

Strengthening NF1 Care: Your guide to what good care looks like

What the NICE Quality Standard for Rare Disease means for NF1 care

This material was developed in collaboration with Nerve Tumours UK and the Childhood Tumour Trust through a co-creation process. Alexion, AstraZeneca Rare Disease provided financial and material support in the development of this resource.

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A guide for families and people living with NF1



What is NF1?



Neurofibromatosis 1 (NF1) is a common genetic condition caused by a 'spelling mistake' in a gene on chromosome 17. It affects around 1 in 2,500 people.¹ NF1 can affect many parts of the body, including the skin, nerves and bones, and can be linked to learning difficulties. The condition varies greatly between individuals, and no two people with NF1 are exactly the same.



Click a stage to find out more



Getting a diagnosis on time

NICE Quality Standard: People referred to a consultant-led service because of concerns about a rare disease undergo diagnostic investigations and have first definitive treatment or a decision on non-treatment in line with national maximum waiting times for non-urgent consultant-led treatment. Under the NHS Constitution in England, people should wait no longer than 18 weeks from GP referral for consultant-led treatment for non-urgent conditions.



What this means for you and your family

01. ○▶▶▶▶

If your GP or paediatric doctor suspects NFI, they should refer you to clinical genetics for further assessment. Genetic testing may form part of the diagnostic process. Following assessment, you should receive a clear outcome, whether that is a confirmed diagnosis, a monitoring plan, a referral to local paediatrics for your child, or the next steps in your care.

02. ○▶▶▶▶

Some people with NFI may be referred to a specialist NFI centre if they have complex health needs and meet the eligibility criteria. Specialist NFI centres are currently based in Manchester or London.

03. ○▶▶▶▶

Because genetic conditions can be complex, timelines may vary. Your healthcare team should keep you informed about what is happening and what to expect next.



Resources and links

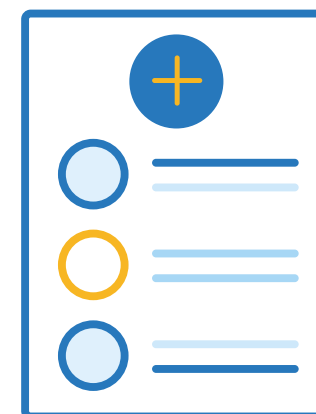


Nerve Tumours UK Specialist NF Advisor helpline if you have questions about referral or diagnosis



If you do not have a diagnosis yet

NICE Quality Standard: People with a suspected rare disease that remains undiagnosed after diagnostic investigations are recognised as a distinct patient group by healthcare services and are on a care pathway that can support future diagnosis.



What this means for you and your family

01. ○▶▶▶▶

If you or your child show some but not all the signs of NF1, you should not be left without support. You should be placed on a regular monitoring pathway with annual reviews. If you or your child begins to develop more features of NF1 or any other unexpected symptoms, your healthcare professional may refer you to Genetics services to be seen more quickly.

02. ○▶▶▶▶

Many of the signs used to diagnose NF1 develop over time. Sometimes a child may have some signs of NF1 but not enough to confirm a diagnosis. Most children with NF1 will meet with diagnostic criteria by around 8 years of age. When uncertain, they should continue to be monitored over time and should not be discharged without a clear explanation of what happens next.

03. ○▶▶▶▶

Your clinician, such as your GP or paediatrician, should track how your or your child's clinical features change over time, including features such as freckling, neurofibromas or Lisch nodules (small harmless spots on the eye).



Resources and links



Childhood Tumour Trust



Genetic Alliance UK



Nerve Tumours UK Specialist
NF Advisor helpline

open Monday, Wednesday and Friday 09.00 - 17.00



Getting the right information

NICE Quality Standard: People undergoing diagnosis for a rare disease, with a newly diagnosed rare disease or with a suspected rare disease that remains undiagnosed after diagnostic investigations, are provided with clear and accurate information to aid self-management.

What this means for you and your family

01. ○▶▶▶▶

From the point of diagnosis, you or your child should receive clear, accessible information about NFI, including what symptoms to look out for and when to seek help.

02. ○▶▶▶▶

Information should be right for your or your child's age. Many people with NFI have learning difficulties, and services should take this into account.²

03. ○▶▶▶▶

Your family and caregivers should also be given information and support. You are not managing this alone.



Resources and links



Nerve Tumours UK Specialist NF Advisor helpline



Childhood Tumour Trust – 'Red Book'* (PCHR) Insert



Childhood Tumour Trust – Your child's been diagnosed with NFI: what now?



Nerve Tumours UK – What is NF?



Childhood Tumour Trust Facebook Support Group



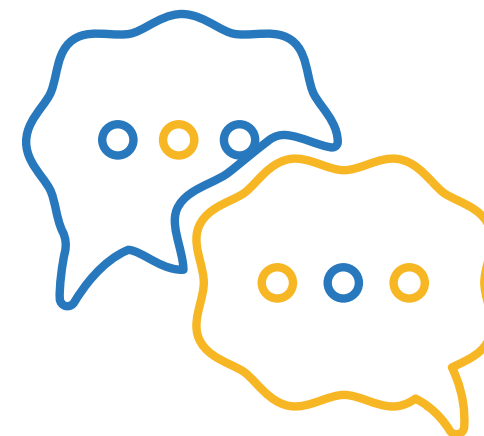
Nerve Tumours UK - Facebook Community Group

*Red Book is not endorsed by Nerve Tumours UK



Having a say in your care

NICE Quality Standard: People diagnosed with a rare disease or with a suspected rare disease that remains undiagnosed after diagnostic investigations are supported to make shared decisions throughout all stages of care.



What this means for you and your family

01. ○▶▶▶▶

You should be supported to make informed decisions at every key point in your or your child's care – whether that relates to monitoring, imaging, surgery, treatment, or future care planning.

02. ○▶▶▶▶

Any treatment options, including for plexiform neurofibromas (a noncancerous tumour that grows on several of the protective coverings around the nerve) must always be clearly explained to you, including the potential benefits, risks, and uncertainties.³

03. ○▶▶▶▶

Decisions should take into account you or your child's quality of life, not just clinical findings. The uncertainty that comes with NF1 should be acknowledged honestly.



Resources and links



Nerve Tumours UK Letter to your GP explaining NF1 and signposting to specialist support



Childhood Tumour Trust Letter to request access to breast cancer screening for women with NF1



Cancer Research UK - Malignant peripheral nerve sheath tumour (MPNST)



Having a named healthcare professional

NICE Quality Standard: People diagnosed with a rare disease or with a suspected rare disease that remains undiagnosed after diagnostic investigations have a named healthcare professional who coordinates their care.



What this means for you and your family

01. ○▶▶▶▶

You should have one clearly identified person who coordinates your or your child's care, you should not have to start again every time you or your child need help.

02. ○▶▶▶▶

Where possible, this should be an NF-experienced nurse or specialist nurse.

03. ○▶▶▶▶

If no regional NF nurse is available, your clinician should communicate clearly who is responsible for your or your child's care and point you towards the Nerve Tumours UK Specialist nurse helpline.



Resources and links



Nerve Tumours UK Specialist
NF Advisor helpline



Nerve Tumours UK – Specialist NF Nurse
Advisor



Support for your whole self

NICE Quality Standard: People diagnosed with a rare disease or with a suspected rare disease that remains undiagnosed after diagnostic investigations have physical, psychological and emotional support needs assessed at key points of the care pathway.



What this means for you and your family

- 01.** Living with NFI affects more than just your physical health. Your or your child's emotional and social needs should be assessed at key points throughout your care – not only during a crisis. The impact of NFI on education, employment, relationships, independence and daily life should be recognised.
- 02.** If you or your child would benefit from psychological support, you should be offered information about available services and how to access them.
- 03.** You and your family should be connected to support networks and peer groups. Connection with others who understand NFI matters.
- 04.** Support should extend to parents, caregivers, siblings and partners, people who are often overlooked.
- 05.** Your or your child's transition points, such as starting school, moving to secondary education, entering employment, leaving home, and transferring from childhood to adult services, should prompt a review of support needs.
- 06.** Where appropriate, you should have access to information about your reproductive choices and alternative pathways to parenthood, as well as being offered psychological support. This should be offered to you regardless of whether you decide to have biological children or seek alternative methods of parenthood.



Resources and links



Childhood Tumour Trust – camps, events and peer support



Nerve Tumours UK Specialist NF Advisor helpline



Nerve Tumours UK mental health support signposting



Children's Tumor Foundation – Family Planning and Reproductive Options in NF



Ready Steady Go Transition Programme



Nerve Tumours UK – Neurofibromatosis type 1 (NFI) a guide for adults and families



Nerve Tumours UK – Help Hub, iNForm



Getting the right treatment, wherever you live

NICE Quality Standard: People with a rare disease have equitable access to treatments when recommended by each nation's health and care guidance body.



What this means for you and your family

- 01.** You or your child should have equal access to appropriate treatments and specialist expertise, regardless of where you live in the UK. Decisions about treatment should be based on clinical need and personal circumstances, not on where you live or whether there is a local specialist service.
- 02.** If you or your child have plexiform neurofibromas (tumours that form a soft, spreading mass involving multiple branches of a nerve rather than a single lump), you should have access to appropriate monitoring, specialist assessment and treatment where needed.⁴
- 03.** Treatment decisions, including any decision not to treat, should always be clearly explained to you, including the benefits, risks and uncertainties involved.
- 04.** If you have questions about your or your child's treatment options, you should know who to contact for advice.
- 05.** Healthcare services should be able to identify people with NFI, particularly those who may benefit from specialist assessment or treatment and ensure they are referred appropriately.



Resources and links



Nerve Tumours UK Specialist NF Advisor helpline, speak to a nurse about your treatment options

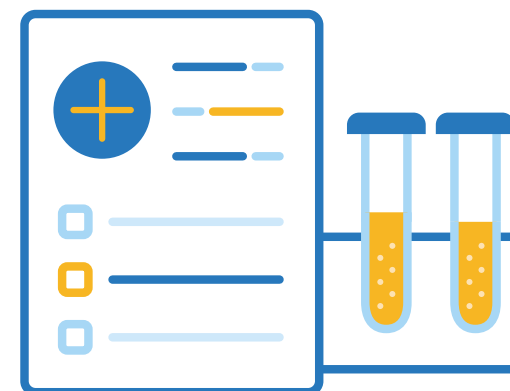


Nerve Tumours UK – Specialist NF Nurse Advisor



Taking part in research

NICE Quality Standard: People with a rare disease are offered the opportunity to take part in clinical research, if available and they are eligible.



What this means for you and your family

01. ○▶▶▶▶

You should be told about relevant research opportunities and supported if you or your child wish to take part.

02. ○▶▶▶▶

Research should be discussed as part of your or your child's routine care, not only at specialist centres.

03. ○▶▶▶▶

You should always have enough time to consider any research invitation, with clear and balanced information to help you decide.



Resources and links



Nerve Tumours UK Research page



NIHR – Be Part of Research



Childhood Tumour Trust – NFI Support & Information Closed Group



Children's Tumor Foundation – NF Registry

References

- ¹ Nerve Tumours UK (2026) *Support for Adults with NFI*. Available at: <https://nervetumours.org.uk/what-are-nerve-tumours/what-is-nfi/support-for-adults-with-nfi> (Accessed: 19 June 2026).
- ² Children's Tumor Foundation. *Learning with NFI: Introduction to the cognitive and social development of children with neurofibromatosis type 1*. New York: Children's Tumor Foundation; 2023. Available from: https://www.ctf.org/wp-content/uploads/2024/02/NFI_Learning_Disabilities_2023_web.pdf
- ³ Mayo Clinic. *Plexiform neurofibroma: symptoms and causes*. Rochester (MN): Mayo Clinic; 2025. Available from: <https://www.mayoclinic.org/diseases-conditions/plexiform-neurofibroma/symptoms-causes/syc-20587495>
- ⁴ Mayo Clinic. *Plexiform neurofibroma: symptoms and causes*. Rochester (MN): Mayo Clinic; 2025. Available from: <https://www.mayoclinic.org/diseases-conditions/plexiform-neurofibroma/symptoms-causes/syc-20587495>





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