



Fast Forward

Fast track to specialist treatments

About muscle-wasting conditions

There are approximately 60 forms of muscular dystrophy and related neuromuscular conditions ('muscle-wasting conditions). The conditions cause the muscles to weaken and waste over time, leading to increasing disability. They may affect not only the muscles in the limbs, but also those of the heart and lungs, sometimes significantly shortening life-expectancy. Approximately 70,000 people in the UK are affected by muscular dystrophy or a related neuromuscular condition.

Many of the conditions are low incidence, rare conditions, with some regarded as very rare or ultra-orphan. Muscle-wasting conditions can be genetic or acquired and, with very few exceptions, there are currently no known effective treatments and currently no cures available.

Family perspectives

The Bennett Family

"For children like our Abbi there is not a moment to lose....Abbi is six now and her condition has already had a big impact on her body. Although currently she can just about walk ten metres, next year it might only be two. As parents we can feel so helpless, with the current system it could be nearly a decade before Abbi could benefit from potential treatments which could radically improve the quality of her life. It is vital that children with progressive conditions have access to these treatments as soon as possible"



Abbi (right), with her family

Sarah Bennett, whose daughter, Abbi, has Ulrich muscular dystrophy

The Robinson Family

"So far, the condition only affects Thomas's mobility - he has trouble standing up, running, jumping and climbing stairs - he can do all these things, but not as fast or steadily as his peers. Without a treatment to stop it, his muscles will weaken and waste as he grows, affecting first his legs, then his arms and eventually his heart and lungs. We are racing against the clock. The earlier we are able to treat him, the more likely we are to stop further damage to his muscles. We need to be ready for when a treatment does become available, and plan ahead now so that a day is not lost before it reaches our children."

Karen Robinson, whose son, Thomas, has Duchenne muscular dystrophy.

1. Executive Summary

The Muscular Dystrophy Campaign is leading the UK campaign to speed up access to potential treatments for muscle-wasting conditions.

Time is not a luxury that people and their families affected by muscle-wasting conditions have. For example, in rapidly progressive conditions such as Duchenne muscular dystrophy there is currently no way of restoring muscle mass and strength once it is lost.

The launch of *Fast Forward* comes shortly after the European Medicines Agency (EMA) recommended conditional approval to Translarna (formerly known as ataluren), a drug designed to treat the 10-15% of boys and young men with Duchenne muscular dystrophy caused by a 'nonsense mutation'.

This is a landmark decision and Translarna will be the first drug treating an underlying genetic cause of Duchenne muscular dystrophy to be made available outside of clinical trial. We must now ensure that other potential drugs to treat muscle-wasting conditions are able to follow as quickly as possible. Our Fast Forward Campaign aims to achieve this by;

- ensuring the European Medicines Agency (EMA), National Institute for Health and Care Excellence (NICE), Medicines and Healthcare products Regulatory Agency (MHRA) and NHS England minimise delay in the regulatory, approval and funding processes for potential treatments for muscular dystrophy or related neuromuscular conditions
- working with our supporters in all four UK Parliaments and Assemblies to highlight the need to Government Ministers for fast and fair access to high-cost drugs for rare diseases and ensure the voices of patients and families are heard
- pressing funding and regulatory bodies to improve clinical trials infrastructure, such as securing statutory funding for patient registries and databases
- encouraging the NHS to support specialist centres and clinics and help them develop into clinical and research centres of excellence. This would enable them to take part in clinical trials, as well as deliver and monitor any forthcoming treatments for muscular dystrophy or related conditions
- working with the NHS to ensure sufficient support is provided to develop multi-disciplinary teams at centres of excellence. This would ensure that patient centred care can be given during one visit, rather than visits to many different hospitals. This should include a neurologist assessment, respiratory assessment and a cardiology assessment as appropriate. Consistently high standards of care would also mean patients' conditions are managed to the best possible level, thereby enabling them to take part in trials for drugs to treat muscle-wasting conditions
- enabling the views of patients to be heard so that regulators fully understand the severity and impact of muscular dystrophy and related neuromuscular conditions when they make decisions on drug licensing

2. Calls to Action - Ten steps towards faster access to specialist treatments

1. Funding for patient registries and databases from statutory bodies, for example NHS England or Public Health England.

“Patient registries and databases for rare conditions are crucial, as without them it is much harder for researchers to monitor the natural history of a condition or have access to enough patients to conduct a large scale clinical trial. However, funding for these resources is fragile and currently reliant on charitable organisations.

We must ensure clinical trials infrastructure is prioritised: I support the Muscular Dystrophy Campaign’s call for statutory funding for registries and databases.”



Professor Kate Bushby, Professor of Neuromuscular Genetics, MRC Centre for Neuromuscular Diseases

2. The NHS to provide support to a national network of Muscle Centres across the country and enable them to develop into clinical and research centres of excellence, allowing them to conduct trials as well as deliver and monitor any forthcoming treatments.

“Research into treatments for muscular dystrophy and related conditions is an exciting and growing area and it is important that we develop a network of centres that can drive this work forward. With the right support, I believe that services such as ours in Southampton can develop into a clinical and research centre of excellence and play a crucial role in trialling, delivering and monitoring any new therapies.”



Dr Simon Hammans, Consultant Neurologist, Wessex Neuromuscular Service

3. The NHS to support work to identify and develop new biomarkers for muscle-wasting conditions, including MRI scanning. This would enable the diagnosis and progression of a condition to be measured more accurately and without the painful and invasive procedure of muscle biopsy.
4. The EMA to ensure that, should exon skipping drugs for Duchenne be licensed in Europe, platform approval is made available so that further such drugs with the same mechanisms of action would not have to go through the same lengthy approvals process from the start.
5. The EMA to grant applicant companies conditional approval whenever appropriate to do so. We urge them to continue to meet with patient groups, expert clinicians and researchers to learn more about the severity and the impact of long term progression of conditions such as Duchenne muscular dystrophy when reaching their decision.
6. The EMA to continue to support pharmaceutical companies during the development of clinical trial design; meeting with researchers early on in the process and taking into account outcome measures.

7. NICE and equivalent bodies in the devolved countries to assess high-cost drugs for rare conditions separately from drugs for more prevalent conditions and not subject them to an inappropriate cost benefit analysis.

'With therapies finally coming to the market, it is clearly vital we gain the necessary approval and funding to get treatments to patients. I wholeheartedly endorse the Muscular Dystrophy Campaign's approach. They are working with regulatory bodies (EMA and MHRA), NICE and NHS England, along with the biopharma industry, to tackle these important issues'.



Professor Dame Kay Davies, DBE, FRS, FMedSci, Dr Lee's Professor of Anatomy, University of Oxford. Deputy Chair, Wellcome Trust

8. The NHS to identify a new model of sustainable funding to help meet the costs of delivering high-cost treatments for rare conditions, given that the ring fenced fund for rare diseases was abolished in 2012.

"Successfully developing an effective treatment is far from the end, with agonising waits for some families through licensing and funding issues. We must focus on ensuring that if treatments for muscle-wasting conditions are proven to be safe and effective, the UK is in a position to license and fund them swiftly."



Dave Anderson MP, Chair of the All Party Parliamentary Group for Muscular Dystrophy, who lost his brother, sister, nephew and niece to myotonic dystrophy

9. NICE to develop clinical guidelines for rare neuromuscular conditions. As well as promoting excellent care, this would also make the UK a more attractive place for clinical trials for rare neuromuscular conditions.

10. The NHS to ensure that optimum standards of care are delivered so that all patients in the UK have access to essential care and support from specialist multi-disciplinary teams. Consistent, high standards of care will also ensure that patients can participate in clinical trials for their particular condition.

'The physical care needed for muscular dystrophy and related neuromuscular conditions is complex and distinct. This should primarily be carried out by multi-disciplinary teams at specialist centres over the course of one visit and include input from a range of specialists. I fully support the Muscular Dystrophy Campaign's call for the NHS tariff to adequately fund this complex assessment so that each patient's condition is managed and monitored as comprehensively as possible.'



Dr Ros Quinlivan, Transition service lead for adolescents and young adults with Neuromuscular Disease at Great Ormond Street Hospital and Queen Square

3. Analysis and Discussion

Clinical trials/infrastructure

Currently, there is no statutory funding in the UK for some key elements of the clinical trials infrastructure, such as patient registries and databases for muscle-wasting conditions. This absence of long term statutory funding can hinder a centre's ability to fully and adequately participate in clinical trials. In addition, reductions to administrative support mean that some centres are finding there is no member of staff to enter data into patient registries. For rare conditions these gaps can be damaging; patient numbers at each centre are small and registries are needed to access greater patient numbers so that trials can take place across many centres.

*We call on statutory authorities to take on responsibility of funding **patient registries**, so that clinical trials are not delayed or prevented by not being able to find and recruit enough patients, and registries are not completely reliant on charitable funding.*

*We call on statutory authorities to provide funding for **clinical databases** so that researchers and clinicians can have access to data that sets out the natural history of a condition. This aids researchers in selecting patients to take part in clinical trials, as well as helping to show the disease modifying impact of potential treatments.*

*The Muscular Dystrophy Campaign currently provides funding for Clinical Research and Training Fellowships. We call on the NHS and hospital trusts to build on this work and support **neuromuscular disease clinical research fellows**, to encourage greater links between research and care.*

We call on NHS England and hospital trusts to develop skills of teams at neuromuscular disease centres so that they are well equipped to take part in clinical trials. For example, physiotherapists will need to know how to effectively use the six minute walk test so as to ensure a consistent set of tests in clinical trials.

Functional indicators in clinical trials

In addition to funding for clinical trials infrastructure, the Fast Forward campaign will also focus on raising awareness amongst regulators of functional indicators that matter to patients. It is encouraging that in clinical trials, some additional indicators are already being used to measure a drug's efficacy, alongside the six minute walk test. We believe further discussions must take place with patients to assess what other indicators can be developed to assess drug's performance in clinical trial.

*We urge the EMA to consult with patients and gather data to support the development of **patient reported indicators** relating to daily living when assessing the effectiveness of treatments.*

Speeding up access to treatments

We call for a wider roll out of the EMA's adaptive licensing pilot scheme to support access to innovative medicines in a responsible and managed way.

Under current rules, each new drug using exon skipping technology would have to be tested and licensed individually because each drug has a slightly different chemical structure, despite having the same mechanics of action. *In exon skipping, once safety and effectiveness of the technique have been satisfactorily proven, it will be important for the EMA to ensure that each drug would not have to go through the same lengthy approvals process from the start.*

*With the pipeline for potential treatments set to grow for muscle-wasting conditions, we call on the EMA to grant applicant companies **conditional approval** wherever appropriate and with safety and effectiveness having been demonstrated. In cases where a company applies for conditional approval for a potential treatment for muscular dystrophy or a related condition, we will provide the EMA with relevant supporting evidence to help it reach its decision. We will also petition the European Parliament whenever there is a compelling case for a drug to receive authorisation.*

We call on the EMA to fully consider the severity of muscle-wasting conditions when appraising potential treatments, including taking into account the experience of clinicians and patient groups.

Appraisal and commissioning process

NICE appraisal process. We meet regularly with NICE, and there is still considerable uncertainty in this area. In particular, there is little clarity on what will constitute a 'highly specialised technology'. This could potentially leave costly treatments for rare conditions being assessed inappropriately with the use of the same criteria as cheaper treatments for more prevalent conditions.

We call on NICE to ensure that drugs for rare diseases are assessed in a fair and transparent way, so that they are not denied simply on the grounds of high cost and patients and families are not disadvantaged.

Funding for highly specialised technologies. A ring fenced fund for highly specialised technologies was abolished following the disbanding of NHS Specialised Services. Without any new initiative to take its place, the refusal to grant approval on the grounds of high-costs could be increased.

We call on the NHS to work with the pharmaceutical industry to look at innovative ways in which drugs for rare diseases could be funded, so that the necessarily high-costs that these drugs entail do not prevent them from receiving funding in the UK.

Care and support

Appropriate care and support are essential to help manage neuromuscular conditions and in some parts of the country support for patients and families falls well short of what is needed. The new NHS commissioning mechanisms, far from increasing equality of care for patients with rare diseases, are threatening to increase regional variation due to local interpretations of the guidance provided.

*We call on the NHS to support a network of at least ten **specialist centres** across the UK and support them to evolve into clinical and research centres of excellence. This would*

ensure that comprehensive and consistent care is delivered by multi-disciplinary teams across the country.

We call on the NHS to provide all individuals and families affected by muscle-wasting conditions with access to a fully funded multi-disciplinary team. Comprehensive support should be delivered over one visit and include a neurological assessment, sleep study/respiratory assessment, cardiologist and echocardiogram, gastroenterologist and a physiotherapist. Consistently high standards of care will also mean patients can take part in trials for potential drugs to treat their particular condition.

We are concerned to hear from leading clinicians that training programmes for neuromuscular diseases are very limited. With people with severe muscle-wasting conditions now surviving into adulthood, we need to ensure that clinicians for the future are identified and trained. *We call on the National Institute for Health Research to fund a programme of training fellowships for neuromuscular diseases.*

In order for patients to receive the care and support they need, it is vital that best practice standards of care are implemented. *We call on the National Institute for Health and Clinical Excellence to adopt **clinical guidelines** for rare neuromuscular conditions. As well as promoting excellent care, this would also make the UK a more attractive place for clinical trials for rare conditions.*

Newborn screening

This is closely linked to the development of treatments; Duchenne will need to be detected early and treatments may need to be administered early in order to have optimum effect. It will be necessary to have a programme of newborn screening in place by the time any treatments reach the market so any boys who could benefit from them are identified.

*Our APPG report has been very successful in enabling us to establish links with the National Screening Committee, and we will ensure we are involved in **compiling evidence in support of a programme** when the National Screening Committee reviews its policy on newborn screening for Duchenne in 2015.*