

The logo consists of the letters 'M' and 'D' in a thick, rounded, teal-colored font. The 'M' and 'D' are connected at the top and bottom, with the 'D' having a curved bottom edge.

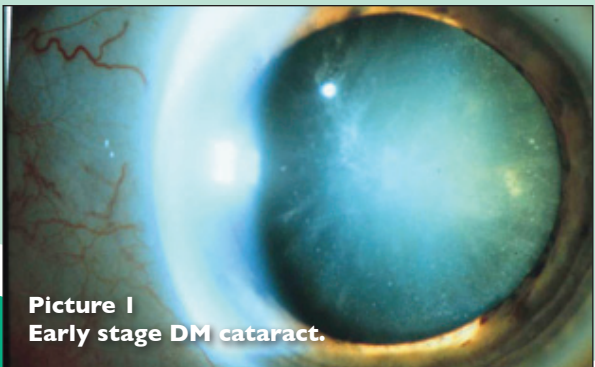
Myotonic Dystrophy
SUPPORT GROUP

Myotonic Dystrophy and the Eye

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Myotonic Dystrophy

The eye is very badly affected by myotonic dystrophy (DM) and almost every part can show signs of disease. The symptoms can include: droopy eyelids (ptosis), weakness in the extraocular muscles, corneal ulcers, abnormal blood vessel formation in the iris, low internal eye pressure (hypotony), and damage to the retina at the back of the eye (retinopathy). It has also been reported by one study that more than 80% of DM patients between the ages of 5-17 had long sightedness (hypermetropia) caused by a shortened eyeball. The lens of the eye appears to be particularly sensitive to the effects of DM and the early appearance of cataract is one of the most common and reliable symptoms of the disease. Like all aspects of DM, however, the presence and severity of eye symptoms are extremely variable and no two patients will appear the same.



Picture 1
Early stage DM cataract.

MD

Myotonic Dystrophy and Cataract

In the general population, cataract is most frequently associated with older people and more than 80% of people over the age of 75 years have cataract. In DM, however, cataract can occur at

a much earlier age and can even appear in the lenses of teenagers. In many individuals with DM the presence of a cataract may



Picture 2
DMI cataract.
(By David Broadway, 2012)

be the only obvious symptom of the disease and it is, therefore, important for ophthalmologists to be aware of the possibility that DM may be involved, particularly if there is a history of cataracts in the family.

The lens of the eye is normally crystal clear so that it can focus what we see onto the retina at the back of the eye, in much the same way the lens of a camera focuses images onto the film. In cataract this perfect clarity is disturbed

Table 1. The Symptoms of Myotonic

Part of the eye affected	Symptoms
Ocular motor function	<ul style="list-style-type: none">• Abnormal saccadic and smooth pursuit eye movements. Strabismus• Altered versions/ductions• Nystagmus
Eye lids	<ul style="list-style-type: none">• Ptosis• Blepharitis
Tears	<ul style="list-style-type: none">• Epiphora
Cornea	<ul style="list-style-type: none">• Corneal lesions• Keratitis
Iris	<ul style="list-style-type: none">• Rubeosis (neovascularisation)• Microhaemangiomas (vascular tufts)
Eye pressure	<ul style="list-style-type: none">• Hypotony• Flare in anterior chamber
Lens	<ul style="list-style-type: none">• Cataracts
Lens after cataract surgery	<ul style="list-style-type: none">• PCO• Fibrosis
Retina	<ul style="list-style-type: none">• Pigmentary retinopathy• Abnormal ERG activity• Reduced photopigment

Dystrophy in the Eye.

	Notes
	Both extraocular muscle and CNS involvement.
	Myotonia and muscle wasting.
	Weepy eyes.
	Ulcers and inflammation.
	Abnormal blood vessel permeability reported.
	Reduced aqueous production. Impaired ciliary function. Detached ciliary reported.
	Posterior subcapsular cataracts. Iridescent or white dust-like opacities. Spokes and cortical cataract when mature.
	Aggressive posterior capsule opacification (PCO) and rhexis contracture reported. Laser treatment required.
	Optic nerve atrophy reported. Butterfly-shaped and reticular dystrophies reported.

and opaque white patches develop in the lens preventing the formation of clear images. If not treated, cataract can cause total blindness in the affected eye.



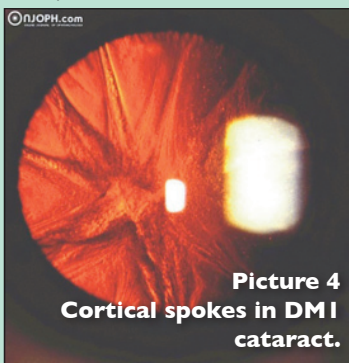
Picture 3
Early stage DM2 cataract.
(By Lori J. Williams, 2008)

In its early stages, DM cataract is quite unusual in appearance and has frequently been used as an early diagnostic indicator. Typically the cataract first appears in the

outer layers at the back of the lens (posterior subcapsular) as very fine dust-like particles that can have a brightly coloured (iridescent) appearance (Pictures 1-3).

At this early stage, it is often called a Christmas tree cataract because of its resemblance to the coloured lights on a Christmas tree. As the cataract develops, however, the number of particles increases and larger spokes and star-shaped white regions appear which cloud the

lens and eventually form a mature cataract (Picture 4) which is indistinguishable from other more common types. Cataract is also a feature of DM2 (Picture 3) and as far as we know develops in the same way as has been observed in DMI. As the cataract grows, a process which can take several years, it spreads from the edges to the centre of the lens where it starts to have a detrimental effect on vision.



The Cataract Operation

When normal clear sight is no longer possible your ophthalmologist will usually recommend that you have an operation. The modern cataract operation is a relatively simple procedure taking less than an hour to perform. After a local anaesthetic has been applied to the eye, a small incision is made in the cornea through which the surgeon can remove the damaged

lens by a process called phacoemulsification. Once removed the damaged lens is replaced with a new, artificial, plastic lens called an IOL (intraocular lens) which restores near perfect vision to the vast majority of patients.

Cataract patients should, however, be aware that there appears to be an increased risk of secondary cataract or PCO (posterior capsular opacification) in DM and patients may be required to return to hospital for corrective laser treatment at a later date.

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MD

Glossary

TERM - DEFINITION

ANTERIOR CHAMBER - The fluid-filled space between the **iris** and the **cornea**.

AQUEOUS - Watery solution.

BLEPHARITIS - Inflammation of the eyelid.

CATARACTS - Cloudy patches in the **lens** of the eye.

CILIARY - The **ciliary** body is at the front of the eye. One of its functions is to hold the **lens** in place.

CNS - Central nervous system: the brain and the spinal cord, which process sensory information.

CORNEA - The clear 'window' at the front of the eye.

CORNEAL LESIONS - Damage to the **cornea**.

CORNEAL ULCER - Open sore on the **cornea**.

CORTICAL CATARACTS - **Cataracts** that affect the outer layer of the **lens**.

DMI - Child-onset, more common form of myotonic dystrophy.

DM2 - Adult-onset, rarer form of myotonic dystrophy, generally with less severe symptoms.

DUCTION - Eye movement involving only one eye.

EIPHORA - Watering eyes, because tears do not drain away properly.

ERG - Electroretinography: a way of measuring the electrical responses of cells in the **retina** when a patient is asked to look at flashes of light, patterns etc.

EXTRAOCULAR MUSCLES - 7 muscles that control eye movement.

FIBROSIS - Usually after injury, body tissues thicken and scar.

FLARE - Proteins leak from the blood into the **anterior chamber** and make the fluid here less clear, thus impeding vision.

HYPERMETROPIA - Long sightedness.

HYPOTONY - Low pressure in the eye.

IRIS - The coloured part of the eye.

KERATITIS - Inflammation of the **cornea**.

LENS - A structure at the front of the eye that lets light in.

MICROHAEMANGIOMAS - Clumps of abnormal, tightly coiled blood vessels in the **iris**.

MYOTONIA - Inability to relax the muscle after muscle contraction.

NEOVASCULARISATION - The formation of new blood vessels.

NYSTAGMUS - Involuntary eye movements.

OCULAR MOTOR FUNCTION - Eye movement.

OPACITY - An opaque area, i.e. an area you cannot see through.

OPHTHALMOLOGIST - A medical doctor specialising in eye health and disease.

OPTIC NERVE ATROPHY - Deterioration of the **optic nerve** which carries electrical signals to the brain when light hits the **retina**.

PCO - Posterior capsule opacification: clouding of the **lens** after **cataract** surgery, which is easily removed using a laser.

PHACOEMULSIFICATION - Cataract surgery where ultrasound equipment is used to remove the **lens**.

PHOTOPIGMENT - A type of pigment found in the cells of the **retina**. **Photopigments** are involved in telling your brain what your eyes are looking at.

PIGMENTARY RETINOPATHY - Deposits of pigment on the **retina**.

POSTERIOR SUBCAPSULAR CATARACTS - **Cataracts** that affect the back of the **lens**.

PTOSIS - Droopy eyelid.

RETICULAR DYSTROPHIES - Degeneration of the **retina**.

RETINA - The layer at the back of the eye that detects light.

RETINOPATHY - Damage to the **retina**.

RHEXIS CONTRACTURE - Contraction of the wound in the **lens**.

RUBEOSIS - New, abnormal blood vessels on the **iris**.

SACCADIC - Fast movement of the eye.

SMOOTH PURSUIT EYE MOVEMENTS - Allows eyes to closely follow a moving object.

SPOKES - **Opacities** that look like spokes in the **lens**. This is a symptom of **cortical cataracts**.

STRABISMUS - Where the eyes are not properly aligned. This can affect vision.

VASCULAR TUFTS - See **microhaemangiomas**.

VERSION - Both eyes moving in the same direction.

Further information about Myotonic Dystrophy can be obtained from the:

Myotonic Dystrophy Support Group

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NG4 3HQ

Tel/Fax: 0115 987 5869

Email: contact@mdsguk.org

Web: www.myotonicdystrophysupportgroup.org

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Other publications available from the Myotonic Dystrophy Support Group:

- Anaesthesia and Sedation for patients with Myotonic Dystrophy
- Basic Information for Midwives
- Bowel Problems in Myotonic Dystrophy
- Congenital Myotonic Dystrophy
- Excessive Daytime Sleepiness and Myotonic Dystrophy
- Facts for patients, family members and professionals
- Myotonic Dystrophy Support Group
- Relatives Information
- The Heart and Myotonic Dystrophy
- Why do we get new families with Myotonic Dystrophy?



Myotonic Dystrophy
SUPPORT GROUP

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