Outer retinal atrophy in a patient with Maternally Inherited Diabetes and Deafness

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Purpose

To report a case of Maternally Inherited Diabetes and Deafness (MIDD).

Case Report

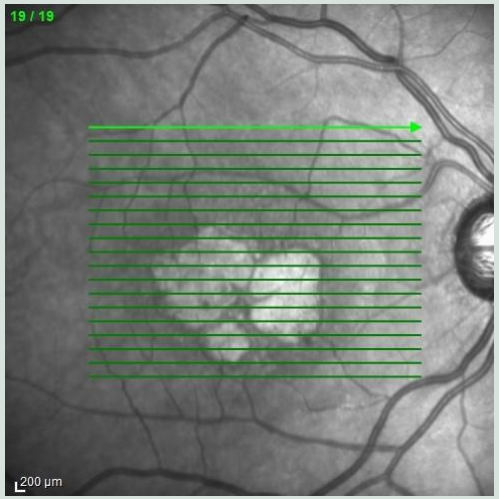
An 84-year-old female patient presented in the Medical Retina department in order to have her regular ophthalmological check-up.

History :

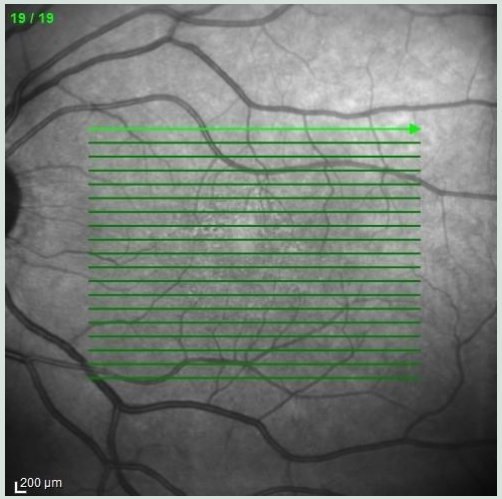
- Hearing impairment
- Diabetes
- Family: hearing loss-daughter and brother, diabetes-daughter
- Ophthalmic: phacoemulsification cataract surgery in both eyes two years ago.

Clinical examination

- BCVA: OD 4/10 OS 7/10
- Fundus examination:



Atrophic areas surrounding the fovea (one papillary diameter).



Mild RPE changes.

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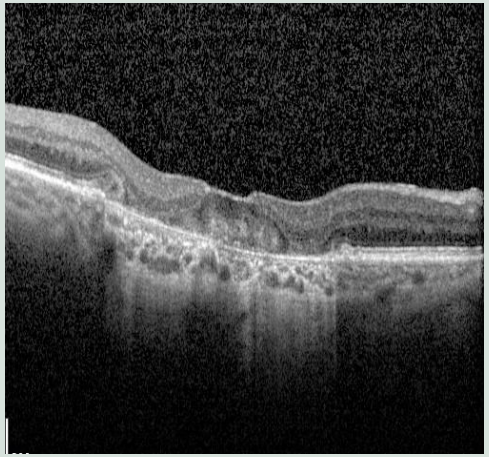
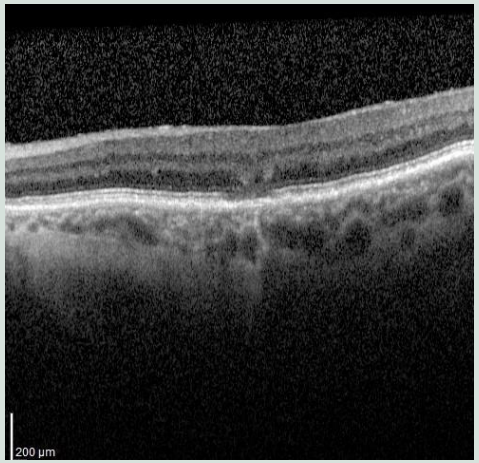
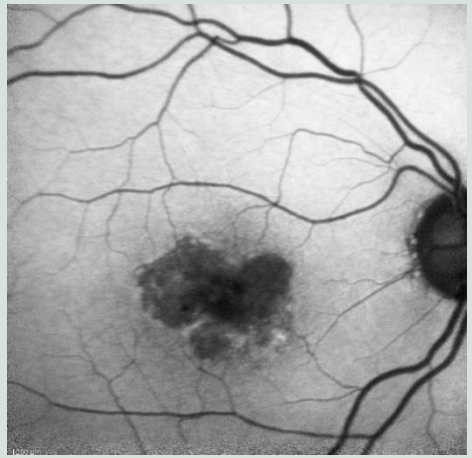


Significant outer retinal atrophy, leading to underlying choroidal hypertransmission.

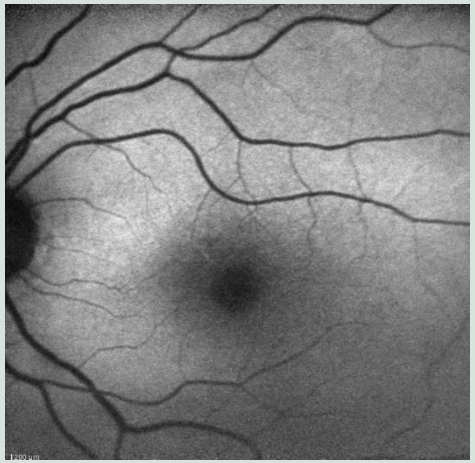


Distinctive focal ellipsoid zone integrity loss located at the foveola.

Autofluorescence



Hypo-autofluorescence corresponding to the atrophic areas surrounded with hyper-autofluorescent borders.



Normal.

The presence of this specific pattern of macular atrophy, in combination with medical and family history, raised the suspicion for MIDD. Patient was sent for screening for a mutation of mtDNA (m.3243A>G), in order to genetically confirm the diagnosis. The results from the molecular screening are anticipated.

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Conclusion

MIDD is a rare genetic disease, related to the m.3243A>G point mutation, first described by J.A.Massen et al. in 1992 [1]. The diagnosis is suspected based on the presence of one or more of:

- Maternal heritability of diabetes.
- Hearing impairment.
- Maculopathy: affects a high percentage, taking the form of a usually bilateral macular pattern dystrophy and/or RPE and outer retinal atrophy. The case presented here has an asymmetrical macular involvement which is less common in these patients.

MIDD is often misdiagnosed, thus it is important to raise awareness about the clinical and imaging manifestations of this entity.

Early diagnosis is important: all first-degree family members should be screened for the mutation and have genetic counseling in order to have the appropriate examinations upon diagnosis.

References:
1.an den Ouweland JM, Lemkes HH, Ruitenbeek W, Sandkuijl LA, de Vijlder MF, Struyvenberg PA, van de Kamp JJ, Maassen JA. Mutation in mitochondrial tRNA(Leu)(UUR) gene in a large pedigree with maternally transmitted type II diabetes mellitus and deafness. Nat Genet. 1992 Aug;1(5):368-71. doi: 10.1038/ng0892-368. PMID: 1284550.