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Iris Localization of Neurofibromatosis Type 1: About 3 Cases

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INTRODUCTION

Von Recklinghausen's disease or neurofibromatosis type I (NF1) is the most common phacomatosis [1].

It is a relatively common genetic disease. The ophthalmological manifestations of type I neurofibromatosis are many and varied.

The purpose of our presentation is to remind the clinician of the appearance of Lisch nodules through an iconographic presentation and their still current role in the identification of neurofibromatosis.

We report three observations of patients with neurofibromatosis type I and in whom Lisch nodules were the only ocular manifestation of the disease.

CASES

Case 1:

This is a 36-year-old patient, in whom the pathology has evolved since childhood with the appearance of multiple hyperpigmented skin macules.

She had more than six café au lait spots on her skin. (Figure 1).

Visual acuity 10/10 p2 and ophthalmic examination reveals more than two Lisch nodules in both eyes (Figure 2a,b).

These are small lesions raised above the surface of the iris, with a very sharp edge, gelatinous, lighter than the pigmentation of the iris, brown.

Funduscopic examination was normal. MRI was unremarkable.

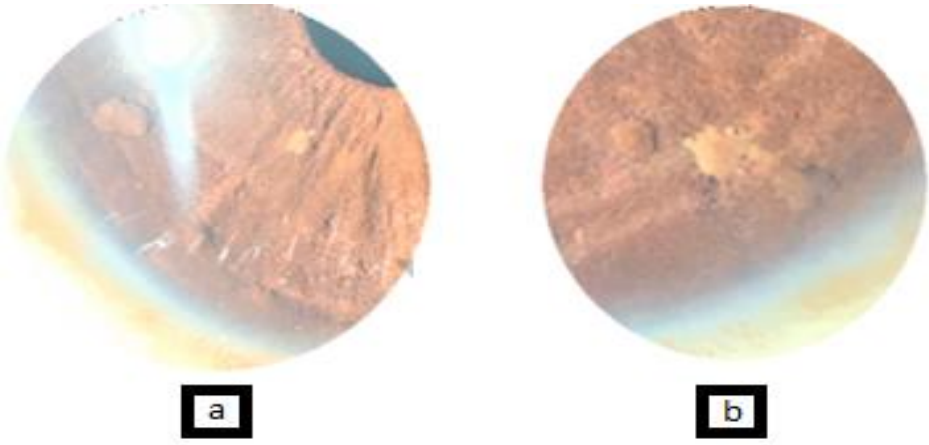

Figure 2a, 2b: Lish's nodule at the slit lamp (a: right eye, b: left eye)

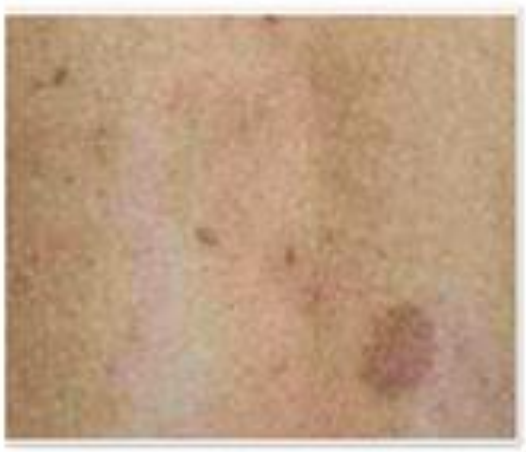

Figure 2: brown spots

Case 2:

This is a 45-year-old patient, referred for ophthalmological examination in search of signs in favor of neurofibromatosis.

The ophthalmological examination objectified an AV 10/10 p2, the LAF examination revealed lisch nodules ODG, These are small lesions raised above the surface of the iris, with a very clear edge, gelatinous, more pigmented than the iris. Intraocular pressure is normal.

The fundus shows a normal retina and papilla.

Cutaneomucosal examination revealed several café-au-lait spots with predominantly neurofibromas on the face.

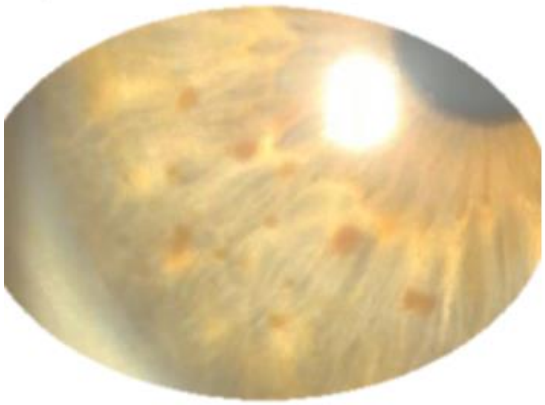

Figure 3: Lish's nodule at the slit lamp

Case 3:

This is a 36-year-old patient, referred for systematic ophthalmological examination in the context of Von Recklinghausen 's disease.

The ophthalmological examination found a VA at 10/10 p2 in both eyes, the slit lamp examination revealed bilateral Lisch nodules. These are small lesions raised above the surface of the iris, with a very sharp edge, gelatinous, more pigmented than the iris (Figure 4).

The fundus was unremarkable.

Furthermore, skin examination revealed several brown spots spots with multiple neurofibromas (Figure 5, 6).

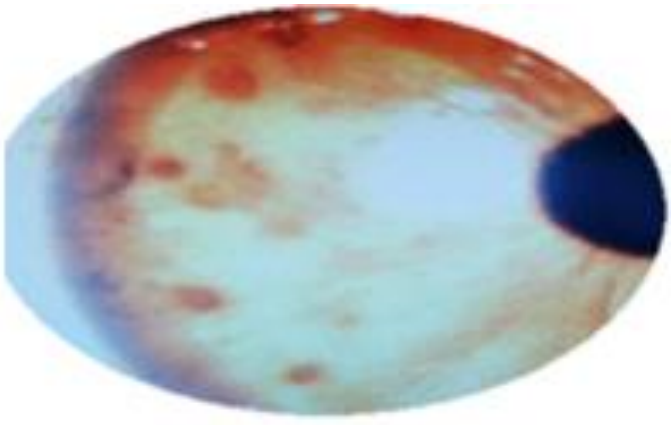

Figure 4: Lish's nodule of the right eye at the slit lamp.

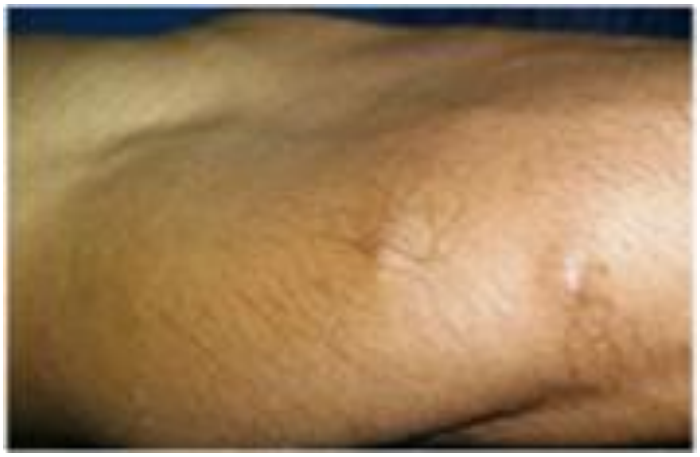

Figure 5: Cutaneous neurofibromas

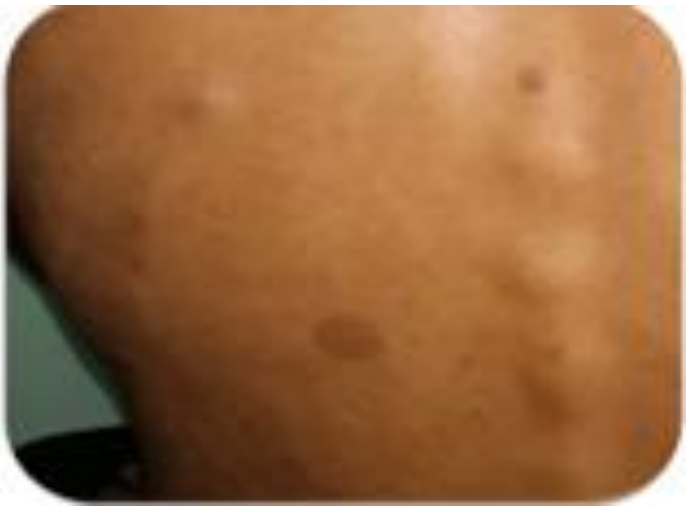

Figure 6: brown spots and neurofibromas

DISCUSSION

One of the rare genetic disorders, neurofibromatosis type 1 (nf1), also known as von recklinghausen's disease, involves the development of multiple benign neurofibromas and areas of abnormal hypo or hyperpigmentation in the skin.

It affects 3 out of 10,000 newborn babies worldwide. It affects women as well as men [1].

Lisch nodules represent the most frequent ocular manifestation of neurofibromatosis type I of which it constitutes a pathognomonic sign.

Several studies have been conducted on the frequency of Lisch nodules in the neurofibromatous patient population to determine their diagnostic role. It varies between 92% and 100% [2].

Under the bio-microscope Lisch's nodules are bilateral, they look like small gelatinous masses, in relief compared to the surface of the iris. Their color varies from unpigmented to highly pigmented depending on the color of the iris [3].

In optical microscopy, the Lisch nodules are intrastromal with poorly defined limits.

They are made up of spindle-shaped cells that are generally larger than the melanocytes of the normal iris. The cytoplasm of some of these cells contains melanin.

Electron microscopy confirms that the iris nodules are composed of melanocytes. These are melanocytic hamartomas

Von Recklinghausen's disease is easy to recognize in its typical and complete form by the association of pigmented spots, skin tumors and nerve tumors [4,5].

On the other hand, the pauci symptomatic forms linked to a weak expressiveness of the disease pose more diagnostic problems.

Lisch nodules then represent an important clinical sign which can in certain cases establish or eliminate the diagnosis of Von Recklinghausen disease.

Lisch nodules should not be confused with other iris nodules.

- On blue and green irises, they appear pale brown with fluffy borders.
- On the brown and black irises, they are creamy brown in color, and with very well defined boundaries.
- They are distributed randomly over the entire surface of the iris, from the periphery to the pupillary margin.

There is no correlation between the number and size of the nodules and the severity of the disease.

CONCLUSION

Lisch nodules are pathognomonic for Von Recklinghausen disease.

Their presence is an important argument to support the diagnosis of the disease which, at present, is still based on a bundle of clinical arguments.

They are easily detectable by bio-microscope examination. The difficulty for the practitioner is to know how to distinguish them from other types of iris nodules.

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