Retinal changes in myotonic dystrophy (DM1): a case report and a review of the literature

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INTRODUCTION

Ocular manifestations of myotonic dystrophy type 1 (DM1) include **cataracts, ptosis, weakness of the ocular muscles, ocular hypotony and retinal changes**. Ophthalmologists are familiar with cataract in DM1 patients, but **retinal involvement is less well known**, although the **1st publication** dated in **1952**.

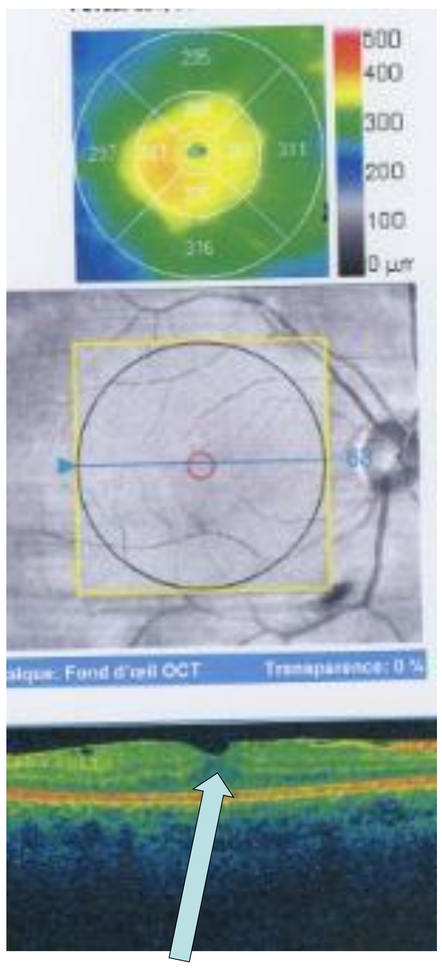
CASE REPORT (Summary)

We report the case of a **55-year-old female**, previously confirmed as DM1 who ,experienced **visual acuity decrease** on both eyes. Slit lamp examination revealed **lens opacity**. Optical coherence tomography (OCT), performed in 2008, 2013 and 2015, showed a slow progression of **bilateral epiretinal membranes (ERM)**.

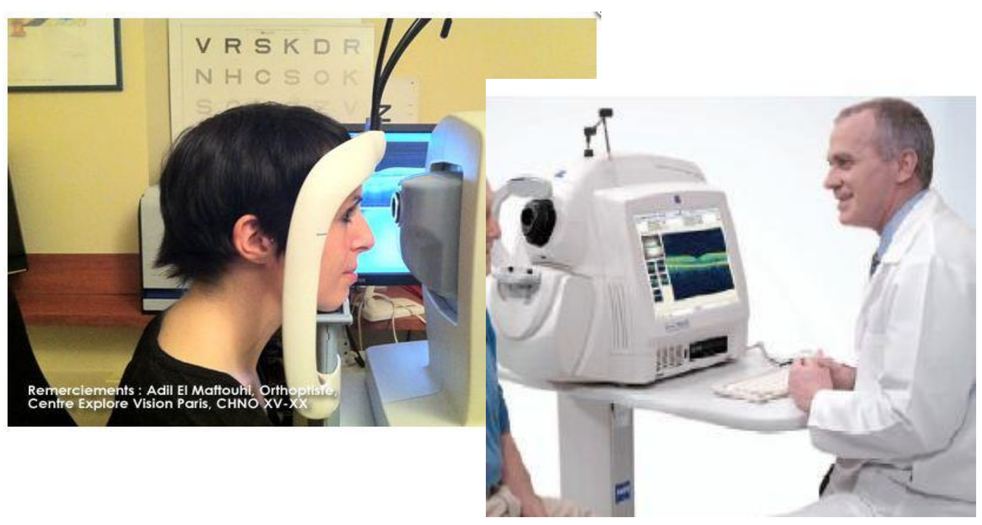
- **Family medical history : Father:** retinal detachmant surgery (boths eyes); **Children (3 sons)** with DM1.
- **Opthalmological medical history**:
- **2001**: **Left** Eye: retinal flange;
  Right Eye: retinal tears 4H 9H, treated with laser, then relapse and laser treatment again.

Optical Coherence Tomography (OCT):

- **2008**: bilateral vitreoretinal traction syndrome (Jaffe syndrome or early stage of ERM)
- **2011**: bilateral ERM, no macular lesion. slight
- **2015** : Right Eye: slight development of the ERM in macular region; small increase of retinal thickness
  Left Eye: no evolution of ERM.

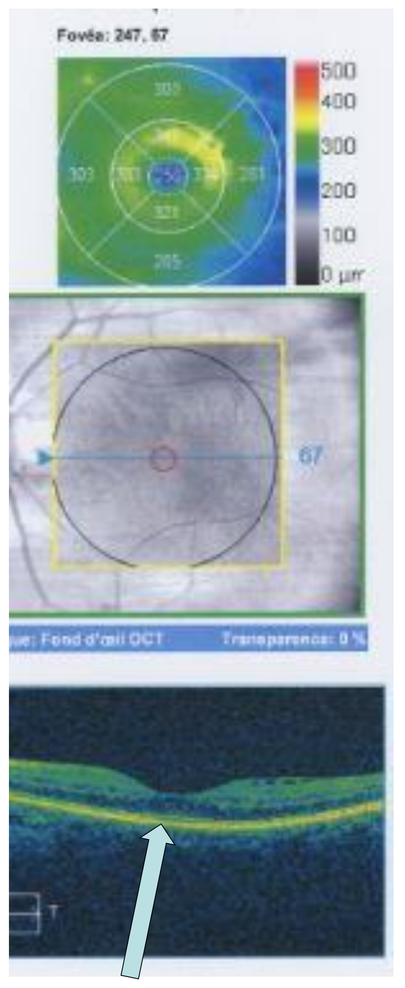


OCT (2015) – Right Eye: macular region (thickening)



Optical coherence tomography (OCT):

- Imaging technology performing a **real-time « optical biopsy »**
- Can identify **early stages** of disease
- OCT is a **rapid**, **noncontact**, and **noninvasive exalination** method of imaging intraocular tissue
- OCT has significantly improved the **understanding** of retinal disease pathogenesis, and **monitoring** of disease progression ans response to therapy.



OCT (2015) - Left Eye: macular region (no evolution)

LITERATURE REVIEW

A searching in PubMed using key-words: « retina » AND « myotonic dystrophy" has identified **about 30** articles reporting cases or case-series of DM1 patients with retinal lesions (**5 papers over the period 2011-2015)**.
According to a study (*Kernsten et al, 2014*) (incuding **30 patients with DM1 et 28 controls)**, OCT examination revealed:

- **EMR** (**48.2% of DM1** patients'eyes eyes vs 12.5% controls' eyes),
- **56.7%** of DM1 patients **had an EMR in at least 1 eye**.
- an average **macular thickness** significantly greater (DM1 patients vs controls: 327.3 µm vs 308.5 µm),

CONCLUSIONS

- In patients with DM1, the visual disturbance could have been caused by both **cataracts** and **retinal degeneration**
- It is of great relevance to analyze the presence of retinal changes that might limit the visual improvement following cataract extraction (*Esteves, 2013*).
- As therapeutic approach is available for epiretinal membrane, OCT should be implemented as part of an ophthalmologic assessment for DM1 with reduced visual acuity (*Kersten, 2014*).
- A better collaboration between ophtalmologists and neurologists from neuromuscular centers coud optimize the eye care management of patients with DM1. This partnership could be facilitated through DM-SCOPE, the longitudinal observational DM1 registry. To date, DM-scope collects information from more than 2,200 patrients.

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