



07

Chapter 07 – Research and clinical trials

Contents	Page No.
In a nutshell – Research and clinical trials for DMD	106
Treatments in the pipeline	108
We’re making progress with DMD research	110
What is a clinical trial?	111
What does it mean to my child and our family?	112



In a nutshell — Research and clinical trials

1 We're helping to develop new treatments for this generation of DMD patients

Duchenne UK are tackling some of the biggest barriers to getting new treatments to this generation.

We're funding dozens of research projects and clinical trials. We're also working with pharmaceutical companies to accelerate the approvals process for new treatments by government regulators.

2 Doctors are making huge progress in treating DMD

In the last five years, the landscape has changed for DMD. There are now clinical trials taking place which are testing gene therapy, steroid alternatives and other approaches, including repurposed medicines, which could all lead to better treatments.

3 We're aiming for a cure

Ultimately, we want to end Duchenne. We want to develop a cure so that everyone diagnosed will have access to life-changing treatments.

So, we are working with doctors and medical researchers on gene therapies, including gene editing. These therapies have the potential to treat the cause of DMD, not just its symptoms.

We're funding dozens of research projects and clinical trials.



Ultimately, we want to end Duchenne.



We want to develop a cure so that everyone diagnosed will have access to life-changing treatments.



4 Your child may be able to join a clinical trial

Your child may be able to participate in a trial where they receive potential treatments which are being tested for DMD.

And they may be able to join a trial where the effects of DMD on their body are observed so doctors can understand the disease better.

5 You can get information and advice about clinical trials

The DMD Clinical Trial Finder has information about every clinical trial in the UK. You can find it at www.dmdhub.org/clinical-trial-finder.

It was set up by Duchenne UK to ensure everyone with DMD has the opportunity to access clinical trials.

You should discuss clinical trials, especially ones that you are considering joining, with your neuromuscular doctor. They will be able to provide information and advice to you.

We recommend you join the DMD Hub by signing up to their mailing list at www.dmdhub.org/join-the-hub/. This will help you to keep up-to-date with the latest clinical trials for DMD in the UK.

Ultimately, we want to end Duchenne.
We want to develop a cure so that everyone diagnosed will have access to life-changing treatments.

Making an impact

Duchenne UK is ambitious and determined.

Since we were established in 2012, we have:

- Spent **£6,500,000** on accelerating research
- Further **£3,500,000** committed to research
- Attracted **US\$42,500,000** of private investment for Duchenne Biotech firm by working with three other charities
- Helped win **US\$12,000,000** of government funding a clinical trial of VBP15, a potential alternative to steroids, along with international charities

Currently, we are funding more than 50 projects, including three clinical trials.

Treatments in the pipeline

Medical researchers are working hard to develop treatments that could maintain children's long-term muscle function without unacceptable side effects.

In the last 10 years, many potential new treatments have emerged.

Some focus on the underlying cause of DMD (which is a lack of dystrophin). Others aim to reduce the symptoms caused by the lack of dystrophin in the muscles.

Some potential new drugs are now being tested on patients with DMD in clinical trials. Others are about to start trials.

In the last 10 years, many potential new treatments have emerged.



These are six of the approaches that are being used to develop new treatments for DMD:

- **Repurposing existing drugs** – Using drugs which were originally developed to treat one condition (e.g. visual impairment) for another condition (e.g. DMD). This can be a much faster way to produce treatments than developing new drugs.
- **Anti-fibrotics** – Drugs which reduce the scar tissue (fibrosis) which replaces muscles when they are injured but cannot repair properly.
- **Utrophin modulators** – Drugs which increase utrophin, a protein which is naturally found in muscles and has the potential to work as a substitute for dystrophin in muscle cells.
- **Myostatin inhibitors** – Drugs which reduce the effect of myostatin, leading to an increase in the size of patients' muscles. (Myostatin is a natural protein that stops our muscles from becoming too big as we grow up).
- **Gene therapies** – A therapy which aims to replace patients' faulty dystrophin genes with different ones so their muscles work much better. It's one of the most exciting treatments being developed for DMD because it aims to target the cause of DMD, not just the symptoms.
- **CRISPR** – An exciting genetic-engineering technique that aims to 'cut out' the faulty part of patients' dystrophin gene so they can make a shortened dystrophin protein which will make their muscles stronger.

Find out more

You can find out more about these approaches and other potential treatments for DMD on the Duchenne UK website:

www.duchenneuk.org

We're making progress with DMD research

Duchenne UK are determined to speed up the process of developing new treatments for DMD patients.

Currently, developing a new drug is a costly and complicated process that can take many years. This can be very difficult to manage for DMD families, for whom every minute counts.

Accelerating the drug development and approval process

Duchenne UK are supporting the development of new treatments for DMD in three ways:

1. Funding new scientific research and clinical trials into drugs and therapies that could:

- Preserve quality of life by minimising damage to patients' muscles and keeping their hearts and lungs healthy
- Change the disease itself by replacing dystrophin, increasing utrophin (a natural muscle protein) or repairing damaged muscles

2. Speeding up clinical trials – By creating an online hub www.dmdhub.org for DMD clinical trials which supports patients, doctors and industry. It provides details of all trials in the UK and provides doctors with resources to help them run their trials better.

Already, the DMD Hub has funded more than 16 critically-needed clinical posts and increased the number of hospitals where trials are being run.

3. Speeding up the approval process for DMD drugs – Through running Project Hercules, www.projecthercules.org, which is a collaboration between a group of leading pharmaceutical companies to fund and develop a single robust evidence base that shows the true economic cost of DMD.

This evidence base will be used by government regulators when they are considering whether to approve a new drug for use in DMD.



What is a clinical trial?

Clinical trials are research studies that explore whether a medical treatment, drug, procedure or device is safe and effective for humans.

Clinical trials are an important part of the evidence-based process for making new treatments available to patients. They produce the best possible data for healthcare decision-makers.

They are required by the Medicines and Healthcare Products Regulatory Agency (MHRA) which licenses new medicinal products in the UK. Doctors can only use drugs, procedures and devices with this licence to treat their patients.

Doctors, other healthcare professionals and patients also use the results of clinical trials when they are creating treatment plans for individual patients. For example, they might look at the results of a clinical trial for steroids to help them decide when a patient should start taking this treatment.

What are the different types of trials?

Doctors use two different types of clinical trials to study diseases and investigate treatments:

- **Controlled trials** – Doctors give a potential new drug, device or therapy to the patients and measure the effects. This helps them work out if the treatment is safe and effective. (Trials where patients receive treatments are also called interventional studies)

For example, doctors might give patients a drug which could be repurposed for DMD. Or they may use different devices for administering steroids on different patients to see which one works best.

- **Observational studies** – Doctors just observe patients and take measurements from them. The patients do not receive any treatments.

For example, the doctor may be interested in comparing the movements of patients who have been taking steroids since they were five years' old and those that have not. So, the doctor will only measure their movement and won't give them any new treatments.



Find out more

You can find out more about the different types of trials on Duchenne UK's clinical trial website:

www.dmdhub.org

What does this mean for my child and our family?

When the time is right, you may like to explore whether your child should take part in a clinical trial.

This is a big decision and a very personal one. It depends entirely on what you feel is best for your child.

You should talk it through fully with your child's doctors. But you should never feel any pressure to take part in a trial.

What are the benefits of taking part in an interventional trial?

Clinical trials are often a family's main hope of accessing potential new treatments that might one day become approved medicines.

If your child takes part in an interventional trial, they could receive a potential new drug, device or therapy much earlier than if they don't take part in the trial.

But please bear in mind that some interventional trials use placebo-controls. Placebos are sugar-pills which look the same as the potential new treatment. Doctors give the potential new treatment to one group of patients and the placebos to another. This helps them identify exactly what effect the treatment has on patients.

So, if your child takes part in an interventional trial, they may receive the potential new treatment or the placebo.

If they do receive the potential new treatment, it

Clinical trials are often a family's main hope of accessing potential new treatments



Placebos are sugar-pills which look the same as the potential new treatment.



If your child takes part in an interventional trial they may receive a placebo.

may help your child by making them stronger and healthier. But, of course, the treatment could also have no effect on them. Or it may even harm them.

These are things that you need to think about carefully and discuss with your doctors.

What are the benefits of taking part in an observational trial?

If your child takes part in an observational study, they will not receive any treatments. They will only be observed and measured.

Your child will help doctors to learn more about DMD. Over the long-term, this will help them to develop and use drugs better for many DMD patients.

But they will not benefit directly from the trial themselves.

How can I find out about clinical trials for DMD?

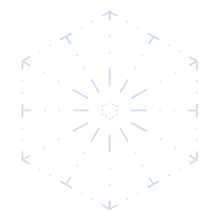
You can find all of the clinical trials for DMD in the UK on Duchenne UK's clinical trial website at www.dmdhub.org/clinical-trial-finder

We set up the Hub to ensure that all patients with DMD, both children and adults, have the opportunity to access clinical research opportunities.

Your consultant won't always tell you about research. So, you should check the DMD Hub website regularly and sign up to their newsletter so you can stay up-to-date yourself.

Your child will help doctors to learn more about DMD

The Hub ensures that all patients with DMD have access to clinical research opportunities







08

Chapter 08 – Emergencies

Contents	Page No.
How should we prepare for emergencies?	116
Who needs to know these emergency procedures?	117
What is an emergency for a child with DMD?	118
What should we do in an emergency?	119
What should you tell the emergency services?	119

