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Neonatal Buphthalmos as the Initial Presentation of Orbitofacial Neurofibromatosis Mimicking a Maxillary Vascular Malformation: A Case Report

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Abstract

Neurofibromatosis type 1 is the most common phacomatosis which is associated with multiple systemic involvement. Secondary congenital glaucoma may be one of the signs of NF1 although very rare. We report a case of 20 days old neonate who presented with buphthalmos who later developed soft tissue swelling involving the temple and was initially radiologically miming vascular malformation with later development of plexiform neurofibroma at the age of 4 month. This case is unique in that the baby was initially misdiagnosed as primary congenital glaucoma and there was a delay in the diagnosis of NF1 especially that radiological imaging supports the diagnosis of vascular malformation.

Keywords: ***Orbitofaical neurofibromatosis; Buphthalmos; Neonates; Vascular malformation***

1. Introduction

Neurofibromatosis type 1 (NF1) is a genetic disorder characterized by the development of benign nerve sheath tumors, which may occasionally undergo malignant transformation [1].

The National Institutes of Health (NIH) Consensus Conference on Neurofibromatosis established the diagnostic criteria for NF1 in 1987, which were subsequently reviewed and reaffirmed in 1997. Diagnosing NF1 in neonates is particularly challenging, as many of the characteristic diagnostic features may not yet be apparent at this age, especially in the absence of a positive family history [1]. Early diagnosis is crucial, as it allows prompt monitoring and anticipation of potential systemic

complications [2]. Orbitofacial neurofibromatosis represents a specific subtype of NF1 and is characterized by progressive disfigurement involving the orbit and temporal region. It accounts for approximately 1%-22% of all NF1 cases. Plexiform neurofibroma, which is considered pathognomonic for NF1, is a hallmark feature of this subtype [3].

Secondary congenital glaucoma has been reported in association with NF1, although it remains a very uncommon manifestation [4]. In most reported cases, glaucoma is preceded by other classical features of NF1. In contrast, in this case, buphthalmos was the initial and presenting manifestation.

2. Case Presentation

This paper reports a case of a 20-days-old neonate who was diagnosed with congenital glaucoma and referred for a second opinion and possible surgical intervention. He was the first child of healthy parents, born full-term, with no reported prenatal complications or family history of glaucoma. At presentation, the neonate was already receiving topical combination therapy with beta-blockers and carbonic anhydrase inhibitors. According to the mother, the condition had been present since birth, when she first noticed enlargement of the left globe. On initial examination, there was evident enlargement of the left globe with questionable minimal proptosis, which could not be reliably assessed clinically at this early age. The corneas were clear in both eyes. Intraocular pressure (IOP), measured using the iCare tonometer, was 16 mmHg in the right eye and 40 mmHg in the left eye.

The patient was prepared for examination under general anesthesia (EUA), and a prostaglandin analogue was added to the left eye pending completion of investigations. Magnetic resonance imaging (MRI) of the brain and orbits was requested to confirm the presence of proptosis and to exclude secondary causes of glaucoma. Additionally, A-scan ultrasonography and ultrasound biomicroscopy (UBM) were performed. MRI revealed a normal brain study with enlargement of the left globe and thickening of the lateral rectus muscle. A-scan ultrasonography demonstrated an axial length of 18 mm in the left eye. UBM showed a normal, open anterior chamber angle in all quadrants with an average corneal thickness.

Full assessment was done and demonstrated normal findings on echocardiography and pelviabdominal ultrasonography, with a clear chest examination and no abnormalities detected on neurological assessment. Examination under general anesthesia revealed an IOP of 14 mmHg in the right eye and 23 mmHg in the left eye while on combined topical beta-blockers and carbonic anhydrase inhibitors. Horizontal corneal diameters measured 10 mm in the right eye and 12.5 mm in the left eye. Anterior segment examination demonstrated clear corneas bilaterally, with prominent iris vessels, particularly in the left eye. Fundus examination revealed a cup-to-disc ratio of 0.5 in the right eye—which more or an average IOP and corneal diameter for age—and 0.9 in the left eye. Additionally, soft tissue swelling was noted in the temporal region.

Based on these findings, a compressive orbital lesion causing secondary glaucoma was suspected. Differential diagnoses included capillary hemangioma, lymphangioma, orbital varices, neuroblastoma, optic nerve glioma, and rhabdomyosarcoma. Given the patient's age, computed tomography was initially avoided. MRI of the brain and orbits with contrast and facial ultrasonography were therefore requested. Facial ultrasonography demonstrated a multicompartmental vascular malformation suggestive of hemangioma, which was supported by contrast-enhanced MRI findings.

Multidisciplinary consultations—including neurology, maxillofacial surgery, vascular surgery, oculoplastics, and oncology—were obtained. No surgical or interventional management was recommended at that stage, and systemic beta-blocker therapy was initiated. The parents were lost to follow-up and returned two months later, when the child was four months old, reporting progressive enlargement of the soft tissue swelling. Vascular and oncology departments advice was to repeat facial ultrasonography which raised suspicion for neurofibromatosis: so, their decision was to do A guided biopsy that demonstrated schwannoma cells, and subsequent computed tomography confirmed the diagnosis by revealing a plexiform neurofibroma.

FIG. 1. **patient at the initial manifestation showing left eye buphthalmous with query proptosis.**

FIG. 2. **Patient at 4 months showing left eye ptosis with S shaped formation and proptosis.**

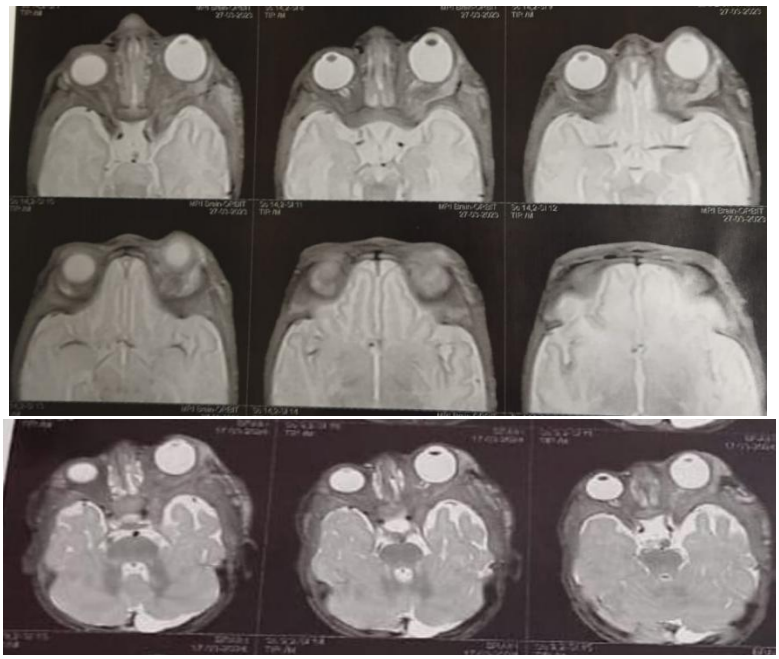

FIG. 3. **MRI showing soft tissue mass lesion in the left ptrygo palatine fossa associated with enlarged left cavernous sinus and enlarged lacrimal gland.**

3. Discussion

Neurofibromatosis type 1 (NF1) is the most common phacomatosis, with an estimated incidence of approximately 1 in 3,000 live births, and follows an autosomal dominant inheritance pattern; however, nearly 50% of cases arise from de novo mutations. Diagnostic criteria for NF1 were initially established by the National Institutes of Health (NIH) and were later revised [5].

Only a few cases of neonatal neurofibromatosis have been reported in the literature. In most of these cases, there was a positive family history, or the diagnosis was made at an older age than in this patient [6-8].

Neurofibromas are classified into three types: plexiform, diffuse, and localized. Among these, NF1 most commonly manifests as plexiform neurofibroma (PN), which is considered pathognomonic for the disease [9].

Although uncommon, early childhood glaucoma associated with NF1 may represent the earliest manifestation of the disease and can precede the appearance of classical systemic features. Unilateral congenital glaucoma accompanied by buphthalmos, proptosis, or ptosis should therefore raise suspicion for NF1, even when initial neuroimaging findings are inconclusive [10].

Grant and Walton proposed four theories in 1964 to explain the etiology of glaucoma associated with NF1: (1) developmental anomalies of the iridocorneal angle; (2) infiltration of the ciliary body or iridocorneal angle by neurofibromas; (3) presence of retrobulbar plexiform neurofibroma; and (4) choroidal neurofibromatosis with secondary involvement of the ciliary body and choroid [11].

To the best of our knowledge, only two cases have been reported in which buphthalmos was the presenting sign of NF1. The first case involved a 3-week-old boy with congenital glaucoma and congenital upper eyelid swelling, who was later found to have a trigeminal nerve neurofibroma, buphthalmos, and an orbital plexiform neurofibroma, leading to the diagnosis of NF1. The authors emphasized that congenital glaucoma associated with eyelid plexiform neurofibroma is an uncommon presentation of NF1 [12].

The second reported case described a 1-month-old boy who presented solely with an enlarged right eye (buphthalmos) and elevated intraocular pressure (30 mmHg). Initial MRI findings were unremarkable; however, by the age of three years, the child developed classic NF1 features, including sphenoid wing dysplasia, eyelid plexiform neurofibroma, and café-au-lait macules. This report highlighted congenital glaucoma as a potential prodromal manifestation of NF1 [13].

This case is unique in that the patient presented with both buphthalmos and minimal proptosis, the latter being difficult to clinically detect at such an early age without imaging. Early MRI was inconclusive and did not provide sufficient information regarding the nature of the underlying pathology. Computed tomography was avoided due to the patient's young age. Axial length measurement revealed an increase to 18 mm; however, this finding alone was not sufficient to conclusively diagnose buphthalmos [10].

Additionally, Abnormal vascular maintenance and repair may also contribute to the pathogenesis of NF-related vasculopathy with abnormalities of neurofibromin expression leading to altered smooth muscle cell responses to endothelial signals [14].

This case demonstrated prominent iris vessels, which may be associated with increased episcleral venous pressure or prolonged elevation of intraocular pressure in infants. In this context, prominent iris vessels may also represent a precursor to the development of Lisch nodules.

Plexiform neurofibroma, which is considered pathognomonic for NF1, has been reported to mimic vascular malformations, further complicating early diagnosis [15].

4. Conclusion

This case demonstrates that buphthalmos can be the initial presenting feature of neurofibromatosis type 1, even before other classical systemic manifestations appear. Clinicians should consider NF1 in infants presenting with unilateral congenital glaucoma and orbital enlargement, and pursue careful imaging and multidisciplinary evaluation to enable early diagnosis and appropriate monitoring for potential systemic complications.

5. Patient Consent

Written informed consent for publication was obtained from the patient's parent.

6. Acknowledgements

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7. Conflict of Interest

The authors declare that there is no conflict of interest.

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