

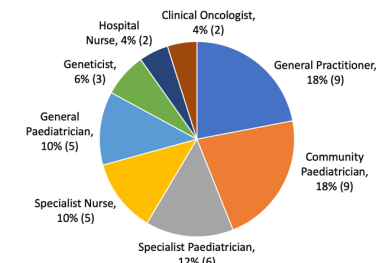
Introduction & Methods

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

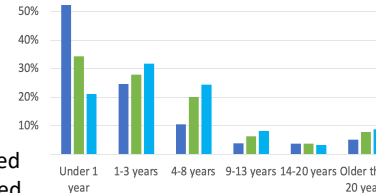


HCP roles also include one each of: Audiovestibular Physician, Child Psychiatrist, Disability Nurse, Health Visitor, Medical Student, Neurology, Ophthalmologist, Optometrist.

Results

Variation in pathway of care

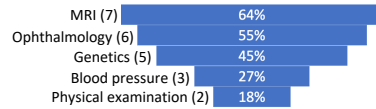
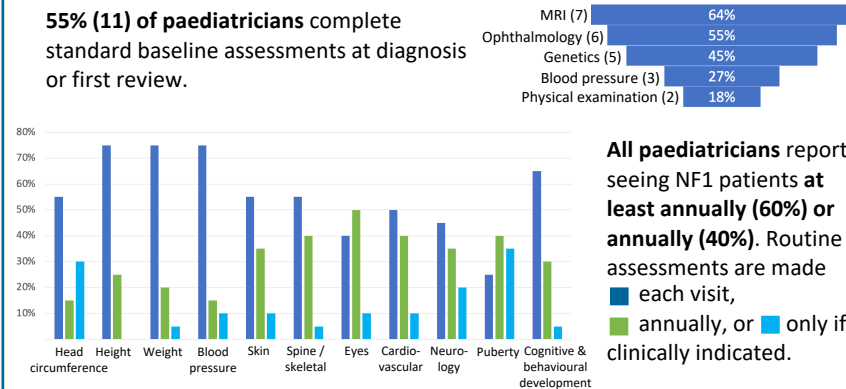
After confirmed or suspected diagnosis, **42% waited 0-6mos** to see a “HCP with NF1 knowledge” (including private healthcare). 15% waited 7-11mos, 1-2yrs, and >2yrs each. 3% have not yet received a formal diagnosis. Variation in age by family, acknowledge by HCP, diagnosed.



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.

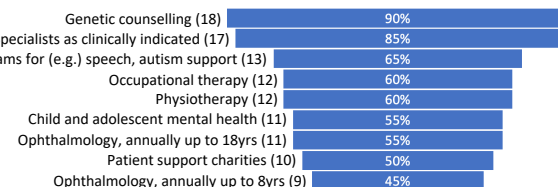
Variation in clinical care from paediatricians

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.

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