Ophthalmologic Screening for Optic Pathway Glioma in Neurofibromatosis Type 1

As pediatric ophthalmologists, we are frequently referred children with neurofibromatosis type 1 (NF1) for ophthalmic examinations. We are usually consulted to aid in the diagnosis by determining whether iris Lisch nodules are present. It is generally accepted that the incidence of these nodules increases by 10% per year of age in the first 10 years of life in children with NF1. Another significant aspect of our care includes examining patients known to have NF1 for the symptoms and signs of optic nerve and chiasmal gliomas. These include poor vision, proptosis, afferent pupillary defects, and optic nerve atrophy or papilledema. The American Academy of Pediatrics (AAP) has published recommendations for patients with NF1 to have annual ophthalmic examinations from ages 1 to 7 years and every 2 years from ages 8 to 18 years. This recommendation is due to the increased risk of optic glioma in this population.

In this issue, Parkhurst and Abboy found an incidence of 4.2% for gliomas in 708 patients. Half of the patients diagnosed presented with symptoms that included vision loss, proptosis, and precocious puberty. The AAP does not suggest routine magnetic resonance imaging (MRI) screening for patients with NF1 because the incidence and morbidity from these slow-growing tumors is low. An MRI should be obtained if there are visual changes, persistent headaches, seizures, marked increase in head size, or a plexiform neurofibroma of the head, or for a small percentage of the NF1 population for whom a deletion of the entire NF1 gene and flanking DNA is found. The authors found that 14 of the 15 children initially found to have an optic glioma by routine screening MRI were asymptomatic and remained so for the duration of the study. Their findings support the AAP position against routine MRI screening. A disturbing finding was that most children with a diagnosis of NF1 in their cohort did not even have one ophthalmologic appointment prior to their diagnosis. Clinical screening for optic glioma in children needs to be consistent. In line with the AAP recommendations, the authors’ data indicate that annual ophthalmologic examination in children with NF1 and screening for precocious puberty are extremely important for early detection.

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REFERENCE