

FOR IMMEDIATE RELEASE

Accelerated approval granted for new drug for muscular dystrophy

The prospect of widespread access to a life-changing drug for children with a rare muscular disorder is a step closer today after the United States Food and Drug Administration (FDA) granted accelerated approval for a new medication.

Eteplirsen, the first drug approved to treat patients with Duchenne Muscular dystrophy (DMD), was originally developed by a UK consortium led by Professor Francesco Muntoni, Director of the Dubowitz Neuromuscular Centre at Great Ormond Street Hospital (GOSH) and the UCL Great Ormond Street Institute of Child Health. Professor Muntoni is one of the theme leads for the NIHR Great Ormond Street Biomedical Research Centre (BRC), the only centre of its kind in the UK dedicated to paediatric research.

Duchenne Muscular dystrophy (DMD) is a devastating condition that affects one in 5,000 male babies, with around 100 new cases diagnosed in the UK each year. The disorder is caused by errors in the dystrophin gene, which affects the production of an essential muscle protein called dystrophin. Boys diagnosed with the condition experience progressive muscle weakness due to the breakdown, and loss, of muscle cells. By age 8-12, boys become unable to walk and by their late teens or early twenties the condition can become severe enough to limit life expectancy.

The newly approved drug, Eteplirsen, will be used to treat patients with a specific subset of mutations of the dystrophin gene affecting 13 per cent of boys living with DMD. The drug, which is given intravenously at regular intervals, 'skips' a part of the gene that makes dystrophin, exon 51. It allows the body to make a shortened form of the dystrophin protein, alleviating some of the symptoms of Duchenne Muscular dystrophy.

Professor Francesco Muntoni said: "It's incredibly welcome news to see Eteplirsen fast-tracked after our work to develop it here at Great Ormond Street Hospital. There are currently very few treatment options for boys with Duchenne Muscular dystrophy, who face severe health challenges. This drug could improve their symptoms and give them enhanced mobility for longer."

The drug was filed by Sarepta Therapeutics for accelerated approval by the FDA and is planned for immediate clinical use in US while the process of approval in the EU is underway.

The findings from the original study on Eteplirsen were reported in the [Lancet](#) in 2011. The first patient to receive the systemic injection of this novel compound was performed in the BRC-supported Somers Clinical Research Facility at GOSH. The study, funded by the Medical Research Council and coordinated by Professor Muntoni, recruited 18 children from all over England and was conducted in two UK sites, London and Newcastle.

Last week the NIHR Great Ormond Street Biomedical Research Centre (BRC) was awarded £37 million in funding to continue to drive forward translational research into rare diseases in children between 2017-2022. The Centre is the only one of its kind in the UK dedicated to paediatric research.

The Centre currently supports another trial coordinated by Professor Muntoni, funded by an EU grant, to develop a novel therapy for another subgroup of boys with Duchenne Muscular dystrophy. For more information visit www.skip-nmd.eu

Contact information

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For out-of-hours media enquiries call the GOSH switchboard on 020 7405 9200.

Notes to editors

About Great Ormond Street Hospital for Children NHS Foundation Trust

Great Ormond Street Hospital is one of the world's leading children's hospitals with the broadest range of dedicated, children's healthcare specialists under one roof in the UK. The hospital's pioneering research and treatment gives hope to children from across the UK with the rarest, most complex and often life-threatening conditions. Our patients and families are central to everything we do – from the moment they come through the door and for as long as they need us.

Great Ormond Street Hospital Children's Charity needs to raise money to support the hospital to give children who need help the most, the best chance for life. The charity funds research into pioneering new treatments for children, provides the most up to date medical equipment, funds support services for children and their families and supports the essential rebuilding and refurbishment of the hospital. You can help us to provide world class care for our patients and families. For more information visit www.gosh.org

About the UCL Great Ormond Street Institute of Child Health (ICH)

The UCL Great Ormond Street Institute of Child Health is part of the Faculty of Population Health Sciences within the School of Life and Medical Sciences at UCL. Together with its clinical partner Great Ormond Street Hospital for Children (GOSH), it forms the largest concentration of children's health research in Europe.

For more information visit www.ucl.ac.uk/ich

About the NIHR Great Ormond Street Biomedical Research Centre

The Biomedical Research Centre (BRC) at Great Ormond Street Hospital (GOSH) and the UCL Institute of Child Health (ICH) enables the translation of basic scientific discoveries in laboratories into 'first in man' or 'first in child' clinical studies. Our research aims to accelerate discoveries into the basis of childhood rare diseases and to develop novel diagnostics, imaging techniques and new treatments, including cellular and gene therapies.

The GOSH BRC, funded by the National Institute of Health Research (NIHR), is the largest provider of financial support for research across GOSH and ICH. It aids various programmes such as the Gene and Cell Therapy Facility, GOSomics, GOSgene and the Somers Clinical Research Facility, as well as

providing financial support to individual researchers and training opportunities. Visit the GOSH BRC website (<http://www.gosh.nhs.uk/research-and-innovation/nihr-great-ormond-street-brc>)

About the Medical Research Council

The Medical Research Council is at the forefront of scientific discovery to improve human health. Founded in 1913 to tackle tuberculosis, the MRC now invests taxpayers' money in some of the best medical research in the world across every area of health. Thirty-one MRC-funded researchers have won Nobel prizes in a wide range of disciplines, and MRC scientists have been behind such diverse discoveries as vitamins, the structure of DNA and the link between smoking and cancer, as well as achievements such as pioneering the use of randomised controlled trials, the invention of MRI scanning, and the development of a group of antibodies used in the making of some of the most successful drugs ever developed. Today, MRC-funded scientists tackle some of the greatest health problems facing humanity in the 21st century, from the rising tide of chronic diseases associated with ageing to the threats posed by rapidly mutating micro-organisms. www.mrc.ac.uk

About The National Institute for Health Research

The National Institute for Health Research (NIHR) is funded by the Department of Health to improve the health and wealth of the nation through research. Since its establishment in April 2006, the NIHR has transformed research in the NHS. It has increased the volume of applied health research for the benefit of patients and the public, driven faster translation of basic science discoveries into tangible benefits for patients and the economy, and developed and supported the people who conduct and contribute to applied health research. The NIHR plays a key role in the Government's strategy for economic growth, attracting investment by the life-sciences industries through its world-class infrastructure for health research. Together, the NIHR people, programmes, centres of excellence and systems represent the most integrated health research system in the world. For further information, visit the NIHR website (www.nihr.ac.uk).