A CASE REPORT: Optic glioma in a child with NF1

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Abstract

A 10 year old female patient presented with progressive right eye proptosis (Fig 1) and skin rash of three years duration was seen at Tikur Anbessa Specialized Hospital, department of pediatrics hematology /oncology unit. Physical examination showed mildly decreased visual acuity and ***cafe*** *au lait spot (Fig 2) AND axillary freckling and Orbital CT (Fig 3) showed right intraorbital mass with an assessment of right optic nerve glioma she is to be started on weekly vinblastine at a dose of 6mg/m2.*

INTRODUCTION

Optic nerve glioma (also known as optic pathway glioma) is the most common primary neoplasm of the optic nerve. Along with reducing visual acuity in the affected eye, the tumor sometimes produces additional symptoms as it grows. A low-grade form of this neoplasm, benign optic glioma, occurs most often in pediatric patients. Another form, aggressive glioma, is most common in adults; it is frequently fatal, even with treatment.[1] Optic-pathway glioma accounts for 1-5% of all brain tumors in children [1]. About half of these cases occur in children with neurofibromatosis type 1 [2]. The diagnosis is usually rendered before age 6 years, although there are some reports of older ages [3,4]. The vast majority of optic-pathway gliomas in children are pilocytic astrocytomas [2,5]. The tumor may arise anywhere along the optic pathway, from just behind the globe to the lateral geniculate body [5,6]. In patients with neurofibromatosis type 1, the tumor is usually smaller than in sporadic (non-neurofibromatosis type 1-associated) cases [5,7].

The clinical presentation is variable. In patients with neurofibromatosis type 1, 40-80% of optic-pathway gliomas are asymptomatic at diagnosis, whereas in sporadic cases, they are symptomatic [3,5,8-10]. The most common signs are vision-related: mainly visual loss, decreased visual acuity, and strabismus. Other findings include endocrine disturbances, signs of increased intracranial pressure, and hydrocephalus [2,5,8,9,10-13]. Ophthalmologic examination may reveal decreased visual acuity, pathologic visual fields, proptosis and more [9,10,13,14]. The generally young age of the children and high prevalence of neurofibromatosis type 1-associated attention deficit hyperactivity disorder render the ophthalmologic examination difficult, and decrease its sensitivity and specificity [2,15]. Early diagnosis is important so that the tumor can be carefully monitored and treatment can be administered early, before visual deterioration. The diagnosis can be made functionally by visual-evoked potentials, but as is the case for eye examinations, their efficacy is limited, and specificity is low [2,16]. Modern neuroimaging modalities provide excellent characterization of optic-pathway gliomas, obviating the need for biopsy [17]. Magnetic resonance imaging was found to be superior to computed tomography for the detection and evaluation of extensive tumor involvement. It has a higher specificity, and can be used to assess disease progression [18,19]. The biological behavior of optic-pathway glioma varies. The tumor may progress rapidly, remain stable for years, or even shrink spontaneously or after biopsy, mostly with clinical improvement [2,5,7,20]. Regrowth after a stable period or after biopsy was also reported [2]. Tumor progression is apparently affected by the presence of neurofibromatosis type 1, patient age, and tumor location [21]. Less progression was evident in patients with neurofibromatosis type1 than in sporadic cases, and in children who were older at diagnosis [2,5,7,9,10,19,22]. Tumors situated at the optic nerve or chiasma tend to grow more slowly and less aggressively than chiasmatic/hypothalamic gliomas, with lower mortality [23,24]. Posterior involvement may also lead to significant morbidity and mortality [6,14,25].The natural history of optic-pathway gliomas in neurofibromatosis type 1 is considered unpredictable [2,26]. Hence deciding whether, or when, to initiate treatment becomes difficult. The presence of an optic-pathway glioma in a patient without neurofibromatosis type 1 is considered an indication for treatment [5]. Although neurofibromatosis type 1 is thought to be relatively benign, given the risk of visual impairment, blindness, neurologic deficits, or death [27,28], patients affected by the tumor should be monitored routinely for its size and visual function, and an adverse change in either should be considered an indication for treatment [2]. If a glioma tends to remain stable, the

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intervals between magnetic resonance examinations can be gradually increased [10]. Nevertheless, the subjective timing of imaging scans and the lack of objective references to identify deviations from normality place patients at risk of either unnecessary or insufficient neuroimaging. An optic-pathway glioma tends to grow along the optic pathways by increasing its width, rather than as one ‘‘concentric’’ mass that grows in all directions [5]. As such, stereotypical patterns of growth as seen on imaging scans of children with optic-pathway glioma are often highly comparable, because the tumor tends to involve the same brain structure, and the manner of growth is very similar.

CASE REPORT

A 10 year old female patient presented with progressive right eye proptosis and skin rash of three years duration was seen at Tikur Anbessa Specialized Hospital, department of pediatrics hematology /oncology unit. Since the last 6 months the proptosis was more progressive to attain the current size. Family history is positive for paternal unilateral loss of vision unrelated to trauma. Physical examination showed mildly decreased visual acquity (OD =6/9, OS =6/6) and *cafe au lait* spot(Fig 2) and axillary freklings other wise no other findings on the musculoskletal and CNS. Orbital CT(Fig 3) showed right intraorbital mass with an assessment of right optic nerve glioma she was started on weekly vinblastine at a dose of 6mg/m2.

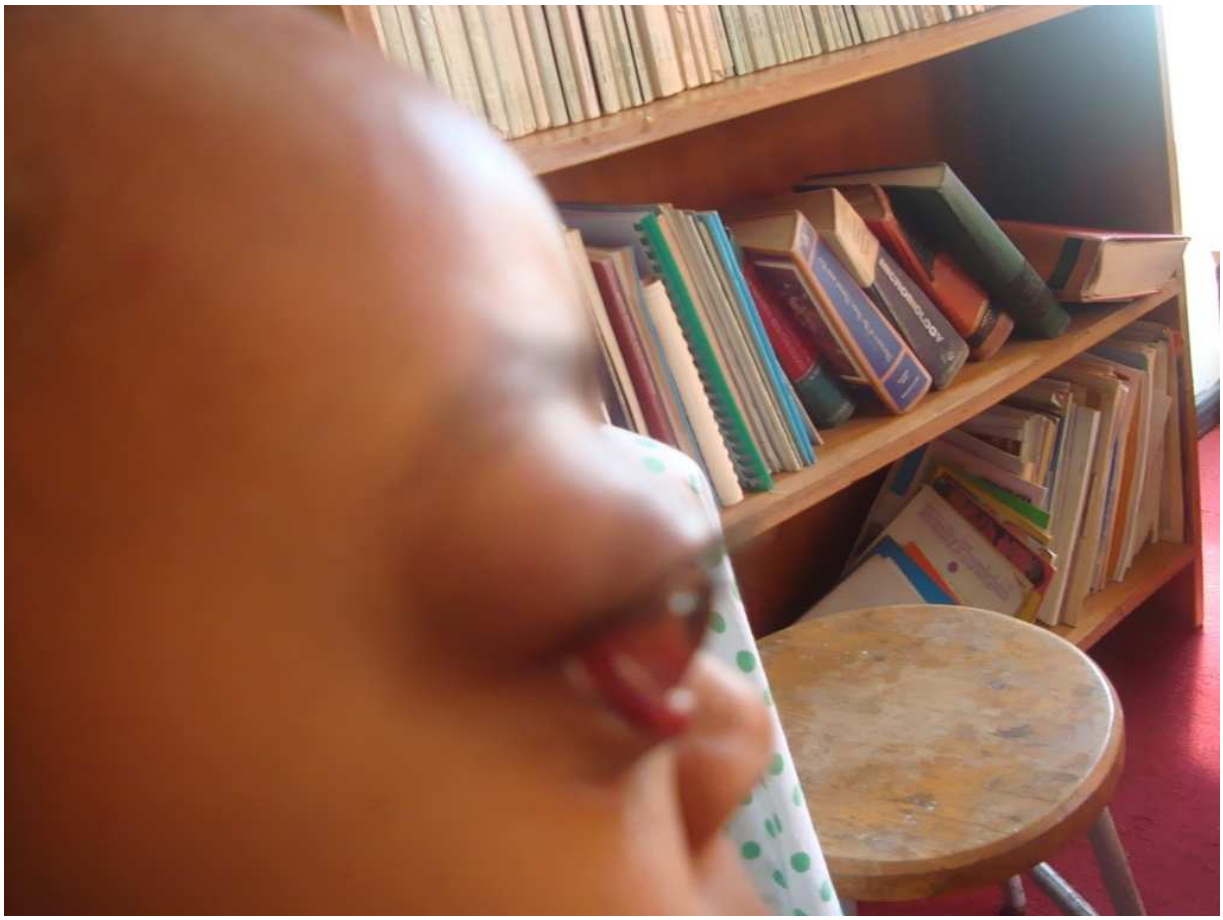

Fig 1

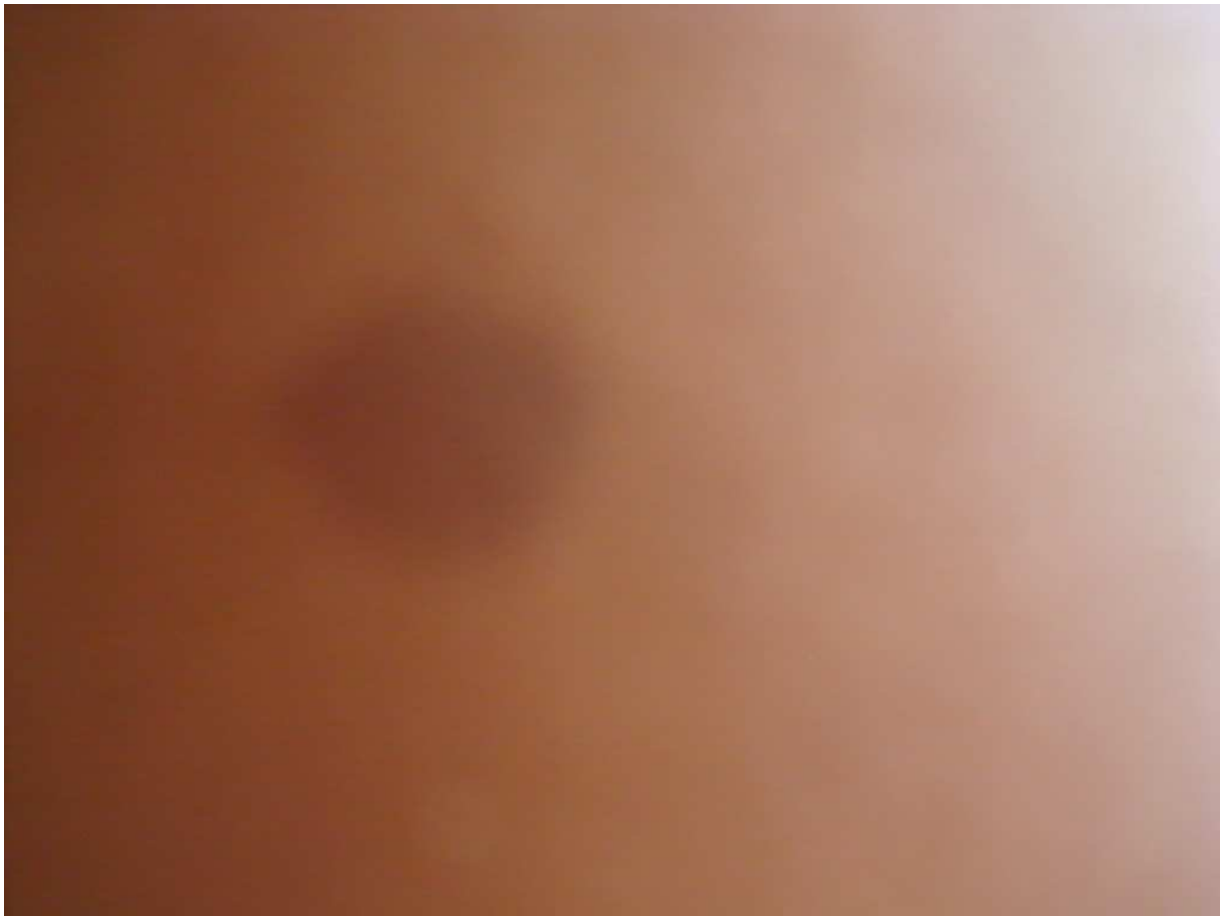

Fig 2

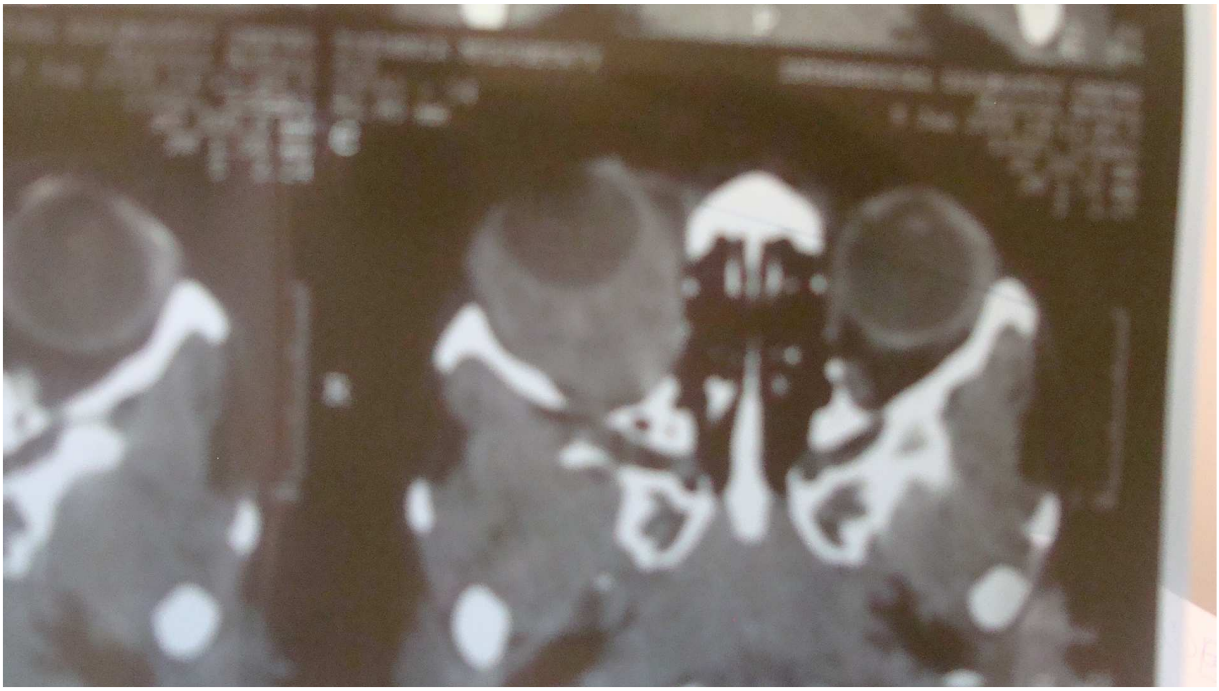

Fig 3

Discussion

This is one of the rarely reported case of a 10 year old Ethiopian child with type 1 neurofibromatosis and right optic nerve glioma. In most young patients with optic glioma the presenting symptom is painless proptosis. Optic atrophy is common, as is reduced visual acuity, although the latter may be a late symptom. A large lesion may compress the optic chiasm, causing nystagmus or other symptoms. Hypothalamic symptoms, such as changes in appetite or sleep, also may occur. Massive lesions may compress the third ventricle, resulting in obstructive hydrocephalus accompanied by headache, nausea, and vomiting also may occur but these findings were not found in this patient. Historically, surgery and radiotherapy have played a primary role in management, however, in the last 15 years, chemotherapy has evolved into the first-line treatment of choice. The case presented was started on weekly Vinblastine at a dose of 6 mg/m2 for one year with regular ophthalmologic and Orbital CT/MRI if feasible.

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