

# 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

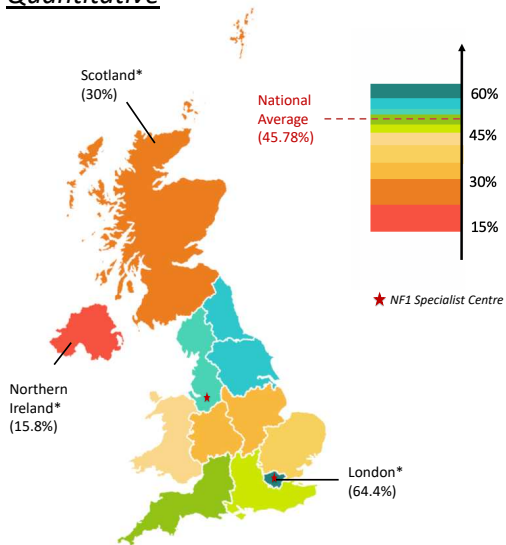
- Conduct an in-depth analysis of PFC's experiences within the NF1 care continuum.
- Identify factors contributing to satisfaction and dissatisfaction.

## 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

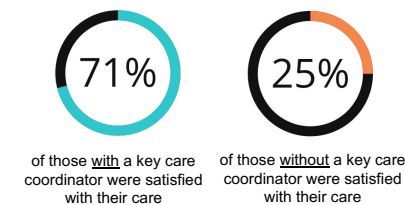


**Figure 1: Heatmap of regional satisfaction rates:** London (64.4%), Northeast and Yorkshire (53.2%), Northwest (52.1%), Southwest (51%), Southeast (47.6%), Wales (42.9%), East England (40.4%), Midlands (40.2%), Scotland (30%), and Northern Ireland (15.8%). \* indicates statistical significance relative to the national average.

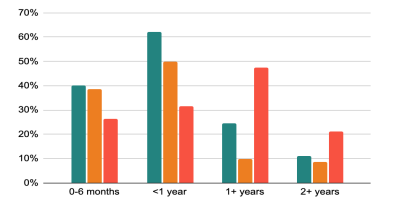
1. Across the UK, **54%** of PFC responses showed **dissatisfaction** with NF1 care.

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.

Acknowledgements: with special thanks to survey correspondents and supporting charities, notably Childhood Tumour Trust, Tumour Support Scotland and Children's Tumor Foundation.

## Introduction & Methods

Neurofibromatosis type 1 (NF1) is a rare genetic condition that causes tumours to grow along nerves, affecting approximately 25,000 people in the UK. Clinical presentation and severity are highly variable, and many patients experience learning and behavioural difficulties. Two NHS England Trusts provide a Highly Specialised Complex NF1 Service; despite their excellent care, the large number and geographical spread of NF1 patients dictate a need to explore how patients can be supported outside the Service. As a first step, in agreement with Service Commissioners, Childhood Tumour Trust initiated a patient led project to learn about the current pathway of care.

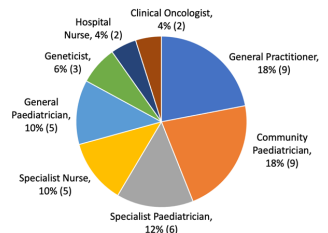
Anonymous online surveys were composed for 1) patients, families, carers and 2) healthcare professionals. Feedback on content and language was gathered from families, NF1 charities, clinical and academic experts, and a trials methodologist. Surveys remained open for 4 months with links cascaded widely through UK patient and healthcare networks via social media, newsletters, email, and relevant websites.

## Respondent Demographics

**Patients, Families, Carers (PFC) Survey:** 1083 responses, 784 completed in full  
**Healthcare Professionals (HCP) Survey:** 94 responses, 49 completed in full



**Region NF1 care is received (■ PFC, n=784) or provided (■ HCP, n=49).**

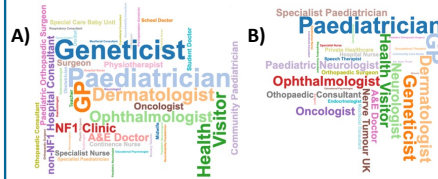
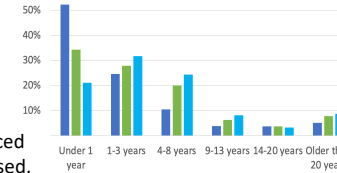


**HCP roles also include one each of:** Audiovestibular Physician, Child Psychiatrist, Disability Nurse, Health Visitor, Medical Student, Neurology, Ophthalmologist, Optometrist.

## Results

### Variation in pathway of care

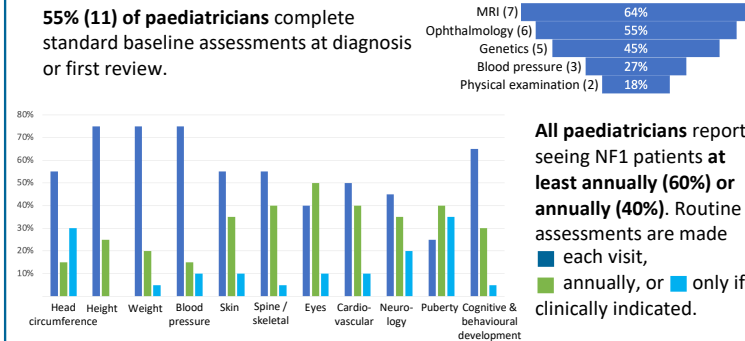
After confirmed or suspected diagnosis, **42% waited 0-6mos** to see a "HCP with NF1 knowledge" (including private healthcare). 15% waited 7-11mos, 1-2yrs, and >2yrs each. 3% have not yet received a formal diagnosis. Variation in age ■ NF1 signs/symptoms noticed by family, ■ acknowledge by HCP, ■ diagnosed.



**Education and healthcare professionals involved in (A) NF1 diagnosis (46 roles; 1-465 mentions) and (B) diagnostic referrals (35 roles; 1-264 mentions).** Word size represents number of PFC & HCP mentions.

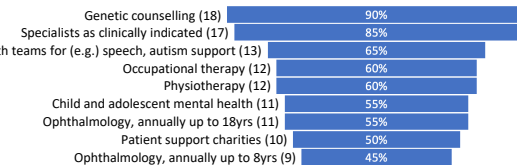
### Variation in clinical care from paediatricians

**55% (11) of paediatricians complete standard baseline assessments at diagnosis or first review.**



**All paediatricians report seeing NF1 patients at least annually (60%) or annually (40%).** Routine assessments are made ■ each visit, ■ annually, or ■ only if clinically indicated.

**Outward referrals for NF1 patients by all paediatricians.**



## Results

### Coordinating care

**72% of HCPs use guidelines for NF1 management.** However, there is little consistency, with respondents using **at least 11 different sources** and 5 different 'local' sources.

**64% of PFCs say pathway of care is unclear,** and **54%** say they are **not satisfied** with the way their NF1 care is managed. Only 30% report being referred to a patient support group or charity.

**40% of PFCs and 51% of HCPs** believe a General or Community **Paediatrician** should be responsible for orchestrating holistic care for children with NF1. **20% of PFCs and 27% of HCPs** believe a **Specialist Nurse** (closely followed by Neurologist and GP) should orchestrate care for adults.

**73% of HCPs** believe these professions need further knowledge, support and financing to provide the desired holistic care.

### Research priorities

Respondents were generally agreed on research priorities. **Special education needs** and **management of plexiform neurofibromas** ranked within the top 3 priorities for both groups. However:

- **66% of PFCs** ranked **benefits of baseline body scan** a top priority, compared to 29% HCPs.
- **61% of HCPs** ranked **monitoring for malignancy** a top priority, compared to 28% PFCs.

## Conclusions

- Rapid uptake and high volume of responses from patients, families, and carers illustrating the desire for change and willingness to be involved in service improvement and new research.
- Opportunity to improve education (especially for early years medics), resources, and a professional network to share knowledge and best practice, leading to improved diagnosis.
- Opportunity to reduce variation of clinical care to improve efficiency, equity and quality of care by implementing standard guidance.
- Opportunity to design patient-centred care networks with key professionals coordinating holistic care and engaging with families.

**Acknowledgements** With special thanks to survey respondents and supporting charities, notably Tumour Support Scotland and Children's Tumor Foundation.

1 Childhood Tumour Trust; 2 Royal Free Hospital; 3 Patient Led Research Hub (PLRH); 4 Manchester University Hospitals (retired) The PLRH (plrh.org) is a collaboration between University of Cambridge and Cambridge Biomedical Research Centre.

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