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Case Report

Lisch Nodules in Schwannomatosis: A New Manifestation

Emanuele Miraglia[1], Chiara Iacovino[1], Giovanni Corbo[2], Fabiana Mallone[2], Antonietta Moramarco[2]* and Sandra Giustini[1]

[1]Department of Dermatology and Venereology, "Sapienza" University of Rome, Policlinico Umberto I, Viale del Policlinico, Rome, Italy

[2]Department of Sense Organs, Sapienza University of Rome, Viale del Policlinico, Rome, Italy

***Corresponding Author:** Antonietta Moramarco, Professor, Department of Ophthalmology, Policlinico Umberto I, Viale del Policlinico, Rome, Italy.

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Abstract

Schwannomatosis is a syndrome characterized by presence of schwannomas in the absence of bilateral vestibular schwannomas and meningiomas. Schwannomas interest frequently peripheral nerves (90%) and spinal nerves (75%).

Schwannomatosis are generally sporadic; in 15 - 25% are familiar. The genes involved are *SMARCB1* (40-50% of familial) and *LZTR1.*

The reported phenotype continues to expand and evolve. We report the case of a patient with Schwannomatosis and Lisch nodules, typical manifestation of NF1.

Keywords: Lisch Nodules; Schwannomatosis; Schwannomas

Introduction

Schwannomatosis is a syndrome characterized by presence of schwannomas in the absence of bilateral vestibular schwannomas and less frequently meningiomas.

Schwannomas interest frequently peripheral nerves (90%) and spinal nerves (75%) such as lumbar spine. It rarely affects cranial nerve (trigeminal nerve). Unilateral vestibular schwannomas can occur (in NF2 vestibular schwannoma is bilateral) [1].

Schwannomatosis are generally sporadic; in 15 - 25% are familiar. The genes involved are *SMARCB1* (40 - 50% of familial) and *LZTR1* [2].

Schwannomatosis is a rare disease (incidence: 0.58/1.000.000 person-years) [3]. Unusual manifestations have already been reported such as malignant peripheral nerve sheath tumors (MPNSTs) [4].

Case Report

We report the case of a patient with Schwannomatosis and an unusual ocular manifestation.

A 41-year-old man with a 4 years history of low back pain. An MRI of the lumbosacral column showed the presence of multiple small rounded areas of altered signal, slightly hyperintense on T2 and hypointense on T1 at L1-L3 levels. Histological examination confirmed the clinical diagnosis.

On physical examination, there were no clinical manifestation of NF1.

On slit lamp examination, there were small multiple hypopigmented elevated lesions over the iris in right eye suggestive of Lisch nodules (Figure 1). There were not any abnormality.

MRI brain was normal.

Figure 1: Lisch nodules in a patient with schwannomatosis.

He had no other characteristic manifestations of NF1 and NF2. In addition, genetic testing for *NF1* gene in the peripheral blood was negative. The genetic examination identified instead the LZTR1 mutation c.523delA (p.Arg175Glyfs*25). There was no family history of other genetic disease.

On the basis of all the manifestations found, we made a diagnosis of schwannomatosis.

Color of Lisch nodules may vary from creamy white (dark irides) to brown (blue and green irides). They may have sharp or jagged margins and in some cases they may coalesce. Histologically, Lisch nodules are composed of melanocytes, fibroblasts and mast cells.

Discussion and Conclusion

Lisch nodules occur predominantly in NF1 and they are the most frequent ocular manifestation in NF1 together with the anomalies of the choroid [5]. Their prevalence increases with age (90 - 95% over the age of thirty) [6].

They are also one of the diagnostic criteria of NF1 but not represent a cause of disability [7].

In our case there were only Lisch nodules while other typical manifestations of NF1 or NF2 were absent.

We report this rare association and underline the presence of Lisch nodules in other rare diseases besides NF1. To our knowledge, Lisch nodules have been reported only in patients with NF1 and segmental neurofibromatosis; ours is the first case of Lisch nodules in Schwannomatosis.

Further studies are needed to understand the mechanisms behind this rare association.

Conflict of Interest

The authors declare that they have no conflict of interest.

Ethical Approval

All procedures were in accordance with the ethical standards of the institutional research committee of Sapienza University of Rome and with the 1964 Helsinki Declaration and its later amendments or comparable ethical standards. This article does not contain any studies with animals performed by any of the authors.

Informed Consent

Informed consent was obtained.

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