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Case Report

Neurofibromatosis Type 1: Monocular Blindness in Children with Plexiform Neurofibromas in Two Cases

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Abstract: Neurofibromatosis type I (NF1) or Von Recklinghausen disease is one of the most frequent genetic diseases with polyvascular manifestations, including the eye. Its prevalence was variously estimated varying between 1/7800 and 1/2000. The diagnosis of NF1 is essentially clinical based on criteria established by the consensus conference of the National Institute of Health (NIH) in 1988. Several complications have been reported by numerous authors in cases of plexiform neurofibromas sometimes associated with congenital glaucoma. We report two cases of monocular blindness, complications of this disease in children, responsible for stigmatization by their peers and school dropout, seen at the National Hospital of Zinder, Niger.

Keywords: Neurofibromatosis type 1, plexiform neurofibroma, monocular blindness, congenital glaucoma, child, Zinder, Niger.

INTRODUCTION

Neurofibromatosis type 1 is an autosomal dominant genetic neurocutaneous disorder in which the gene responsible for the mutation was located on chromosome 17 in the 17q11.2 region in 1990 [1, 2]. It was in 1882 that Friedrich Daniel Von Recklinghausen described for the first time the nervous origin of the tumors observed in the disease that would later bear his name [3]. Its penetrance is close to 100% around the age of 5 years and de novo mutations occur in about 50% of cases [4]. The diagnosis of neurofibromatosis type 1 is clinical based on criteria (Table I) established in 1988 by the consensus conference of the National Institute of Health (NIHCDC) [5]. Several complications could occur during the course of Von Recklinghausen disease and the eye is not spared. We report in this work two cases of monocular blindness due to NF1 seen at the National Hospital of Zinder.

Observation 1

M.B., female, 8 years old, monophthalmic right eye, with a family history of neurofibromatosis. She was dropped out of school in the first grade because of the stigma attached to her disease by children her age. She had undergone evisceration of her left eye at the age of 5 years after failed trabeculectomy treatment at the age of 2 years for unilateral congenital glaucoma of the left eye (Figure 1A, red arrow). On ophthalmologic examination, the distance visual acuity on the right eye was 10/10 and there was no light perception on the left eye. The adnexa examination, noted a purulent conjunctivitis and a total ptosis covering an anophthalmic orbital cavity (Figure 1B), normal adnexa on examination of the right eye, a clear cornea, a quiet anterior chamber, Lish's iridal nodules with a reactive pupil (Figure 1D). Dermatological examination noted several café au lait spots, numerous skin nodules, and a plexiform neurofibroma on the left upper eyelid resulting in total mechanical ptosis (Figure 1C yellow arrow). A CT scan of the orbitocerebrum was requested and was unremarkable except for the palpebral mass. The diagnosis of neurofibromatosis type 1 was retained, treatment with eye drops and antibiotic ointment was administered and the patient was referred for reconstructive surgery.

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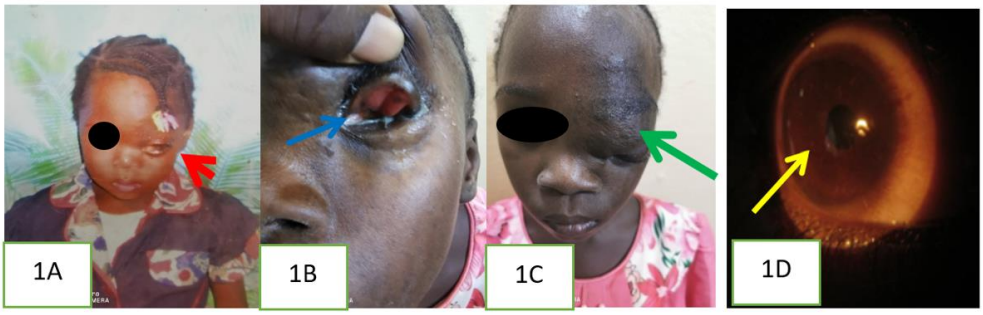

Figure 1: 1A: Congenital glaucoma left eye (red arrow), 1B: anophtalmia and conjunctivitis left eye (blue arrow), 1C plexiform neurofibroma on the left upper eyelid resulting in total mechanical ptosis (green arrow), 1D: Lish Nodules right eye (yellow arrow)

Observation 2

A.B, male, 15 years old, monophthalmic with right eye, having dropped out of school because of stigmatization by his schoolmates and engaging in begging, was brought by his mother to the ophthalmology department of the Nation Hospital in Zinder for swelling in the orbit and hanging from the left hemiface, with facial dysmorphia. (Figure 2A red arrow). No family history of disease was reported. On ophthalmologic examination, visual acuity was 10/10 on the right and see move hand. Biomicroscopic examination of the right eye revealed normal adnexa, a healthy cornea with a good quiet anterior chamber, Lish nodules (Figure 2B blu arrow) with a normal fundus. Examination of the left eye, hampered by swelling, revealed conjunctivitis, a 360° corneal pannus with a corneal opacity, all of which made examination of the anterior segment difficult. The dermatological examination noted a plexiform neurofibroma hanging from the orbito-periorbital area and involving the left hemiface (Figure 2A), café au lait spots, skin nodules and axillary lentigines. The diagnosis of neurofibromatosis type 1 was retained, a treatment based on antibiotic eye drops was administered and the patient was referred for maxillofacial surgery.

Table I

Panel : NIH consensus criteria for diagnosis of neurofibromatosis type 1 (1988) [5] *Two or more the following clinical features are sufficient to establish a diagnosis of neurofibromatosis type 1*
1. Six or more café-au-lait macules (> 0-5 cm at largest diameter in a prepubertal child or > 1-5 cm in post-pubertal individuals)
2. Axillary freckling or freckling in inguinal regions
3. Two or more neurofibromas of any type or one or more plexiform neurfibromas
4. Two or more Lish nodules (iris harmatomas)
5. A distinctive osseous lesion (sphenoid wing dysplasia, lon-bone dysplasia)
6. An optic pathway glioma
7. A first degree relative with neurofibromatosis type 1 diagnosed by the above criteria
NIH : National Institute of Health

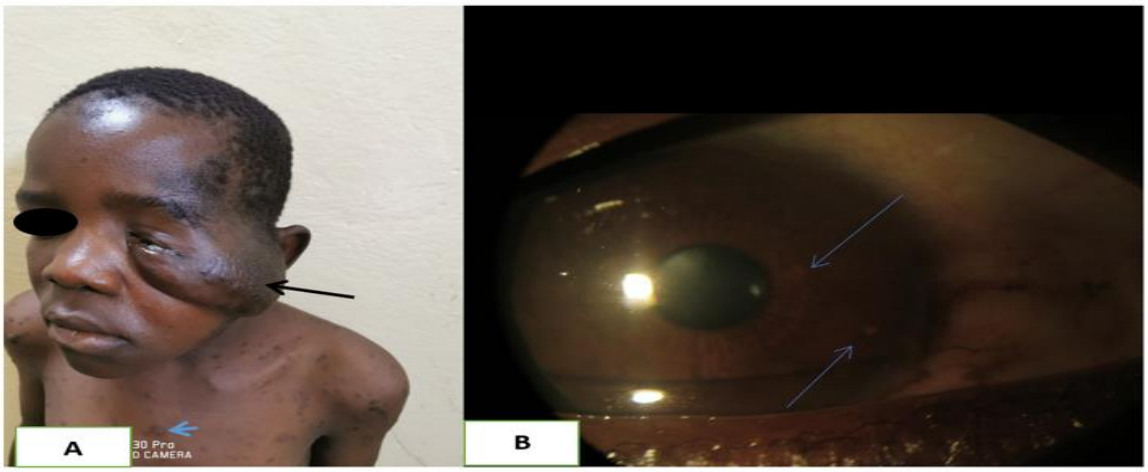

Figure 2: 2A plexiforme neurofibroma orbito-periorbital suspended from the left temporal area with facial dysmorphia (black arrow), 2B Lisch nodules (bleu arrow)

DISCUSSION

Neurofibromatosis type I (NF1) is a heterogeneous group of diseases characterized by a genetically determined predisposition to the development of benign or malignant polyvisceral tumors, including ocular ones [6]. Its prevalence was variously estimated ranging from 1/7800 to 1/2000. It is one of the most frequent genetic diseases [7-10]. Its diagnosis is essentially clinical based on criteria established by the NIH (Table 1). Several complications have been reported by numerous authors in cases of plexiform neurofibromas. These occur in 30-50% of patients with NF1 [11, 12]. These tumors were reported in our two patients with various localizations but all involving the upper eyelids of the left eye. Congenital glaucoma is a complication of NF1 [13]. It can be bilateral or unilateral as was the case in our 1st patient. She presented the Jules François syndrome described by the author who bore his name in 1956 [14] this syndrome associates an early congenital glaucoma, a plexiform neurofibroma of the homolateral upper eyelid and a facial hemihypertrophy. The etiopathogenic mechanisms are multiple: infiltration of the Schwann sheath of the ciliary nerves (neurofibromas) that block the iridocorneal angle, malformation or immature development of the iridocorneal angle, uveal ectropion ... [13, 14] etc.... The treatment of congenital glaucoma in NF1 is complex. It requires surgery with disappointing results, as in the case of our patient whose eye was eventually eviscerated, thus rendering the patient monophthalmic. In the majority of cases, this glaucoma occurs on the same side as the palpebral plexiform neurofibroma with the secondary appearance of buphthalmia [14]. In the case of plexiform neurofibroma, exophthalmos was present in 25% of cases. It was found in our 2nd patient (50%), responsible for facial dysmorphia and left unilateral blindness, blindness being defined by the World Health Organization (11th International Classification of Diseases 2018) as a visual acuity of less than 3/60) [15]. Orbital involvement due to plexiform neurofibroma growth is rare and considered pathognomonic of NF1 with several designations: Orbitotemporal neurofibromatosis, orbitofacial neurofibromatosis, cranioorbital neurofibromatosis, cranioorbital-orbito-temporal neurofibromatosis and recently a working group had suggested the term orbital-periorbital plexiform neurofibromas to encompass all sites of localization [14, 16]. Cognitive and behavioral disorders occur in 46-80% of children with NF1. Specific learning disabilities affecting performance on visuospatial tasks, reading and writing skills, and oral and written expression often lead to poor school performance [17]. Both of our patients dropped out of school due to stigmatization and marginalization and both were engaged in begging. This stigma would negatively impact the quality of life of these patients who would feel inferior. In a study of 129 patients with NF1, the authors noted a greater degree of psychological distress in patients with NF1 than in the general population [18]. All diagnoses taken together, psychiatric pathologies are much more frequent in neurofibromatosis type 1 than in the general population with a prevalence of 33% [19], even if some authors estimated that these pathologies were under-diagnosed.

CONCLUSION

Von Recklinghausen disease is a genetic disease with polyvisceral manifestations. The ophthalmologist has a central role not only in the diagnosis and management of NF1, but also in the detection of certain ocular complications, some of which may jeopardize the visual prognosis through blindness that is often irreversible. In the chain of care of NF1, which is meant to be holistic, psychologists and psychiatrists are not left out because of the stigmatization of NF1 patients.

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