Muscular Dystrophy UK

Fighting muscle-wasting conditions

Here for you: Research, care and information

Saturday 21 April 2018

Granta Centre, Granta Park, Great Abington
Cambridge CB21 6AL

#MusclesMatter

MDUK CONFERENCES 2018

National Conference 2018
13 October in London

Scottish Conference 2018
3 November in Glasgow

Save the dates for these opportunities to meet others and hear all the latest news and updates. Email us at infoday@musculardystrophyuk.org and we'll let you know when registration goes live.

Accelerating steps to treatments and cures

I am delighted to welcome you warmly to our Information Day here in Cambridge. Many thanks for joining us. We trust you'll have an excellent day of information, news and – perhaps most importantly – the chance to meet and make connections with others living with muscular dystrophy.

We're working to accelerate the steps to treatments and cures to improve lives today and transform those of future generations. Together, we're pressing for faster access to potential drugs that could transform lives and we're driving change to secure better care and support to help people stay active, independent and connected.

Throughout the day, you'll have the opportunity to connect with each other. Families often tell us how valuable it is to meet with other people, to share experiences and to make lasting friendships. Muscular Dystrophy UK events are always a great place to do that.

We know for many individuals and families living with muscle-wasting conditions that expert clinical care, independence and the best possible quality of life are all important goals. We're committed to supporting you at every step, by working closely with clinical teams in centres and in the community, and by providing information and advice.

Today's programme looks really exciting. You'll hear from world-leading researchers who will tell us about progress in their research into a range of muscle-wasting conditions. Health professionals will share with you information and advice about living with a muscle-wasting condition, whether that is through physiotherapy, exercise, equipment or adaptations to your home.

You'll also hear what we are doing, as a charity, to speed up the rate of progress in the fight to beat all types of muscular dystrophy. Every day counts when you're living with a progressive and potentially life-threatening muscle-wasting condition.

Enjoy this Information Day, and thank you for your valued support.

Robert Meadowcroft
CEO, Muscular Dystrophy UK

Join in the Twitter conversation!
Use #MusclesMatter and @MDUK_News

Here for you: Research, care and information
Accelerating steps to treatments and cures

9am – 10am

Registration begins
Tea/coffee available

10am – 10.15am

Lecture Room 1

Welcome and introduction
Robert Meadowcroft, Chief Executive, MDUK

10.15am – 10.45am

Lecture Room 1

National Institute for Health Research (NIHR) BioResource
Prof Patrick Chinnery, Professor of Neurology

10.45am – 11am

Lecture Room 1

Leading the fight
Nic Bungay, Director of Campaigns, Care and Information, MDUK

11am – 11.15am

Lecture Room 1

Research progress
Dr Jenny Sharpe, Research Communications Manager, MDUK

11.15am – 11.30am

Foyer
Tea/coffee break

Networking time: MDUK staff will be available to answer any of your questions

11.30am – 12.15pm

Workshop 1
(see next pages for details)

12.15pm – 1.30pm

Restaurant
Lunch

Networking time: MDUK staff will be available to answer any of your questions

1.30pm – 2.15pm

Workshop 2
(see next pages for details)

2.15pm – 3pm

Workshop 3
(see next pages for details)

3pm – 3.15pm

Foyer
Tea/coffee break

Networking time: MDUK staff will be available to answer any of your questions

3.15pm – 4pm

Workshop 4
(see next pages for details)

4pm

Questions and close
(in your workshop room)

4.30pm

Lecture Room 1

Optional workshop – Share your views, help shape MDUK's three-year strategy

Nic Bungay, Director of Campaigns, Care and Information, MDUK

Keynote speaker

National Institute for Health Research (NIHR) BioResource

Professor Patrick Chinnery, Professor of Neurology, University of Cambridge

Patrick qualified in medicine at the University of Newcastle in 1992 before undertaking a PhD on mitochondrial disease. Following further clinical training and postdoctoral research in Newcastle and London, he was appointed Lecturer at Newcastle University and in 2004 became Professor of Neurogenetics. He was Director of the NIHR Newcastle Biomedical Research Centre (2008-2015), and the Institute of Genetic Medicine at Newcastle University (2010-2015). In 2015, he moved to the University of Cambridge as Professor of Neurology and Head of the Department of Clinical Neurosciences. He jointly chairs the NIHR Rare Diseases Translational Research Collaboration.

Photographs may be taken, for use in our print and online materials, on social and other media. If you don't wish your or your family's photographs to be used, please tell one of our staff members or email brand@musculardystrophyuk.org

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TRAILBLAZERS
Muscular Dystrophy UK
Fighting muscle-wasting conditions

JOIN TRAILBLAZERS

We're a growing network of young disabled campaigners. When you join us, you'll get to make friends and campaign on issues that are important to you.

www.musculardystrophyuk.org/trailblazers

Muscular Dystrophy UK, 61A Great Suffolk Street, London SE1 0BU
Registered Charity No. 205395 and Registered Scottish Charity No. SC039445

Get involved now

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Muscular Dystrophy UK Trailblazers

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MDUK_Trailblazers

Limb girdle muscular dystrophy

Room: Lecture Room 2

11.30am – 12.15pm
Gene therapy
Dr Isabelle Richard, Director of the Progressive Muscular Dystrophies research group, Généthon

1.30pm – 2.15pm
Equipment and adaptations
Cathy Szeplaki, Occupational Therapist

2.15pm – 3pm
Physiotherapy for neuromuscular conditions
Andrew Rose, Care Advisor

3.15pm – 4pm
Questions and answers on LGMD
Dr Charlotte Brierley, Consultant Neurologist
Dr Max Damien, Consultant Neurologist
Andrew Rose and Andrea Russell, Care Advisors

4pm – 4.30pm
Questions and close

Speakers

Gene therapy
Dr Isabelle Richard, Director of the Progressive Muscular Dystrophies research group, Généthon

Since her PhD studies in 1991, Isabelle has been working on limb girdle muscular dystrophies (LGMDs). Her research has identified new genes, the function of their corresponding proteins, and potential drug targets. She has also developed animal models of LGMDs and gene therapies, which are undergoing preclinical testing. She joined Généthon in 1996 and currently leads its translational muscular dystrophy gene therapy programme.

Equipment and adaptations
Cathy Szeplaki, Occupational Therapist, Harrogate and District NHS Foundation Trust

Cathy qualified as an Occupational Therapist (OT) from St Loye's School of OT, Exeter in July 1989 and completed her MSC in Professional Health Studies in 2010 from York St John University. She is currently the Team Leader for Children's OT in Harrogate. Her previous role was Neuromuscular Occupational Therapist based within the Regional Paediatric Team at Leeds Children's Hospital, where she worked for 13 years. Cathy was involved with the revision of MDUK's *Adaptations manual* published in 2017.

Physiotherapy for neuromuscular conditions
Andrew Rose, Care Advisor, Addenbrooke's Hospital

Andy qualified as a physiotherapist in 2002, and started working in the neuromuscular field as part of the clinical research teams at Great Ormond Street Hospital and Queen Square at the start of 2010. Since the end of 2012, he's been working as one of three neuromuscular care advisors for the East of England, based at Addenbrooke's Hospital in Cambridge. In addition to being a member MDUK Research Grants Lay Panel, Andy has published research papers on topics such as falls in inclusion body myositis, advanced care discussions in life-limiting muscle conditions and the development of outcome measures in neuromuscular conditions.

Questions and answers on LGMD
Dr Charlotte Brierley, Consultant Neurologist, Addenbrooke's Hospital

Charlotte is a full-time neurology consultant. She works at Addenbrooke's Hospital where she runs the neuromuscular service, and at the West Suffolk Hospital. Charlotte runs muscle clinics at both centres along with consultant Dr Max Damien and the Neuromuscular Care Advisors, Andy Rose, Carol Wood and Andrea Russell. The Addenbrooke's Neuromuscular Service has been recognised as an MDUK Centre of Excellence.

Facioscapulohumeral muscular dystrophy (FSHD)

Room: Lecture Room 1

11.30am – 12.15pm
Myostatin therapies
Dr Julie Dumonceaux, Principal Research Associate

1.30pm – 2.15pm
Equipment and adaptations
Cathy Szeplaki, Occupational Therapist

2.15pm – 3pm
Physiotherapy for neuromuscular conditions
Andrew Rose, Care Advisor

3.15pm – 4pm
Questions and answers on FSHD
Dr Chris Turner, Consultant Neurologist

4pm – 4.15pm
Scapular fixation research
Phil Cammish, FSHD Registry Curator

4.15pm – 4.30pm
Questions and close

Speakers

Myostatin therapies

Dr Julie Dumonceaux, Principal Research Associate, UCL Institute of Child Health

Julie has been working in the neuromuscular field since 2002, mainly on Duchenne, Becker and facioscapulohumeral muscular dystrophies. She moved to the UK in 2016 and is currently the co-head of the Translational Myology Laboratory based at University College London. The group mainly focuses on understanding the molecular mechanisms leading to muscular dystrophies, and the development of innovative therapeutic approaches. In addition, they investigate general aspects of muscle health such as myostatin mechanisms and how these can be exploited to reverse or slow muscle wasting.

Equipment and adaptations

Cathy Szeplaki, Occupational Therapist, Harrogate and District NHS Foundation Trust

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Questions and answers on FSHD

Dr Chris Turner, Consultant Neurologist, The National Hospital for Neurology and Neurosurgery

Chris graduated from Oxford University and undertook his clinical training in Oxford, Newcastle and London hospitals. He was awarded a Wellcome Clinical Research Training Fellowship and completed a PhD in 2009. In 2007, he became a consultant at the National Hospital for Neurology and Neurosurgery at the MRC Centre for Neuromuscular Diseases. Chris runs general neurology clinics at UCLH and neuromuscular clinics at Queen Square, including a specialist clinic for FSHD. His research interest is in myotonic dystrophy and the genetics of neuromuscular conditions.

Myotonic dystrophy

Room: Abington

11.30am – 12.15pm
Developing drugs to treat myotonic dystrophy
Prof David Brook, Professor of Human Genetics

1.30pm – 2.15pm
Improving quality of life in myotonic dystrophy – the OPTIMISTIC trial
Dr Grainne Gormann, Consultant Neurologist

2.15pm – 3pm
Questions and answers on myotonic dystrophy
Dr Chris Turner, Consultant Neurologist

3.15pm – 4pm
Eating, drinking and swallowing in myotonic dystrophy: what changes and what can help?'
Jodi Allen, Highly Specialist Speech and Language Therapist

4pm – 4.30pm
Questions and close

Speakers

Developing drugs to treat myotonic dystrophy
Professor David Brook, Professor of Human Genetics, University of Nottingham

David completed his PhD at the University of Edinburgh in 1983, before undertaking post-doctoral research in Cardiff, Wales, and Massachusetts, USA. In 1992, he was appointed Senior Lecturer at the University of Nottingham, where he is now Professor of Human Genetics. David's research group is interested in the molecular cause of neuromuscular and heart conditions, in particular myotonic dystrophy. He is currently being funded by MDUK to develop tools to measure the toxic RNA in cells with myotonic dystrophy, which will help to identify promising drugs.

Improving quality of life in myotonic dystrophy – the OPTIMISTIC trial
Dr Grainne Gorman, Consultant Neurologist, Newcastle University

Grainne qualified from the Royal College of Surgeons in Ireland in 1997. After completing her specialist neurology training, she moved to Newcastle to further her interest in neuromuscular conditions. She is now a Consultant Neurologist and contributes to the mitochondrial disease clinical service at the Wellcome Trust Centre for Mitochondrial Research. Grainne's research focuses mainly on evaluating exercise interventions and reducing fatigue in people with neuromuscular conditions. She has led several clinical studies in mitochondrial disease and has been involved in the first international multi-centre myotonic dystrophy trial called OPTIMISTIC.

Questions and answers on myotonic dystrophy
Dr Chris Turner, Consultant Neurologist, The National Hospital for Neurology and Neurosurgery

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Eating, drinking and swallowing in myotonic dystrophy: what changes and what can help?'
Jodi Allen, Highly Specialist Speech and Language Therapist, The National Hospital for Neurology and Neurosurgery

Jodi works at the National Hospital for Neurology and Neurosurgery, where she specialises in the assessment and management of people living with speech and swallowing difficulties, secondary to neuromuscular conditions. She provides specialist input to patients visiting the neuromuscular complex care centre at Queen Square. She has completed a Masters of Clinical Research, looking at assessment of swallowing.

Mitochondrial disease

Room: The Granta

11.30am – 12.15pm
Activity and exercise for mitochondrial disease
Dr Grainne Gormann, Consultant Neurologist

1.30pm – 2.15pm
Clinical presentations and novel approaches for therapy in mitochondrial disease
Prof Rita Horvath, Professor of Neurogenetics

2.15pm – 3pm
Available treatments for mitochondrial disease
Dr Rob Pitceathly, Senior Clinical Research Associate

3.15pm – 4pm
Prevention of mitochondrial disease
Prof Robert McFarland, Clinical Senior Lecturer and Consultant Paediatric Neurologist

4pm – 4.30pm
Questions and close

Speakers

Activity and exercises for mitochondrial disease
Dr Grainne Gorman, Consultant Neurologist, Newcastle University

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Clinical presentations and novel approaches for therapy in mitochondrial disease
Professor Rita Horvath, Professor of Neurogenetics, Newcastle University

Rita completed her neurology training at the Semmelweiss Medical School in Budapest, Hungary, in 1992. After finishing her PhD on mitochondrial encephalopathies, she moved to Munich where she established a new mitochondrial genetic diagnostic service. In 2007, she was appointed Lecturer in the Mitochondrial Research Group at Newcastle University. In 2013, she was made Professor of Neurogenetics. Rita's research focuses mainly on identifying the underlying cause of mitochondrial diseases, which will ultimately help to develop potential treatments.

Available treatments for mitochondrial disease
Dr Robert Pitceathly, Senior Clinical Research Associate, UCL Institute of Neurology

Robert completed his clinical training in Manchester before undertaking a PhD in mitochondrial diseases at the UCL Institute of Neurology. He was appointed as an NIHR academic clinical lecturer and in 2017 was made Senior Research Associate. Robert is actively involved in the NHS England nationally-commissioned mitochondrial clinic, in addition to research studies in mitochondrial diseases. His research combines clinical observations with laboratory approaches to develop understanding of the molecular basis underpinning mitochondrial diseases, with the ultimate aim of developing treatments.

Prevention of mitochondrial disease
Professor Robert McFarland, Clinical Senior Lecturer and Consultant Paediatric Neurologist, Newcastle University

Bobby trained in paediatrics and paediatric neurology in London and Newcastle. He began his research career studying the molecular consequences and clinical problems associated with mutations of mitochondrial DNA. Since then, Bobby's research has included the identification of mutations causing mitochondrial disease and understanding more about the underlying disease process. He has also been involved in several clinical studies, including the development of assessment tools, drug trials and more recently, bringing together over 1,500 people with confirmed mitochondrial disease as part of the MRC Mitochondrial Disease Patient Cohort Study.

Duchenne muscular dystrophy (DMD)

Room: The Grove

11.30am – 12.15pm
Nanodelivery of drugs
Prof Alessandra Ferlini, Professor of Medical Genetics

1.30pm – 2.15pm
New bone and endocrine standards of care in DMD and a focus group on osteoporosis clinical trials
Dr Jarod Wong, Senior Clinical Lecturer and Consultant Paediatric Endocrinology

2.15pm – 3pm
New treatments in paediatric DMD
Dr Deepa Krishnakumar, Clinical Lead for Paediatric Neuromuscular Service

3.15pm – 4pm
Physiotherapy and Duchenne muscular dystrophy
Felicity Vann, Senior Specialist Paediatric Physiotherapist

4pm – 4.30pm
Questions and close

Speakers

Nanodelivery of drugs
Professor Alessandra Ferlini, Professor of Medical Genetics, University of Ferrara, Italy

Alessandra graduated in medicine and surgery at the University of Bologna in 1983. After specialising in neurology, she undertook a PhD in genetics at Imperial College London. She is currently Director of the Medical Genetics Unit at the University of Ferrara and honorary visiting Professor at University College London. She is also actively involved in the European Reference Network for Neuromuscular Diseases, the Treat NMD network and the European NeuroMuscular Centre. Alessandra is Principal Investigator for several innovative trials for muscular dystrophy and is involved in gene discovery research. Her research group also focuses on developing new methods for drug delivery.

New bone and endocrine standards of care in DMD and a focus group on osteoporosis clinical trials
Dr Jarod Wong, Senior Clinical Lecturer and Consultant Paediatric Endocrinology, Royal Hospital for Children Glasgow

Jarod is based at the Royal Hospital for Children Glasgow and his major research interest is the impact of childhood chronic conditions on skeletal development. With funding from the Scottish Government, MDUK and Action Duchenne, Jarod is currently involved in research into bone morbidity including vertebral fractures in boys with Duchenne muscular dystrophy, and using novel methods of high resolution MRI to study osteoporosis.

New treatments in paediatric DMD
Dr Deepa Krishnakumar, Clinical Lead for Paediatric Neuromuscular Service, Addenbrooke's Hospital

Deepa has been working as a Consultant Paediatric Neurologist at Addenbrooke's Hospital since October 2011. She holds two dedicated multi-disciplinary neuromuscular clinics each month. She organises the East of England Neuromuscular study days along with colleagues – for local paediatricians, trainees, physiotherapists and nursing staff. Deepa has been involved with various neurology research studies and most recently taken on as Principal Investigator at Addenbrooke's Hospital for STRIDE NIHR research study on Translarna in Duchenne. She also set up the expanded access programme for Nusinersen IT therapy for children with SMA, at Addenbrooke's Hospital.

Physiotherapy and Duchenne muscular dystrophy
Felicity Vann, Senior Specialist Paediatric Physiotherapist, The Evelina London Children's Hospital

Felicity studied physiotherapy at The University of Birmingham and worked at Birmingham and Bristol Children's Hospital before moving to London to work at The Evelina London Children's Hospital to specialise in paediatric neurology. Felicity enjoys working at The Evelina, 'a fun and innovative hospital that is focused on up-to-date research'.

Friends of
Muscular
Dystrophy UK
Fighting muscle-wasting conditions

Become a Friend of MDUK

Join us in the fight against muscle-wasting conditions, while enjoying a whole range of great benefits for you and your family.

Sign up today!

We make a difference today, helping people to live as independently as possible. We're making a difference for tomorrow, progressing research to find treatments and cures.

We couldn't do this without your support. Please join us.

Get involved.

www.musculardystrophyuk.org/friends

Muscular Dystrophy UK

Fighting muscle-wasting conditions

Muscular Dystrophy UK is the charity bringing individuals, families and professionals together to beat muscle-wasting conditions. We provide a range of services and opportunities to help people live as independently as possible.

We're here for you at the point of diagnosis and at every stage after that, and can:

- **tell you about – and help you get – the services, equipment and support you're entitled to**
- **let you know of progress in research into muscle-wasting conditions**
- **send you accurate and easy-to-understand information about your muscle-wasting condition, ways to manage it and tips to help you day-to-day**
- **put you in touch with other families living with the same muscle-wasting condition, who can tell you about their experiences and provide peer-to-peer advice.**

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www.musculardystrophyuk.org
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