Sir,
Screening for symptomatic optic pathway glioma in children with neurofibromatosis type 1

We would like to clarify some important issues raised in the debate about screening children with NF1-associated optic pathway glioma (OPG).^{1,2} The principles of population mass screening are not applicable to the detection of OPG in young children with NF1—a significant complication in a high-risk group of patients. Ophthalmological assessments aim to detect visual impairments resulting from a symptomatic OPG rather than to identify all NF1-associated OPG, at least half of which will never cause signs or symptoms.³ Age-appropriate visual testing is recommended throughout the first decade of life to identify children requiring increased surveillance.³

MRI does not predict clinical behaviour or preclude the later development of an OPG; moreover, incidentally identified NF1-associated OPGs rarely progress or require treatment.⁴ In the absence of refractive error, reduced visual acuity or proptosis in a child with NF1 is highly predictive of an OPG, while cataracts and retinal disease are rarely detected in this population. The significant causes of non-correctible visual loss in NF1 children are OPG, glaucoma secondary to a plexiform neurofibroma involving the eyelid, or proptosis and optic nerve damage due to a retro-orbital plexiform neurofibroma. While the latter two may be visible, detailed ophthalmological examination including fundoscopy is necessary to detect a symptomatic OPG.

In current practice, the goal of annual age-appropriate visual assessments in children with NF1 is to identify abnormalities in visual function attributable to an OPG. These assessments should continue until at least 7 years of age, when the risk of visual impairments from OPG is significantly reduced.^{4,5} The finding of non-refractive decreased visual acuity warrants neuroimaging for identification and localisation of OPG, followed by serial visual testing and neuroimaging. Visual progression, defined as a two-line decrement in visual acuity, is an indication for chemotherapy.

Recently, the National Commissioning Group funded a 'Complex NF1' service in London and Manchester. A cardinal aim is to work with local ophthalmologists and optometrists to perform annual visual screening for OPG in NF1 children under 8 years. This prospective assessment will facilitate a nationally cohesive screening programme for OPG in a high-risk group of children.

Conflict of interest

All the authors lead large NF1 services and manage children with NF1-associated OPG. The corresponding author is lead clinician in the National Commissioning Group Complex NF1 service at Guy's and St Thomas' NHS Foundation Trust, London.

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Sir,
Response to Gutmann *et al*

We are pleased that the publications have provoked open debate and thank Gutmann *et al* for their comments.¹

We concur that the current recommendations of ophthalmology appointments for children with NF1 do not constitute screening, and should not be labelled as such. A recent audit of the NF1 OPG screening service in Manchester revealed that screening appointments were poorly attended and no OPGs were detected during annual ophthalmology screening over a 7-year, 128-episode period.²

The term 'age appropriate screening' remains problematic on several levels. At least 40% children with NF1 have developmental delay or learning disability and have difficulty performing the tests.³ Learning disability is associated with cerebral visual impairment (CVI); CVI and amblyopia reduce vision. If there are no 'normal' data for visual development in children with NF1, how are we to define the 'reduced vision' that is 'highly predictive of OPG'?¹

Perhaps what we should all be working towards is 'developmentally appropriate screening' and use the ongoing national project to collect the 'normal' data we need, so that we may be better informed when making the next round of recommendations.

Conflict of interest

The authors declare no conflict of interest.