OCT provides a better understanding about retinal damage in two rare neuromuscular diseases

The facial - scapular - humeral muscular dystrophy (FSHD) and myotonic dystrophy type 1 (DM1) are neuromuscular diseases with systemic clinical manifestations. Although rare, they are genetic myopathies most frequently encountered in adults. The diagnosis is confirmed by molecular biology by identification of the genetic defects. A literature review was performed, focusing on retinal damage in FSHD and DM1, with the goal of promoting retinal monitoring by OCT.

Fig. 1. OCT: the results are presented on the screen.

OCT stands for "Optical Coherence Tomography" and is similar to ultrasound. Instead of sound waves, a special laser light and totally harmless is used. Light (optical) laser consists of waves of coherent light (coherence) for producing sectional image (tomography) of ocular tissues. Cross-sectional visualization is an extremely powerful tool in the identification and assessment of retina abnormalities. Appeared in 1996, OCT is the most valuable advance in retinal diagnosis imaging since the introduction of fluorescein angiography in 1959. OCT technology has continually evolved. It allows to visualize with high accuracy the structure and thickness of the cells of the retina and the optic nerve (nerve that route the visual information to the brain). It can identify very subtle retinal interface pathologies, such as fine epiretinal membranes. The ability to perform volumetric and retinal thickness analysis provides a quantitative and repeatable method to evaluate disease progression. To help the practitioner to better see small details, the results are presented with artificial colors (false color) (Fig. 1.).

OCT is a non-invasive technique. There is no contact with the eye and OCT requires no anesthesia. The test is absolutely painless. A measurement takes a few seconds. Measuring

cycles take between 1-10 minutes, depending on the issue. Images are printed and explained in detail and given to patients. Furthermore, the images are stored and available for subsequent investigations.

In DM1, besides muscle symptoms, a wide range of ocular symptoms has been described. The ocular manifestations such as cataracts, ocular hypotonia and retinal damage are frequently reported. The literature review indicates that 56.7% of DM1 patients had epiretinal membranes in at least one eye. The peripheral retinal pigment clumps were reported in 50% of patients with DM1 patients. A monitoring beyond 5 years in 9 patients showed that retinal pigmented lesions had rather slow evolution.

In FSHD, most patients develop mild to moderate abnormalities of the retinal vasculature (telangiectasia, microaneurysm), with no clear relation with age and severity of the myopathy. Most of retinal lesions were asymptomatic.

OCT is a tool of choice for monitoring retina of patients with FSHD and DM1, who generally have pupil difficult to dilate. As OCT allows to examine the eye fundus without dilatation, earlier detection of retinal lesions can be performed.

A close collaboration between ophthalmologists and neurologists is necessary for monitoring patients with FSHD and DM1. When the diagnosis is known, eye examination is essential as treatments to limit visual impairment may be proposed. Otherwise, some ocular signs, sometimes discrete, need to be recognized by ophthalmologists as clues for identifying these rare myopathies.

A disease or disorder is defined as rare in Europe when it affects less than 1 in 2,000.

Most rare diseases (also referred to as orphan diseases) are genetic, and thus are present throughout the person's entire life, even if symptoms do not immediately appear.

For rare diseases, exchanges between experts from different medical specialties are facilitated by using common databases. For DM1, the collaboration is facilitated thanks to DM-scope, the French registry that collects so far data relating to more than 2,200 patients.

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Publication

[Literature review of the importance of retinal examination in two genetic neuromuscular diseases (DM1 and FSHD). Potential clinical applications].
[Article in French]

Brignol TN.
J Fr Ophtalmol. 2015 Dec