

Neurofibromatosis – eye screening

Information for Referrers

Recommended Assessments Whilst Waiting For Ophthalmology Appointment

There is currently a substantial wait for an initial appointment with a consultant ophthalmologist at Women's and Children's Hospital and therefore we recommend that you arrange the following whilst the patient awaits an appointment.

- 1. MRI Head and Orbits with Gadolinium** – the department recommend all patients with Neurofibromatosis (suspected and genetically confirmed) undergo MRI scan as evidence shows that children with negative MRI screening at age 15 months or later did not develop symptomatic optic nerve glioma (reference below).
- 2. Optometry assessment annually age 4 and over** – focusing on assessment of unocular visual acuity, colour vision, pupil responses, anterior segment assessment (to detect presence of lisch nodules in cases who have not yet had genetic testing), retinal check and optic nerve check.

MRI Reference for Clinicians: Prada CE, Hufnagel RB, Hummel TR, Lovell AM, Hopkin RJ, Saal HM, Schorry EK. The Use of Magnetic Resonance Imaging Screening for Optic Pathway Gliomas in Children with Neurofibromatosis Type 1. *J Pediatr.* 2015 Oct;167(4):851-856

What to look out for in NF1:

Potential Eye Findings in NF1

- Lisch Nodules
- Plexiform Neurofibromas of the lid
- Optic Nerve Glioma – affecting approximately 15% of patients ([Source](#)).

Rare Eye Findings in NF1

- Retinal Hamartomas
- Choroidal Hamartomas
- Retinal Vasoproliferation Tumours
- Prominent Corneal Nerves
- Heterochromia
- Conjunctiva Chroistoma
- Multifocal Chroidal Nevi
- Uveal Melanoma
- Infantile Glaucoma

For more information

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Figure 1 - Lisch nodules on a blue iris ([Source](#)). These are gold, tan or brown in colour and are dome-shaped gelatinous benign lesions on the surface of the iris (melanocytic hamartomas). They rarely cause any issues



Figure 2 - Plexiform Neurofibroma of right upper eyelid causing visually significant ptosis ([Source](#))

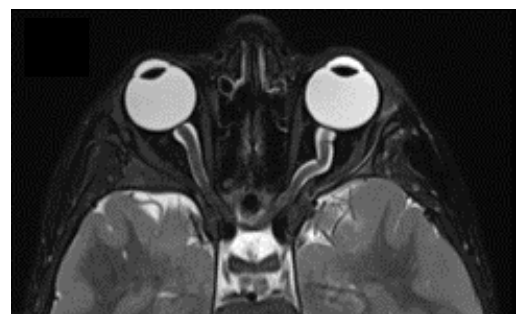


Figure 3 - Left optic nerve glioma shown on coronal T2-weighted MRI with visible kinking of the nerve ([Source](#))



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