Transforming lives through research
Contents

Executive summary .................. 4
Introduction ......................... 6
What we have achieved .......... 7
  Research support .............. 8
  Clinical trial support .... 10
  Working in partnership .... 12
  Large strategic investments 14
Key themes underpinning our work 17
  Partnerships and innovation 17
  Research excellence ........ 19
Our broad aims for the next three years 20
Our key priorities for the next three years 23
  To harness the power of genetics 24
  To understand disease mechanisms 27
  To facilitate treatment development 29
  To improve quality of life and well-being 34
Conclusion ....................... 38
Every day Muscular Dystrophy UK is working towards a future with effective treatments and ultimately cures for all muscle-wasting conditions and where there are no limits in life for people who are affected. We believe this as passionately now as we did when we were established 60 years ago. It drives everything we do.

It is this vision that unites us all – from the researchers we support to the health professionals we train; from the people we help through our information and advocacy services, to our supporters who inform, guide and campaign with us to make change happen.

It is our vision that has been the driving force behind our research strategy for 2019–2022. Here we present the key objectives and priorities that will guide our decision-making, and explain the research portfolio that critically underpins our aim to help everyone with a neuromuscular condition live their life to the full.

There have been great advances in neuromuscular research, and today there are over 150 clinical trials globally for a number of neuromuscular conditions. Looking to the future we see even more trials in the pipeline, and Muscular Dystrophy UK is committed to supporting researchers and clinicians to carry out the best quality research to underpin them.

There is, of course, still so much more to do. We recognise that our vision may take time to come to fruition, but we are committed to deepening our understanding of neuromuscular conditions, with a view to developing ever more accessible treatments and, in the longer term, working towards cures that will ultimately transform outcomes for people with neuromuscular conditions.

We face many challenges, but we live in exciting and hopeful times.
Executive summary

Muscular Dystrophy UK works to improve the lives of people living with a neuromuscular condition. High-quality research plays a key role in our ambition, helping us to better understand these conditions and maximise treatment improvements.

We have five key aims for the next three years, which are at the heart of our research strategy:

- to deepen our understanding of neuromuscular conditions
- to support more studies into ultra-rare conditions
- to expand patient access to clinical trials
- to encourage new researchers to join the field, and
- to support allied healthcare professionals.

To help us achieve our aims, we will focus on four priority areas:

- to harness the power of genetics
- to understand disease mechanisms
- to facilitate treatment development, and
- to improve quality of life and well-being.

The theme of innovation and partnership underpins all that we do and enables research to be given every chance to succeed. By supporting research in the UK and across the world, we link researchers and research projects, thereby accelerating progress and helping to improve lives as quickly as possible.

We will encourage and support scientists and clinicians with new ideas and technologies to come to this area of research, building on existing scientific studies to develop a new generation of therapies with the ultimate, long-term goal of being able to treat anyone, whatever their neuromuscular condition. We will also seek to provide early career researchers with the support they need to gain independence and become the next generation leading research in this exciting field.

Over the next three years, Muscular Dystrophy UK will continue to invest in high-quality research to understand the underlying causes of disease, develop potential treatments and improve the quality of life of people living with neuromuscular conditions.
We will encourage and support scientists and clinicians with new ideas and technologies to come to this area of research.
Introduction

In the UK today, it is estimated that there are over 70,000 people living with neuromuscular conditions, and, among over 60 known neuromuscular conditions, there are many that are rare or ultra-rare. The 60 or so conditions encompass groups such as: muscular dystrophies, myotonic disorders, congenital myopathies, mitochondrial myopathies, spinal muscular atrophy, myositis, distal myopathies, metabolic myopathies, hereditary neuropathies, inflammatory and autoimmune neuropathies, and disorders of the neuromuscular junction.

At Muscular Dystrophy UK, our vision is of a world with effective treatments and, ultimately, cures for all of these conditions, and with no limits in life for affected individuals and families.

We work tirelessly to improve the life of anyone living with one of these conditions, and high-quality research plays a key role in this aim, helping us better understand these conditions and maximise treatment improvements.

Over the last few years there have been a number of promising new therapies coming to the fore, and we have seen an explosion of clinical trials for neuromuscular conditions. Globally today there are almost 150 active clinical studies for muscular dystrophies, and there has been a surge in the number of genetic therapies entering the clinic.

Some drugs are showing encouraging results, e.g. nusinersen (or Spinraza) for spinal muscular atrophy, and ataluren (or Translarna) for Duchenne muscular dystrophy. And while some treatments can be incredibly expensive, especially some forms of gene therapy, by working with and influencing our relevant partners (such as regulators, NICE, the NHS, and pharmaceutical and biotech companies), we aim to develop innovative solutions to deliver to patients the drugs that they need to treat their condition.

We have listened carefully to individuals affected by neuromuscular conditions, together with their families, researchers and clinicians. And we are committed to making sure as many people as possible with experience of a neuromuscular condition are involved in helping us set our priorities and the approach that we take going forward – whether from the level of our Board of Trustees, or the level at which recommendations are made on the research that we fund.

Great advances have been made in science and there is a pipeline of clinical trials that would have been unthinkable even just 10 years ago. But there’s so much more to do.
What we have achieved

In our last strategy report, published in 2013, we set out our goals to develop potential therapeutic approaches; promote clinical trial readiness; support clinical trials and pilot studies; understand the cause of neuromuscular conditions; build clinical and scientific capacity; improve quality of life; foster sharing of knowledge and networking; and build partnerships. Over the past four years, and thanks to our investments, we have come a long way.

We are now investing in a gene-therapy clinical trial for Duchenne muscular dystrophy; supporting registries for a number of neuromuscular conditions; supporting several clinical trial co-ordinators so that more trials can be carried out; supporting research fellows; supporting projects that seek to improve the quality of life; providing continued support for scientific meetings, such as the UK Annual Translational Research Conference; and joining with other charities to support research projects in ultra-rare conditions.

All that we do and strive for is built around two key themes: partnership and innovation and research excellence.
Muscular Dystrophy UK continues to support research across a broad range of areas, and there have been some exciting developments.

Of particular note are key studies into exon skipping, gene therapies, cell therapies, natural history studies, patient registries, novel magnetic resonance imaging studies, and studies that help us understand the cellular and molecular bases of many neuromuscular conditions.

Four research projects supported by Muscular Dystrophy UK are highlighted opposite.

None of this work could have been achieved without the support of people with neuromuscular conditions and their families, many of whom have played key roles in several of the research projects through participation in studies. Others have helped shape our research portfolio through their participation in our Lay Research Panel – an integral part of our grant-making process.

Funding research that matters to patients

Our allocation of grants goes through a rigorous peer-review process, a key part of which is listening to the voices of people with experience of neuromuscular conditions. For this, we rely on the work of two panels: a Lay Research Panel, and our Medical Research Committee (MRC).

Our Lay Research Panel comprises people directly or indirectly affected by a neuromuscular condition, a physiotherapist, an occupational therapist and a scientific advisor. They review each application for its importance and potential for benefit to patients in the long term.

The MRC comprises a committee of scientific experts, who are able to focus on the scientific quality of the application and the potential for the research to better our understanding of neuromuscular conditions or for development of a new treatment.

Applications from researchers are sent out to experts across the world who provide a written assessment of the project. Applicants are given the opportunity to comment on these assessments prior to further review by two panels at separate face-to-face meetings.

In this way, Muscular Dystrophy UK, together with our supporters and our Trustees, can have confidence that the research that we support is of the highest quality and relevant to patients living with neuromuscular conditions.

Muscular Dystrophy UK’s Lay Research Panel brings an important patient perspective.
UNITE-DMD
Professor George Dickson,
Royal Holloway

For over 20 years, Muscular Dystrophy UK has supported Professor George Dickson in work to design, test and refine a gene therapy for Duchenne muscular dystrophy, which is now ready to be tested in boys with this condition for the first time. Professor Dickson and a team of researchers from the UK and France are in the process of setting up a study that includes a clinical trial, called ‘UNITE-DMD’, and hope to start recruiting patients in 2019.

As principal funder of the UK part of UNITE-DMD, we played a key role in making this possible. We very much look forward to seeing how the trial progresses.

Cell-based screening for myotonic dystrophy
Professor David Brook, Nottingham University

In 2008, Muscular Dystrophy UK supported Professor David Brook in his development of a cell-based screening method for myotonic dystrophy. With funding from us and from the Medical Research Council, Professor Brook conducted the screen and identified a promising type of drug, enabling him to secure a £3.1m award from the Wellcome Trust. This will allow him to build on this work and develop more selective versions of the drug that could potentially be put forward for clinical trial.

This is a great example of how a small grant can help scientists to apply for much larger grants to further progress their research. We are proud to have played a small part in Professor Brook’s research and very much hope it will be successful in developing new drugs for myotonic dystrophy.

ACTMus
Dr Chris Graham, former PhD student at King’s College London

During his PhD at King’s College London, Dr Chris Graham became interested in how adults with neuromuscular conditions perceive their quality of life. He piloted a psychological therapy called Acceptance and Commitment Therapy (ACT) to see whether this could improve quality of life. The study was small, but its results were encouraging and provided the basis for further funding from the National Institute of Health Research (NIHR) to support a larger trial: Acceptance and Commitment Therapy for Muscle Disease (ACTMus).

Upon completion of his PhD in 2013, Dr Graham trained as a clinical psychologist and went on to become an Academic Fellow in Behavioural Medicine at the University of Leeds. He is still very much involved with the neuromuscular field through the ACTMus trial.

Dr Graham was able to gain his valuable knowledge and experience thanks to the support of Muscular Dystrophy UK. To support neuromuscular research in the long term, we need to train and inspire the next generation of scientists and clinical researchers.

Exome sequencing
Dr Lizzie Harris, former clinical fellow at Newcastle University

During her clinical fellowship, funded by Muscular Dystrophy UK, Dr Lizzie Harris used a technique called exome sequencing to identify mutations in 25 families with previously genetically undiagnosed neuromuscular conditions. She also discovered a new gene associated with a congenital myopathy, and published 14 papers in academic journals.

Muscular Dystrophy UK is delighted to have supported Dr Harris in her research, which has not only given families more accurate information about their condition, but has also increased our knowledge of the genetic causes of neuromuscular conditions, and demonstrated that exome sequencing is quicker and more accurate than current genetic testing methods.
Clinical trial support

Muscular Dystrophy UK actively supports clinical studies for neuromuscular conditions through the provision of clinical trial co-ordinators and our work establishing databases and registries.

Clinical trial co-ordinators oversee the effective and safe management of trials in a number of centres across the UK. Over the last year, these posts have helped to set up and run 45 different studies, two thirds of which have been testing interventions or treatments. We know that the demand will increase given the pipeline of clinical trials, and we know that centres can find it difficult to support all aspects of clinical trials, owing to a lack of capacity. Therefore, Muscular Dystrophy UK is working to develop a network of researchers across the UK (including clinicians, physiotherapists and others), who will have the skills and capacity to carry out clinical trials across multiple neuromuscular conditions in children and adults. And we are now working with the National Institute for Health Research in England and other key stakeholders to provide sustainable support for these vital roles.

We also support several patient registries (often in partnership with other charities and funders), natural history studies and databases. These vital infrastructures allow researchers to follow the progression of a condition and provide access to patients that can be recruited to clinical trials.

An example of two important registries supported by Muscular Dystrophy UK is highlighted opposite.

Many of our research projects could not happen without the involvement of patients, their families and carers.
Registries are a vital tool to help advance the research and development of treatments, therapies and care for all those diagnosed with neuromuscular conditions. In addition to being a resource for researchers to plan, recruit for and conduct research, they also work to facilitate research to improve standards of care and best practice. Studies from registries help to understand less well characterised symptoms of conditions.

The UK FSHD Registry

Facioscapulohumeral muscular dystrophy (FSHD) is a rare condition. In the UK it is estimated that between 2,000 and 2,500 people have FSHD, so it can take years to identify enough eligible people to take part in clinical trials and research studies. The UK FSHD Registry collects clinical, genetic and demographic data about people with FSHD across the UK and currently holds data on over 900 people. The registry has been running since 2013 and is funded by Muscular Dystrophy UK, with support from the John Walton Muscular Dystrophy Research Centre, TREAT-NMD Alliance and the MRC Centre for Neuromuscular Diseases. To date we have committed over £130k to the registry.

For example, in a study conducted at Newcastle University, registry participants were invited to fill out questionnaires. Of the 398 people who completed the questionnaire, 88.6% reported pain at the time of the study, with 51% of people reporting chronic pain. The study showed that pain in FSHD type 1 (FSHD1) is frequent and strongly impacts quality of life. It highlights the importance of pain management and will help to improve the care of FSHD patients.

The UK Myotonic Dystrophy Patient Registry

Myotonic dystrophy type 1 (DM1) is a rare condition that is estimated to affect 1 in 8,000 people. It can therefore take years to identify enough eligible people to take part in clinical trials and research studies. The UK Myotonic Dystrophy Patient Registry collects clinical and genetic data from people with DM1 in the UK and currently holds data on over 700 people. The registry has been running since 2013 and is funded by Muscular Dystrophy UK and the Myotonic Dystrophy Support Group, with support from the John Walton Muscular Dystrophy Research Centre, TREAT-NMD Alliance and the MRC Centre for Neuromuscular Diseases. To date we have committed £35k to the registry.

For example, in a study conducted at Newcastle University, registry participants were invited to fill out questionnaires. Of the 398 people who completed the questionnaire, 88.6% reported pain at the time of the study, with 51% of people reporting chronic pain. The study showed that pain in FSHD type 1 (FSHD1) is frequent and strongly impacts quality of life. It highlights the importance of pain management and will help to improve the care of FSHD patients.

Registries can be used in research both nationally and internationally and have been utilised to recruit for trials and studies.

Examples include: OPTIMISTIC, an international clinical trial that investigated the impact of Cognitive Behavioural Therapy (CBT) and increased activity on fatigue and quality of life of people with myotonic dystrophy type 1; and AMO Pharma’s AMO-001-02 phase II study of Tideglusib for people with juvenile-onset or congenital myotonic dystrophy type 1.
Working in partnership

We could never achieve all that we do by working alone. Over the years we have developed some extraordinary partnerships with other funders. We have worked with major UK funders such as the Medical Research Council and the Chief Scientist Office in Scotland to support researchers. We have partnered with international funders, such as Cure CMD, to support studies in ultra-rare conditions.

In addition, we partner with UK charities to support patient registries and databases, which allow medical specialists and researchers to learn more about a particular condition, track symptoms over time and, ultimately, improve care. They also help scientists speed progress towards a treatment, by making it easier to find participants for drug trials.

Partnership with the Medical Research Council

To support medical graduates in clinical training, we partnered with the Medical Research Council in 2014 for a joint clinical research training fellowship funding scheme. The scheme is fully administered by the Medical Research Council and candidates are funded on the basis of a highly competitive selection process. Clinical research and training fellowships are an important way of encouraging clinicians into neuromuscular research, which is an essential component of future translational research. Funding the clinical fellows jointly spreads the cost, and grantees enjoy benefits such as opportunities to engage with families and supporters through Muscular Dystrophy UK as part of their prestigious Medical Research Committee award. Through this scheme we are co-funding one clinical fellow in Edinburgh, Dr Clare Wood, who is studying how bone development is affected in Duchenne muscular dystrophy.

Partnership with Cure CMD

In 2017, we teamed up with US charity, Cure CMD, to co-fund research into LMNA congenital muscular dystrophy (LMNA-CMD). This condition causes progressive muscle weakness of the skeletal, respiratory and cardiac muscles and is estimated to affect around 50 people in the UK. As it is such a rare condition, there is little research into it and a high unmet need. For this reason, we decided to partner with Cure CMD to pool our resources and research networks. LMNA-CMD researchers from around the world were invited to apply for funding and the applications were evaluated and judged against others to our grant round. As a result, a £150k award was made to Dr Gisèle Bonne at INSERM, Paris, to investigate the role of genetic modifiers in modulating the severity of LMNA-CMD.
Muscular Dystrophy UK and ENMC

Muscular Dystrophy UK is a member of the executive of the European Neuromuscular Centre (ENMC), which was founded in 1992 by a group of European patient associations. The mission of ENMC is to encourage and facilitate communication and collaboration in the field of neuromuscular research, with the aim of improving diagnosis and prognosis, finding effective treatments, and optimising standards of care to improve the quality of life of people affected by neuromuscular disorders. The organisation achieves this by financing and organising workshops.

Our work with ENMC allows us to work closely with other European charities focusing on neuromuscular conditions, to share ideas, and to increase our understanding of the challenges faced by neuromuscular researchers in other countries. Our membership of ENMC also allows us to support clinicians and researchers to work together to gain a common understanding of the state of the art and, where possible, to agree actions on how to take aspects of the field forward and work collaboratively to obtain grant funding.

For example, at a workshop on GNE myopathy in September 2018, participants reviewed the current medical and scientific knowledge relevant to the condition to achieve a better understanding of its epidemiology, phenotype and genetics. GNE myopathy is a rare neuromuscular condition caused by mutations in the GNE gene, which codes for an enzyme that has a key role in the production of sialic acid in the body. A deficiency of the enzyme in muscle cells almost always leads to increasing disability from young adulthood. The 26 participants formed a multidisciplinary group from ten countries, comprising basic and clinical researchers and four patient representatives. At the workshop they agreed on a standard of care for GNE myopathy patients, discussed strengths and weaknesses of the currently available animal models, and the biochemical consequences of the GNE defect on muscle tissue leading to muscle damage and weakness.

This was an excellent opportunity for world leaders in the field to gather to discuss and reach consensus on several important areas for the condition.
Large strategic investments

We have continued to invest in areas where we are looking to see change in the foreseeable future. Three large, strategic investments we have made have been in:

- gene therapy (UNITE-DMD)
- enabling translational research (at the MDUK Oxford Neuromuscular Centre), and
- clinical trial readiness (through the NorthStar database).

**UNITE-DMD**

Muscular Dystrophy UK is a long-standing supporter of research into gene therapy. This technology has advanced significantly over the years and we are delighted to be taking a bold step: to test its safety in people living with neuromuscular conditions in the UK.

UNITE-DMD is an international collaboration with colleagues in France working on viral production for gene therapy. Once pre-clinical studies are complete, the collaboration will assess the safety of a gene therapy for Duchenne muscular dystrophy in a phase I/II clinical trial. Although this particular gene therapy will be designed to treat Duchenne muscular dystrophy, its development will refine and improve the gene-therapy technique generally, which will help in the development of gene therapies for other neuromuscular conditions in the future.

UNITE-DMD is a four-year project taking place in the UK and France. The UK investigators are Professor George Dickson and Dr Linda Popplewell at Royal Holloway, University of London; Professor Francesco Muntoni (chief investigator for the clinical trial element of the study) at the University College London Great Ormond Street Institute of Child Health; and Professor Volker Straub at the John Walton Muscular Dystrophy Research Centre, Newcastle University.

We are delighted to be the lead funder of the UK side of the project and have committed £1.6m, supported by Action Duchenne. The French Muscular Dystrophy Association (AFM-Téléthon) is funding the French arm of the project.
NorthStar Clinical Network

Muscular Dystrophy UK has been supporting the NorthStar Clinical Network for over a decade and has invested £1.7m in the programme. It is a programme that is collecting natural history data of boys with Duchenne muscular dystrophy, aiming to help clinicians and researchers understand more about the condition, and care and treatments. Collecting data from patients across 23 centres in the UK, it has had measurable impact for boys with the condition, and it is expanding to include men. NorthStar data showed the benefits of steroid treatment, an intervention that is now used widely across the UK, and the database is currently being used as a way to collect post-marketing data on the Duchenne drug, ataluren (Translarna). A rating scale that is used to measure functional motor abilities in ambulant children with Duchenne muscular dystrophy, called the NorthStar Ambulatory Assessment, has been developed by the network. It is used to monitor the condition progression and treatment effects and is used as a primary or secondary outcome measure in the majority of clinical trials.

MDUK Oxford Neuromuscular Centre

The number of clinical trials being undertaken is increasing, but across the UK centres are struggling to meet demand owing to inadequate capacity – such as a lack of infrastructure and expertise. This risks a reduction in opportunities for patients to be enrolled in clinical trials. We are addressing the need for additional capacity so that the UK can remain one of the key countries for clinical trials for neuromuscular conditions.

We are working with the University of Oxford to establish a world-leading centre aimed at bringing new treatments to patients more quickly. Working alongside the major centres in Newcastle and London (at University College London Great Ormond Street Institute of Child Health), this third centre will be a game changer for individuals and families living with neuromuscular conditions in the UK. With an investment of £1m from Muscular Dystrophy UK, and a significant financial commitment from the university, new resources including start-up seed funding for research costs for clinical lecturers, as well as a number of new staff posts, the MDUK Oxford University Neuromuscular Centre will carry out translational research and boost capacity for clinical trials.
Key themes underpinning our work

All that we do and all that we strive to achieve at Muscular Dystrophy UK is built around two key themes: partnership and innovation, and research excellence.

Partnerships and innovation

We cannot achieve our aims by working alone. To pursue our huge ambitions beyond our relatively limited resources, we join forces with partners, including other funders and charities, the NHS, the National Institute for Health Research (NIHR), devolved nations and regulators. In the coming years, we intend to strengthen these partnerships and forge new ones.

Key partnerships

We already work in partnerships in a variety of ways to support research into neuromuscular conditions.

- Many of our research projects could not happen without the involvement of patients, their families and carers. These individuals are our most valued partners.
- We co-fund research projects through bespoke calls, e.g. with Cure CMD, Duchenne muscular dystrophy charities, the Chief Scientist Office in Scotland and the Medical Research Council.
- Partnering with the French charity AFM Téléthon, Genethon and supported by Action Duchenne, we are funding the UNITE-DMD clinical trial.
- We are on the executive committee of the European Neuromuscular Centre (ENMC), giving us a close working relationship with a group of European patient associations to support researchers and clinicians to discuss focused challenges in neuromuscular conditions.
- We are a key partner in an EU Horizon 2020 project called PREFER, which looks at how and when it is best to perform and include patient-preference in decision-making during the drug life cycle. As patient stakeholders, we are involved in a work package within the project that is testing different methods for preference elicitation in clinical case studies, in particular in neuromuscular disorders. The end result will be recommendations to support the development of guidelines for industry, regulatory authorities and health technology assessment bodies.

Over the next three years we will seek specific partnerships with other charity funders in the UK and overseas, particularly in areas where there is a need to increase research capacity (e.g. in ultra-rare conditions), or in areas that intersect with other areas of expertise (e.g. in research of comorbidities or engineering).
**Key innovative developments**

We recognise that we need to develop new ways of funding research areas that do not fit traditional models. In particular, we want to support the development of areas, particularly in the field of quality-of-life research, that might be hard to fund without an initial proof of concept. In addition, we wish to ensure that we support early career researchers to help them develop their independence and become the leaders of tomorrow. To begin with, over the next year, we will develop two funding streams to support research in new ways:

- **The first funding stream will provide seed funding for research projects covering areas that are currently under-served or lacking from our research grant portfolio. Examples include research related to quality of life (e.g., in the area of technology development, either in the digital or engineering space), where the aim is to develop an intervention or tool that will benefit people with neuromuscular conditions.**

- **The second stream will provide a mechanism for us to provide funding to researchers who are in the process of spinning out a company, start-ups or small biotech companies seeking investment for their technology or compound that is in early stages of development. We are exploring opportunities to engage with stakeholders (researchers, partner funders, investors, etc.) in this area.**

**Technology and data**

Huge advances in technology are not only enabling people with even the most complex disabilities to manage their environment, participate in leisure and employment, and stay connected with their communities, but also helping to collect information and build invaluable data resources to aid ongoing research and improve treatment options.

Particular advances in wearable devices and apps have enabled the recording of activity, behaviour and movement patterns. Remote monitoring, by researchers and clinicians, has the potential to bring new insight into neuromuscular disorders. And the monitoring of movement/ambulation at home and in real-world settings can facilitate early detection of changes and be used in clinical trials.

Data collected from wearable devices and apps can also be linked to other data sets to enable research on a scale not previously possible in patients with neuromuscular conditions; informatics tools can provide a route to develop and validate objective outcome measures (e.g. wearable devices for patient-reported outcome measures).

Artificial Intelligence is being used to facilitate rapid screening for the identification of new drugs, and for the mining of data to seek drugs that could be repurposed to treat neuromuscular conditions.
Research excellence

It is often noted that the capacity of the UK neuromuscular research workforce is sub-optimal. The ambitions identified within this research strategy rely on the strengths of researchers and clinicians from the neuromuscular community, which is relatively small in the UK.

Muscular Dystrophy UK is working to expand research excellence in the field of neuromuscular conditions, whether through encouraging early career researchers already within the field, or by encouraging researchers from other disciplines to move into the field. These other disciplines may include (but are not limited to): cell biology, genetics, muscle biology, cardiology, psychology and physiotherapy.

In contact with learned societies and professional bodies, we will work to highlight the opportunities in the field. And as part of our commitment to encouraging new researchers into the field, we will be reviewing our grant schemes for early career researchers in 2019.
Our broad aims for the next three years

In the case of many neuromuscular conditions, it must be acknowledged that only a few scientists worldwide undertake research into the role that mutated genes play in causing the muscles not to function properly. Therefore, one of our aims at Muscular Dystrophy UK is to facilitate communication and collaboration in the global scientific community to ensure researchers share their knowledge and expertise.

Our commitment to find treatments, and ultimately cures, through research remains as strong as ever. Over the next three years, we aim to maximise our impact and accelerate progress towards finding treatments for neuromuscular conditions. Through research that we support, we strive to understand more about the mechanisms of neuromuscular conditions, support the development of new treatments, and enhance existing treatment paradigms to help people living with neuromuscular conditions. We will do this by supporting proof-of-principle studies and project grants, while also working to facilitate clinical trials for children and adults with neuromuscular conditions.
To encourage new researchers to join the field

It is important that we help expand the existing pool of dedicated and talented researchers working in the field of muscular dystrophy. We already support fantastic academics (scientists, medics and physiotherapists) across the UK, but we want to ensure that there are sufficient neuromuscular researchers for the years to come. Therefore, we will look to support researchers early on in their careers who may be looking for exciting opportunities, and encourage experienced researchers from other fields to collaborate with experienced neuromuscular scientists, bringing new experience to the field.

To support allied healthcare professionals

Allied healthcare professionals (including, but not limited to, physiotherapists, orthotists, occupational therapists, and speech and language therapists) and specialist nursing staff are key to driving high-quality research into quality of life for people with neuromuscular conditions.

Allied healthcare professionals can apply for Muscular Dystrophy UK’s grants. In addition, through the refresh of our grant schemes, we will provide further opportunities for individuals and teams who wish to carry out quality-of-life research.

To deepen our understanding of neuromuscular conditions

To ensure that there is a pipeline of potential new treatments for trials, we will continue to support excellent scientists in carrying out first-class research – from understanding the fundamental pathological underpinnings of a condition, right through to looking for drug targets.

To support more studies into ultra-rare conditions

We want to support more studies in ultra-rare neuromuscular conditions. In order to do this we will work with international funders and charities to ensure that we bring research skills from across the globe. International collaboration will also provide researchers with access to larger numbers of patients, thereby speeding up the journey to develop new treatments.

To expand patient access to clinical trials

More patients across the UK should be able to take part in clinical trials for neuromuscular conditions. We are aware that only a limited number of sites in the UK are able to carry out clinical trials for neuromuscular conditions, and in some of these sites there is a lack of capacity – in terms of trained staff – to help support these trials. In the first half of 2019, we will be reviewing ways in which capacity could be strengthened, and over the next three years we will work to ensure that the centres across the UK are able to support trials.
Our vision is of a world with effective treatments and cures for *all* neuromuscular conditions.
Our key priorities for the next three years

To help us plan how we can best support people living with neuromuscular conditions, we held a number of workshops with patients and families, alongside national and international experts. These workshops helped us identify four key priority areas that now form the basis of our research and innovation strategy for the next three years:

- To harness the power of genetics
- To understand disease mechanisms
- To facilitate treatment development
- To improve quality of life

Partnerships and innovation

Research excellence

All these priorities stem from our vision of a world with effective treatments and cures for all neuromuscular conditions, and with no limits in life for affected individuals and families. Many apply to several different neuromuscular conditions, while some are more specific to a smaller number of rarer conditions. All are important, given their significant impact on any individual life affected.
1. To harness the power of genetics

These days, reliable diagnostic tests for neuromuscular conditions are available, and thanks to increased investment and improvements in technology, many more mutations are being identified. However, because of the sporadic nature of neuromuscular conditions, there remain significant challenges around accurate and timely diagnosis. For some people, the underlying cause remains unidentified.

Accurate diagnosis is a priority. We are engaged at all levels in discussions around the inclusion of genetic and genomic testing for some neuromuscular conditions. New education systems are needed for GPs, general neurologists and trainees, as well as improved pathways of communication between multidisciplinary teams of medical professionals in the community and in hospitals. Resources and platforms, such as RD-Connect® – a resource linking different data types such as genomics, clinical information, patient registries and biobanks from across the globe – provide an excellent repository for researchers to share their data.

A person’s genome is their complete set of DNA, including all of their genes. Our genes comprise only a small part of our genome, so by knowing the sequence of all of our DNA, doctors and scientists can identify changes anywhere that might cause a disease. The Department of Health and Social Care set up Genomics England in 2013, with the aim of delivering the 100,000 Genomes Project, an ambitious project to sequence the whole genome of thousands of people. Building on the success of the 100,000 Genomes Project, the NHS Genomic Medicine Service will transform the genetics diagnosis for patients with rare diseases (including neuromuscular conditions) in the coming years.

In all these ways, we are striving to enable more rapid and more accurate diagnosis, and more effective clinical management, for all patients with neuromuscular conditions.

Muscular Dystrophy UK has worked extensively to raise awareness of the needs and challenges experienced by patients with neuromuscular conditions among healthcare professionals such as GPs and physiotherapists, so that people are referred to centres of excellence more quickly. We will continue to do this and provide access to our GP and physiotherapist online training modules. In addition, we will develop further training modules for other allied healthcare professionals.

### Biobanking

A biobank is a collection of biological and tissue samples that can be used for research purposes. Several biobanks hold tissue from patients with neuromuscular conditions, but there can be challenges in discovering information about the samples held and accessing the materials.

Working in partnership with organisations and researchers, Muscular Dystrophy UK supports investigation into ways in which the storage of samples can be promoted; making sure samples are appropriately banked and made accessible for research purposes benefits the whole research community.
Accurate diagnosis

Thanks to the increased knowledge gained from accurate genetic diagnosis, researchers are developing bespoke interventions to target specific genetic mutations that cause neuromuscular conditions.

Muscular Dystrophy UK is supporting Professor Volker Straub and his team at Newcastle University, who are using a faster and more accurate genetic diagnostic technique, called next generation sequencing, to identify new genetic changes that lead to limb girdle muscular dystrophy (LGMD). This will provide a genetic diagnosis to a number of people with genetically undiagnosed LGMD, as well as improving the understanding of the condition and helping to inform the development of new treatments.

We are funding Professor Carsten Bönnemann and his team at the National Institute of Neurological Disorders and Stroke in Maryland, NIH USA, to develop a new mouse model of Ullrich congenital muscular dystrophy (UCMD), which they will use to test a molecular patch that could be a potential treatment for people with the condition (see ‘Antisense oligonucleotides and exon skipping’ on page 33). UCMD is caused by mutations in genes that produce a protein called collagen VI. Professor Bönnemann and his team recently identified a new mutation in the COL6A1 gene, which is thought to be one of the most common causes of UCMD. This particular mutation creates an extra, unwanted exon in the COL6A1 gene and this unwanted exon could potentially be excluded using a therapeutic approach called exon skipping. Designing the specific molecular patch will be carried out in collaboration with Professor Francesco Muntoni and his team at University College London Great Ormond Street Institute of Child Health who are currently developing molecular patches targeting several UCMD mutations. None of this work would have been possible without an accurate genetic diagnosis of the cause of the condition.

We are striving to enable more rapid and more accurate diagnosis, and more effective clinical management, for all patients with neuromuscular conditions.
**Variants of unknown significance and deep phenotyping**

Different individuals with the same condition (even within the same family) can be affected differently or present with a different severity of symptoms.

Technological advances in genomics enable us to observe many small changes and variations in genes, but the significance of these in relation to the symptoms observed is often poorly understood. While many of these small genetic changes may not be the direct cause of a neuromuscular condition, they may be highly relevant in terms of regulating or modifying the function of the disease-causing gene, so we need to understand more about them.

With a detailed clinical picture of an individual's symptoms that are linked to their genetic information, we can better understand the impact of genetic changes. It is therefore vitally important to gather details about disease manifestations (phenotypes) in an individual and fine-grained way, known as deep phenotyping. By linking data (such as detailed clinical and genetic information) from as many patients as possible, researchers will be able to reach a greater understanding of the significance of these small changes, even in people who have the same mutation in the disease-causing gene. This, in turn, could have a significant impact on the success of clinical trials.

Having access to large databases of genetic and biological information allows researchers to investigate these phenomena across conditions. This kind of research creates opportunities for better understanding biological mechanisms of diseases and for finding treatments that could be effective in more than one condition. For example, drugs that are used to treat similar symptoms in other conditions could be repurposed for neuromuscular conditions.

- **Muscular Dystrophy UK promotes the collection of genetic and phenotypic information in recognised platforms such as Phenotips® and RD-Connect. We therefore expect that grantholders make available the results of research that we have funded in platforms and/or formats that can be discovered, shared and available for further analysis.**
2. To understand disease mechanisms

Having knowledge of the genetic mutations that cause neuromuscular conditions tells us only part of the story. To better understand how the muscles (and indeed the whole body) are affected by a mutation, scientists need to study the underlying biology. They can do this using animal models and/or cells taken from patients.

Model systems

While there are animal models for many neuromuscular conditions, these are often not representative of how conditions present or progress in humans. The symptoms of several neuromuscular conditions can take decades to develop fully in humans; animals typically used as model systems, such as mice and zebrafish, have considerably shorter lifespans and are unlikely to live long enough to exhibit many of the later symptoms.

Nevertheless, animal models can tell us a lot about aspects of biological mechanisms, and are used not only to test potential drug candidates, but also to help predict the safety and efficacy of drugs that are planned for human clinical trials. Animals therefore play a valuable role in drug development, but further research is required for the development of additional and alternative model systems.

- The availability of gene-editing tools provides the opportunity to generate mutation-specific cellular and animal models that can be used to assess biological mechanisms. Muscular Dystrophy UK has supported research to develop in vitro mini-muscles, with the ultimate aim of using these in drug development. We welcome further applications for research into these and other model systems such as ‘muscle on a chip’ that can be used as fast-throughput assays to screen drug candidates.

Muscular Dystrophy UK funds research using established animal models and will continue to do so. Additionally, we will fund research into the development or utilisation of alternative model systems, e.g. inducible pluripotent stem cells, cell lines. However, we will only support the creation and phenotyping of new animal models where no alternative is available. This is a costly endeavour and beyond the scope of our response mode funding. We will mandate (as far as is possible) that new model systems that are generated through Muscular Dystrophy UK-funded research are made freely available to the scientific community.
Common pathways

There is clinical, genetic and biochemical evidence that similar molecular pathways are relevant in different neuromuscular conditions. Researching common pathways within and between conditions could therefore provide insight into underlying mechanisms, and lead to new treatment approaches. This kind of research is exploratory and often considered highly risky. But ambitious, innovative and collaborative research is needed to better understand the fundamental links between different conditions. Finding common pathways would be attractive to industry because companies may already have treatment development pipelines in related areas, and would ultimately benefit patients.

- Through its annual grant rounds, Muscular Dystrophy UK will support research that seeks to gain understanding of pathways common across conditions. We will, where appropriate, work in partnership with other funders to support this.

Molecular pathways

In addition to what we know of changes to the molecular pathways and machinery in muscle cells, in order to design effective therapies, we also need to understand what happens to other body systems when specific genetic mutations occur. For some conditions, we have a comprehensive knowledge base; in others, more laboratory research is required to understand the molecular pathways that are affected. This is particularly true of conditions with multi-systemic effects. We know that respiratory, cardiac, pain, as well as psychological and cognitive functions (e.g. anxiety, depression, sleep disturbance, learning disability, emotional regulation) can all be affected in neuromuscular conditions, and symptoms in the brain, heart, respiratory system or the gut can be as challenging for patients, and their families, as the effect on the muscles.

- Muscular Dystrophy UK has provided support for basic research to understand the molecular pathways that contribute to the symptoms associated with neuromuscular conditions, with the aim of providing targets for future drug development. We will continue to do this through our annual grant rounds.

In addition, we will seek opportunities to work in partnership with other funders, such as other condition-specific charities working in this area, to better understand other systems affected by neuromuscular conditions, such as respiratory, cardiac and cognitive systems.
3. To facilitate treatment development

Over the last few years we have seen a growth in the number of clinical trials for several neuromuscular conditions, such as Duchenne muscular dystrophy and spinal muscular atrophy. There are other trials in the pipeline for several other conditions, such as limb girdle muscular dystrophy, Charcot-Marie Tooth disease and myotubular myopathy. Thanks to its excellent clinical research base and the NHS, the UK is a natural location for the development and trial of new treatments. It is therefore vital that the foundations are in place to understand how conditions progress over time, to allow the selection and recruitment of patients for trials, to measure disease progression and develop new treatments. Muscular Dystrophy UK has a long history of supporting treatment development and will continue to do so.

Natural history studies

Natural history studies – studies that follow a group of people with a certain condition over time – provide crucial information on how that condition progresses. In the case of rare conditions where the causes are poorly understood, these studies can provide useful data for researchers. They play a valuable role in evaluating whether a treatment is effective, by comparing someone having received treatment with a matched control who has not received that treatment.

For slow-progressing conditions, where data may need to be collected over several years, supporting this type of study presents some challenges, requiring sustained commitment both financially and on the part of the researchers and the patients.

- Muscular Dystrophy UK has already provided significant funding for the collection of natural history data for several conditions, and we will continue to do this. We will also explore the feasibility of expanding these neuromuscular databases to cover additional conditions by working in partnership with other funders, clinicians, researchers and, where appropriate, companies interested in carrying out clinical trials. We will seek to develop a business model that could help support the database running costs in the longer term.

Where we support new studies, we will look for a harmonised approach across the field to ensure that relevant and sensitive measures are used. To collect relevant and useful data, it is important that there is international collaboration and communication, such as is promoted through the neuromuscular network, TREAT-NMD. Studies should be multi-centre and of sufficient length (minimum two to three years) and size. They also need to have multiple outcome measures (and where possible include multi-system measures – cardiac, gut, etc.) to be of value for the participants, researchers and the funders.
Registries

Patient registries collect information, such as clinical and genetic details, so that eligible patients can be found and easily contacted by researchers conducting clinical trials. Registries are not only an important resource to facilitate research, but they act as a communication tool with the patient community.

Muscular Dystrophy UK funds the maintenance costs of several registries, which have been useful for research and for identifying patients who might benefit from participation in clinical trials. These national registries are maintained by TREAT-NMD, which ensures the core data set can be integrated into international registries. International registries are useful when planning clinical trials, in helping to identify sites where trials can be undertaken. It should be noted that many patient organisations have established condition-specific registries, but often these do not contain an agreed set of data points and their accuracy may be variable.

- Muscular Dystrophy UK monitors the value of the registries that we support. We will work in partnership with other funders and charities where we see the need to develop a new registry or modify our support for existing registries.

Biomarkers and outcome measures

When translating potentially therapeutic interventions from preclinical to clinical studies (i.e. from in vitro studies to animal models and then to humans), we need reliable biomarkers to identify whether an intervention is effective, and outcome measures to better understand the progression of a condition, and treatment efficacy.

A biomarker is an objective, reliable and reproducibly measurable indicator of a physiological or disease state. Biomarkers may not always correlate with a patient’s experience because they are measuring quantifiable biological characteristics, e.g. the expression level of a protein in a muscle cell. Even if a mutation in the gene for the protein is the causative factor for a neuromuscular condition, changes in the level of the protein (as the result of an intervention) may not equate to clinically measurable change in the patient. In other words, biological changes may not result in functional changes for the patient. Therefore, it is important that we have separate measures that can indicate biological and clinical or functional outcomes.

An outcome measure is the result of a test that is used to objectively determine the baseline function of a patient at the beginning of treatment, and against which progress and treatment efficacy can then be measured. The change in outcome measures from baseline to the end of treatment is then defined as the treatment effect.

There is a need for the identification and development of sensitive outcome measures and biomarkers to help measure response to treatments. Physical outcome measures need to be validated and standardised to assess specific conditions and the result of specific interventions. And measures to assess the psychosocial impact of neuromuscular disorders in individuals and their families where appropriate should also be considered.
Drug targets, delivery and time of intervention

Drug targets are typically proteins or nucleic acids within cells whose function can be changed or blocked by a drug (usually a small molecule) or other nucleic acid constructs. Such targets can be discovered by undertaking more laboratory-based research to increase our understanding of what causes a condition. Similarly, working in partnership with the pharmaceutical industry and other funders, researchers may be able to carry out high-throughput screens of libraries of compounds (simply a series of lots of chemicals that might have drug-like properties) against biological targets likely to be the cause of the condition.

Alongside knowing drug targets, safe and effective drug delivery is also crucial for ensuring a treatment gets to the site of action. And it is important to understand the optimal time for delivery of an intervention (e.g. daily morning or night, single dose, etc.). Further studies are required to understand this better for many conditions.

Muscular Dystrophy UK will continue to support research to evaluate biomarkers and outcome measures for neuromuscular conditions through our grant funding. We will also work with researchers, clinicians, patients, regulators and NHS partners to ensure that meaningful and functional outcome measures are part of clinical trials.
Muscular Dystrophy UK has a long history of supporting treatment development and will continue to do so.
Drug repurposing

Drug repurposing involves applying the knowledge gained from the development and use of existing drugs into the use of those drugs to treat different diseases. This has a number of advantages. For example, toxicity, safety issues and side effects are largely known for an existing drug, and the formulation for the particular method of delivery is also already known (although changes may be required if the previous target was not a neuromuscular condition).

Clinical trials can be significantly cheaper and speedier than they might be if one were starting with a new drug. It typically costs over £1bn and over a decade to get a new drug to market, whereas it can cost as little as £500k and take less than five years for a repurposed drug.

Repurposing is a promising avenue for some aspects of treatment, but, of course, it is not without risks. The drug in question may not have been given to people with neuromuscular conditions before, it may not be effective, or it could be harmful and the trial fails. Currently there may be few incentives for pharmaceutical companies to take repurposing work forwards if a drug is out of patent or generic.

Companies and the NHS will need to work together to create innovative incentives to get repurposed drugs to patients, particularly those with rare conditions.

- Working in partnership with other charities where appropriate, Muscular Dystrophy UK will support research into repurposing drugs that are licensed for other conditions and apply them to neuromuscular conditions.

Antisense oligonucleotides and exon skipping

Exons are sections of DNA that code for a protein. They are interspersed with introns, which do not code for proteins and are therefore cut out and discarded in the process of protein production, to leave just the exons. For example, in Duchenne muscular dystrophy an exon, or exons, are deleted from the dystrophin gene, which interferes with the rest of the gene being pieced together. The dystrophin protein normally sits in the membrane that surrounds muscle fibres (like a skin) and protects the membrane from damage during muscle contraction. Without dystrophin, muscle fibre membranes become damaged and eventually the muscle fibres die.

Dystrophin is a very large protein; its middle section consists of lots of repeated segments. Mutations that cause exons to be lost result in missing segments, but we know that the protein can still carry out its function if its two ends are joined up, even if some of the middle segments are missing. Using a ‘molecular patch’, scientists are able to join the exons together, ignoring the missing ones – a process called ‘exon skipping’. These patches are made of small, synthetic pieces of DNA called antisense oligonucleotides (ASOs), and these could help to address the underlying genetic cause of several neuromuscular conditions. Results from clinical trials suggest that the efficacy of this therapy may be limited, owing to poor delivery to target tissues. One strategy to improve delivery is to add short, cell-penetrating peptides to the ASOs, and this approach has been pioneered by Professor Matthew Wood at the University of Oxford, who is developing CPP-linked antisense therapies for myotonic dystrophy type 1, spinal muscular atrophy and Duchenne muscular dystrophy. Muscular Dystrophy UK has supported Professor Wood in his work for over eight years. Findings from this research will help to enhance the technology generally, which will be valuable for researchers looking into treatments for other conditions.
4. To improve quality of life and well-being

The effect of neuromuscular conditions on quality of life is significant and wide-ranging, impacting not just on mobility but also on functional activities of daily living like feeding and washing oneself, psychological needs, sleep disturbances, pain and fatigue, and affecting patients, spouses and families. There are nuanced differences in these experiences across different conditions, but well-designed studies are needed to investigate what factors impact quality of life and how it can be improved.

The imperative for the provision of mental health support is also well recognised – for example, parents who receive a diagnosis of a neuromuscular condition at their child’s birth can require a great deal of psychological support in order to be able to remain positive for their child. Similarly, an adult who receives a diagnosis in late adulthood and loses the ability to do other things, and their family roles, may require support to grieve the loss of role and function and adapt to a new reality.

Measuring quality of life

A major challenge in quality of life research is that there are few accurate, meaningful, reliable and sensitive research tools for the study of activity, orthotics, cognition, mental health, pain and fatigue of people with neuromuscular conditions.

Measures of quality of life are often subjective and outcomes can be influenced by factors unrelated to the neuromuscular condition. There are already a number of scales and questionnaires in use to measure and investigate various elements of ‘quality of life’, but these are often not specific to neuromuscular conditions. The challenges this presents are highlighted in the interpretation of outcomes from cognitive tests requiring the movement of objects; it can be difficult to differentiate between the muscular and cognitive effects.

- Muscular Dystrophy UK will support the improvement of outcome measures and research tools, with a particular focus on patient-reported outcome measures (PROMs) and patient-reported experience measures (PREMs). Understanding the expectations of individuals with neuromuscular conditions is crucial. What do people consider quality of life to be? What do children or affected adults and their partners/carers/parents consider quality of life to be? And what improvements do they expect and want?

Researchers who seek to develop tools will be expected to carry out focus groups to refine research needs and priorities. In addition, researchers should review the international landscape and, where possible, build on learning from existing measures.
Understanding the expectations of people with neuromuscular conditions is crucial. What do people consider to be quality of life?
Areas of particular interest include:

- **Nutrition** – the benefits of dietary intervention are largely unknown, with many contradictory reports in circulation. This can cause confusion and frustration for patients and families. More evidence – and more robust evidence – is needed to provide meaningful advice to patients.

- **Psychological well-being for individuals and families** – the psychological impact of living with long-term progressive neuromuscular conditions on the individual and family cannot be underestimated. More research is needed to understand this better and to investigate ways of managing these effects.

- **Activities/exercise** – there is a strong interest from patient communities as to the benefits of activity such as exercise, physiotherapy and hydrotherapy in the management of their conditions. For example, there is emerging evidence of the benefits of aerobic exercise being beneficial and improving quality of life of people with facioscapulohumeral muscular dystrophy (FSHD). However, it is still not clear as to the optimal type or duration of exercise and how this could be translated to the ‘real world’ such that it is of ‘everyday’ benefit. The impact and benefits of exercise is likely to vary for different neuromuscular conditions, therefore more research is needed to better understand the benefits of exercise for people with neuromuscular conditions.
Impact of pain and fatigue – more research is needed into the management of fatigue and pain, for example, chronic pain and/or fatigue are often reported by people with FSHD, myotonic dystrophy, limb girdle muscular dystrophy, Charcot-Marie-Tooth disease, inclusion body myositis, Pompe disease and myasthenia gravis and can have a strong impact on their quality of life. Both aerobic exercise and cognitive behavioural therapy have been shown to reduce the severity of chronic fatigue in FSHD and have a favourable effect on the rate of muscular deterioration. Nevertheless, further investigation into the impact of fatigue and pain on quality of life and how it can be best managed, in addition to improved measures to study quality of life, are required for all neuromuscular conditions.

Muscular Dystrophy UK will support research into the development of robust studies into the quality of life. We are particularly interested in studies that are co-produced with people with experience of neuromuscular conditions. In addition, within the next 12 months we will support a workshop to bring together relevant experts (including allied healthcare professionals) and people with experience of neuromuscular conditions. The workshop will allow participants to create and enhance connections across existing specialisms in order to share best practice and generate well-designed research studies for neuromuscular conditions. The workshop will also inform the advice and guidance that we provide on our website.
Conclusion

Within our research strategy we have identified areas with existing momentum where Muscular Dystrophy UK can focus its efforts over the next three years. We will invest in exciting and innovative research that will deepen our understanding of neuromuscular conditions and help develop new treatments for people living with neuromuscular conditions including ultra-rare conditions. We will also work to ensure that the UK has the capacity to carry out clinical trials to test new treatments in patients with neuromuscular conditions.

We recognise that it is important to seek solutions to help those living with neuromuscular conditions to live well and as independently as possible. Therefore, we will be investing in research that will improve the quality of life for individuals with neuromuscular conditions.

Muscular Dystrophy UK already supports high-quality research into neuromuscular conditions. Awards will continue to be made according to scientific quality and importance to patients, from basic and pre-clinical science to clinical trials and quality of life studies. Through our grant support we aim to encourage new researchers to join the field to ensure that there are sufficient academics for the future. We also aim to support allied healthcare professionals to build research portfolios in the area of neuromuscular conditions.

With our investments we will be transforming lives through research. Through these activities we will further our mission to support research to drive the development of effective treatments and cures because we know that for people living with neuromuscular conditions, every day counts.

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We know that for people living with neuromuscular conditions, every day counts.
Our work relies on voluntary donations from individuals, groups, companies and grant-making bodies. We receive no statutory funding.