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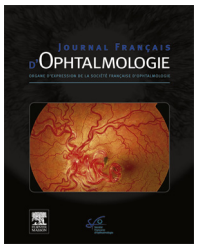

EDITOR'S CHOICE

Corneal dystrophies☆

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Endothelial dystrophy;
Classification

Summary Degenerative or hereditary corneal diseases are sometimes difficult to discriminate. Corneal dystrophies affect approximately 0.09% of the population. They are identified by the IC3D classification based on their phenotype, genotype and evidence gathered for their diagnosis. In practice, the ophthalmologist manages functional symptoms such as recurrent erosions, visual loss and amblyopia, photophobia, foreign body sensation, and sometimes pain and aesthetic concerns. Medical treatments consist of drops to promote healing, ointments, hyperosmotic agents and bandage contact lenses. Less invasive surgical treatments are used as second line therapy (phototherapeutic keratectomy, lamellar keratectomy). More invasive procedures may eventually be utilized (lamellar or penetrating keratoplasty). Anterior lamellar or endothelial keratoplasty are now preferred to penetrating keratoplasty, although the latter still remains the only possible option in some cases. Some rare dystrophies require coordinated and comprehensive medical care.

☆ See this article unabridged, illustrated and detailed, with electronic enhancements in EMC-Ophtalmologie: Bourges JL. Les dystrophies de cornée. EMC — Ophtalmologie 2017 [Article 21-200-D-30].

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Definition

A corneal dystrophy is an abnormality of one or several layers of the cornea of genetic or hereditary epigenetic origin. Its occurrence is not dependent on environmental, inflammatory or systemic factors, although these may sometimes influence its progression [1].

A corneal degeneration is a change, which causes the cornea to lose one or more of its normal properties.

The border between dystrophy and degeneration is not always clear. In fact, technological and biological advances allow the description of common familial characteristics for conditions previously thought to be degenerative. One typical example is keratoconus.

Epidemiology

Corneal dystrophies affect 0.09% of the population [2]. Approximately 60% are of endothelial origin.

Classification

It is possible to classify corneal dystrophies by two major approaches: the phenotypic approach and the genotypic approach.

Phenotypic approach

This offers two sub-classifications, anatomic and/or descriptive.

The anatomic-histologic classification describes disease by the involved corneal layer(s). The involvement may be epithelial, epithelial-stromal, stromal or endothelial-Descemetic. The anatomic-descriptive classification separates the dystrophies by the appearance, which they take on in the affected corneal layers. They are granular, lattice, ''honeycomb'', mosaic, amyloid, etc. (Table 1).

Genetic approach

This combines Mendelian genetics with molecular genetics. The principle of molecular identification uses the description of the affected chromosomal region, the mutated gene, identification of the mutation and its affected product.

The international classification proposed by the International Committee for Classification of Corneal Dystrophies (IC3D) is currently the most utilized and functions as the standard [3,4]. It combines the phenotypic and genetic axes. Its 2015 revision shows how the classification of dystrophies is changing and open for discussion. It is transcribed in Table 1. It attributes one category to each dystrophy, which translates the level of proof established for its diagnosis.

Specifics of the clinical and paraclinical examination

History

This verifies that a supposition of heredity does exist. It allows the establishment of a mode of transmission and a census of the subjects. It looks for systemic involvement. It may be independent or associated with a corneal dystrophy. The functional history attempts to objectify the visual effects of the dystrophy. It establishes progression. This progression determines the management strategy and suggests the prognosis.

Biomicroscopic examination (SLE)

The entire cornea must be examined anatomically. The examination must be bilateral, since the phenotype is generally bilateral. It is of primary importance to observe all the layers of the cornea. The illumination goes successively from diffuse to a broad slit, then narrow, and then retroillumination. Observation angles should be en face and with an oblique slit. Epithelial and endothelial specular reflection should be analyzed. Dyes such as fluorescein allow the evaluation of epithelial integrity and regularity of the corneal surface. Superficial corneal sensitivity is tested over the entire corneal surface. Pachymetry and its regularity are evaluated. Similarly, the corneal curvature and power must be known so as to organize the treatment strategy. Finally, biomicroscopic examination should be performed any time it is possible on family members who are present or who can be called in.

Useful ophthalmologic testing for corneal dystrophies

It is useful to document the biomicroscopic examination with photos, which should be repeated as the disease progresses.

Corneal elevation topography allows the evaluation of pachymetry at every point on the cornea and the assessment of the curvature and refractive power so as to adapt treatment. The posterior elevation values are unreliable when corneal transparency is highly altered.

OCT allows for visualization of the location, shape, extent and depth of deposits or dystrophic corneal anomalies.

In vivo confocal microscopy (IVCM) characterizes the intracorneal deposits or tissue abnormalities, as well as endothelial cell quality and density.

General therapeutic principles

We cannot currently treat the genetic cause of these conditions. Treatment is therefore aimed at relieving functional signs, which vary with the dystrophy and its stage of progression. They are primarily recurrent epithelial erosions, decreased visual acuity, photophobia, edema or pain.

The treatment strategy takes into particular account the following:

- dystrophies regularly affect young, immunocompetent, active and/or atheletic patients, thus, corneal biomechanics must be preserved, as well as the possibility of future treatments;
- dystrophies progress, and the rate of progression is different for each dystrophy;
- dystrophies involving deposits recur after treatment, sometimes more densely than before, in a less granular and more diffuse fashion;