Neurofibromatosis type 1 (NF1) - onward care in adults

Depending on complexity of the patient's NF1, their care is transferred to the appropriate service at transition by the paediatrician or transition clinics:

Non-complex NF1 - under the care of primary care physicians
Complex NF1 - under the care of NF1 services Complex service

Every patient should have an annual blood pressure review

CLINICAL ISSUES	FEATURES	ACTION
Cardiovascular	Hypertension diagnosis and management as per NICE hypertension guidance - confirmed by ambulatory or 24-hour monitoring	Refer all patients with hypertension for investigation of secondary causes (e.g phaeochromocytoma and renal artery stenosis)
Skin	Urgent - any lump that's rapidly growing, persistently painful and/or changes in appearance or texture	Urgent 2 week wait referral
	Routine- any lumps causing discomfort, cosmetic concerns, irritation, infection or severe psychiatric distress	Routine referral as per local policy for skin lesions
	Pruritus/Neuralgia- neurofibromas may become itchy.	Manage as per normal practice. (e.g. titration of antihistamines for itch)
Pain	Pain persisting for >2 weeks, that causes nighttime waking or affecting activities of daily living e.g. rapidly growing intra-abdominal nerve tumours	Urgent referral (to appropriate specialty depending on site)
Neurology	New or changing neurology - Headaches, changes to vision, balance, personality, weakness or seizures	2 week wait referral +/- direct access CT/MRI
	Non-motor peripheral neurological symptoms- paraesthesia or numbness	Referral to neurology (urgency depends on the severity and rate of progression of symptoms)
Eyes	Changes to visual acuity, colour vision, visual fields, proptosis or new squint	Urgent referral to ophthalmology
		Annual optometry review + onward referral if necessary
Skeletal	Osteoporosis, osteopenia, vitamin D insufficiency/deficiency.	Recommend supplemental vitamin D +/- blood tests as per regional policy and further investigation if indicated
Reproductive issues	Pre pregnancy	Clinical Genetics referral to be sent as early as possible pre-conception - for counselling and onward planning
	Routine antenatal care - monitor BP and neurofibromas for growth	Routine antenatal care
Women	Increased breast cancer risk- as per NICE moderate risk guidance	Screening and management as per the NICE guidance for moderate risk factors for breast cancer.

REFERRAL TO THE COMPLEX NF1 TEAM

Criteria for referrals found at: Neurofibromatosis Type 1 | Manchester Centre for Genomic Medicine (mangen.co.uk)

If criteria is not met completely the complex NF1 team are available for advice/discussion of cases or imaging at MDT meetings via the above contact details

NHS
North East and Yorkshire
Genomic Medicine Service

NF1 features to be aware of:

- Learning difficulties and behavioural disorders e.g. ADHD and autism - NF1 patients are at higher risk
- Psychological issues
- Fatigue
- Generalised muscle aches and pains
- Scoliosis, congenital bowing or pseudoarthrosis of long bones usually tibia
- Short stature

Red flags

- Vision: changes to visual acuity, visual fields etc. - may indicate an optic glioma or CNS tumour
- Central neurology: headaches, seizures, vision disturbance, gait - may indicate a CNS tumour
- Skin: painful, rapidly growing or changing skin lesions - may indicate malignant peripheral nerve sheath tumour
- Cardiovascular: hypertension, episodic headache, sweating, tachycardia - may indicate phaeochromocytoma
- Pain: new onset, severe in nature, waking from sleep, not resolving after 2 weeks - may indicate malignant transformation of internal neurofibroma

Resources:

- Nerve Tumours UK | Home | Nerve Tumours UK
- Childhood Tumour Trust
- Neurofibromatosis Type 1 | Manchester Centre for Genomic Medicine (mangen.co.uk)
- NICE Hypertension in adults: diagnosis and management | Guideline NG136
- NICE breast cancer guidance. (NICE CG164)