

The Pathways of Care for Neurofibromatosis Type 1 (NF1) in the UK: A Mixed-Methods Survey Study

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BACKGROUND

Neurofibromatosis Type 1 (NF1) is a genetic neurocutaneous disorder characterised by the development of neurofibromas, affecting an estimated 25,000 individuals within the UK. The heterogeneity of NF1 presentation poses substantial challenges in the standardisation of care protocols. Data derived from a public consultation led by Childhood Tumour Trust - comprising 1083 responses from NF1 patients, families and carers (PFCs) and 94 responses from healthcare professionals (HCPs), respectively - revealed widespread dissatisfaction and a lack of standardised guidelines and unified care pathways across the UK.

AIMS

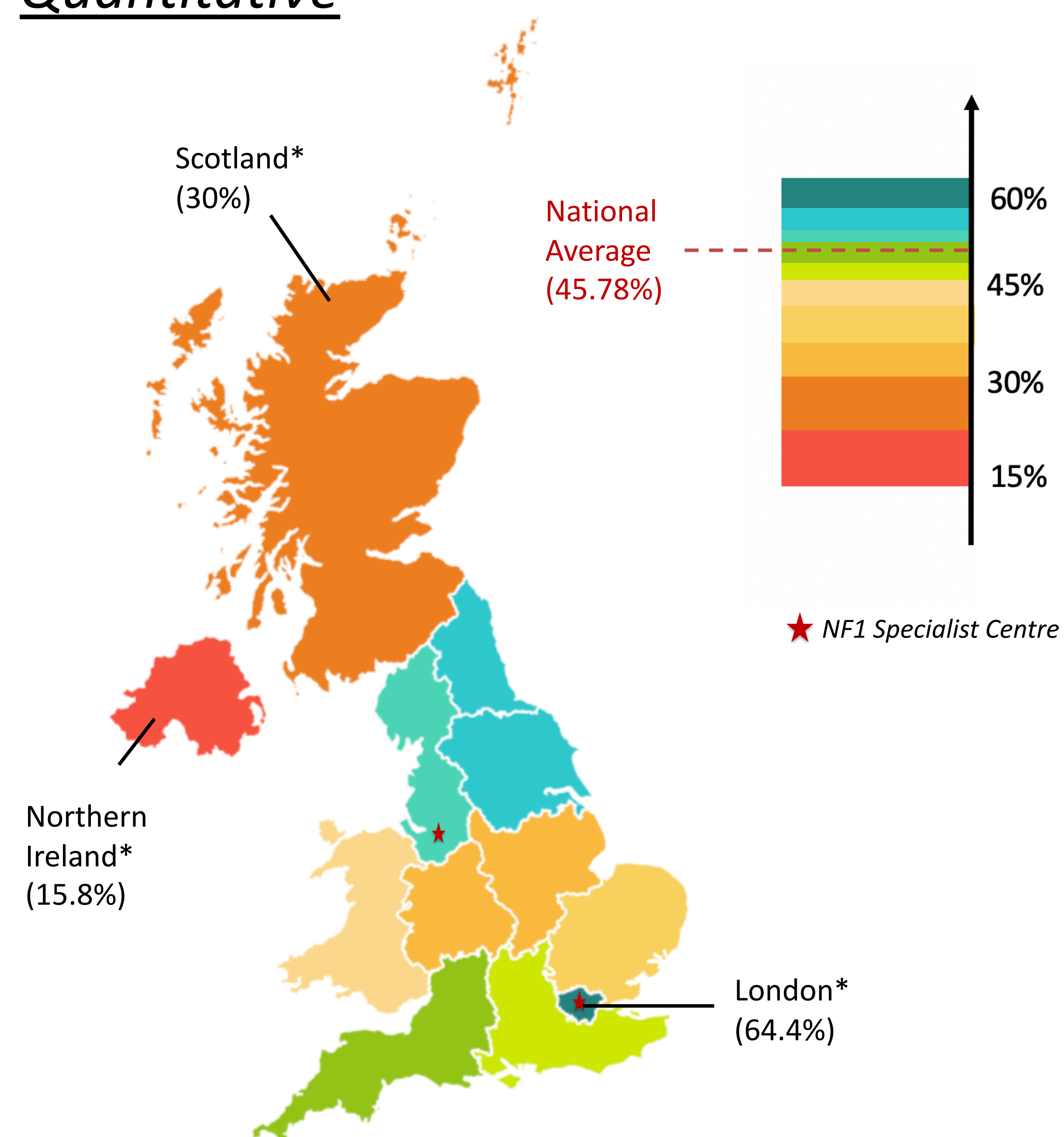
- Conduct an in-depth analysis of PFC's experiences within the NF1 care continuum.
- Identify factors contributing to satisfaction and dissatisfaction.

METHODS

- *Quantitative*: used descriptive statistics to illustrate key points in the pathways and chi-squared tests to investigate correlations with satisfaction.
- *Qualitative*: used thematic analysis of free-text responses to identify specific challenges along the pathways.

RESULTS

Quantitative



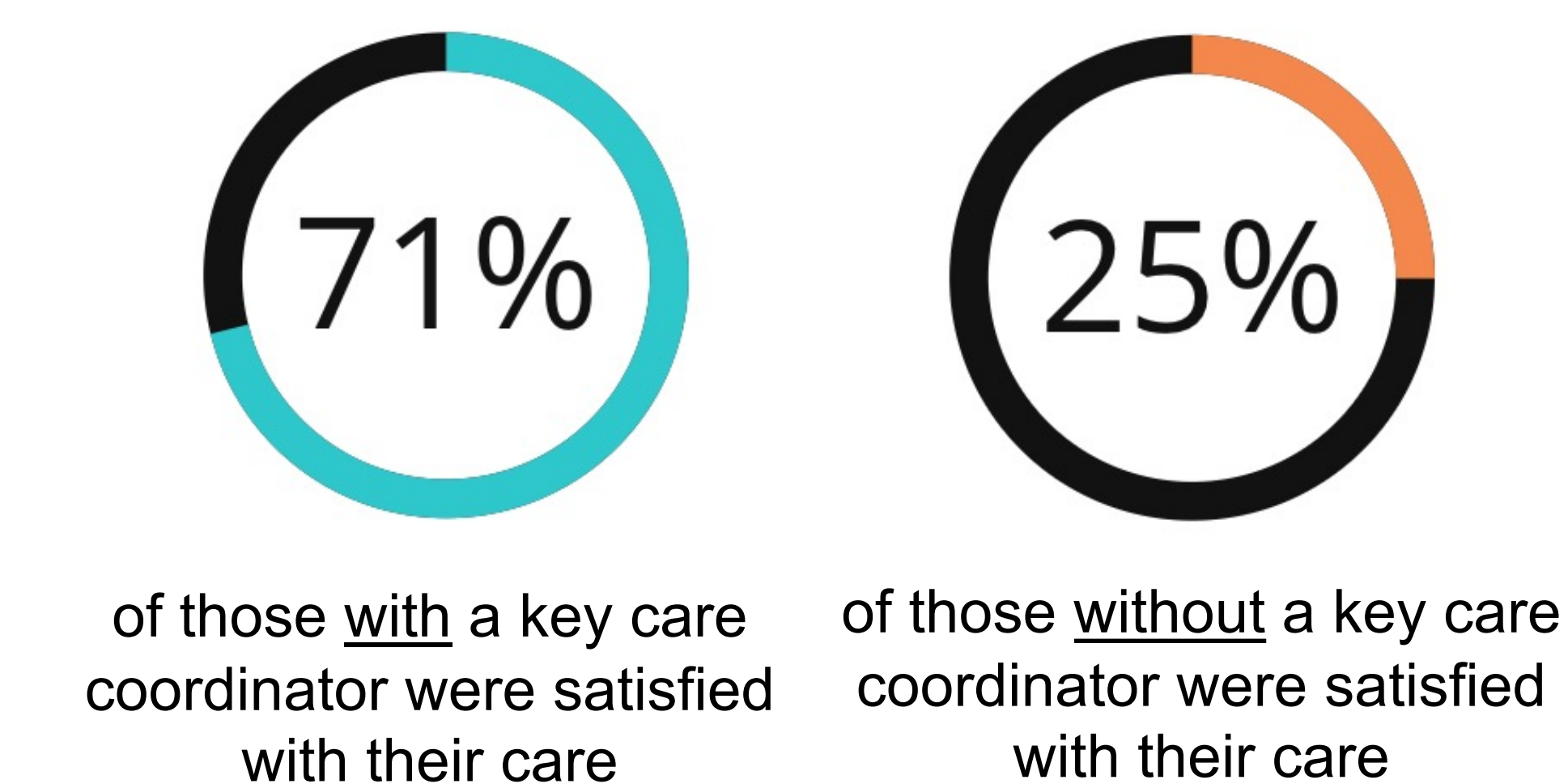
1. Across the UK, **54% of PFC** responses showed **dissatisfaction** with NF1 care.

2. **Regional differences** were notable: **London** had significantly higher satisfaction rate than the national average, while **Scotland's** and **Northern Ireland's** rates were significantly lower.

3. **Factors contributing to satisfaction**

i) **Wait time:**

62% are being seen by a NF1 specialist within 1 year in London, 50% in Scotland and 32% in Northern Ireland.



ii) **Care coordination:** having a coordinator of care positively impacts PFC satisfaction.

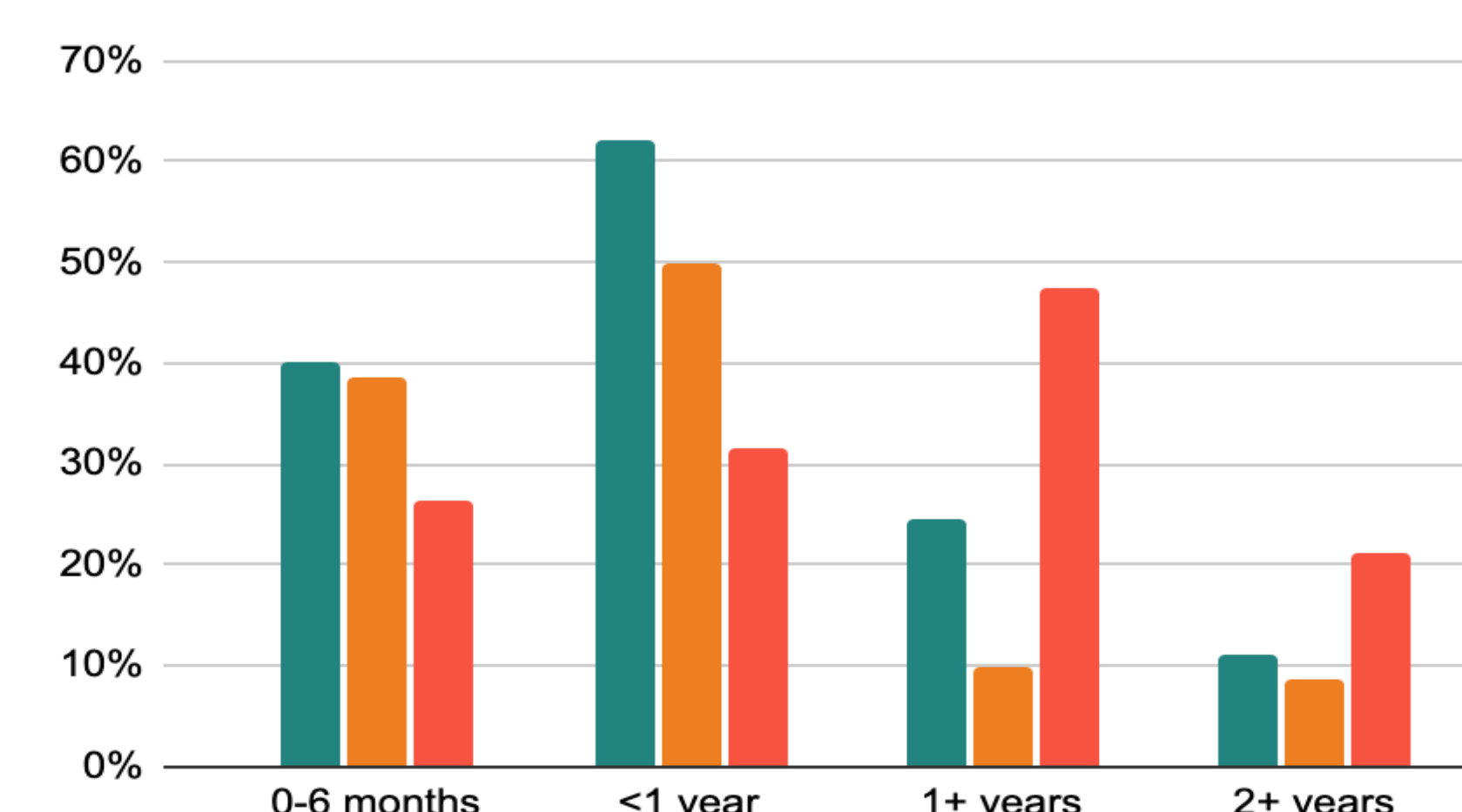


Figure 2: Cumulative proportions of wait times to be seen by a NF1 specialist.

iii) **Support groups and charities:**

69.4% were **NOT signposted**

Proportions of PFCs signposted were significantly smaller in East England and Northern Ireland.

Qualitative

1. **PFC lack of NF1 awareness and understanding:** Difficulties communicating with HCPs and the lack of accessible educational resources led some PFCs to rely on charities for information.
2. **HCP lack of NF1 knowledge and training:** The lack of NF1 knowledge among HCPs, particularly GPs, and their failure to acknowledge PFCs' concerns call for improved training and holistic care.
3. **Regional disparities:** Access to specialist care, resources and funding vary widely by region, creating a 'postcode lottery' for quality care.
4. **Disconnected care:** Care pathways are unclear and uncoordinated, leaving PFCs feeling unsupported, particularly during critical transition periods.
5. **Need for holistic care:** Comprehensive care that caters to the diverse health needs of patients, including their mental health and educational requirements, is crucial.

CONCLUSIONS: The study reveals nationwide dissatisfaction with the NF1 care model, showcasing regional disparities. It highlights the need for establishing standardised national guidelines - with an emphasis on optimising key points along care pathways, including access and care coordination - and improving HCP training and patient education.