

**International consensus guidelines set out best-practice care for Duchenne muscular dystrophy**

A major international consensus document setting out best practice in care for Duchenne muscular dystrophy (DMD) is now available as an e-publication from the *Lancet Neurology* journal ahead of its printed release in January 2010. The product of an extensive review process by 84 international experts representing 20 disciplines across DMD diagnosis and care, this document is a unique guide for individuals, care providers and families to comprehensive healthcare management for individuals with DMD.

The drafting of these guidelines is the result of a three-year-long project guided by the US Centers for Disease Control (CDC) using a rigorous review process that required expert panels to consider more than 70 thousand different care scenarios. The preparation of the guidelines was supported by advocacy groups worldwide and by TREAT-NMD ([www.treat-nmd.eu](http://www.treat-nmd.eu)), an international network formed to advance diagnosis, care and treatment for people with neuromuscular diseases. In a close collaboration between TREAT-NMD, patient advocacy groups and healthcare professionals, the full academic publication is also being transformed into a comprehensive “family guide”, which will be made available at the start of 2010.

There is still no cure for DMD, but it is recognised that receiving the best care can dramatically improve the quality of life and life expectancy of individuals with the condition, enabling them to lead fulfilling, independent lives into adulthood. The importance of care recommendations such as these therefore cannot be underestimated. The international guidelines, which cover the diagnostics, cardiovascular, neuromuscular, gastroenterology and nutrition, orthopaedic and surgical, psychosocial, rehabilitation and respiratory fields, can be used by doctors, patients and families worldwide as a guide to the treatment that individuals with Duchenne should receive at each stage of the disease. They are also a valuable tool for lobbying at a national level to enable incorporation of these recommendations into national health systems.

“What is really significant about these guidelines is the weight of international expert opinion behind them,” explains Kate Bushby, managing editor of the *Lancet Neurology* article, coordinator of the TREAT-NMD network and herself a doctor specialising in DMD and related neuromuscular conditions. “Guidelines containing really quite similar recommendations have been produced before, but always by a much smaller group of authors or an individual patient advocacy group, which has meant they have been easier to ignore. This document represents real international consensus including both the medical and the patient advocacy perspectives and can be used across the world as a powerful tool to recognize those centres where best practice is already in place and to identify gaps in care.”

Most of the recommendations in the document are not for especially expensive or hard-to-obtain treatments, or indeed for care that is not already available in many of the best centres worldwide. What is stressed is the importance of a multidisciplinary approach – the necessity for patients to see specialists in all the fields that are involved in DMD, and for those specialists to talk to one another to ensure a coordinated approach to the care of each individual. With this authoritative document behind them, TREAT-NMD will work with patient advocacy groups, healthcare professionals and health authorities across the world to establish the best ways of implementing these recommendations and ensuring that all individuals with Duchenne have access to best-practice care.

## **Document details**

Bushby K et al, *Diagnosis and management of Duchenne muscular dystrophy part 1* and *Diagnosis and management of Duchenne muscular dystrophy part 2*, The Lancet Neurology, In press, 2009

**Link to the Lancet Neurology article (parts 1 and 2 combined) hosted on the TREAT-NMD website:**

<http://www.treat-nmd.eu/diagnosis-and-management-of-dmd>

**Links to the Lancet Neurology article via Science Direct (please copy and paste into your browser):**

Part 1: [http://dx.doi.org/10.1016/S1474-4422\(09\)70271-6](http://dx.doi.org/10.1016/S1474-4422(09)70271-6)

Part 2: [http://dx.doi.org/10.1016/S1474-4422\(09\)70272-8](http://dx.doi.org/10.1016/S1474-4422(09)70272-8)

**Note:** A comprehensive family guide will be made available in January 2010. Anyone interested in receiving further details or translating the guide into their own native language is invited to contact TREAT-NMD at [info@treat-nmd.eu](mailto:info@treat-nmd.eu).

## **About TREAT-NMD**

TREAT-NMD is an international network that was formed to facilitate collaborative research in neuromuscular disease and create the infrastructure to ensure that the most promising new therapies reach patients as quickly as possible. The network brings together the key players in the neuromuscular field and is developing the resources that industry, clinicians and scientists need to bring novel therapeutic approaches through preclinical development and into the clinic, as well as helping to establish best-practice care for neuromuscular patients worldwide.

For more information, visit [www.treat-nmd.eu](http://www.treat-nmd.eu)

## **About the document creation process**

The document was created using a rigorous review process (known as the RAND/UCLA Appropriateness Method or RAM) that required 84 international experts to rate interventions and assessments used in the management of DMD for “appropriateness” or “necessity” at different stages of the condition. The US Centers for Disease Control and Prevention (CDC) supported its development and it was funded under the US MD-CARE Act. The managing editor was Professor Kate Bushby of Newcastle University in the UK.

## **About DMD**

Duchenne muscular dystrophy (DMD) is one of the most common genetic disorders affecting children and young adults. It is a severe muscle wasting condition affecting 1 in 3,500 newborn boys worldwide, with onset in early childhood and the ensuing progressive muscle weakness and wasting leading to affected individuals becoming wheelchair bound by their early teens. Without treatment, the condition leads to death by the early twenties. DMD is caused by mutations in the DMD gene that lead to a failure to produce a functional muscle protein called dystrophin. Although several possible treatments are currently in clinical trial, there is currently no cure, but care interventions ranging from psychosocial, cardiac and nutritional care and steroid treatment to respiratory and orthopaedic interventions can have a dramatic effect on quality of life and life expectancy.