**Congenital Glaucoma with Neurofibromatosis Type-I: A Rare Occurrence**

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Neurofibromatosis 1 (NF1) is a rare genodermatoses in which there is involvement of the skin, eye, peripheral nervous system and skeletal system. Congenital glaucoma occurs in 1 in 300 NF1 patients.

A 6 year old boy presented to us with defective vision in the left eye since 4 years. On examination the child had café-au-lait spots, reduced visual acuity, Lisch nodules, increased corneal diameter, with glaucomatous cupping of optic nerve. Based on these findings, a diagnosis of congenital glaucoma with NF1 was made. The child was started on anti glaucoma medication for which he was non compliant and hence trabeculectomy was performed and the pressures are being maintained till date. Hence, if the diagnosis of NF1 is made, eye examinations are recommended in children for early detection of increased intraocular pressure or other ophthalmological manifestations and to reduce visual morbidity.

**Keywords:** neurofibromatosis, congenital glaucoma, trabeculectomy

**Introduction**

Neurofibromatosis 1 (NF1), also known as the von Recklinghausen’s disease (birth incidence varies from 1 in 2500 to 1 in 4–5000) is a rare genodermatoses with an autosomal dominant inheritance, characterized by a myriad of symptoms and signs that mirror the involvement of skin, eye, peripheral nervous system and skeletal system.¹ The National Institute of Health (NIH) Consensus Development Conference proposed the term neurofibromatosis 1 (NF1) and formulated the current diagnostic criteria to describe the same in 1987.² Accordingly, a patient is considered to have NF1 if at least 2 of the following 7 criteria are fulfilled³:

- Six or more café-au-lait macules with a diameter >5 mm in pre-pubescent individuals and >15 mm in post-pubescent individuals.
- Two or more neurofibromas of any type or one plexiform neurofibroma.
- Freckling in the axillary or inguinal regions.
- Optic nerve glioma.
- Two or more iris Lisch nodules.
- A distinctive osseous lesion (e.g., sphenoid wing dysplasia, long bone cortical thinning with or without pseudoarthrosis).
- A first-degree relative who meets the criteria for NF1.

However, in pediatric patients, it is often difficult to make an early diagnosis of NF1 because several clinical manifestations with diagnostic and prognostic prominence may not be present at birth and may appear as the child grows. This can be frustrating because children often have better outcomes with earlier intervention when subtle clues of NF1, such as congenital glaucoma (accounting for 1 in 300 NF1 patients) are recognized early in life.

Congenital glaucoma, a globally prevalent disease entity with a relative incidence of 1 in 3300 from the state of Andhra Pradesh, India accounts for 4.2% of childhood blindness. It poses a great diagnostic and therapeutic challenge to the ophthalmologist.⁴ Recent studies have made a distinction between Neurofibromatosis type 1 patients with or without orbital facial involvement. In patients with orbito-facial involvement, glaucoma was present in 23% of patients.⁵ A thorough annual ophthalmologic evaluation, carried out at least until the age of seven years, can be a distinguished screening tool for identifying increased Intraocular Pressure (IOP) or other ocular disorders; this would help in the early detection of ocular disorders including Primary Congenital Glaucoma (PCG), which can be the first symptom of NF1.² Here, we report a case of PCG associated with NF1 in a young child that may provide an insight into these diseases, especially the significance of early diagnoses and intervention to curb the morbidity.

**Summary**

A thorough annual ophthalmologic evaluation, carried out at least until the age of seven years, can be a distinguished screening tool for identifying increased Intraocular Pressure (IOP) or other ocular disorders; this would help in the early detection of ocular disorders including Primary Congenital Glaucoma (PCG), which can be the first symptom of NF1.² Here, we report a case of PCG associated with NF1 in a young child that may provide an insight into these diseases, especially the significance of early diagnoses and intervention to curb the morbidity.

**Keywords:** neurofibromatosis, congenital glaucoma, trabeculectomy

**Figure 1:** café-au-lait spots
Case Report

A 6 year old boy presented to the Outpatient Department of Ophthalmology in a tertiary care hospital with complaints of gradual painless diminution of vision in left eye over the past four years. On general examination, the child had multiple (>10) light brown coloured macules of varying sizes ranging from 2mm to 70mm with a smooth regular border distributed over the chest, anterior abdomen and back; the appearance being consistent with café-au-lait macules. On ocular examination, the left eye appeared to be larger than the right eye. Best Corrected Visual Acuity (BCVA) in the right and left eyes was 6/6 and 3/60 respectively. Examination of the left eye showed the following findings: dilated conjunctival episcleral vessels, edematous cornea (grade 2), increased diameter of cornea (13mm) and a deep (Van Hericks grade 4) and quiet anterior chamber. Iris showed multiple dome shaped brown coloured nodules consistent with Lisch nodules. Relative afferent pupillary defect was seen and the lens was clear. Fundus examination of the left eye showed cupping of disc (0.8) with bipolar neuroretinal rim loss and retinal nerve fiber layer defects. Foveal reflex was normal. On examination of the right eye, both the anterior and posterior segment examinations appeared to be normal. Intraocular pressures measured by the applanation tonometry were 20 and 40mm of Hg in the right and left eyes respectively. Gonioscopy revealed open angles in both the eyes. Refraction by automated refractometry showed OD: -1.25X150; OS: -14.5/-3.50X1800. Axial length of the right and left eyes was 20.1mm and 24.52mm respectively. The father of the child also had multiple café-au-lait spots and Lisch nodules. He also underwent surgery for the removal of tumour (glioma) from his brain five years back. A provisional diagnosis of primary congenital glaucoma of the left eye with Neurofibromatosis type 1 was made in the child. The child was started on Misopt (dorzolamide 2% w/v, timolol 0.5 % w/v) eye drops twice daily and Tovaxo (travoprost 0.004% w/v) eye drops once daily in the left eye. The child was subsequently sent to a neurosurgeon for further evaluation where MRI was done. The MRI showed hamartomas in basal ganglion region and it was suggested that they did not need any active intervention at that time. After two weeks of follow up, the IOPs were 16 and 24 in the right and left eyes respectively. However, the patient was non compliant for treatment; hence a decision to perform trabeculectomy was reached and it was performed under general anesthesia, the surgery being uneventful. A good elevated bleb was seen on the first post operative day. On follow up visit, IOP was 14 and 16mm in the right and left eyes respectively and the pressures were being maintained till date.

Discussion

Neurofibromatoses is a rare condition characterized by the development of hamartomas of neural crest origin, the prevalence of NFI being about one in 3,500 live births. The transmission modality is autosomal dominant; however, approximately 50% of cases are caused by spontaneous mutations, and there is considerable variability of expression between affected families. Diagnosis is usually performed clinically and is based on specific diagnostic criteria. Neurofibromatoses has been divided into varying types, the chief ones being type 1 (NFI or von Recklinghausen syndrome- 85%) and type 2 (NF2, acoustic NF, or central NF). The present case fulfilled the following clinical features...
of NF1 criteria: more than six café au lait spots (>5 mm in diameter) on the chest, anterior abdomen and back, Lisch nodules and a first degree relative with NF1. The right eye was normal. In the left eye, the horizontal and vertical corneal diameters were 13 mm each. The right eye corneal horizontal and vertical diameters were 11 mm each. The axial lengths of the right and left eye were 20.1 mm and 24.52 mm, respectively. The left eye had 0.8 glaucomatous cupping of 0.8:1.

Congenital glaucoma is usually managed surgically. When an NF1-associated congenital glaucoma is diagnosed, visual loss due to glaucoma is common and should be aggressively treated. Goniotomy or trabeculotomy are the preferred techniques for IOP control in younger individuals, but if they fail or cannot be performed, trabeculectomy, with or without supplemental therapies (i.e., adjunctive antifibrotic agents, glaucoma drainage devices [GDDs], cyclodestructive procedures), can be considered. In this case, we performed trabeculectomy.

**Conclusion**

Our rare case is about PCG with NF1 and PCG may be a prelude to NF1. Once the diagnosis of NF1 is made, eye examinations are recommended in children for early detection of increased intraocular pressure or other ophthalmological manifestations. And it is an example to us about the importance of continuous follow up. Also, we should raise our awareness of the combined condition and to improve chances for an early diagnosis. The importance of continued ophthalmologic and neurological monitoring was stressed to the family before they were sent home.

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