Challenges in the Transition From Pediatric to Adult Healthcare Faced by Patients With Neurofibromatosis Type 1

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This white paper reviews challenges and unmet needs in patients with neurofibromatosis type 1 (with a focus on those with plexiform neurofibromas [NF1-PN] where applicable) as they transition from pediatric to adult healthcare. It includes important insights from experts in NF1—including healthcare practitioners, patients, and patient advocacy group members—who were convened to provide their perspectives. The white paper summarizes the main topics from that discussion, as well as results from a premeeting survey sent to participants. Call-out text boxes are not direct participant quotes but are paraphrased from the live discussion.

Challenges in the Transition of Patients with NF1 from Pediatric to Adult Care

A roundtable discussion explored the challenges that patients with NF1 face when transitioning from pediatric to adult healthcare, focusing on four key areas and recommending potential solutions.

Attendees included healthcare providers (HCPs), adult patients (>18 years of age) with NF1, and patient advocates from US-based national NF1 organizations.

1

Improving patient access and care

Recommendations

- More NF1 clinics and/or specialists who understand how to treat NF1
- Guidelines that establish standards of care
- Dedicated contacts with insurance companies for patients and HCPs
- Improved access to electronic medical records for patients and HCPs

2

Reducing the number of patients lost during transition

Recommendations

- Early transition support and education for young patients (as early as 12 years of age)
- Mental health care/support for patients
- Funding for care coordinators in HCP offices for adult patients
- Communication between pediatric HCPs/care coordinators and their adult care counterparts

3

Preventing/decreasing gaps in treatment during transition

Recommendations

- Patient assistance regarding insurance options when aging out of parents'/caregivers' coverage
- Approval of drug treatments for adult patients with NF1-PN
- Communication between HCPs and working patients regarding time commitments for care visits

4

Educating HCPs and patients/caregivers about NF1

Recommendations

- Easy-to-use, up-to-date online resources and simple, straightforward printed materials for patients
- Early and repeated education of young patients on the why's and how's of the transition of care process
- Patient education on the importance of maintaining preventative healthcare throughout adulthood
- Improved education for adult care HCPs on NF1 and stronger connections with their pediatric counterparts

Discussions on NF1 should continue between various groups—including HCPs, patients, advocacy groups, insurance representatives, government representatives, and pharmaceutical companies—to improve the patient journey from childhood and beyond

Introduction

Neurofibromatosis type 1 (NF1) is a genetic condition affecting 1 in 3,000-3,500 people around the world.[1,2] Patients with NF1 are at risk of developing a variety of tumors, including neurofibromas, which occur on the peripheral nerves.[1,2] NF1 may negatively affect one's emotional, physical, and social functioning due to pain and complications from tumor growth, such as neurologic deficits, visual/hearing impairments, difficulties with speech or mobility, and changes in one's physical appearance.[1,3,4] Plexiform neurofibromas (PNs) are common types of benign tumors that grow along nerve fibers, affecting 30%-60% of patients with NF1.[3] NF1-PN is a rare, complex disease with unpredictable progression, variable severity, and a wide range of signs, symptoms, and comorbidities.[1,5] Management of NF1-PN primarily consists of the treatment of pain, surgical intervention, and/or medical therapy; treatments include mitogen-activated protein kinase kinase inhibitors (MEKi) to reduce tumor volume.[3,5-7]

Most patients with NF1 are diagnosed in childhood[8] and require lifelong healthcare from a multidisciplinary team that can include specialty areas, such as oncology, neurology, neurosurgery, neuropsychology, medical genetics, ophthalmology, dermatology, orthopedics, plastic surgery, general surgery, and cardiology.[1,5]

Transitioning out of pediatric care and into adult healthcare can be challenging for patients without proper education and guidance. According to the National Survey of Children's Health, only 18% of all children aged 12-18 years in the US in 2021-2022 received the services needed for transitioning to adult healthcare.[9] Adolescent and young adult patients with chronic conditions, such as NF1, can face additional challenges when they transition such as limited knowledge of their condition. They may also have lack of adequate education regarding the transition process and limited healthcare access, which can contribute to patients dropping out of the healthcare system.[10-13]

The purpose of this report is to discuss the challenges of the transition of care (ToC) from pediatric to adult care and maintenance of continuity of care through adulthood in patients with NF1, with a focus on *actionable challenges* that have potential solutions. These actionable challenges can lay the foundation for future discussions to establish a roadmap of solutions to improve patient care.

Methods

This report was developed through insights derived from multiple stakeholders (5 healthcare providers [HCPs], 2 adult patients [aged >18 years], and 3 national patient advocacy group members) who have knowledge of and experience with NF1 and the ToC process. Insights were captured via premeeting surveys and a virtual expert roundtable discussion held in October 2024. The HCP specialties were pediatric oncology (n=2), pediatric neurology (n=1), and adult neurology/neuro-oncology (n=2). Participants also included patient advocacy group members (NF Network; Children's Tumor Foundation), who may also be caregivers to patients with NF1. Participants discussed the current challenges of ToC specific to NF1, with a focus on NF1-PN when applicable. Discussions were focused on actionable challenges that fell under 4 key topics (**Figure 1**).

Abbreviations

CT, computed tomography
EMR, electronic medical records
FDA, Food and Drug Administration
HCP, healthcare provider
MEKi, mitogen-activated protein kinase kinase inhibitors
MRI, magnetic resonance imaging
NF1, neurofibromatosis type 1
PN, plexiform neurofibroma
ToC, transition of care

Figure 1. Roundtable topics of discussion

1. Optimal patient access/care
2. Patient retention
3. Treatment continuity
4. Education (HCP/patient/caregiver)

1. Optimal Patient Access and Care

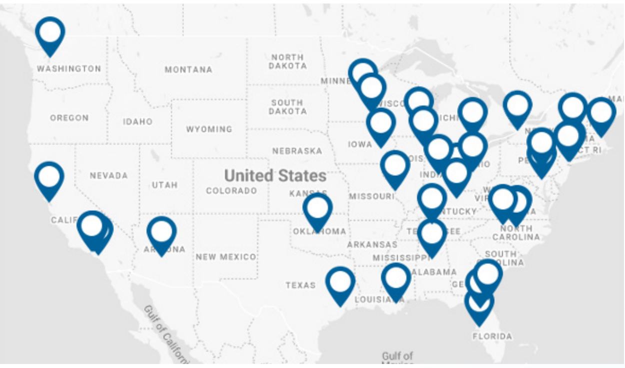
In the transition to adult multidisciplinary care, there are many challenges faced by patients with NF1 and the healthcare providers (HCPs) who treat them. A published survey of patients with NF1 found that only half of pediatric patients and around a third of adult patients had attended an NF clinic (clinics that provide comprehensive medical care to patients with NF1) in the preceding 3 years.[14] Major barriers to visiting an NF clinic for medical appointments were lack of insurance and long travel distances due to the limited numbers of NF1 clinics, especially for adult patients (**Figure 2**).[14] There is also a lack of adult-care HCPs who have knowledge of or specialize in treating NF1.[11]

> *West of the Mississippi River is an NF desert in terms of clinics. That geographic access really makes a difference, especially once patients graduate to adult care and have a job that won't let them get away.*
>
> – NF Network and NF North Central Patient Advocacy Group member

Figure 2. Clinics treating patients with NF1 in the US[15]

62 Clinics Treating Pediatric Patients (52 with PN specialty)

36 Clinics Treating Adult Patients (31 with PN specialty)



Clinics recognized by the NF Clinic Network as providing comprehensive medical care to individuals with NF.

Insurance and travel

Roundtable attendees agreed that insurance and geography/travel are serious limitations to a patient's access to care. Insurance may require justification as to why a patient would need to see an out-of-network HCP at an NF clinic rather than an in-network HCP elsewhere who may not be familiar with NF1. Some patients may find that insurance will cover an initial out-of-network visit to an NF clinic/specialist but not follow-up visits. Many patients must travel long distances to visit one of the limited numbers of NF clinics (**Figure 2**) or see specialists with expertise in NF. The 2 patients who attended the roundtable noted having to travel more than 100 miles to see their primary NF HCP. HCP attendees noted that patients sometimes need to travel out of state to see HCPs or specialists; however, Medicaid and some insurance companies do not cover out-of-state care, leaving patients with limited or no access to HCPs who have training and knowledge in treating NF1.

Clinical management guidelines

Another challenge is that clinical practice is regularly evolving, but insurance guidelines are not developed at a matching pace. An example is the current practice of using MRI (regional or whole-body) for baseline screening and surveillance of PNs in pediatric/adolescent patients with NF1.[3,16,17] Similar guidelines have not yet been established for PN surveillance in adult patients with NF1, which may affect their insurance approval and coverage of such scans. Roundtable attendees stated that there is a need for evidence-based management guidelines to establish standards of care (particularly in adult patients) that will demonstrate to insurance companies the importance of supporting specialized care, including tests, imaging, and treatments.

Telehealth

Telehealth in the management of NF1 was also discussed. Virtual visits are a potential way to improve patient access[18] for those who live far from an NF clinic or HCP, live in rural areas, and/or cannot take time off from work or school. There was a general consensus among roundtable attendees that once an initial in-person visit has occurred, telehealth can be a useful tool to reduce the cost and time required for travel to follow-up visits (**Figure 3**), allowing patients to see an HCP who has expertise in NF to whom they otherwise would not have access. Virtual visits, however, were not considered useful for certain types of signs or symptoms that need to be evaluated in person (eg, new neurologic symptoms, new growth of existing tumors, new tumor development, etc). In addition, there is a need for government support (eg, Congress and Medicaid) to continue telehealth use in this patient population and facilitate telehealth access. Overall, it is important to advocate for telehealth in care maintenance, but virtual visits alone are not sufficient to provide comprehensive care to patients with NF1, and patients must learn when virtual visits are appropriate.

Figure 3. Premeeting survey results regarding telehealth

Electronic medical records

The role of electronic medical records (EMRs) in the care of patients with NF1 was the final topic covered by roundtable attendees in this section. EMRs are considered critical for both patients and HCPs to improve healthcare delivery[18] and to avoid duplication of care, especially for patients who require a multidisciplinary team of HCPs. Only 1 of the 2 patient roundtable attendees noted having access to their EMRs. HCPs expressed concerns that even when they have access to EMRs, imaging records (eg, MRIs, CT scans, etc) are not always available, with only a radiological report available instead. Additionally, there can be multiple EMR databases rather than a central source, which can make finding/accessing the needed information difficult. Although challenging in our current healthcare system, a centralized source for EMRs that interact with each other and contain all patient records, including imaging, would help optimize healthcare delivery.

Actionable Challenges for Optimal Patient Access and Care

Recommendations

- More NF1 clinics and/or specialists with expertise in NF1
- Evidence-based guidelines that establish standards of care
- Dedicated insurance liaisons working with pediatric and adult care teams
- Improved access to EMR for patients and HCPs

2. Patient Retention

Many patients with NF1 believe that their NF1-related healthcare needs will decrease as they age, while in reality, their care needs may increase. There are increased health risks that come with age in patients with NF1, such as malignant tumors, hypertension, osteoporosis, and neuropathy; additionally, patients who wish to start a family may encounter reproductive risks or complications. Finally, in patients with NF1-PN, tumor progression and associated healthcare needs can be unpredictable.[13]

In the premeeting survey, patient roundtable attendees noted several challenges they have encountered in maintaining their healthcare into adulthood, such as trouble finding or retaining an HCP knowledgeable/experienced with treating NF1 and insurance coverage (**Figure 4**). Furthermore, the majority of the HCP attendees estimated that they had lost up to a quarter of patients with NF1 in the ToC process in the past 5 years (**Figure 5**).

Figure 4. Roundtable pre-meeting survey results from patients

What is the biggest challenge for you in maintaining your care in adulthood?

- **Current HCPs retiring**
- **Insurance coverage**
- **Can't find an HCP with NF1 expertise**

Figure 5. Roundtable premeeting survey results from HCPs

18–26 y →	Age at which their patients with NF1 typically transition to adult care
11%–25% →	Percent of patients with NF1 estimated lost to ToC in the past 5 years by the majority of HCP attendees

Early and slow transition

HCP roundtable attendees noted that treatment is more challenging with adult patients who drop out of care and only return after new or severe symptoms emerge, as it may be difficult to diagnose quickly what is causing these symptoms. Thus, maintaining multidisciplinary care throughout adulthood is a necessity for preventive care. Roundtable attendees proposed that one way to improve retention is to start the transition to adult care during late adolescence, with patients being involved in taking ownership of their care throughout the ToC process. This could be accomplished with regular communications between HCPs and patients, early patient education, and periodic patient questionnaires that determine readiness to transition.

While the introduction to adult care should start earlier, it is best not to rush the ToC process. All roundtable attendees agreed that easing patients into their ToC could help retain them and reduce anxiety. Adolescent/young adult patients with NF1 can have years of experience with healthcare, but they are new to the adult healthcare system. Attendees agreed it is unrealistic to assume these patients have the experience or knowledge to navigate the adult healthcare system successfully. Starting the ToC early and taking the time to ensure adolescent patients fully understand the process gives them time to adjust and prepare for being in charge of their own care and may reduce some of their anxiety. Patient attendees agreed that being eased into the ToC process would have been helpful for them. They also noted that introducing pediatric patients to their adult HCPs in advance could help reduce the anxiety that comes from being treated by someone new or unknown to them.

> *It's easy to assume young patients know what's going on with their care. They should be talked to like they're new instead of someone who has 10 years experience doing this.*
>
> – Patient with NF1-PN
>
> *I felt blindsided by my transition. A lead-up would be helpful and an earlier discussion of what transition would look like. The hardest part is feeling your security is going away.*
>
> – Patient with NF1-PN

One pediatric HCP attendee advocated for providing patients recommendations or care guidelines for NF1 treatment. These are not only for the patient's own knowledge but also for them to share with new HCPs to ensure these HCPs understand the standard of care that is needed.

Mental health support

Also briefly discussed at the roundtable was the need for mental health support. Pediatric/adolescent patients with NF1 suffer from anxiety, depression, poor coping skills, and difficulty socializing; in addition, some may have learning disabilities.[13,19] In a recently published survey of adult patients with NF1, over half reported having a mental health diagnosis, and over a quarter who did not have a diagnosis suspected they had an undiagnosed mental illness.[4] In another survey, adult patients reported high levels of stress and anxiety related to their condition.[11] All roundtable attendees agreed that mental health issues can negatively affect the retention of patients with NF1. A patient attendee stated that healthcare with NF1 can be overwhelming, causing some patients to take a break from their care or give up on care altogether.

> *Are some patients giving up on themselves? Maybe their health is getting bad, and they feel like nothing can be done about it. Psychological care could be beneficial for them.*
>
> – Patient with NF1-PN

Care coordinators

While there are many people who may help patients maintain healthcare through and beyond the ToC process, the importance of care coordinators was also noted at the roundtable (**Figure 6**). Care coordinators are individuals at a medical practice who help organize patient care (appointments with HCPs, etc) and communicate with all individuals involved with the patient's care. Attendees discussed how the personal touch of a care coordinator with whom patients are already familiar may reduce anxiety and help them maintain regular care into adulthood. If patients do miss appointments, care coordinators can ask them in a nonjudgmental manner why they missed their appointments and try to find a way to get past any barriers. Unfortunately, while pediatric NF clinics often have care coordinators, adult NF clinics may not.[13] Attendees acknowledged that finding funding for adult care coordinators who can provide ToC support within an institution is a serious challenge. Attendees also noted the need for HCPs to take a more proactive approach to retaining patients through more frequent appointment reminders, which may be another area where a care coordinator could provide vital services.

> *If patients feel the provider is engaged, you know they will come back.*
>
> – Adult HCP

Figure 6. Roundtable premeeting survey results from patients

Top 3 things that would help you as a patient maintain at least annual visits to HCPs:

- **Help with insurance**
- **Care coordinator or social worker**
- **Appointment reminders from HCPs**

Pediatric and adult HCP communication

Finally, communication and coordination between pediatric HCPs and care coordinators with their adult care counterparts was deemed an essential component of successful ToC and patient retention. Two attendees—a pediatric HCP and an adult care HCP who have clinics in different healthcare systems located in the same city—noted that their coordinators work together to ensure that patients transition properly and maintain regular care.

> *My coordinator and the adult clinic coordinators working together makes a huge difference when tracking patients down who don't show.*
>
> – Pediatric HCP

Actionable Challenges for Patient Retention

Recommendations
Early transition support and education for young patients (as early as 12 years of age)
Mental health care/support for patients
Funding for care coordinators in HCP offices for adult patients
Communication between pediatric HCPs/care coordinators and their adult care counterparts

3. Treatment Continuity

Treatment continuity throughout the patient's lifetime was considered a challenge by all attendees. Many patients may lose access to care when they turn 26 and are no longer eligible for coverage under their parents' or caregivers' health insurance plans (**Figures 5 and 6**). These patients may not be aware of alternative options, such as applying for disability benefits, using the health insurance marketplace for subsidized coverage (per the Affordable Care Act), or applying for Medicaid, and may drop out of care due to financial costs.

> *Young people don't realize there are ways around the problems of not having insurance, like being on disability. Trying to research this yourself is very complicated.*
>
> – Patient with NF1-PN

In addition, the only treatment approved in the US for patients with NF1 who have symptomatic, inoperable PNs is indicated for pediatric patients.[6] This leaves adult patients without an approved treatment for NF and could lead to treatment interruption for young patients when they transition to adult care. For the treatment of adults with NF1-PN, there are several drugs currently under investigation in the US,[20] with mirdametinib receiving a US Food and Drug Administration (FDA) Priority Review in August 2024.[21]

Insurance coverage

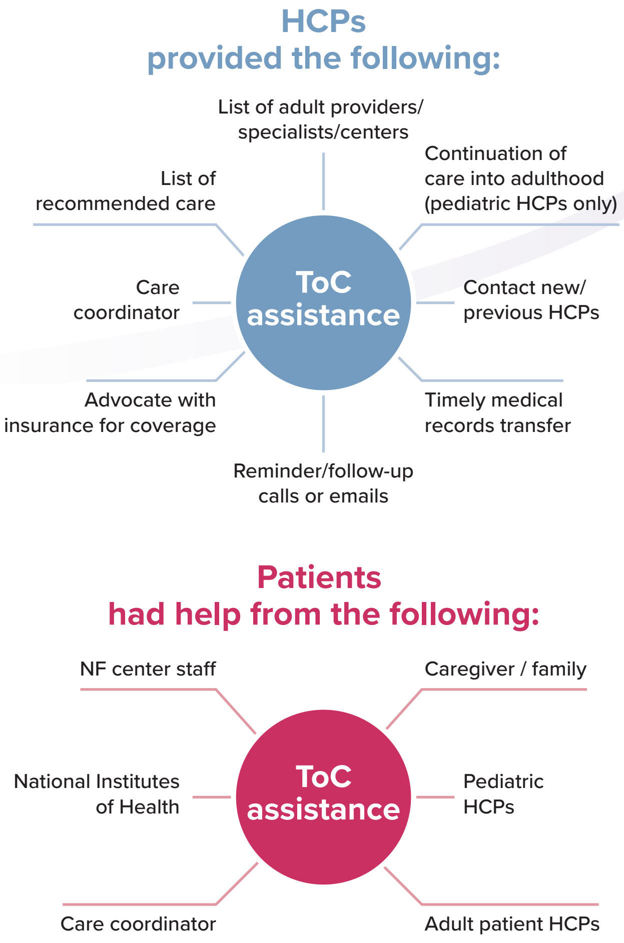
HCPs and coordinators should help patients who will soon transition to adult care make a plan to navigate insurance coverage so that care is not interrupted (**Figure 6**). Additionally, attendees noted that when patients age out of pediatric care, change insurance, or move away, there can be a gap between the last visit with the previous HCP and an initial visit with the new HCP. Any extended gap in care (eg, weeks or months) can cause a loss of drug treatment if authorization for a prescription runs out before patients can see a new HCP. Also, if there was a change in insurance coverage, these patients may no longer be able to get refills from their previous HCP to cover the gap. Attendees agreed that this is another area where care coordinators are needed as they can contact the new HCP or insurance company to try to prevent or reduce a lapse in care.

Time commitments

Difficulty in patients taking time off from work for care visits was also identified as a challenge by attendees. Many employers do not offer flexible work schedules, and many patients with chronic conditions cannot afford to take unpaid leave.[22] Patient attendees mentioned that care visits can vary unpredictably in length from a few hours to a whole day, making it difficult to provide estimates to their employers on how much time off they will need. All attendees confirmed that clear communication between HCPs and patients can reduce the burden on working adult patients and may encourage them to continue with regular care visits.

Premeeting survey results from the roundtable showed that HCP attendees offered a variety of types of assistance for ToC to maintain access and treatment continuity and that patients had received help during ToC from various medical professionals and family, as well as from the National Institutes of Health (**Figure 7**).

Figure 7. Roundtable premeeting survey results from patients and HCPs



Actionable Challenges for Treatment Continuity

Recommendations
Patient assistance regarding insurance options when aging out of parents'/caregivers' coverage
Approval of drug treatments for adult patients with NF1-PN
Communication between HCPs and working patients regarding time commitments for care visits

4. Education

All attendees agreed that education is an essential component of empowering patients with NF1 to take charge of their healthcare. Attendees discussed starting education early to fully prepare patients for the ToC. Because ToC is psychologically difficult for patients, it is critical for pediatric clinics to start the educational process early (~12 years of age). Pediatric patients need to understand that their care team will change and that they will become responsible for their own care as they reach adulthood. Educational materials that specifically focus on this ToC period are important to share with patients and their families. Existing resources for patients, caregivers, and HCPs are included in **Table 1.**

Table 1. NF1 resources for patients, caregivers, and healthcare providers

For Patients and their Caregivers	Website
Finding an NF1 specialist or clinic	https://www.ctf.org/find-a-doctor/
	https://www.nfnetwork.org/understand-nf/find-doctor/
What is NF1?	https://www.ctf.org/understanding-nf-answering-questions-shedding-light/
	https://www.nfnetwork.org/understand-nf/what-is-nf/
NF1 educational materials	https://www.nfnetwork.org/resources/educational-materials/
	https://www.ctf.org/resources/
	My NF Guide – planned for early 2025
What is a MEK inhibitor?	https://www.ctf.org/first-drug-approved-for-nf/
Transitioning from pediatric to adult healthcare	https://nfcollective.org/transitioning-to-adult-care
	https://www.gottransition.org/
Clinical trials	https://www.ctf.org/clinical-drug-pipeline/
Other resources (education, financial aid, insurance, legal aid, etc)	https://www.nfnetwork.org/resources/other-resources/
For Healthcare Providers	**Website**
Transition Readiness Assessment Questionnaire (TRAQ)	https://www.etsu.edu/com/pediatrics/traq/
Transitioning patients from pediatric to adult healthcare	https://www.gottransition.org/
Conferences and education	https://www.ctf.org/conferences-education/
Diagnostic criteria and updates	https://www.ctf.org/diagnostic-criteria/
Other resources (education, financial aid, insurance, legal aid, etc)	https://www.nfnetwork.org/resources/other-resources/

The importance of transition of care

Attendees also agreed that it is necessary for patients to understand why they need to go through ToC, as well as the importance of maintaining regular, preventive care into adulthood. In a published survey of adolescent patients with NF1, many said they were uncertain how NF1 would affect them in the future.[1] Both patient attendees agreed this was an important point and noted that their HCPs had discussed with them the health challenges they may face later in life.

> *Patients are surprised when I mention preventive care for cancer, high blood pressure, heart disease, etc. It's important patients know why they transition and why they need specialists when they become adults.*
>
> – Adult HCP

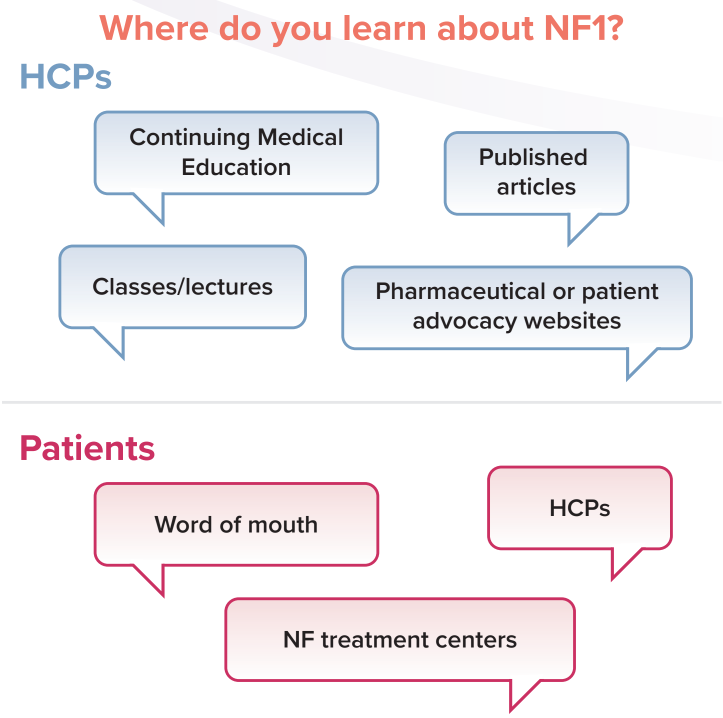
Distributing NF1 information

Premeeting survey results indicate that patients and HCPs receive their educational information in different ways (**Figure 8**). The challenges of disseminating educational materials were discussed. All roundtable attendees noted that there are pros and cons of using technology to share educational information. Patient advocacy group member attendees addressed the variety of published resources and upcoming guides for patients that can be linked across HCP and patient advocacy group sites to provide better access and visibility. Patients explained that some websites they had accessed for information about NF1 were not helpful because they were outdated. They felt that keeping online resources easy to use, well-maintained, and up to date are all important considerations. Patients also noted that some websites and forms are optimized for viewing/use on laptops and cannot easily be accessed or filled out on a phone. This can be a problem for patients who do not have laptops or have older phones. Additionally, some patients and caregivers may not have access to the internet. As such, all attendees agreed that printed materials that are simple and straightforward may be better for many patients. Materials that exist online, such as information about

NF1, ToC checklists/guides, or treatment guidelines, can be printed by HCPs and given to patients at visits. HCP attendees noted additional considerations for any printed materials for patients included having separate versions written at an appropriate level for patients or caregivers who might have learning disabilities, which are common in NF1, or for younger patients (eg, below 16 years of age).

> *People may not have advanced technology. They may have a really crappy old phone. You should assume websites and online forms may not work.*
>
> – Patient with NF1-PN

Figure 8. Roundtable premeeting survey results from patients and HCPs



Note: word of mouth does not include social media.

HCP education

HCPs also need education about NF1. A published survey of adult patients with NF1 about their healthcare concerns showed patients found it difficult to locate HCPs who had adequate experience with and knowledge of NF1.[11] HCP roundtable attendees agreed that there was a lack of knowledgeable HCPs to treat adult patients with NF1. Attendees also felt that more education on NF1 for HCPs is needed, with a particular focus on those primary care and subspecialist adult HCPs who demonstrate interest in treating NF1. Pediatric specialists could help in the educational process by making connections and identifying adult HCPs in their institutions and in the local community who have an interest in treating NF1. Finally, it was noted that introductions to other HCP NF1 specialists, pharmaceutical companies, professional societies, and NF patient advocacy groups could be made so that the adult HCPs can continue to learn about NF1 and be involved with these patients.

Actionable Challenges for Education

Recommendations
Easy-to-use, up-to-date online resources and simple, straightforward printed materials for patients
Early and repeated education of young patients on the why's and how's of the transition of care process
Patient education on the importance of maintaining preventative healthcare throughout adulthood
Improved education for adult care HCPs on NF1 and stronger connections with their pediatric counterparts

5. Summary

Roundtable attendees discussed the challenges faced by patients with NF1 as they transition from pediatric to adult healthcare, with a focus on those actionable challenges with potential solutions. For optimal patient access and care, there is a need for evidence-based guidelines to determine the standards of care, dedicated insurance liaisons to help patients navigate insurance systems, and improved access to EMRs for patients and HCPs. In terms of patient retention, patients need early ToC support, as well as education and mental health support; there is also a need for strong communication between pediatric and adult care HCPs, as well as for funding for institutions to be able to provide adult care coordinators. For treatment continuity, FDA-approved treatments for adults are lacking, and adult patients may need assistance finding insurance coverage options. Finally, for education, there needs to be dissemination of both online and printed materials that are up to date and simple, in addition to early and repeated education of patients on both the ToC process and the importance of maintaining regular care throughout adulthood. There is a need for more adult care HCPs who are interested in and knowledgeable about NF1, which may be addressed through stronger connections with their pediatric counterparts.

6. Future Directions

Identification of these challenges is only the first step. Future discussions need to occur with various stakeholders (eg, HCPs, patients, patient advocacy groups, insurance representatives, and healthcare plan/system administrators) to develop a roadmap of solutions that will ultimately improve healthcare and the ToC process for pediatric and adult patients with NF1.

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