Ocular Manifestations of Neurofibromatosis Type 1; 2 Case Reports

Sebbata S* and Abdellaoui T

Ophthalmology Department, Military Hospital Mohamed V, Rabat, Morocco

***Corresponding author:** Soundous Sebbata, Ophthalmology Department, Military Hospital Mohamed V, Rabat, Morocco

Citation: Sebbata S, Ocular Manifestations of Neurofibromatosis Type 1; 2 Case Reports, Ann Surg Onco Treat, 2024; 1(1): 1-3.

Published Date: 10-08-2024 **Accepted Date:** 05-08-2024 **Received Date:** 25-07-2024

Keywords: Neurofibromatoses; Schwannoma; Iridocorneal; Iridocorneal

1. Introduction

Neurofibromatosis 1 (NF1) also known as von Recklinghausen is a rare disease, that is part of the spectrum of neurofibromatoses. Its genetic and clinical aspects differ from one entity to another. Alongside NF1, we find NF2 and Schwannoma. Its incidence is estimated at 1/3000 cases. In 50% of cases, NF1 appears de novo in patients with no family history of neurofibromatosis. We report the observations of two young female patients who presented with Lisch nodules as part of their disease.

2. Observations

2.1. Clinical case n°1:

This 22-year-old patient had been followed since childhood for NF1. General examination revealed café-au-lait spots on her both arms and her back, aside from cutaneous neurofibromas on the face (Figure 1). She was referred to us for ophthalmological investigation in the context of her disease. Examination of the right eye revealed a corrected visual acuity of 10/10. Slit-lamp examination revealed 4 small iris elevations with slightly downy margins corresponding to Lisch nodules, measuring 1, 1.5, and 2 mm respectively, and located at 2, 3, 4, and 7 o'clock (Figure 2a). In the left eye, corrected visual acuity was also 10/10, with the same types of iridocorneal nodules located at 9 and 11 o'clock, measuring 0.5 and 1 mm respectively (Figure 2b). The iridocorneal angle was open through 360° without individualization of nodules. The photomotor reflex was preserved in both eyes, both direct and consensual. Intraocular pressure was 14mmHg and fundus examination was without abnormalities, notably no optic nerve damage.

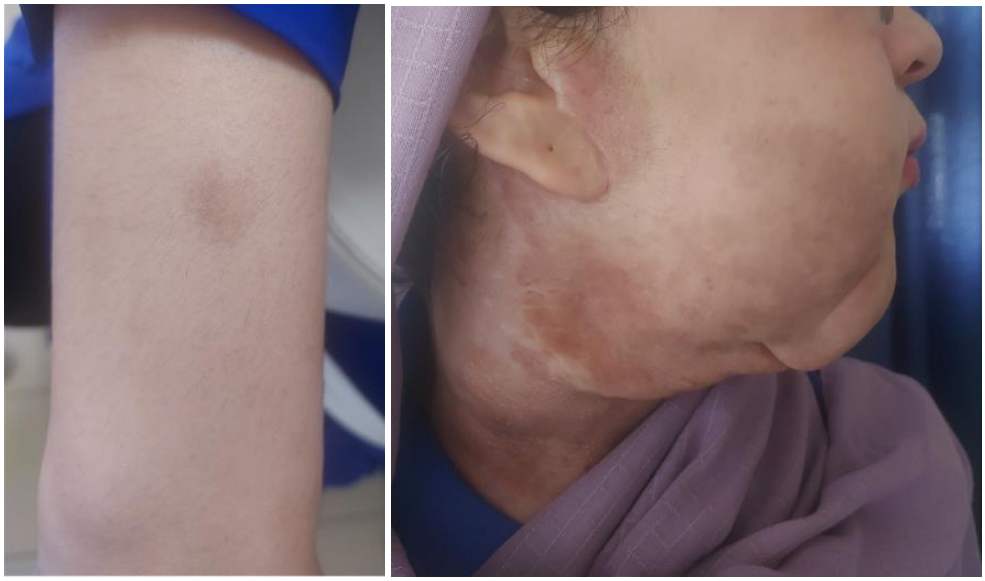

Figure 1: Café-au-lait stains of the forearm and face and cutaneous neurofibromas of the face.

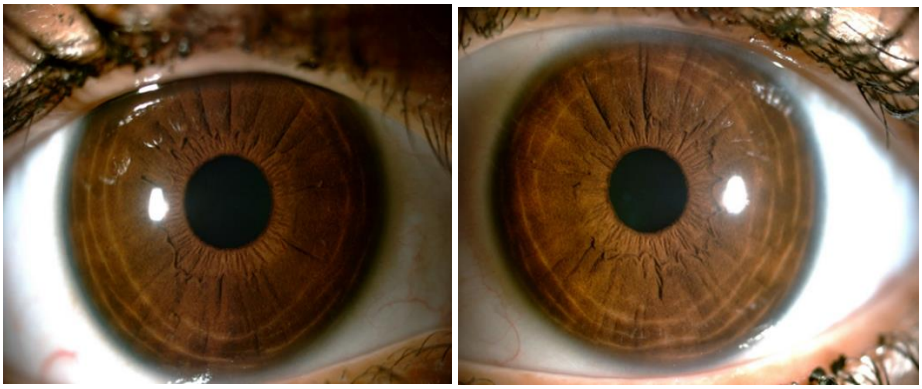

Figure 2: a-Lish nodules of the right eye, b- Lish nodules of the left eye

2.2. Clinical case n°2:

The 2nd patient is an 8-year-old girl whose family history included a father with NF1 who died of a central nervous system tumor (a complication of his disease). General examination revealed cafe-au-lait lesions on the face, neck, both arms and chest. Visual acuity was preserved in both eyes at 10/10. The anterior segment showed three Lisch's nodules on the right eye and one on the left. The direct and consensual photomotor reflex was preserved. Examination of the iridocorneal angle was normal in both eyes. Fundus examination revealed no optic nerve damage.

3. Discussion

Ocular manifestations of Von Recklinghausen disease are frequent and may reveal the disease. An ophthalmological examination should always be carried out if neurofibromatosis is suspected. In the anterior segment, the lesions to look for are Lisch nodules. They were described in 1937, as small pigmented hamartomas most often found in the lower iris [1]. They can be spotted without a high magnification by using a slit lamp to study the iris stroma. They are primarily seen in NF1 [2]. Their prevalence is higher in sporadic cases than in familial cases [1]. They represent the most frequent ocular manifestation of NF1 as they are present in over 90% of adult neurofibromatosis patients where they generally appear between the ages of two and six [3].

The number of nodules increases with age but is not correlated with disease severity [1,2]. Their prevalence increases progressively from 0% at birth to 75% in adolescence and 90% in adulthood [4]. Lisch nodules can be particularly useful in the diagnosis of NF1 within young children with multiple café-au-lait spots and no family history of neurofibromatosis, as these lesions appear before neurofibromas [3]. Their presence is one of the major criteria in the positive diagnosis of neurofibromatosis type 1, as described in Table 1 [5]. In the absence of other signs of NF1, their presence is highly suggestive of NF1 [4]. Therefore, there is no correlation between the presence of these nodules and the severity of the disease [6].

Table 1: Diagnostic criteria for neurofibromatosis: The positive diagnosis of neurofibromatosis is based on the presence of at least two of the above-mentioned criteria.

	Criterias
1.	Six or more café au lait macules whose largest diameter is > 5 mm in prepubertal individuals and > 15 mm in postpubertal individuals
2.	Two or more neurofibromas of any type or one plexiform neurofibroma
3.	Freckles in axillary or inguinal areas.
4.	Optical glioma
5.	Two or more Lisch nodules
6.	A distinctive bone lesion such as sphenoid dysplasia or thinning of the cortex of the long bones, with or without pseudarthrosis.
7.	A first-degree relative (parent, sibling or offspring) with NF-1 according to the above criteria

Less frequent lesions can be found in the posterior segment. Astrocytic hamartomas, typically seen in tuberous sclerosis, are also described in NF1. Capillary hemangiomas and combined retinal and pigment epithelial hamartomas may also be seen. Vision-threatening complications such as retinal detachment, neovascular glaucoma, and vitreous hemorrhage have been associated with

the disease [7]. 99% of patients with NF1 acquire café-au-lait macules before the age of one year, making them the most prevalent dermatological finding [5]. These hyperpigmented lesions range from light to dark brown and are the result of increased melanocyte proliferation.

Optic nerve gliomas can be visually devastating. This is the most common central nervous system tumor to occur in NF1 [6]. Optic nerve astrocytoma is the most frequent type [1].

Neurofibromas are feared in the disease. Their number varies from one individual to another [8]. The sex ratio is 1:1 [9]. They are at risk of malignant degeneration with metastatic risk compromising the vital prognosis, as was probably the case for the father of the 2nd patient.

4. Conclusion

Ocular manifestations in neurofibromatosis are easily assessed by slit lamp. Lisch nodules are one of the most frequent manifestations, unrelated to the severity of the disease. Prognosis is compromised by the presence of central nervous system gliomas or neurofibromas, with a risk of degeneration.

References

1. Theos A, Boyd KP, Korf BR. The Neurofibromatoses. Harper's Textb Pediatr Dermatology Third Ed. 2011; 2: 1-14.
2. Arigon V, Binaghi M, Sabouret C, Zeller J, Revuz J, Soubrane G, et al. Usefulness of systematic ophthalmologic investigations in neurofibromatosis 1: A cross-sectional study of 211 patients. Eur J Ophthalmol. 2002; 12(5): 413-8.
3. Antoniolli LP, Milman L de M, Bonamigo RR. Dermoscopy of the iris: identification of Lisch nodules and contribution to the diagnosis of Neurofibromatosis type 1. An Bras Dermatol [Internet]. 2021; 96(4): 487-9.
4. Ragge NK, Frank RE, Cohen WE, Murphree AL. Images of Lisch nodules across the spectrum. Eye. 1993; 7(1): 95-101.
5. Chernoff KA, Schaffer J V. Cutaneous and ocular manifestations of neurocutaneous syndromes. Clin Dermatol. 2016; 34(2): 183-204.
6. Millichap JG. Lisch Nodules in Neurofibromatosis Type 1. Pediatr Neurol Briefs. 1991; 5(6): 41.
7. Kinori M, Hodgson N, Zeid JL. Ophthalmic manifestations in neurofibromatosis type 1. Surv Ophthalmol. 2018; 63(4): 518-33.
8. Cimino PJ, Gutmann DH. Neurofibromatosis type 1. 1st ed. Handbook of Clinical Neurology. Elsevier B.V. 2018; 148; 799-811.
9. Gutmann DH, Ferner RE, Listernick RH, Korf BR, Wolters PL, Johnson KJ. Neurofibromatosis type 1. Nat Rev Dis Prim. 2017; 3: 1-18.