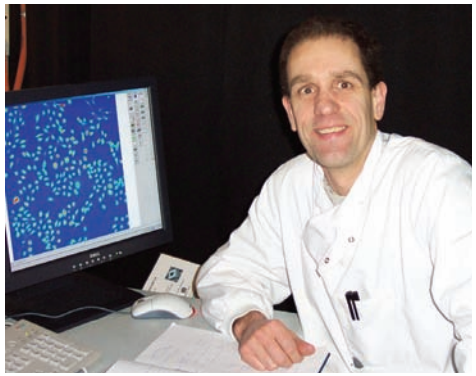
If you wish to donate a sample please contact Dr. Jeremy D. Rhodes at the address shown below. He will need to know the following information:

1. The date of your cataract operation — it is important to allow enough time for him to arrange collection of the sample and postage back to his laboratory, so please contact him at least 4 weeks before hand if at all possible.
2. The name and contact details of your Ophthalmologist/ Surgeon.
3. The details of your myotonic dystrophy and cataract if you know them.



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Tel: 44(0)1603 592252
Email: j.Rhodes@uea.ac.uk

Useful Reading and Web Links

1. Harper, PS, *Myotonic Dystrophy the facts* 2002: Oxford University Press
2. International Myotonic Dystrophy Organization www.myotonicdystrophy.org
3. Muscular Dystrophy Association www.mdausa.org
4. Athena Diagnostics www.athenadiagnostics.com
5. Washington University in St. Louis http://neuromuscular.wustl.edu/
6. National Institute of Neurological Disorders and Stroke http://www.ninds.nih.gov/

For more information

Please visit our website www.myotonicdystrophy.org

Testing and carrying an alert card is important for everyone diagnosed with Myotonic Dystrophy. You can request a free medical alert card by emailing your name and address to info@myotonicdystrophy.org.

International Myotonic Dystrophy Organization (IMDO) promotes and advances the welfare, care, treatment, and research of persons with Myotonic Muscular Dystrophy or Congenital Myotonic Dystrophy. It educates the persons with Myotonic Dystrophy or Congenital Myotonic Dystrophy on their disease, as well as educates the general public, and expedites the discovery of a cure for Myotonic Muscular Dystrophy and Congenital Myotonic Dystrophy by funding projects that will assist IMDO in meeting its goals.

International Myotonic Dystrophy Organization, Inc.

P.O. Box 1121
Sunland, CA 91041-1121
Toll Free: 866.679.7954
Fax: 818.352.6474

6353 Corte Del Abeto, Ste. B102
Carlsbad, CA 92011
Tel: 760.918.0377
Fax: 760.444.2716

www.myotonicdystrophy.org

International Myotonic Dystrophy Organization, Inc.

Cataract in Myotonic Dystrophy

www.myotonicdystrophy.org

Leading the way to management and treatment

What is Myotonic Dystrophy?

Myotonic Dystrophy, also known as Steinert's Disease or Dystrophia Myotonica (DM), is the most common inherited muscle disease, affecting about one in every 8,000 people. It is the most common form of Muscular Dystrophy in adults.

In general, DM consists of muscle weakness and myotonia (slow relaxation of muscles after contraction), which gets more severe over time. Specific problems in other systems of the body can also occur. Since DM can affect many tissues and organs, it is called a "multi-systemic" disorder.

Myotonic Dystrophy is an extremely variable condition. In fact, it is one of the most variable disorders know, which can lead to difficulties in getting an accurate diagnosis.

DM can vary in severity in the systems of the body it affects and in the age of onset. DM has equally severe effects on most other organs of the body and the symptoms include: cataracts, heart defects, breathing difficulties, mental impairment, testicular atrophy, diabetes and premature balding.

The discovery of the gene alteration, which causes DM1 and DM2, helps in the accurate diagnosis of DM1 and DM2. The discovery of the gene alteration, which causes DM, helps explain this conditions' incredible variation. The discovery was commercialized by Athena Diagnostics in 1992.

Myotonic Dystrophy and the Eye

The eye has a unique involvement in myotonic dystrophy, which makes it an important and useful organ in which to study the mechanism of this complex disease. Symptoms of DM in the eye include: retinal degeneration, low intraocular pressure, ptosis, epiphora and extraocular myotonia. The lens appears to be particularly sensitive to this disease and the early appearance of a characteristic cataract is one of the most reliable symptoms of DM1 and DM2. In many individuals the presence of a cataract may 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.

Myotonic Dystrophy and Cataract

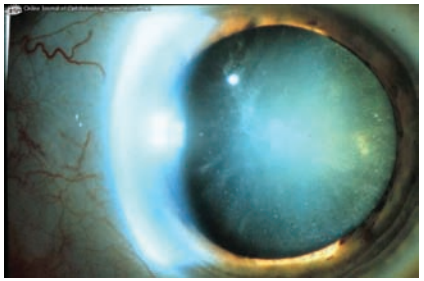

figure 1

DM cataract has a very unusual development, particularly in its early stages when it is often confused with a Christmas tree cataract. The cataract first appears in the outer layers of the lens (cortical and subcapsular) as fine dust like opacities that can have a highly coloured, iridescent appearance (Figure 1). As the cataract develops the number of these increases and cortical spokes appear in the lens (Figure 2). In mature DM cataracts the dust like coloured opacities become less obvious as larger areas of damage appear that are difficult to distinguish from more general types of cortical cataract. There is an excellent description of the development of myotonic dystrophy cataract in the book by Sir Peter Harper.[1]

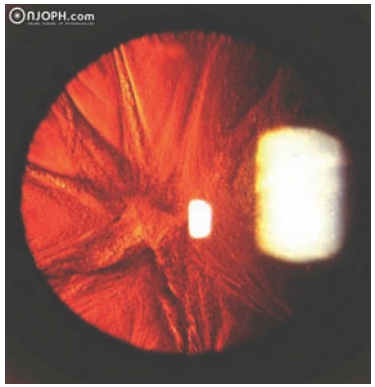

figure 2

Cataract Tissue Samples

The cataract operation provides us with a unique opportunity to obtain lens tissue samples, which would otherwise be discarded by the surgeon. In order to remove the cataract a small disc of the capsule that encloses the lens is removed by the surgeon. This tiny piece of tissue (the capsulorhexis sample) holds enough living cells on its surface to enable us to perform our experiments. One of the most important experiments we can do with these cells is to measure how they grow in the laboratory compared to the cells of patients with other non-DM types of cataract. In Figure 3 is a photograph of DM lens cells growing from a lens sample onto a plastic dish.

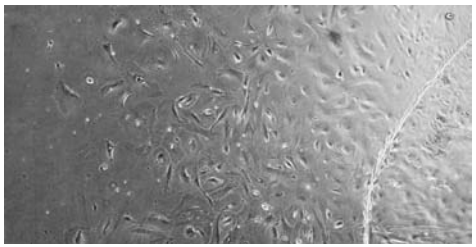

figure 3

Myotonic Dystrophy and Cataract Research

The Norwich Eye Research Group (University of East Anglia, UK), founded by Professor George Duncan (1943–2007), is one of the world's foremost centres for lens research and we are the only group, worldwide, that is currently researching the causes of cataract in Myotonic Dystrophy. There is currently no cure for DM and the ultimate aim of my research is to identify ways in which it will be possible to treat not only the eye but other organs affected by this extremely disabling disease. A significant challenge, when studying a relatively rare disease, is obtaining sufficient tissue samples for our experiments. To address this we have been working in conjunction with the International Myotonic Dystrophy Organization, Inc. (IMDO) to alert patients and clinicians worldwide to the need for samples, without which this important work would not be possible.