Poster 5542

Healthcare Providers' Perspectives on the Management of Adults With Neurofibromatosis Type 1 and Plexiform Neurofibroma: Insights From a US Survey

Phioanh L. Nghiemphu[1,2,*]; Abby Crites[3]; Alyssa Bowling[4]; Xiaoqin Yang[5]; Theresa Dettling[6]
*Presenting author

[1]UCLA Neuro-Oncology Program, David Geffen School of Medicine, University of California, Los Angeles, CA, USA; [2]Department of Neurology, David Geffen School of Medicine, University of California, Los Angeles, CA, USA; [3]IQVIA, Durham, NC, USA; [4]Global Medical Affairs, Alexion, AstraZeneca Rare Disease, Boston, MA, USA; [5]Merck & Co., Inc., Rahway, NJ, USA; [6]US Health Economics and Outcomes Research, Alexion, AstraZeneca Rare Disease, Boston, MA, USA.

STUDY OBJECTIVE

- The aim of this qualitative-quantitative (quali-quant) study was to better understand experiences, approaches, and challenges in treating and managing adults with neurofibromatosis type 1-associated plexiform neurofibroma (NF1-PN) from the perspective of US healthcare providers (HCPs) (**Figure 1**)

CONCLUSIONS

- From the perspectives of HCPs, adults with NF1-PN encounter numerous difficulties, including high rates of reported pain
- Adults with NF1-PN have limited treatment options to manage NF1-PN
- Additionally, HCPs reported that adults have a high overall burden of disease and face multiple challenges when transitioning from pediatric to adult care

PLAIN LANGUAGE SUMMARY

Why did we perform this research?
Researchers conducted this study to understand the experiences of doctors treating adults with NF1-PN. They wanted to identify the main challenges faced by adults with NF1-PN. Finally, the study also aimed to highlight the biggest challenges that young adults with NF1-PN face when they are transitioning to adult care.

How did we perform this research?
The study involved a survey and interviews with 41 doctors. The survey collected data on the doctors' experiences in treating adults with NF1-PN. Four HCPs also answered further questions about their experiences, providing deeper insights.

What were the findings of this research and what are the implications?
HCPs noted that neurofibromas are the most common symptom in adults with NF1. HCPs reported that the symptoms with the greatest burden on adults with NF1-PN are brain stem gliomas (tumors on the brain stem), visual impairment (loss of sight), optic gliomas (tumors that affect the optic nerve), and neuropathic pain (pain associated with the nervous system). HCPs frequently use physical exams and imaging to monitor NF1-PN. Management of NF1-PN often involves physiotherapy and medications like pregabalin and gabapentin, which are medications that are used to treat nerve pain. Many HCPs noted the lack of effective treatment options as a challenge faced by adults with NF1-PN. The findings of this study highlighted the need for better guidelines and treatment options for NF1-PN in adults. Improved transition care guidelines could help manage challenges during the shift from pediatric to adult care.

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Poster | Plain language summary | Supplementary material

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Poster presented at Children's Tumor Foundation (CTF) NF Conference, Washington DC, USA, June 21–24, 2025.
Presenting author: Dr. Phioanh L. Nghiemphu (pnghiemphu@mednet.ucla.edu)

BACKGROUND

- NF1 is a heterogeneous, multi-system genetic disorder characterized by a wide range of signs and symptoms that often arise from birth/childhood and may progress into adulthood[1,2]
  - PN are peripheral nerve sheath tumors that can be associated with pain, motor and sensory dysfunction, and disfigurement, which can impact a person's quality of life[1-5]
- For patients with NF1, the transition of care from childhood into adulthood can be challenging
  - There are few official guidelines on how this process should be managed

METHODS

Figure 1. Study Design

Study summary
- This was a US-based quali-quant study, designed to better understand **HCP perspectives, experiences, approaches, and challenges** when managing the treatment of adults with NF1-PN
- HCPs were asked for their perspectives on unmet needs in the diagnosis, management, and treatment of adults with NF1-PN
  - A subset of HCPs participated in quali-quant interviews to provide additional context to their responses

Population
To participate in the study, HCPs were required to have:
- been practicing for 3–35 years
- spent ≥50% of their time in direct patient care
- treated ≥100 patients in the past 12 months for any condition, including ≥3 adults (aged ≥18 years) with NF1-PN

Data collection
- Quantitative data were collected through 25-minute online surveys
- In quali-quant interviews, a qualitative healthcare moderator asked HCPs further questions on their responses as they completed the survey
- Surveys were carried out between July 27 and August 23, 2023

HCP, healthcare provider; NF1, neurofibromatosis type 1; PN, plexiform neurofibroma; quali, qualitative; quant, quantitative.

RESULTS

Demographics
- Overall, 41 HCPs completed the survey; 4/41 HCPs participated in quali-quant interviews (**Figure 2**)
  - Details of HCP demographics are presented in **Supplementary Table 1**

Figure 2. HCP demographics

HCPs had spent an average of 19 years in practice

HCPs managed an average of 22 patients with NF1-PN

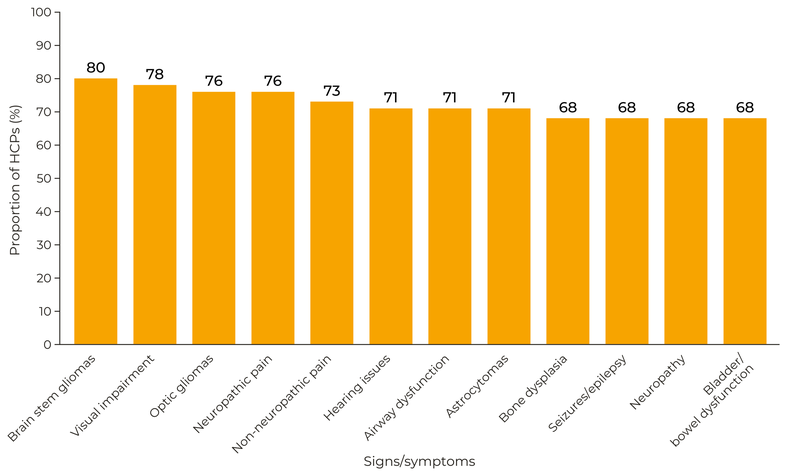
Approximately one-fifth (22%) of HCPs treated adult patients only...

The remaining HCPs (78%) treated both adult and pediatric patients with NF1-PN

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

NF1-PN diagnosis
- HCPs reported the most common manifestation of NF1 in adult patients prior to PN diagnosis was neurofibromas (66%; **Supplementary Figure 1a**)
  - HCPs reported that neuropathic pain and neuropathy are more common in adult than pediatric patients
  - The most commonly reported NF1 manifestations present prior to PN diagnosis in pediatric patients are presented in **Supplementary Figure 1b**
- The most burdensome manifestation experienced by adult patients was brain stem gliomas (80%; **Figure 3**)
  - The most commonly reported (≥65% HCPs) NF1 manifestations with the greatest burden, as reported by HCPs, for pediatric patients are presented in **Supplementary Figure 2**

Figure 3. Most commonly reported (≥65% HCPs) NF1 manifestations with the greatest burden (n=41)



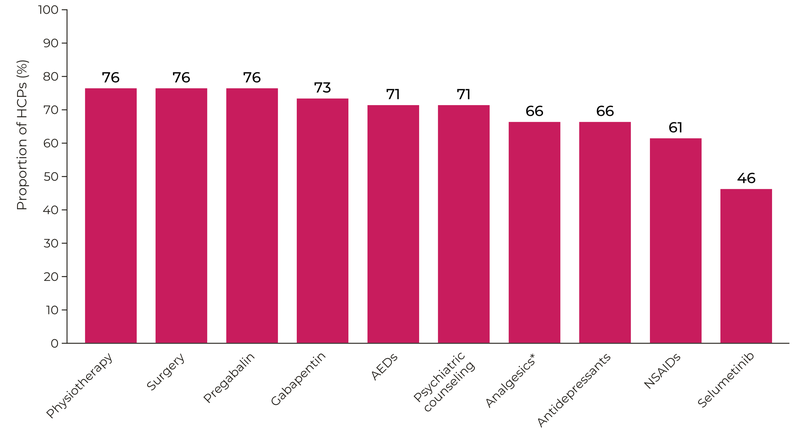
HCPs were asked 'Which symptoms of NF1 PN do you believe place the greatest burden on pediatric patient's daily activities?'.
HCP, healthcare provider; NF1, neurofibromatosis type 1; PN, plexiform neurofibroma.

The majority of HCPs (68%) reported scheduling most adult patients for follow-up appointments every 2–3 months

The most common methods of PN monitoring for adult patients were physical examination of the skin (90%) and diagnostic imaging (76%)

NF1-PN symptom management
- The most common treatments for manifestations of PN in adults included physiotherapy, surgery, and pregabalin (76% each) (**Figure 4**)
  - For pediatric patients, these were anti-epileptic drugs (78%) and physiotherapy (75%)
  - The top 10 most commonly prescribed treatments for pediatric patients with NF1-PN are presented in **Supplementary Figure 3**
- Types of pain medication for adult patients included pregabalin (76%), gabapentin (73%), analgesics (66%), and non-steroidal anti-inflammatory drugs (61%)
- Over two-thirds of HCPs highlighted both the limited (69%) and lack of (67%) effective treatment options as the greatest challenges in the management of adult NF1-PN
- Selumetinib was approved by the U.S. Food and Drug Administration for the treatment of pediatric patients with symptomatic, inoperable NF1-PN prior to the study period; 46% of physicians reported use of selumetinib in adult patients and 50% of physicians reported use of selumetinib in pediatric patients
  - During the study period, there were no approved medications for the adult patient population
  - As of June 2025, selumetinib is not approved for the treatment of adults with NF1-PN

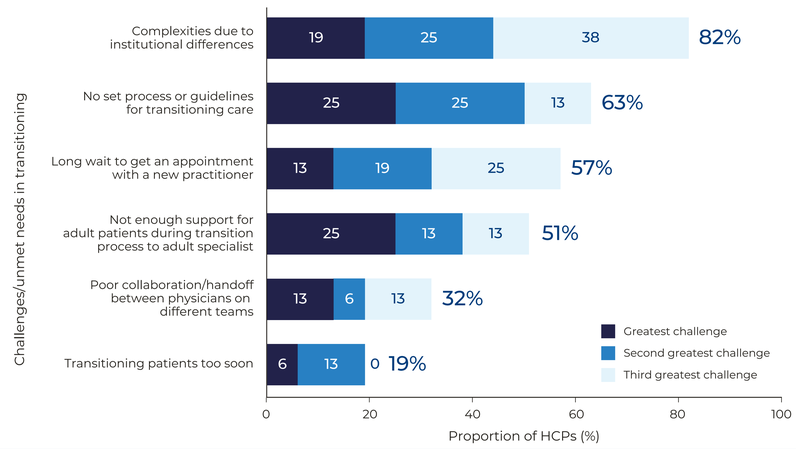
Figure 4. Top 10 most commonly prescribed treatments for NF1-PN symptom management in adults, as reported by HCPs (n=41)



HCPs were asked 'Which treatments are used to manage symptoms of NF1 PN patients?'.
*Analgesics other than NSAIDs, such as opioids.
AED, anti-epileptic drug; HCP, healthcare provider; NSAID, non-steroidal anti-inflammatory drug.

NF1-PN patient management and transition of care
- Regarding continuity of care, HCPs reported that 17% of patients fall out of care when transitioning from pediatric to adult care and attribute this to lack of transition guidelines/standards (**Figure 5**)

Figure 5. Greatest challenges and unmet needs when transitioning pediatric patients with NF1-PN to adult care (n=16)



HCP, healthcare provider; NF1-PN, neurofibromatosis type 1-associated plexiform neurofibroma.

Funding
This study was funded by Alexion, AstraZeneca Rare Disease and Merck Sharp & Dohme LLC, a subsidiary of Merck & Co., Inc., Rahway, NJ, USA.

Acknowledgements
Medical writing support for the development of this poster, under the direction of the authors, was provided by Connie Feyerherm, MSci, of Helix, OPEN Health Communications, London, UK, and funded by Alexion, AstraZeneca Rare Disease, in accordance with Good Publications Practice (GPP) guidelines (www.ismpp.org/gpp-2022).

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. 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. 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. Theresa Dettling and Alyssa Bowling are employees of, and own stocks in, Alexion, AstraZeneca Rare Disease.

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