Vision

Signs and Symptoms

Blurred Vision

Visual impairments in patients with DM1 and DM2 are most often caused by cataracts. Posterior subcapsular iridescent lens opacities represent an initial phase of cataract formation in myotonic dystrophy and are detectable only with slit lamp biomicroscopy. These opacities are usually found in patients who have not developed any visual symptoms. The presence of these types of lens opacities and more mature cataracts may be the only sign of the disease. Posterior subcapsular iridescent lens opacities are highly diagnostic of DM1 and DM2 although not pathognomonic. Glare and blurriness of the vision develop as the progression of the lens opacities into stellate cataracts and eventually mature cataracts, which are indistinguishable from usual cataracts. Cataracts in DM1 and DM2 may progress faster than usual cataracts, and thus patients with DM1 and DM2 may be presented with early-onset cataracts.

Retinopathy

Retinopathy is often detected by electroretinogram (ERG), but seldom causes clinically significant visual impairments. In rare cases, gradual progressive changes in the pigment epithelium of the retina can be detected with indirect ophthalmoscopic examination.

Bilateral Blepharoptosis (Ptosis)

Bilateral ptosis is a frequent feature of DM1 but seldom seen in patients with DM2. It is often found in DM1 patients with characteristic hatchet facies. In severe cases, ptosis can obstruct vision and require intervention.

Ocular Hypotension

Reduced ocular pressure is detected by ocular tonometry as an incidental finding during a routine eye examination.

Ocular Myotonia

Unlike other myotonic disorders caused by muscle chloride (Thomsen's and Becker's myotonia congenita) and sodium (paramyotonia) channel gene mutations, DM1 and DM2 do not cause overt ocular myotonia (often detected as delayed eye opening after forceful eye closure). Lid lag is also usually absent in patients with DM1 and DM2. Although saccadic eye movements may be affected by myotonia, they are generally of no clinical significance.

Diagnosis

Annual ophthalmological examination

Annual eye examinations should be done on every myotonic dystrophy DM1 and DM2 patient to assess above-described eye problems.

Slit-lamp biomicroscopic examination

Slit-lamp biomicroscopic examination must be performed to diagnose these early lens opacities. A general assessment for lens opacities and cataracts may be done during a regular eye exam, but ophthalmologists and optometrists often do not recognize the iridescent lens opacities unless prompted.

Electroretinogram (ERG)

A moderate-to-advanced cataract can interfere with the diagnosis of retinopathy. ERG is not routinely performed unless retinopathy is suspected by routine eye examinations.

Treatment

Prevention of lens opacities

There is no proven therapy to slow or prevent the progression of lens opacity once it develops. Reducing UV ray exposures by wearing sunglasses is generally recommended.

Cataract surgery

Surgical removal of the opaque lens with intraocular lens implantation is indicated when cataracts interfere with the patient's ability to meet the needs of daily living. It is no longer necessary to wait for "ripeness" (vision impairment severe enough to absolutely require surgery). Similarly, there is no technical or medical advantage to taking out a cataract sooner; later treatment does not cause adverse outcomes, since pre-operative visual acuity has no bearing on the outcome of cataract surgery.

Modern microsurgical techniques

Techniques such as standard extracapsular cataract extraction and phacoemulsification (also called small incision surgery) allow cataract surgery to be performed under local anesthetic on an outpatient basis. The surgery is a low risk procedure, but careful pre- and post-operative evaluation is nevertheless important, particularly since myotonic dystrophy patients have elevated risks associated with anesthesia and often have other chronic medical conditions. General anesthesia is necessary only for patients who cannot be counted on to cooperate under local anesthesia, such as those who are significantly cognitively impaired or very young.

Blepharoplasty

The following interventions may be warranted when ptosis is severe and obstructs vision. (Surgery is often delayed as long as possible in patients with muscle disease because repeated procedures will likely be required due to the progressive nature of the disease.)

- Crutches. Eyelid crutches inserted into eyeglasses should be tried before blepharoplasty is considered.
- Frontalis suspension of eyelids. When severe bilateral ptosis and poor levator function are present, frontalis suspension surgery may be performed. A sling is formed which lies below the skin surface and connects the upper eyelid to the frontalis muscle.
- Cosmetic surgery. Surgery may also be considered for cosmetic reasons, but patients should be aware of the potential complications. The most common troubling complication of ptosis surgery is lagophthalmos or failure of the eye to close completely. This in turn may lead to dry eye and exposure keratopathy.