

Adult Patient and Caregiver Perspectives on the Impact of Neurofibromatosis Type 1-Associated Plexiform Neurofibroma (NF1-PN): Insights From a US Qualitative Survey

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STUDY OBJECTIVE

- This qualitative study aimed to improve understanding of the disease burden, healthcare and treatment experiences, as well as unmet needs of adults with NF1-PN in the USA, from the perspective of patients and caregivers (**Figure 1**)

CONCLUSIONS

- This study demonstrates that NF1-PN has a substantial negative impact on the quality of life in adult patients, despite variability in manifestations
- Pain-related disorders (e.g. fibromyalgia and neuropathy) were the most common conditions experienced by adults with NF1-PN
- Patients identified several unmet needs related to their NF1-PN, including the availability of effective treatment options
- Patients highlighted a need for new medications that could stop or slow the growth of PN and reduce the size/severity of PN
- Patients reported the desire to be more informed about their care
- There was variability in patients' experiences with the process of transition from pediatric to adult care
- More support and resources focused on disease and/or treatment education may equip patients to make informed treatment and management decisions throughout the course of the disease

PLAIN LANGUAGE SUMMARY

- Why did we perform this research?** Researchers performed this study to better understand the experiences of adults with NF1-PN. They wanted to identify the disease burden, healthcare experiences, and unmet needs of these patients. Current management options for NF1-PN are limited, especially for adults. The study aimed to highlight gaps in care and potential areas for improvement.
- How did we perform this research?** Researchers conducted a qualitative study involving 11 adult patients with NF1-PN and two caregivers. Participants were surveyed about their healthcare experiences, treatment settings, and knowledge levels. The study collected data on patients' demographics, disease characteristics, and the impact of NF1-PN on their lives. Researchers analyzed the responses to identify common themes and unmet needs.
- What were the findings of this research and what are the implications?** The study found that NF1-PN negatively affects patients' quality of life, mental health, and daily activities. Most patients experienced pain, psychiatric disorders, and migraines. Many patients desired better treatment options, particularly treatments that could stop or slow the growth of PN and reduce the size/severity of PN, and more information about their condition. Transitioning from pediatric to adult care posed challenges for some patients, including finding appropriate providers and feeling unheard. These findings highlight the need for improved healthcare and treatment options for adults with NF1-PN. Healthcare providers should offer support and resources to help patients manage their condition.

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Poster



Plain language summary

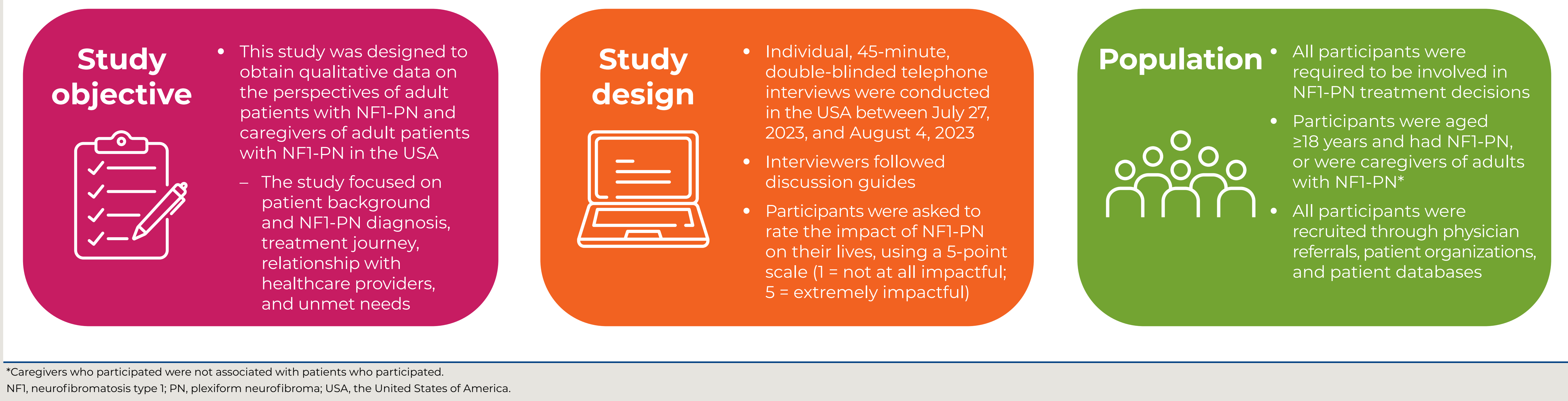
Poster presented at Children's Tumor Foundation (CTF) NF Conference, Washington DC, USA, June 21-24, 2025.
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BACKGROUND

- Neurofibromatosis type 1 (NF1) is a genetic condition caused by mutations in the *NF1* tumor suppressor gene, with an estimated prevalence of approximately 1 in 3000 to 1 in 4000 people^{1,2}
- NF1 can lead to the development of plexiform neurofibroma (PN) in up to 50% of affected individuals^{3,4}
 - PN may be associated with significant clinical symptoms, including pain, disfigurement, motor dysfunction, airway dysfunction, visual impairment, and bladder/bowel dysfunction^{5,6}
- Current management recommendations for adults with NF1 reflect the diversity of clinical manifestations and their extensive impact, as well as the need for ongoing medical care^{5,6}
- However, inequalities in NF1 management have been identified between pediatric and adult patients, and transition of care into adulthood remains challenging⁷⁻⁹
 - Limited approved treatment options and a lack of specialty clinics offering coordinated care specifically for adults with NF1 can significantly impact this patient population^{6,8-10}

METHODS

Figure 1. Study Design



RESULTS

Patient demographics

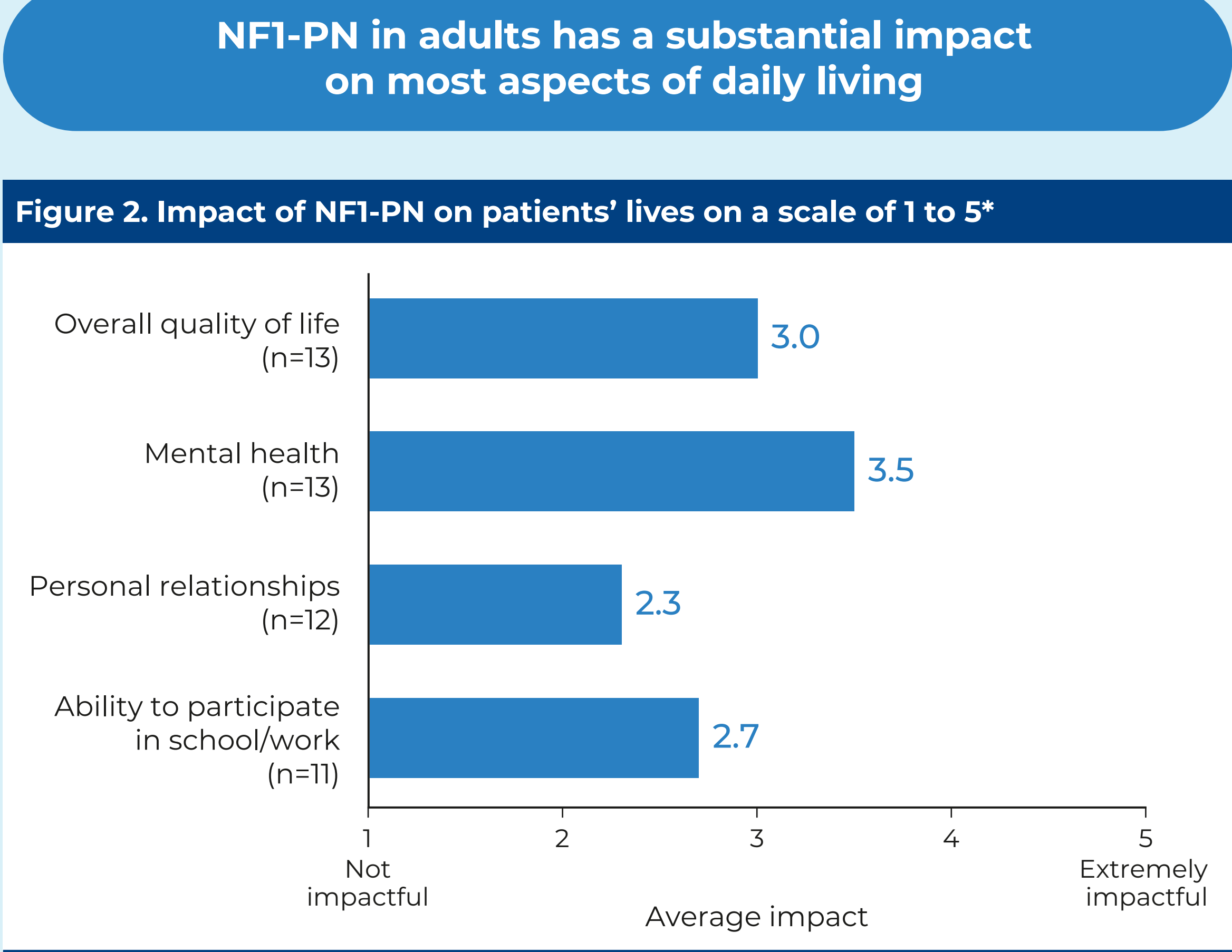
- The study included 11 adult patients with NF1-PN, and two caregivers of adult patients with NF1-PN (**Table 1**)

Table 1. Demographics and disease characteristics at the time of the survey	
Characteristic, n (%)	Patients (N=13)
Current age, years	
18–34	5 (38)
≥35	8 (62)
Region of USA	
Midwest	4 (31)
South	4 (31)
Northeast	3 (23)
West	2 (15)
Time of NF1-PN diagnosis	
Childhood	11 (85)
Adulthood	2 (15)
Level of knowledge of patients (self-reported)	
n=11	
Extremely knowledgeable	3 (27)
Very knowledgeable	4 (36)
Somewhat knowledgeable	4 (36)
Level of knowledge of caregivers (self-reported)	
n=2	
Extremely knowledgeable	1 (50)
Very knowledgeable	1 (50)
Somewhat knowledgeable	0 (0)
Current treatment setting	
HCP's office	6 (46)
Hospital setting	4 (31)
NF1 Center of Excellence	2 (15)
Not currently seeing HCP	1 (8)
No response provided	2 (15)
Insurance type	
Private (PPO/HMO)	6 (46)
Medicare	4 (31)
Medicaid	2 (15)
Not insured/self-pay	1 (8)

HCP, healthcare professional; HMO, health maintenance organization; NF1, neurofibromatosis type 1; PN, plexiform neurofibroma; PPO, preferred provider organization; USA, United States of America.

Impact of NF1-PN

- NF1-PN had a substantial impact on patients' lives, affecting overall quality of life, mental health, personal relationships, and ability to participate in school or work (**Figure 2**)



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Conflicts of interest

Phioanh L. Nghiemphu has received grants paid to her institution from the Children's Tumor Foundation, ERASCA, SpringWorks Therapeutics, and Recursion. Phioanh L. Nghiemphu also declares receiving consulting fees for participation in advisory boards for Alexion, AstraZeneca Rare Disease, CIRM, and SpringWorks Therapeutics. Phioanh L. Nghiemphu has also received payment or honoraria for presentations from SpringWorks Therapeutics. Conrad L. Cordova has received honoraria from the Arizona Nurses Association to present at the Nurse Practitioner Conference 2020, and has served as president of The Southern Arizona Advanced Practice Nurse Society. Abby Crites is an employee of IQVIA and has conducted market research studies with patients, healthcare professionals and payers on behalf of Alexion, AstraZeneca Rare Disease. Theresa Dettling and Alyssa Bowling are employees of, and own stocks in, Alexion, AstraZeneca Rare Disease. Xiaoqin Yang is an employee of Merck Sharp & Dohme LLC, a subsidiary of Merck & Co., Inc., Rahway, NJ, USA, and owns stocks in Merck & Co., Inc., Rahway, NJ, USA.

References

1. Barnett C, Canidio E, Chen B, et al. *Orphanet J Rare Dis*. 2022;17(1):323.
2. Hirbe AC, Gutmann DH. *Lancet Neurol*. 2014;13(8):834–843.
3. Ferner RE, Huson SM, Thomas N, et al. *J Med Genet*. 2007;44(2):81–88.
4. Friedman JM. *GeneReviews*[¹] [Internet]. Seattle (WA): University of Washington, Seattle; 1996 [updated April 3, 2025];1993–2025.
5. Carlton C, Evans DG, Blanco I, et al. *EClinicalMedicine*. 2023;56.
6. Stewart DR, Korf BR, Nathanson KL, et al. *Genet Med*. 2018;20(7):671–682.
7. Crawford H, Barton B, Wilson M, et al. *Clin Genet*. 2016;89(3):385–391.
8. Gregory TA, Molina PSB, Phillips GD, Henson JW. *Neurooncol Pract*. 2022;9(3):229–235.
9. Rietman AB, van Helden H, Both PH, et al. *Am J Med Genet A*. 2018;176(5):1150–1160.
10. U.S. Food & Drug Administration. (2025) FDA approves mirdametnib for adult and pediatric patients with neurofibromatosis type 1 who have symptomatic plexiform neurofibromas not amenable to complete resection. Available at <https://www.fda.gov/drugs/resources-information-approved-drugs/fda-approves-mirdametnib-adult-and-pediatric-patients-neurofibromatosis-type-1-who-have-symptomatic>. Accessed June 04, 2025.

NF1-associated conditions

- Patients and caregivers reported multiple conditions that they associated with NF1; the most common of which were pain-related disorders (n=10), psychiatric disorders (n=7), and chronic migraines (n=6)

Frequency of medical care

- Most patients (77%; n=10/13) reported receiving routine healthcare annually
 - Patients with lapses in care (n=3/13) provided reasons including the perception that there is no available treatment/cure for NF1-PN (n=3), lack of healthcare insurance (n=2), and the lack of local physicians (n=1)

Medication management

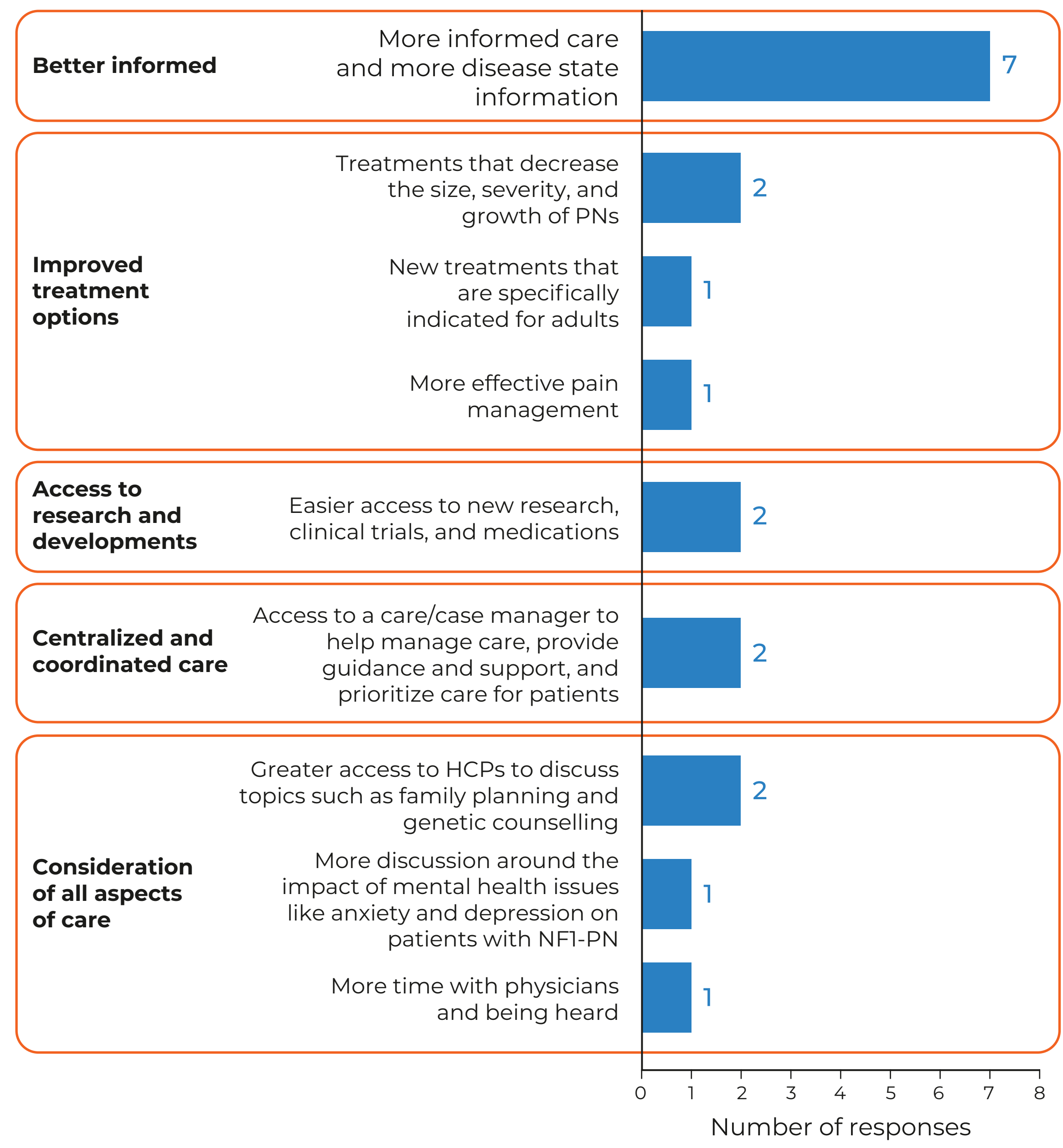
- Medications used for NF1-PN symptom management for adults included both over-the-counter (n=9) and prescription pain medications (n=3), vitamin supplements (n=1), and skin creams/oils (n=2), as well as prescription products for headache, anxiety/depression, blood pressure, sleep disorders, fibromyalgia, attention deficit hyperactivity disorder (ADHD), and cancer

Unmet needs

- When asked what could be changed about the way NF1-PN is managed, patients and caregivers identified being more informed about their care and disease state, and improved treatment options for NF1-PN as key needs (**Figure 3**)

Adults with NF1-PN wish to have access to improved treatment options and to be more informed about their care

Figure 3. Patient and caregiver responses when asked what could be changed about the way NF1-PN is managed



HCP, healthcare professional; NF1, neurofibromatosis type 1; PN, plexiform neurofibroma.

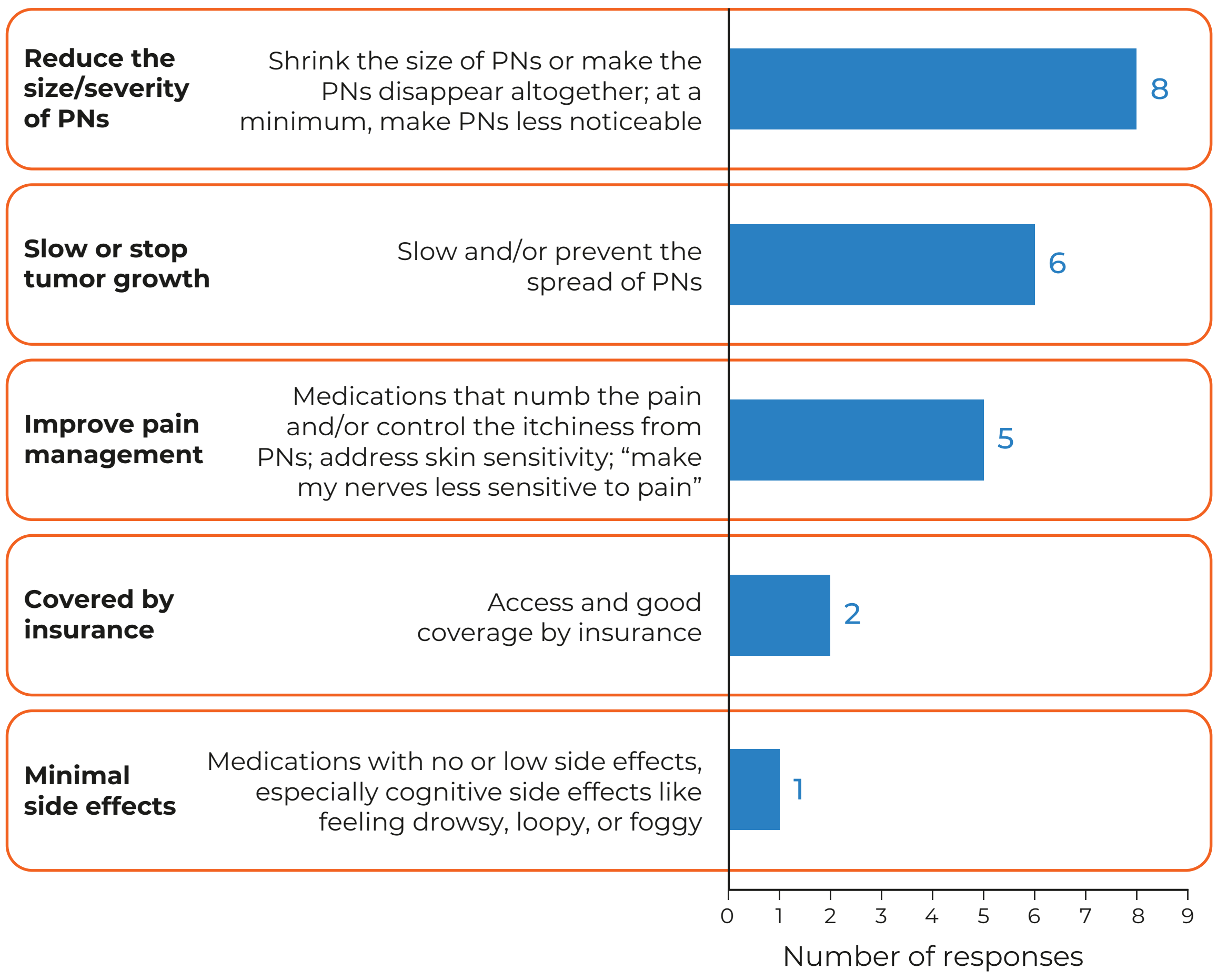
Medication needs*

- The patients with NF1-PN in this study noted that they are highly open to new medications

Patients highlighted a desire for new medications that could stop or slow the growth of PN and reduce the size/severity of PN (Figure 4)

*At the time of the study, there were no approved medications for the treatment of PN in adults with NF1

Figure 4. Patient and caregiver responses when asked about new medications

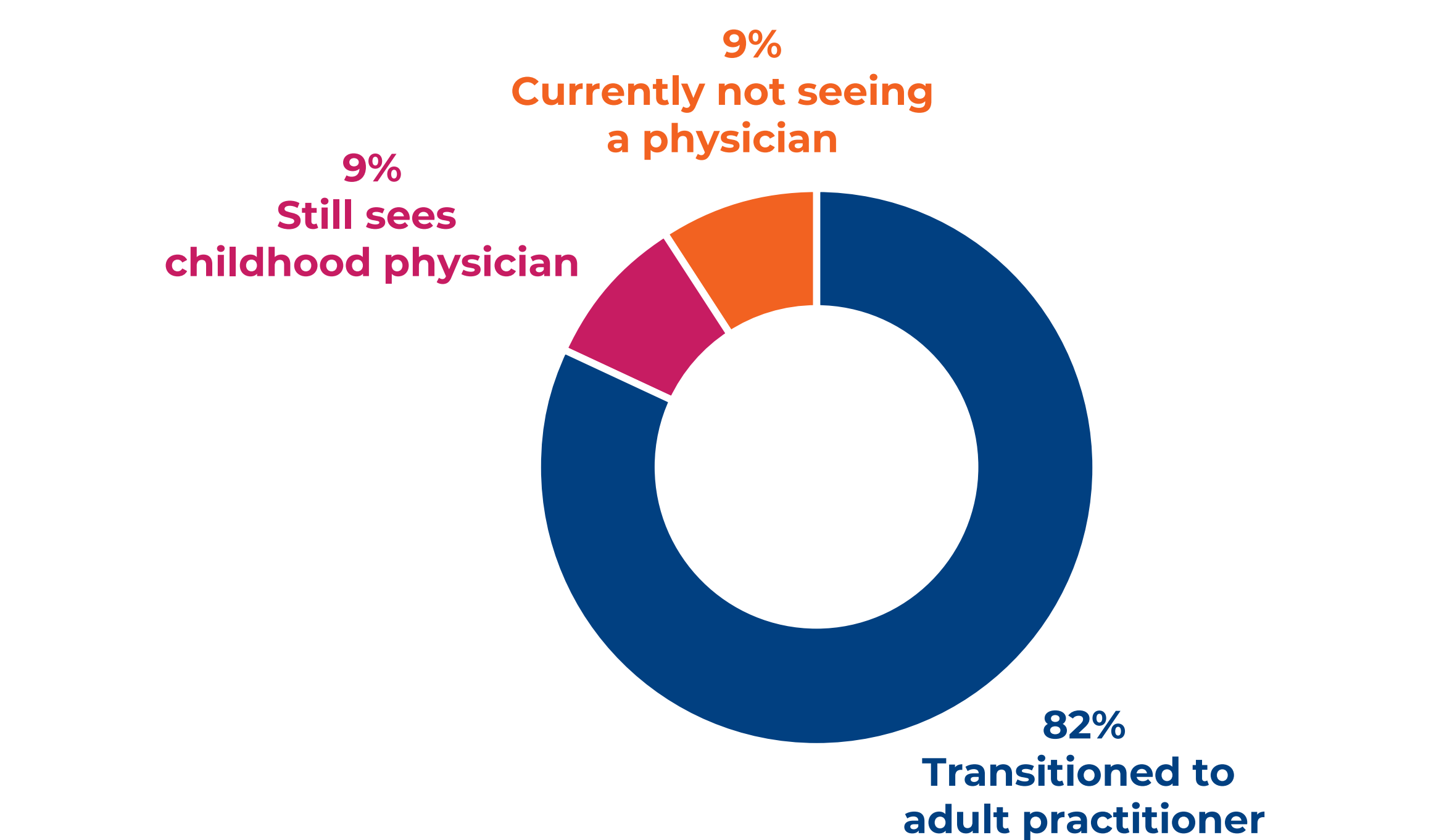


PN, plexiform neurofibroma.

Transition of care

- Of the 11 patients who were diagnosed during childhood, 82% (n=9/11) had been transitioned to an adult practitioner (**Figure 5**)
 - Transition to adult care occurred at the age of 18–19 years for most patients, and was typically driven by the pediatric care team (n=6/9)
 - While the transition process was uncomplicated for 56% of patients (n=5/9), the remainder experienced challenges, including finding an appropriate provider (n=3), feeling that they were not being heard (n=2), and frustration with level of care and understanding of NF1-PN symptoms (n=1)

Figure 5. Care status among patients diagnosed as a child (n=11)



LIMITATIONS

- Although some findings were consistent across adult patients with NF1-PN and caregivers of adult patients with NF1-PN, considerable data variability was observed
- The small sample size limited the ability to identify differences based on patient age or time of transition to adult care
- The inclusion of only two caregivers limited the generalizability of study findings to the wider population caring for adults with NF1
- Patients and caregivers self-selected, which could have introduced some bias into the study findings
- Future studies are needed to examine the experience of patients who have poor or moderate levels of health insurance coverage during the transition from pediatric to adult care