

# Real-World NF1-PN Treatment Patterns in US Adults With NF1-PN

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## OBJECTIVES

- The objectives of this study were to:
  - Describe demographic and clinical characteristics of adult patients (≥18 years old) in the US with symptomatic, inoperable PNs
  - Describe treatment patterns and healthcare resource utilization (HCRU) in the management of symptomatic, inoperable PNs in adult patients with NF1-PN in the US

## CONCLUSIONS

- All patients that met the eligibility criteria had monitoring over time, and most received PN-associated treatment, which included pharmacotherapy and surgery
  - The majority of patients were treated for PN-associated pain, and pain medication use increased between first and last visit
  - Of patients who underwent debulking surgery and who had 12 months of follow-up (n=11), 6 patients experienced recurrence within 12 months (54.5%). The use of adjuvant systemic therapy to prevent recurrence should be studied in adults with NF1-PN
  - Hospitalizations and emergency department visits were common, with nearly 1 emergency department visit per patient per year
  - Patients underwent regular monitoring via imaging, with 95.3% of patients receiving MRIs, at an average of 3 MRIs per year
- These findings underscore the substantial disease burden and complex healthcare needs among adults with NF1-PN prior to the approval of pharmacologic therapies for NF1-PN in adults

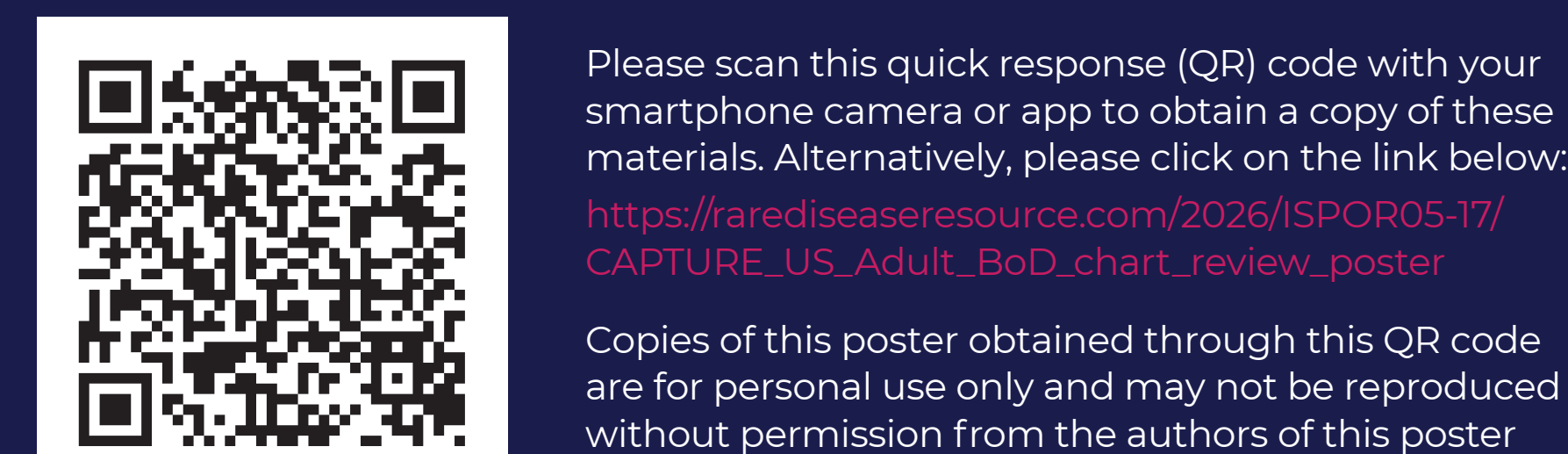
## PLAIN LANGUAGE SUMMARY

- Why did we perform this research?** To explore monitoring and management of NF1-PN in the US in adult patients and to better understand their treatment journey
- How did we perform this research?** We looked at the medical charts of patients with NF1-PN who were older than 18 years of age across 4 clinic locations in the United States
- What were the findings of this research and what are the implications?** NF1-PN can be a significant burden on patients, with high pain medication use and many healthcare visits

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## INTRODUCTION

- Neurofibromatosis type 1 (NF1) is a genetic disease that manifests in early childhood and can affect multiple body systems (e.g., skin, skeletal, nervous) with clinical manifestations such as skin changes and bone deformities<sup>1,2</sup>
- 30%-60% of patients with NF1 develop plexiform neurofibromas (PNs), nonmalignant tumors associated with morbidity and risk of malignant transformation<sup>3,4</sup>
- Prior to 2025, there were no therapies approved by the United States (US) FDA for use in NF1-PN for adults<sup>5</sup>

## METHODS

- A retrospective chart review of adult patients aged ≥18 years with NF1 and ≥1 inoperable PN who had ≥3 clinical visits between January 2010 and September 2024 was conducted across 4 NF1 centers within the US

- Study data were abstracted by physicians or their appointed proxies for the first, middle, and last documented patient visits
- Institutional review boards at the participating sites reviewed and approved the study protocol
- Charts from 85 patients who met the study selection criteria were included

## LIMITATIONS

- Data collection was limited to the available contents of patient medical records; data were entered directly into the case report form by the treating physicians/qualified assistant staff
  - Patients may have received care outside specialty care centers that was not captured in this study

## RESULTS

### Patient Characteristics

- Patient characteristics are reported in **Table 1**
- Median length of time from first to last study visit at participating clinics: 38.9 (range, 12.0-175.3) months

### PN Characteristics

- PNs were most common on the lower extremities and the spine (**Figure 1**)
- Pain (n=64, 75.3%) and disfigurement (n=33, 38.8%) were common complications of NF1-PN (**Figure 2**)
- The majority of PNs (n=62/94, 66.0%) seen over the course of the study period were treated (e.g., pharmacotherapy or partial resection)

### Treatment Patterns

- Majority of patients used pain medication during the study period (60%) (**Table 2**)
  - Between the first and last visit, pain medication use increased (**Figure 3**)
- During the study period, 18 patients underwent debulking or partial resection surgeries on symptomatic, inoperable PNs (**Table 3**)
  - Among patients with at least 12 months of postsurgical follow-up (n=11), PNs recurred for 6 patients (54.5%)

### HCRU

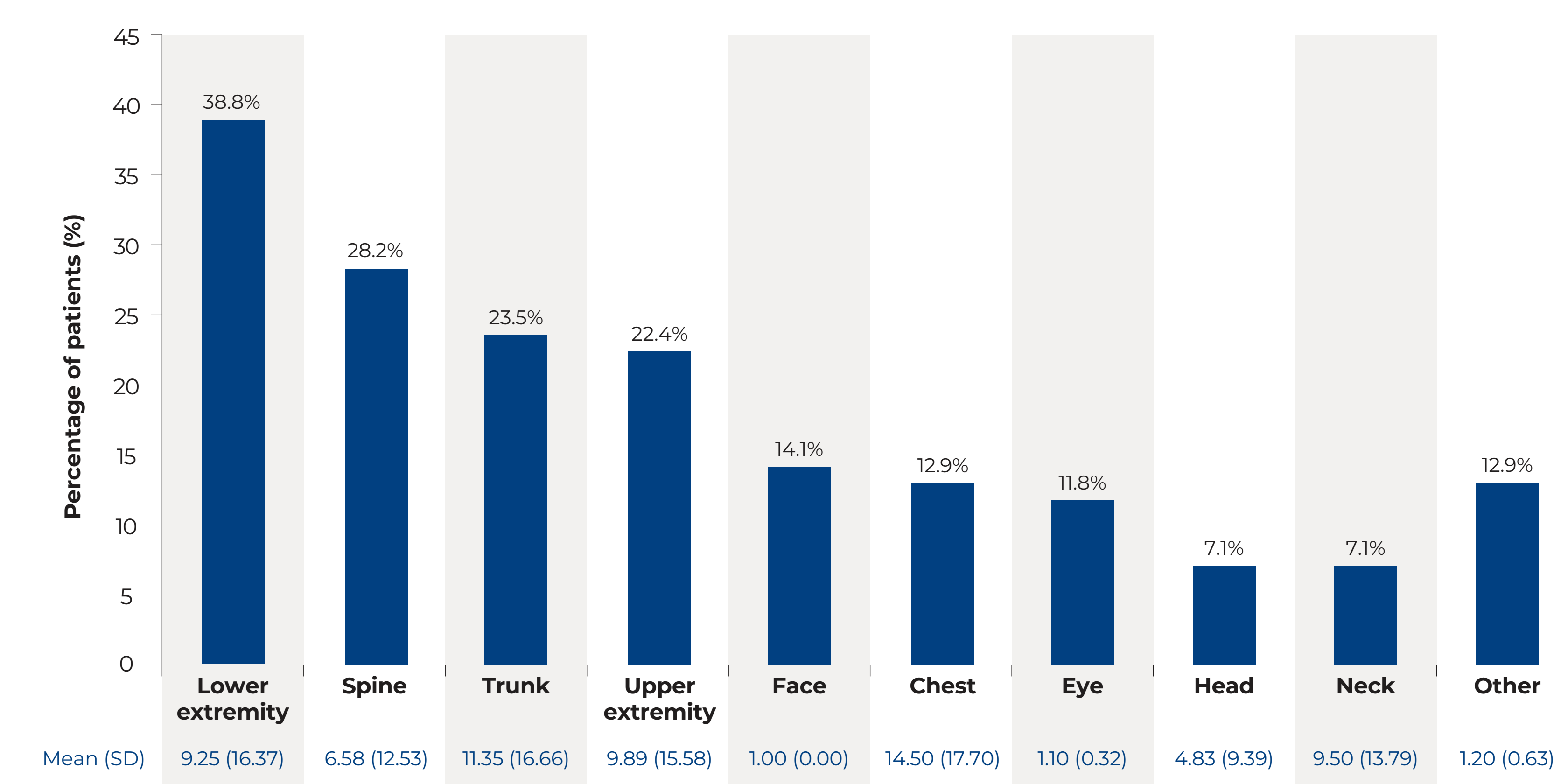
- 18 patients (21.2%) were hospitalized and 36 patients (42.4%) visited the emergency department or urgent care at least once during the study period (**Table 4**)
  - Over the course of the study, patients visited the emergency department nearly once a year, on average
- Patients with NF1-PNs underwent regular imaging to monitor their condition; 95.3% of patients received MRIs during the study period, with an average of 3 per patient per year

**Table 1: Patient Characteristics (N=85)**

Patient characteristics	
<b>Age in years, mean (SD)</b>	
NF1 diagnosis	8.0 (13.1)
Diagnosis of first symptomatic inoperable PN	22.3 (16.7)
First adult visit for NF1 in the study period	34.5 (13.6)
<b>Female, n (%)</b>	
	51 (60.0)
<b>Race, n (%)</b>	
White	71 (83.5)
Black/African American	12 (14.1)
Asian, Native Hawaiian, Other Pacific Islander	2 (2.4)
Other/Unknown	2 (2.4)
<b>Hispanic or Latinx ethnicity, n (%)</b>	
	4 (4.7)
<b>Family history of NF1, n (%)</b>	
	33 (38.8)

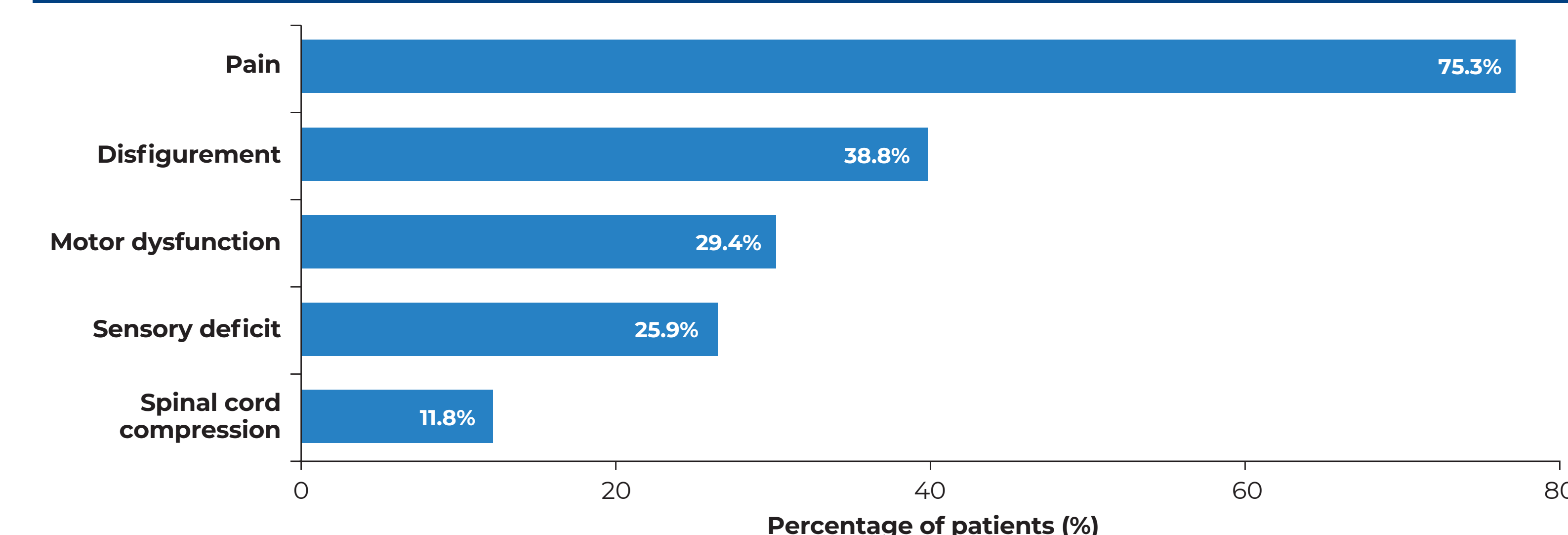
SD = standard deviation

**Figure 1: Anatomical Location of All Symptomatic, Inoperable PNs Observed at the End of the Study Period (N=94)**



Note: Multiple responses allowed; thus, sum may be greater than 100%

**Figure 2: PN-Related Complications (N=85)**



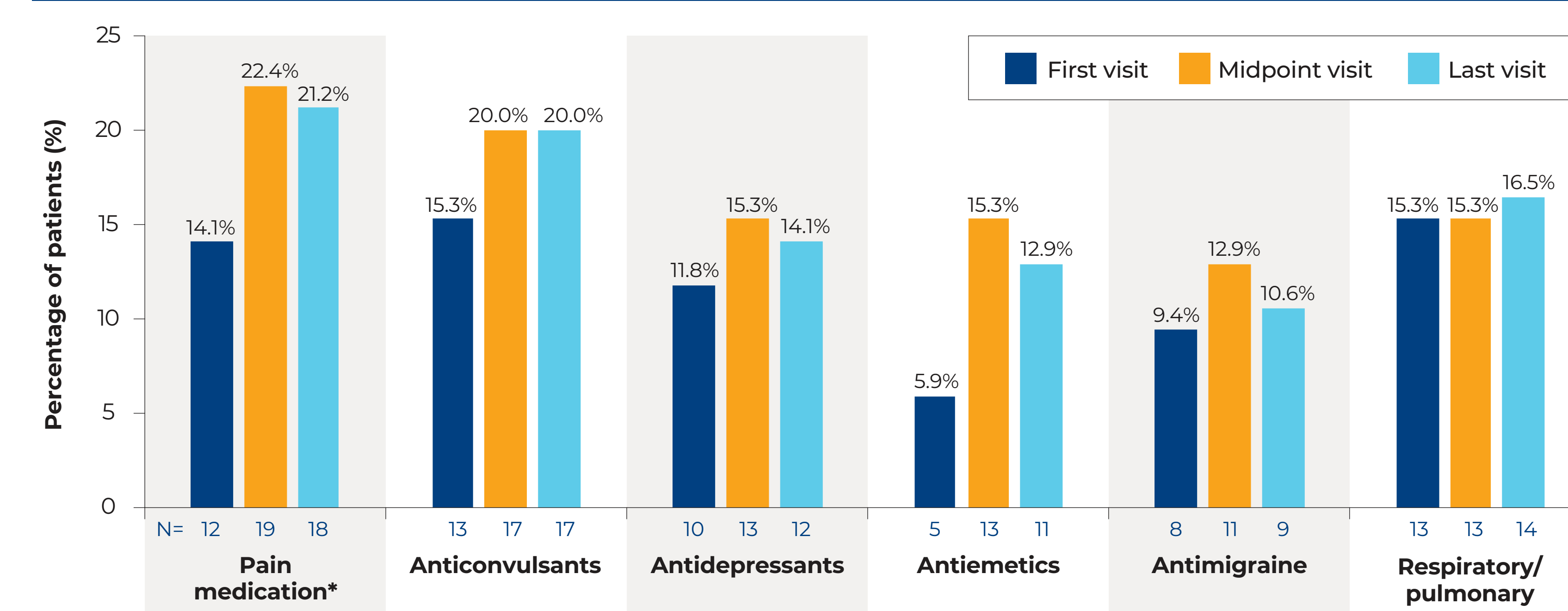
Note: Multiple responses allowed; thus, rows may add up to greater than 100%

**Table 2: Treatments Received During the Study Period (N=85)**

Treatment category, n (%)	n (%)
Pain medication	51 (60.0)
NSAIDs	36 (42.4)
Neuropathic pain medication	35 (41.2)
Opioids/narcotics	20 (23.5)
MEK inhibitor	28 (32.9)
Debulking surgery	18 (21.2)
Physical therapy	16 (18.8)
Other surgeries	2 (2.4)

MEK = mitogen-activated protein kinase kinase; NSAID = nonsteroidal anti-inflammatory drug  
Note: Multiple responses allowed; thus, rows may add up to greater than 100%

**Figure 3: Medication Use Over Time (N=85)**



\*Pain medication specifically refers to analgesics in this context

**Table 3: Surgical Patterns and Outcomes (N=85)**

Number of unique debulking and/or partial resection surgeries that were performed on symptomatic, inoperable PNs	n	Mean (SD)
Before the study	33	2.2 (1.1)
During the study period	18	1.7 (1.1)
n		%
Number of debulked PNs with 12 months of follow-up	11	61.1
Number of PNs with recurrence/progression following surgery	6	54.5

**Table 4: HCRU Related to NF1-PNs (N=85)**

Healthcare resource	Number (%) of patients who received care	Mean (SD) per patient per year
<b>Healthcare visits</b>		
Emergency department or urgent care	36 (42.4)	1.0 (1.2)
Hospitalizations	18 (21.2)	0.8 (0.9)
<b>Imaging</b>		
MRI	81 (95.3)	3.0 (4.3)
CT	33 (38.8)	1.1 (2.0)
X-ray	24 (28.2)	1.4 (2.5)

CT = computed tomography; MRI = magnetic resonance imaging