B13/S(HSS)/a

2013/14 NHS STANDARD CONTRACT
FOR COMPLEX NEUROFIBROMATOSIS TYPE 1 SERVICE (ALL AGES)

SECTION B PART 1 - SERVICE SPECIFICATIONS

Service Specification No.	B13/S(HSS)/a
Service	Complex neurofibromatosis type 1 service (All Ages)
Commissioner Lead	
Provider Lead	
Period	12 months
Date of Review	

1. Population Needs

1.1 National/local context and evidence base

Neurofibromatosis 1 (NF1) is an inherited neurocutaneous disease that predisposes affected individuals to the development of benign and malignant tumours. The disease involves the skin and nervous system predominantly but people with NF1 can developed a range of rare complications that may affect almost any of the body systems.

Neurofibromatosis 1 has a birth incidence of 1 in 3,000 and a prevalence of approximately 1 in 4,500. Based on a population of 50.7 million there are 11, 267 individuals with NF1 in England. The clinical manifestations of complex NF1 are uncommon. It is anticipated that there will be approximately 130 new referrals per year.

2. Scope

2.1 Aims and objectives of service

The overall aim of the service is to provide the highest quality of care to people with Complex Neurofibromatosis Type 1 (NF1). Specifically the Complex NF1 service aims to provide:

- specialist workup of NF1 patients with suspected complex complications of the disease. Provision of accurate diagnosis of unusual phenotypes and other diseases that can be mistaken for NF1 by genetic testing and counselling
- co-ordinated care from a specialist multidisciplinary team when NF1

complications behave differently from the general population
- monitoring the risk of NF1 related malignancy and tumour progression
- long term monitoring to evaluate need for surgery e.g. the cervical cord compression group.

Note: Surgery, chemotherapy and radiotherapy are not covered in the designation of this national service.

Objectives

- to provide an exemplary and comprehensive service that co-ordinates care for patients with complex complications of NF1
- expert diagnosis of complex NF1 phenotypes and complications utilising the most up-to-date validated diagnostic tools and knowledge
- expert management of patients with Malignant Peripheral Nerve Sheath Tumours (MPNST) shared care arrangements with sarcoma services and through the use of the most up-to-date clinical protocols
- effective monitoring of patients to ensure optimal functioning for the patient with regards to complex complications relating to their NF1
- to operate a rolling programme of clinical audit to test current practice and inform the evolution of care in complex complications of NF1
- to provide care with a patient and family centred focus to maximise the patient experience of care within the nationally designated providers
- to be seen as the leading clinical services and a source of expert advice for the diagnosis and management of Complex NF1 and complications within the NHS
- to support local healthcare providers to manage patients with complex complications in NF1 whenever it is clinically appropriate and safe to do so
- provide high quality information for patients, families and carers in appropriate and accessible formats and mediums
- to develop the experience, knowledge and skills of the multi-disciplinary team (MDT) to ensure high quality sustainable provision.

2.2 Service description/care pathway

The national complex NF1 service is for the medical management of patients with complex complications of NF1 (surgical and oncological treatment provided outside of this specification). The service provides multidisciplinary outpatient in order to:

- work up NF1 patients with suspected complex complications of the disease and give accurate diagnosis of unusual phenotypes or diseases that appear to be NF1 but aren't, through genetic testing, supported with counselling; onward referral and signposting patients to appropriate services
- co-ordinate care where complicating diseases behave differently in NF1 compared with the general population
- monitor risk of malignancy and progression tumours.

Patients are referred predominantly for diagnosis of NF1, support and education, assessment of NF1 related neurological complications, assessment of symptomatic plexiform neurofibromas and the diagnosis of MPNST and central nervous system tumours.

The service consists of a multidisciplinary team of senior clinicians who are conversant with the complications of NF1. In addition the team will have strong links with other disciplines and units that may be needed to provide care for patients with complex NF1.

The multidisciplinary team will consist of a neurologist (adult and paediatric), geneticists (including genetics counselling) and clinical nurse specialists. There are formal links to:

- neurosurgery (adult and paediatric)
- spinal surgery
- plastic surgery
- oncology (adult and paediatric)
- ophthalmology, neuropsychiatry
- respiratory physicians
- positron emission tomography (PET)
- radiology (musculoskeletal and neuroradiology)
- pathology
- peripheral nerve surgery
- sarcoma units
- craniofacial unit
- orthopaedic units

Risk management

Care delivered by the complex NF1 service providers must be of a nature and quality to meet the care standards, specification and agreement for the service. It is the trust's responsibility to notify the commissioner on an exceptional basis should there be any breaches of the care standards. Where there are breaches any consequences will be deemed as being the trust's responsibility.

Patients must be managed in line with the specification and care standards. Any deviation from these which has not been approved by NHS England is at the trust's risk both clinically and financially. It is the trust's responsibility to inform the commissioners of any such non-approved deviations on an exceptional basis.

Where a patient's presentation challenges the assumptions that underpin the specification, service standards and contractual arrangements it is the trust's responsibility to inform the commissioners on an exceptional basis, prior to any treatment (except for emergency treatment) so that the implications of the patient's requirements can be considered. This does not affect situations where the individual funding application process applies.

Days/hours of operation

The service operates Monday to Friday, 0900 to 1700 hrs throughout the year apart from public holidays.

Discharge planning

Patients with complex NF1 may require life-long follow-up, with the frequency of follow-up dependent on the severity of the NF1. However patients with non-complex NF1 will have their care transferred to the appropriate local team as required.

Patients with pseudarthrosis of the long bone and with disfiguring tumours will be evaluated and referred to a specialist unit that is conversant with the management of these complications in people with NF1.

Patients assessed and diagnosed with aqueduct stenosis or disfiguring plexiform neurofibromas will be treated by their local clinical team however they will be continued to be monitored for malignancy in symptomatic plexiform neurofibromas and for need for surgery

2.3 Population covered

This service covers patients registered with an English GP, resident in the European Union (including Republic of Ireland) and eligible for treatment in the NHS under reciprocal arrangements. Patients from Scotland, Wales and Northern Ireland are not part of this commissioned service and the trust must have separate arrangements are in place.

2.4 Any acceptance and exclusion criteria

Referral criteria, sources and routes

Referrals will typically come from consultants but GP referrals will also be accepted. The receiving clinician at one of the providers may request the referrer to carry out further investigations to aid the proper diagnosis of the patient's condition.

Referrals will be accepted by the nationally designated providers via the clinical lead for the complex NF1 service.

The role of the national complex nf1 service is to co-ordinate the care for patients with NF1 with complex complications. The most frequent complex complications which occur in NF1 are:

- **Malignant Peripheral Nerve Sheath Tumours** MPNST (~prevalence 1.5%), is difficult to diagnose and high grade tumours have a 16% five year survival rate. Appropriate clinical assessment PET and, MR/CT imaging should be

performed. The NF1-MPNST MDT should discuss the need for referral to a specialist sarcoma unit. The NF1-MPNST unit and the sarcoma unit will discuss the need for biopsy and the surgical and post-operative treatment in the context of NF1. The treatment required is dependent on diagnosis and stage of the disease. The aim is complete excision of the tumour with tumour free margins. Radiotherapy is used for control of local disease and chemotherapy for metastatic MPNST.

- **Symptomatic optic gliomas pathway (OPG) -** 7% of optic gliomas are symptomatic and most cases become indolent in adulthood. Yearly visual screening should be performed in children to detect patients with symptomatic OPG that may require treatment. The treatment of choice is chemotherapy and should be carried out in collaboration with the local paediatric oncology unit. Radiotherapy is not recommended in children with NF1.
- Patients with the following conditions will be monitored to evaluate the need for surgery:
    - **Symptomatic plexiform neurofibromas with respiratory problems** - NF1 patients with symptomatic plexiform neurofibromas causing respiratory symptoms will be evaluated by the respiratory team;
    - **High cervical cord compression -** NF1 patients with cord compression (prevalence 0.5%) due to neurofibroma in the upper cervical region require expert neurological and neurosurgical assessment. Some people have radiological signs of cord compression without associated signs and symptoms and should be kept under close surveillance. The decision to undertake surgery is based on a combination of clinical and radiological progression.
    - **Sphenoid wing dysplasia** (2%) - NF1 patients with sphenoid wing dysplasia will be evaluated by the team for the need for surgery.
- **Neurofibromatous neuropathy** (<1% uncertain) - Neurological assessment is necessary to characterise and monitor weakness and sensory changes, to differentiate neurofibromatous neuropathy from other treatable forms of neuropathy and from lesions in cauda equina. Treatment is usually pain relief and foot care. Individuals should be monitored for the possible development of MPNST.
- **Multiple Sclerosis (MS)** prevalence in NF1 condition is uncertain. It is anticipated that 20 individuals in the UK with NF1 and MS and we anticipate <1 diagnosis per year. Patients should be monitored jointly by NF1 and MS services because clinical symptoms and signs should be carefully evaluated to determine whether they are related to neurofibromatous or Multiple Sclerosis. Careful deliberation will be needed when considering immunosuppression therapy as NF1 is a tumour predisposing condition.
- **Central nervous system gliomas** (prevalence 1.4%) - Patients will be monitored and managed in collaboration with oncology units
- **Epilepsy** – management of patients who have a dysembryoplastic neuroepithelial tumour (DNET), glioma or focal cortical dysplasia that causes refractory epilepsy.

Patients with pseudarthrosis of the long bone and with disfiguring tumours will be evaluated and referred to a specialist unit that is conversant with the management of these complications in people with NF1.

Exclusion criteria

The national designated complex NF1 service is for the medical management of complex manifestations of NF1 is not commissioned to provide treatment for patients with a confirmed diagnosis of a non-complex NF1.

Response time & detail and prioritisation

The 18-week target applies to this service.

Accessibility/acceptability

The service is accessible to all patients with a suspected or confirmed diagnosis of NF1 regardless of sex, race, or gender**.** Providers require staff to attend mandatory training on equality and diversity and the facilities provided offer appropriate disabled access for patients, family and carers. When required the providers will use translators and printed information is available in multiple languages.

The provider has a duty to co-operate with the commissioner in undertaking equality impact assessments as a requirement of race, gender, sexual orientation, religion and disability equality legislation

Those patients found to have a clinical picture and complications consistent with complex NF1 will remain within the national service. Patients who do not fit this profile will be managed by the provider under alternative commissioning arrangements and/or an effective and safe transition to other services will be managed by the provider.

2.5 Interdependencies with other services

Internally the complex NF1 team will link with multiple clinical and administrative teams as a result of the composition of the broad MDT.

External to this the nationally designated complex NF1 providers are the leaders in the NHS for patient care in this area. They provide a direct source of advice and support when other clinicians refer patients into the nationally designated providers. This support will continue until the patient is transferred into the nationally designated provider or it becomes apparent that the patient does not have a confirmed diagnosis of NF1.

The nationally designated providers also provide education within the NHS to raise and maintain awareness of NF1 and its complication and their management.

The national providers will form a relationship with local health and social care providers to help optimise any care for complex NF1 provided locally for the patient. This may include liaison with consultants, GPs, community nurses or social workers

etc.

The national providers form part of the network site-specific group for sarcoma across England.

3. Applicable Service Standards

3.1 Applicable national standards e.g. NICE, Royal College

The nationally designated complex NF1 providers must be fully integrated into their trust's corporate and clinical governance arrangements.

4. Key Service Outcomes

The overall outcome measure for the national complex NF1 service is the reduction of the impact of complex complication on patients and therefore clinical outcomes are different depending on the specific clinical presentations. Outcomes the service collects are:

- new diagnosis/change diagnosis
- new NF1 complication identified
- new non-NF1 complication
- number of OPGs detected on visual screening / magnetic resonance imaging (MRI)
- surgery/intervention avoided
- surgery/intervention facilitated
- deaths.

5. Location of Provider Premises

The service is delivered by two designated centres based in the north and south of England for the national caseload and provides outreach support into major cities. Designated services are based at:

- Guys and St. Thomas' NHS Foundation Trust, London (GSTT)
- Central Manchester University Hospitals NHS Foundation Trust (CMFT)

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