help AYA patients through HCT or to provide adult care to these patients.<sup>82–85</sup> Adult primary care doctors who are comfortable caring for complex patients are also in short supply, and these physicians report needing more support and training to care for this population.<sup>86,87</sup> A survey by Patel et al<sup>87</sup> compared comfort in the transfer of patients with chronic childhood disease to adult-oriented health care between internal medicine and pediatric residents. The authors reported a higher level of comfort among the pediatric trainees and suggested that training should be included in internal medicine programs. Becoming familiar with the HCT process as well as gaining knowledge and experience with NF1 can help build confidence in the HCT of AYA with NF1.

## Billing and Reimbursement

The lack of reimbursement for the billing and care of AYA with special healthcare needs is a hurdle faced by healthcare providers.<sup>55</sup> GT has numerous resources and research on reimbursement for providers to use and consider along with recommendations and the argument for value-based payment models.<sup>88</sup> Healthcare providers should consider contacting their internal billing department to review the process of billing for services such as care coordination and readiness assessment so that transition services can be appropriately reimbursed.

**Table 5** NF1-Specific Transition Tools and Resources

Tool	Access	Audience
<b>Just for Teens: Living with NF1</b> <i>Children's Tumor Foundation</i>	<a href="https://www.ctf.org/images/uploads/resources/NF-Just-For-Teens_Living-with-NF.pdf">https://www.ctf.org/images/uploads/resources/NF-Just-For-Teens_Living-with-NF.pdf</a>	Patient English and Spanish
<b>Living with Neurofibromatosis: A Guide for Teens and Young Adults</b> <i>Children's Hospital St. Louis, Washington University in St. Louis</i>	<a href="https://nfcenter.wustl.edu/wp-content/uploads/2011/06/living-with-NF-clean.pdf">https://nfcenter.wustl.edu/wp-content/uploads/2011/06/living-with-NF-clean.pdf</a>	Patient English
<b>Transitioning to Adulthood: A Guide for Individuals with NF1</b> <i>Children's Hospital St. Louis, Washington University in St. Louis</i>	<a href="https://nfcenter.wustl.edu/wp-content/uploads/2011/06/Transitioning-to-Adulthood-Brochure.pdf">https://nfcenter.wustl.edu/wp-content/uploads/2011/06/Transitioning-to-Adulthood-Brochure.pdf</a>	Patient English
<b>Talking about Neurofibromatosis: A Guide for Teens</b> <i>Children's Hospital St. Louis, Washington University in St. Louis</i>	<a href="https://nfcenter.wustl.edu/wp-content/uploads/2010/10/SLC6906_NeurofibromatosisTeenBrochureRI.pdf">https://nfcenter.wustl.edu/wp-content/uploads/2010/10/SLC6906_NeurofibromatosisTeenBrochureRI.pdf</a>	Patient English
<b>The Guide for NF Patients and Caregivers Transitioning (PACT) to Adult Care</b> <i>NF Collective</i>	<a href="https://nfcollective.org/uploads/general/Transitioning_to_Adult_Care_1-19b.pdf">https://nfcollective.org/uploads/general/Transitioning_to_Adult_Care_1-19b.pdf</a>	Patient English
<b>Living with Neurofibromatosis type 1: A Guide for Adults</b> <i>Children's Tumor Foundation</i>	<a href="https://www.ctf.org/images/uploads/documents/CTF-Adults_with_NF_Brochure_2022.v10_FINAL_.pdf">https://www.ctf.org/images/uploads/documents/CTF-Adults_with_NF_Brochure_2022.v10_FINAL_.pdf</a>	Patient English and Spanish
<b>Living with Neurofibromatosis type 1: A Guide for Adults</b> <i>Children's Hospital St. Louis, Washington University in St. Louis</i>	<a href="https://nfcenter.wustl.edu/wp-content/uploads/2012/04/Adult-NF-Brochure.pdf">https://nfcenter.wustl.edu/wp-content/uploads/2012/04/Adult-NF-Brochure.pdf</a>	Patient English
<b>Telling People you have Neurofibromatosis Type 1</b> <i>Children's Hospital St. Louis, Washington University in St. Louis</i>	<a href="https://nfcenter.wustl.edu/wp-content/uploads/2011/10/Disclosure-Brochure.pdf">https://nfcenter.wustl.edu/wp-content/uploads/2011/10/Disclosure-Brochure.pdf</a>	Patient English
<b>Preparing for Your Clinic Visit</b> <i>Children's Tumor Foundation</i>	<a href="https://www.ctf.org/images/uploads/documents/Preparing_for_Your_Clinic_Visit.pdf">https://www.ctf.org/images/uploads/documents/Preparing_for_Your_Clinic_Visit.pdf</a>	Patient English
<b>Reproductive Options for People with NF1, NF2, and Schwannomatosis</b> <i>Neurofibromatosis Network</i>	<a href="https://nfnetwork.org/data/uploads/nf1-educational-materials/reproductive-options-for-people-with-nf.pdf">https://nfnetwork.org/data/uploads/nf1-educational-materials/reproductive-options-for-people-with-nf.pdf</a>	Patient English
<b>Intimacy and NF1: A Resource Exploring the Impact of NF1 on Intimacy and Relationships</b> <i>The University of Alabama at Birmingham</i>	<a href="https://sites.uab.edu/nf1intimacy/">https://sites.uab.edu/nf1intimacy/</a>	Patient English
<b>Neurofibromatosis type 1 - Transition Readiness Assessment Questionnaire (NF1-TRAQ)</b> <i>A non-validated tool to assess NF1-specific components of transition readiness.<sup>24</sup></i>	<a href="https://pubmed.ncbi.nlm.nih.gov/32969737/">https://pubmed.ncbi.nlm.nih.gov/32969737/</a>	Provider English

**Note:** Several transition tools and resources exist for patients, families, educators, and providers.

## Healthcare Transition and NF1 Resources and Support

Many tools and resources to facilitate effective transition are available; some apply to general healthcare transition (Table 4), while others are NF1-specific (Table 5). Formal transition programs which are often part of large pediatric medical centers are also excellent referral sites for transitioning patients.

In addition, several organizations exist that provide support and education to patients and families impacted by NF1, advocate for NF1 awareness and improvement in care, and promote NF1 research and clinical trials (Table 6). The organizations also provide a list of NF1 providers throughout the US and Canada,<sup>61,62</sup> including many comprehensive clinics that are part of the Children's Tumor Foundation NF Clinic Network, a network established in 2007 to improve clinical care and support best practices for those living with NF1.<sup>89</sup>

**Table 6** Support and Advocacy Organizations for Neurofibromatosis Type I (NF1)

Organization	Access
B The Difference, The Brandon Merritt Charitable Foundation	<a href="http://www.bthedifference.org">www.bthedifference.org</a>
Children's Tumor Foundation	<a href="http://www.ctf.org">www.ctf.org</a>
Littlest Tumor Foundation	<a href="http://www.littlesttumor.org">www.littlesttumor.org</a>
Neurofibromatosis Midwest	<a href="http://www.nfmidwest.org">www.nfmidwest.org</a>
Neurofibromatosis Network - regional groups include: <ul style="list-style-type: none"> <li>• NF Arizona/Southwest</li> <li>• NF California</li> <li>• NF Central Plains</li> <li>• NF North Central</li> <li>• NF Northeast</li> <li>• NF Michigan</li> </ul>	<a href="http://www.nfnetwork.org">www.nfnetwork.org</a> <ul style="list-style-type: none"> <li>• <a href="http://www.nfnetwork.org">www.nfnetwork.org</a></li> <li>• <a href="http://www.nfcalifornia.org/">www.nfcalifornia.org/</a></li> <li>• <a href="http://www.nfcentralplains.org/">www.nfcentralplains.org/</a></li> <li>• <a href="http://www.nfnorthcentral.nfnetwork.org/">www.nfnorthcentral.nfnetwork.org/</a></li> <li>• <a href="http://www.nfnortheast.org">www.nfnortheast.org</a></li> <li>• <a href="http://www.nfmich.org/">www.nfmich.org/</a></li> </ul>
NF Collective	<a href="http://www.nfcollective.org">www.nfcollective.org</a>
NF Team Foundation	<a href="http://www.thenteam.org">www.thenteam.org</a>
Texas Neurofibromatosis Foundation	<a href="http://www.texasnf.org">www.texasnf.org</a>

**Notes:** Several organizations offer information and support for individuals and families affected by NF1. Services provided may include local or regional support and activities, scholarships and funding, and fundraising for NF awareness and research.

## Conclusion

NF1 is a complex multisystemic disorder with a variety of manifestations including a predisposition to tumor development, potential for disease-associated morbidity, and in some cases, cognitive or mental health impairments. A misconception among AYA individuals with NF1 is that their NF1-related health concerns may decrease as they age, rather than NF1 being a progressive condition with additional risks of adult medical complications. A committed clinician and a structured HCT program are necessary to effectively transition AYA with NF1 from a pediatric to an adult care model with the goal of building independence, improving quality of life, and decreasing associated medical complications. This review aimed to introduce clinicians to the concept of formal HCT programming for AYA with NF1 and provide valuable resources for program implementation and patient and provider education.

## Future Directions

Although the need for effective HCT programs and interventions is well recognized, there is a lack of rigorous evidence-based longitudinal studies evaluating interventions, especially in AYA with special healthcare needs. There is a national research agenda currently proposed by Okumura et al<sup>90</sup> that addresses three key areas of interest including the optimal development and implementation of HCT service models, evaluation and outcome measures for HCT, and fiscal policies to incentivize the HCT process and continuity of care. Further research is needed in these areas which can be used as a model for the care of AYA with NF1 in order to best implement HCT services in this population.

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