IOSR Journal of Dental and Medical Sciences (IOSR-JDMS)
e-ISSN: 2279-0853, p-ISSN: 2279-0861.Volume 22, Issue 6 Ser.11 (June. 2023), PP 36-38
www.iosrjournals.org

Congenital Glaucoma And Skin Manifestations In The Context Of Neurofibromatosis Type 1

T. Elongo, L. Kora, M. Adoudou, C. Baqadir, M. Elalami, G. Daghouj, L. Elmaaloum, B. Allali, A. Elkettani
Pediatric ophthalmology department. 20 August 1953 Hospital. CHU Ibn Rochd
Faculty of Medicine and Pharmacy of Casablanca. Hassan II University

Abstract:
Neurofibromatosis 1 (NF1), also known as Von Recklinghausen disease, is a rare genodermatosis with autosomal dominant inheritance. Congenital glaucoma is a rare ophthalmological complication. We report a case of congenital glaucoma in the context of type 1 neurofibromatosis. We will discuss the pathophysiology, the modalities of therapeutic management and the prognosis of congenital glaucoma in the context of vonRecklinghausen's disease.

Key words: *Congenital glaucoma, neurofibromatosis type 1, trabeculectomy*

Date of Submission: 13-06-2023 Date of Acceptance: 23-06-2023

I.Introduction

Neurofibromatosis 1 (NF1), also known as von Recklinghausen's disease (birth incidence ranges from 1 in 2,500 to 1 in 4,000), is a rare genodermatosis with autosomal dominant inheritance, characterized by multi systemic: cutaneous, ocular, peripheral nervous and skeletal(1). The clinical diagnostic criteria for NF1 were defined in 1988 by the National Institutes of Health.

The diagnosis of NF1 is based on at least two of the criteria listed in Table 1. (2). Congenital glaucoma is a rare ophthalmological complication. It poses a great diagnostic and therapeutic challenge for the ophthalmologist. A thorough annual ophthalmological evaluation, performed until at least the age of seven, can be a screening tool to identify increased intraocular pressure (IOP) or other eye disorders; this would allow early detection of eye disorders, including primary congenital glaucoma, which may be the first symptom of NF1.

II. Observation

This is a 4-month-old infant, with no particular pathological history, whose history of the disease dates back to birth when the parents observed exophtalmia associated with unilateral tearing of the right eye. The clinical examination shows a hemi-hypertrophy of the face with cutaneous spots of coffe with milk color all over the body (figure 1 and 2). The ophthalmological examination done under sedation found buphthalmos with megalocornea in the right eye (corneal diameter is 14 mm), corneal edema, habb streaks, a deep anterior chamber; ocular tone was 10 mmHg; the fundus shows an 8/10 papillary excavation; Gonioscopy did not find any other abnormalities of the iridocorneal angle. Examination of the left eye was unremarkable.

Cranio-orbital MRI revealed grade III proptosis, with the appearance of multiple neurofibromas in the right cavernous compartment involving the cisternal portion of the trigeminal nerve, V2 and V3 (figure 3).

Diagnostic criteria for NF1: Diagnosis retained in the presence of at least 2 clinical criteria

≥ 6 coffe with milk spots > 5 mm in greatest diameter in prepubertal patients or > 15 mm after puberty
≥ 2 neurofibromas regardless of type
≥ 2 Lisch nodules (iris hamartomas)
≥ 1 characteristic bone lesion: sphenoid dysplasia, thinning of the cortex of the long bones with or without pseudarthrosis
≥ one 1st degree relative with NF1 according to the above criteria

≥ 1 plexiform neurofibroma Axillary or inguinal ephelides (lentigines)
1 optic pathway glioma

The child underwent a trabeculectomy and peripheral iridectomy with application of mitomycin 0.2%. A control examination under general anesthesia was carried out 1 month post-operatively and showed corneal clearing in the right eye, an ocular tone of less than 6 mmHg and a excavation at 6/10.

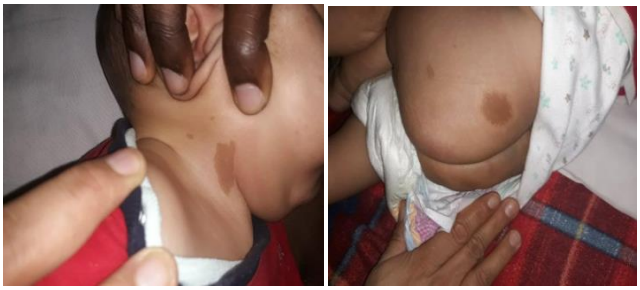

Figure 1: coffee with milk stains on the patient's body

Figure 2: Right facial hemi-hypertrophy with buphthalmos of the right eye

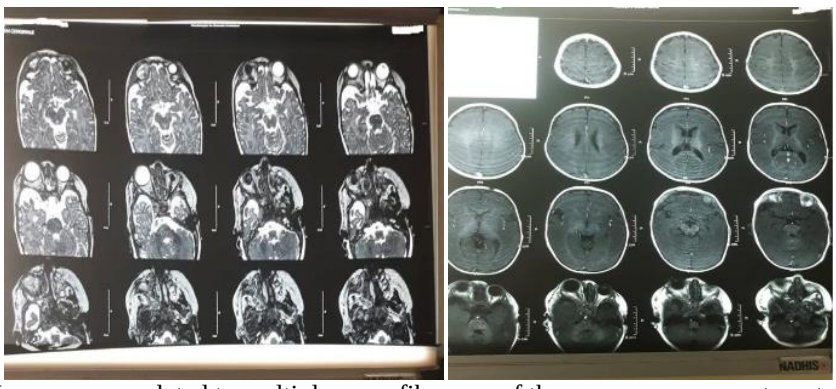

Figure 3 : MRI appearance related to multiple neurofibromas of the cavernous compartment and the cisternal portion of the trigeminal nerve with grade II right proptosis

III. DISCUSSION

Neurofibromatosis is a rare disease characterized by the development of hamartomas from the neural crest , with the prevalence of NF1 being approximately one in 3500 births. The mode of inheritance is autosomal dominant; however, approximately 50% of cases are caused by mutations, and there is considerable variability in expression between affected families. Diagnosis is essentially clinical and is based on specific diagnostic criteria (table 1). Neurofibromatosis has been divided into different types, the main ones being type 1 (NF1 or von Recklinghausen 85%) and type 2 (NF2, acoustic NF or central NF) (1).

The NF1 gene whose mutations predispose to the disease is located on the long arm of chromosome 17 (14) and results in uncontrolled activation of the MAP kinase pathway with deregulation of the mechanisms of cell proliferation and differentiation.

The observation of congenital glaucoma in the context of NF1 remains rare.

It occurs in 1% to 2% of cases (3,4) An increase in intraocular pressure is probably due to a combination of potential mechanisms such as abnormalities of the angle, pigmentary disturbances, secondary closure of the angle by anterior synechiae or following to infiltration by a neurofibroma interfering with the outflow of aqueous humor (5,6). Gonioscopy in NF1 patients may show a normal angle or subtle changes such as a slight anterior insertion of the iris or a thin or absent ciliary band (7); however, even when these signs are present, glaucoma is still uncommon. Grant and Walton estimated that NF1-associated glaucoma occurs in 1 in 300 patients with congenital glaucoma (5). Morales and colleagues found that 23% of NF1 patients with orbitofacial dysmorphism had glaucoma. Furthermore, Hoyt and Billson (8) and Morales and colleagues (6) found normal ocular pressure in patients with buphthalmos, suggesting that it is independent of ocular hypertension. (2) Ectropion uveal is common in NF1 patients with orbitofacial involvement; it occurs in more than half of patients with glaucoma. The association between uveal ectropion and glaucoma is known in patients with or without NF1. Thus, patients with congenital uveal ectropion should be followed for the development of neurocristopathy and glaucoma. (2) There is no consensus for the management of glaucoma in NF1. Treatment is often the same as for congenital glaucoma. Goniotomy or trabeculotomy are the preferred techniques when observed angular anomalies are similar to those of primary congenital glaucoma.

Glaucoma surgery in these cases can be made difficult by corneal opacity preventing good visibility. If this fails, trabeculectomy, glaucoma drainage devices, or cyclodestruction can be used. Trabeculectomy frequently fails due to growth endothelial at the filtration site, which remains similar to that seen in endothelial iridocorneal syndrome; Thus the use of a drainage device or a cyclodestructive procedure might be preferred. Glaucoma drainage devices are preferred in bupthalmic eyes with sclera thinned, or when there are conjunctival scars due to previous filtration surgeries. (9)

The visual prognosis of glaucoma in NF1 is poor due to several factors, including retinal detachment, optic nerve glioma, severe amblyopia caused by anisometropia, and glaucomatous optic neuropathy. Early surgical management alone would improve the prognosis. (10)

Patients with NF1, especially those with uveal ectropion, should be monitored for risk of developing glaucoma (11;12) need an annual ophthalmological evaluation until the age of 7 years, followed by a complete ophthalmological examination every 2 years, for the detection of glaucoma, and the search for gliomas of the optical pathway. (9 ; 13).

IV. Conclusion

In the face of any suspicion of neurofibromatosis, an ophthalmological examination is required to look for glaucoma or any other ophthalmological complication related to the disease; this allows early treatment that can improve the vital prognosis

REFERENCES

[1]. VankireddiMahathi, SairaniKaranam, MalleswariMedikonda. Congenital Glaucoma with Neurofibromatosis Type-I A Rare Occurrence. Delhi J Ophthalmol 2018;28;64-6;

[2]. Michael Kinori, MDa, Nickisa Hodgson, MDb, Janice LaskyZeid, MDa, Ophthalmic manifestations in neurofibromatosistype 1. survey o f ophthalmology xxx (2 0 1 7) 1e1 6

[3]. Huson SM, Harper PS, Compston DA. Von Recklinghause neurofibromatosis. A clinical and population study in southeast Wales. Brain. 1988;111(Pt 6):1355e81

[4]. Wright KW, Spiegel PH. Pediatric ophthalmology and strabismus. New York NY, USA, Springer-Verlag; 2003, p 766
Grant WM, Walton DS. Distinctive gonioscopic findings in glaucoma due to neurofibromatosis. Arch Ophthalmol. 1968;79(2):127e34

[5]. Morales J, Chaudhry IA, Bosley TM. Glaucoma and globe enlargement associated with neurofibromatosis type 1. Ophthalmology. 2009;116(9):1725e30

[6]. Quaranta L, Semeraro F, Turano R, Gandolfo E. Gonioscopic findings in patients with type 1 neurofibromatosis (Von Recklinghausen disease). J Glaucoma. 2004;13(2):90e5

[7]. Hoyt CS, Billson FA. Buphthalmos in neurofibromatosis: is it an expression of regional giantism? J PediatrOphthalmol. 1977;14(4):228e34

[8]. Alisa T. Thavikulwat, Deepak P. Edward, Abdulrahman AlDarrab,Thasarat S. Vajaranant. Pathophysiology and management of glaucoma associated with phakomatoses. J Neuro Res. 2018;1–13.

[9]. Nazih Tzili1,&, Hamza El Orch1 , Fatiha Bencherifa1 , Mohammed Charif Chefchaouni1 , El Hassan Abdallah1 , Amina Berraho.

[10]. Le glaucome congénitale et la neurofibromatose type 1. Pan AfricanMedical Journal. 2015
Edward, D. P., Morales, J., Bouhenni, R. A., Patil, J., Edward, P. R., Cummings, T. J., . . . Alkatan, H. (2012). Congenital ectropion uvea and mechanisms of glaucoma in neurofibromatosis type 1: New insights. Ophthalmology, 119(7), 1485–1494.

[11]. Ritch, R., Forbes, M., Hetherington, J., Harrison, R., &Podos, S. M. (1984). Congenital ectropion uveae with glaucoma.Ophthalmology, 91(4), 326–331.

[12]. Hersh, J. H., & American Academy of Pediatrics Committee on Genetics. (2008). Health supervision for children with neurofibromatosis. Pediatrics