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CASE REPORT

Ultrasound Biomicroscopy (Ubm) Characteristics Of Lisch Nodules In Neurofibromatosis Type 1

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ABSTRACT

Lisch nodules are the most common type of ocular involvement in NF-1. Lisch nodules are melanocytic hamartomas, usually clear yellow to brown in color, that appear as well-defined, dome-shaped elevations projecting from the surface of the iris. Lisch nodules are the most common clinical finding in adults older than 20 years with NF-1. We did Ultrasound biomicroscopy (UBM) of lisch nodules.

Key words: Lisch nodule, neurofibromatosis, Ultrasound biomicroscopy

Introduction

Neurofibromatosis type 1 (NF1) is one of the most common autosomal dominantly transmitted neurocutaneous disorders known to man, with an estimated prevalence of about 1 in 3500 individuals [1]. One of the diagnostic criteria for NF1 is the presence of two or more Lisch nodules (iris hamartomas) [2]. Lisch nodules are well-defined, avascular, smooth, regular, dome-shaped elevations of the iris surface, having a yellow to brown color [3]. We described UBM findings of these Lisch nodules.

Case Report

A 32-year-old lady presented to our department with history of drooping of left eyelid and a mass over the upper left lid.

On ocular examination, we found she had a mass on the left lid, which was causing mechanical ptosis with poor levator function [Table/Fig 1]. Anterior segment revealed presence of multiple lisch nodules [Table/Fig 1]. Her systemic examination revealed café au lait spots and freckles all over her body. A diagnosis of Neurofibromatosis type 1 was made. Ultrasound biomicroscopy (UBM) (HF35-50 High Frequency Ultrasound, OTI, Toronto, Canada) was done on both the eyes.

Discussion

Lisch nodules are melanocytic hamartomas, usually clear yellow to brown in color, that appear as well-defined, dome-shaped elevations projecting from the surface of the iris [3]. They can be clearly distinguished from iris naevi, which show a more flattened appearance. Lisch nodules may be seen without magnification, but a slit lamp examination may be necessary to differentiate them from nevi on the iris, which present as flat or minimally elevated, densely pigmented lesions with blurred margins. Unlike café au lait spots, multiple nodules are

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specific for peripheral neurofibromatosis/NF-1. They usually arise in the first decade; virtually all patients with NF-1 have Lisch nodules by age 20 years[2],[3],[4].

Ultrastructural, histological and cellular studies of the Lisch nodules have mainly been done by light and electron microscopy revealed two lesional compartments in these nodules: a superficial plaque of compact spindle-like cells, and an underlying stromal accumulation of similar but loosely organized cells. However, a recent histological and ultrastructural analysis of a Lisch nodule revealed three main cell types: pigmented cells, fibroblast-like cells and mast cells, which resemble the neurofibroma cell population.

Table/Fig 1

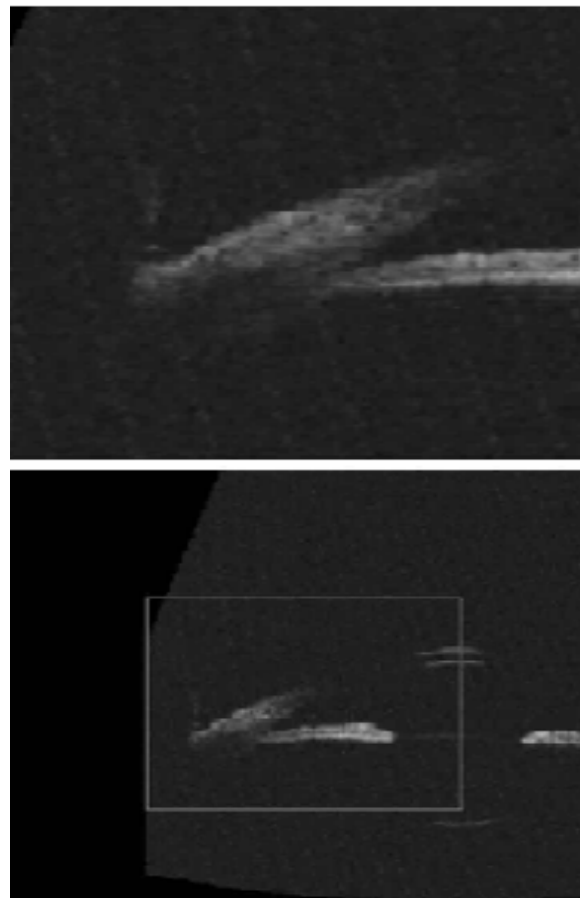


Shows upper lid neurofibroma in the upper picture and lisch nodules on the iris in the lower picture.

Ultrasound biomicroscopy (UBM) revealed small elevation associated with slight depressions around it [Table/Fig 2]. Also, A scan revealed a higher spike than the normal iris tissue. The nodules represented elevations in an otherwise smooth architecture of the iris or with some depression representing the crypts in the iris.

We believe that this is the first ever report of UBM findings in case of Lisch nodule. This being a non-invasive technique could reveal and help in further differentiation of such nodules any prognostic signs; this would require further research.

Table/Fig 2



The upper photograph shows the magnified view of the inset of the lower one. Note the small elevations over the surface of the iris on an otherwise smooth iris surface

Conflict of Interest: None declared

Photographs: All images were obtained after informed consent of the patient.

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