DMD FAMILY FOLDER

SUPPORT AND GUIDANCE FOR NEWLY-DIAGNOSED PARENTS

Important information to help you navigate a DMD diagnosis

Chapter Overview

01

Chapter 01 – Introduction

Dear Parents and Friends

If you're reading this, chances are that either your child, or a child close to you, has been diagnosed with Duchenne Muscular Dystrophy (DMD).

Both of us, our sons and our families, have lived with this disease since 2011. It has caused us unspeakable pain and, at times, nearly broken us. But it has also united us. And it has made us strong. And it has made us fight for our sons in ways we never thought possible. And yes, we are able to smile, and laugh, and enjoy life with our children. And, in time, you will too.

Diagnosis can be an emotionally devastating time. A time when the hopes, dreams and plans that you had for your child and your family, now seem to have no place. Instead, you're faced with a new reality of grief, despair, loneliness, a feeling of losing control, a deep sadness for your child and the end of a carefree life.

But the more you learn about Duchenne Muscular Dystrophy, the more you will hear about a global army of doctors, scientists, researchers and drug developers who are absolutely committed to fighting this disease for all our children.

There are many reasons for optimism now. Promising new therapies are being tested. Many more are being developed. Yes, we've had some tough times in this community. But we can learn from those times. And be better. And be stronger for the future. Because we owe it to our children. Because they deserve safe and effective treatments. And they need them now.

Please use this folder and our website to help you navigate the early months of diagnosis. You will find lots of information to help you - information about DMD and its causes and how you can best look after your child.

Understanding your child's condition will empower you and give you back a sense of control. And it will help you lobby for your child and get them the best possible care. We believe in the power of science and medicine to change lives. And of the strength of communities like ours to move mountains.

Together, we can change the future for everyone living with Duchenne Muscular Dystrophy.

Emily and Alex

Emily Crossley and Alex Johnson
Co-Founders and Joint CEOs of Duchenne UK

The first few months

The first few days, weeks and months can be overwhelming and distressing as you try to absorb this diagnosis and learn everything you can about DMD.

But you are not alone. Duchenne UK are here to support you on your journey.

No one will ever forget the day of diagnosis. Everything in your world changes forever, and yet so much will stay the same. Families find themselves grieving for things that are not yet lost, grieving for the futures they thought their child had. And grief can be all-consuming and exhausting.

When your child or children are diagnosed with DMD, it affects siblings, grandparents, extended family members and friends as well as parents. Coping with other people's distress, as well as your own, can be extremely difficult.

Everybody copes with upsetting situations in their own way. There is no right or wrong way to feel at this time. So, you may find that you, your partner, your children and other family members are all reacting in very different ways.

> You are not alone.
> **Duchenne UK are here to support you on your journey.**

> Don't overwhelm yourself with too much information straight away.
>
> Just focus on getting through the next few days and weeks rather than racing ahead to what the future may hold.
>
> There is lots of hope and the future will likely be very different for your child.

Where you can get help

Support from other Duchenne parents

Parental Support Network – you can find other DMD parents at the Duchenne Parent Zone on Facebook.

We can also put you in touch with other parents and families from our network. Please get in touch with us at **support@duchenneuk.org.**

Emotional support

Your GP, paediatrician and neuromuscular consultant can provide referrals to counselling services for your family.

Information about DMD

You can get reliable information about DMD from your neuromuscular consultant and the Duchenne UK website, **www.duchenneuk.org**

While the internet has a lot of high-quality information, it can also be a source of misleading information and false hope. So, please rely on your neuromuscular doctor for expert and professional advice.

Medical research and clinical trials for DMD

You can find out about research and trials on the DMD Hub website (which is run by Duchenne UK) at **www.dmdhub.org**

In a nutshell — What do we do now?

In the first few months after diagnosis, there are only a few things you need to do.

This is a serious situation. But it's not an emergency. So, you don't need to change everything right away.

These are the things that DMD parents recommend you do in the next one to six months so you can best look after your child and your family:

1 Look after yourself and each other

Give yourself time to cope with the diagnosis. Be gentle with yourselves. You've had a terrible shock and it will take a while before you get your emotional balance back.

Accept all the help that is offered to you. It will make things easier for you.

2 Tell your family and friends - and ask for their support

Sharing the diagnosis with grandparents, extended family and friends can be tough. They are likely to be shocked and upset too.

They may ask what they can do to help. We encourage you to ask them to help with practical things, like cooking, shopping, housework and childcare. We found that these things made a big difference to us during first few months.

If you need time off work, you may also need to speak to your employer and ask for compassionate leave.

Give yourself time to cope with the diagnosis.

Accept all the help that is offered to you.

Ask family and friends to help with practical things.

If you need time off work you can ask for compassionate leave from your employer.

3 Talk to your GP - and ask them for referrals

Tell your GP about your child's diagnosis – if they don't know already.

Your child will get the best treatment if they see a neuromuscular doctor in a multi-disciplinary clinic.

Ask your GP for a referral to a North Star centre if possible as they are DMD experts.

You will also need support from your local authority's health, education and social care services. It's best to start working with your local authority soon because some professionals are very busy.

So, also ask your GP for referrals to a social worker and an occupational therapist. (You can also refer yourself to your local authority if that's easier for you).

4 Tell your child's teachers

You'll need to talk to your child's teachers at nursery or school.

You'll need to make a few changes quickly to protect your child's muscles (which are explained in the school section of this folder). And, in time, you'll need to plan their long-term care and support together.

Your child will be entitled to support at school. You can find information about what you need to do to set this up in the school's section of this folder.

5 Begin learning and talking about DMD

The more you know about DMD, the better you'll be able to look after your child.

You'll find helpful information in this folder and on our website. Your neuromuscular doctor will be able to advise you too.

Guides to DMD on our website

As DMD parents, we know from experience that it can be difficult to explain the condition to other people – especially when you are still in shock from the diagnosis yourself.

So, we've produced special guides to DMD to help you have conversations with:

- **Your family and friends**
- **Your employer**
- **Your child's teachers**
- **Your babysitters**

www.duchenneuk.org/resourcesforparents

Support is available

It is difficult and upsetting to read these things about your child for the first time.

Please remember Duchenne UK and our Parent Support Network are here to support you.

Please get in touch with us at:

support@duchenneuk.org

What is Duchenne Muscular Dystrophy (DMD)?

Duchenne Muscular Dystrophy (DMD) is a genetic disease that causes muscle weakness and wasting. It is the most common form of muscular dystrophy in children.

Children born with DMD cannot produce dystrophin, a protein that is vital for muscle strength and movement.

Dystrophin makes muscles stronger and more stable. It acts like a shock absorber and protects muscles from injury as they contract and relax. Without the shock absorber from dystrophin, muscles are weaker and more likely to get torn and damaged.

Children with DMD cannot produce dystrophin because they have a fault on their dystrophin gene.

This fault is often called a mutation by doctors and scientists.

DMD is not contagious. So, people cannot 'catch' DMD from someone who has it.

The name Duchenne comes from the doctor who first described the condition.

Our Parent Support Network can help

One in 3,500 boys born worldwide will have DMD

Only 1 in 50M girls will have DMD

Who gets DMD?

DMD almost always affects boys. It knows no boundaries and affects people of every race, culture, income level and background.

One in every 3,500 boys born worldwide will have DMD. It is estimated there are 2,500 boys and men in the UK and 300,000 worldwide living with DMD.

Very rarely, DMD can also affect girls. Only 1 in 50 million girls will have DMD. More boys than girls are affected by DMD because of the way the genes are passed down from parents to children.

Children tend to be diagnosed before they are five years' old.

How does DMD affect the body?

DMD gradually weakens the muscles throughout the body. This results in disability and a shortened life expectancy.

Muscle weakness starts in early childhood. At this stage, the weakness mainly affects the 'proximal' muscles which are near the trunk of children's bodies, around their hips and shoulders.

Parents and doctors usually notice the symptoms first when the children are between 2 and 5 years' old. Often, the first signs are when the children have difficulty getting up from the floor, walking or running.

The children's muscles get progressively weaker as they go through childhood and adolescence.

Some children will also have learning and behavioural challenges. This is because dystrophin plays an important role in cell-to-cell communication in the brain. But this is not inevitable. And, if it is present, it does not get worse over time.

Eventually, DMD affects all the muscles the children use for moving and breathing, and their heart muscle too.

In general, children experience a steady decline in muscle strength between the ages of 8 and 12. By early teens, many children will be full-time wheelchair users. By their late teens, most young people with DMD lose the ability to move their arms and experience more problems with their lungs and heart.

Adults with DMD do have shortened lives. It is unusual for someone with DMD to live beyond their 40s at the moment.

However, life expectancy is increasing because medical advances are improving the way we manage DMD. More young people with DMD are able to lead independent lives and go to university, get jobs and live on their own.

Is there a cure?

There is no known cure. But don't despair.

Extensive medical research and clinical trials are underway to find an effective treatment.

There are some current treatments which are already improving the quality and length of life for children and adults living with DMD.

Great advances have been made in treatments that protect children's breathing and hearts and steroid treatments that protect their muscles.

These help us to keep our children in the best possible condition, so they can take advantage of new treatments when they become available.

Duchenne UK is investing millions of pounds in medical research and clinical trials. This is bringing us ever closer to effective treatments for DMD.

Advice and resources

Duchenne UK can help you to access all the available information and resources so you can make informed choices for your child and your family.

Causes of DMD

Duchenne muscular dystrophy is a genetic disease.

It is caused by a mistake in the dystrophin gene that either occurred spontaneously in the child or was inherited from their mother.

How genes work in our bodies

The human body is made up of trillions of cells. Our cells include all the instructions our body needs to develop, live and reproduce.

These instructions are made of DNA. Our long strands of DNA are made up of smaller sections called genes.

Most genes are instructions for making proteins. Proteins tell our cells what sort of cell it is and what it should do. Genes have regulators which turn them on and off – effectively telling cells when to start making their proteins and when to stop.

Our long strands of DNA, which contain most of our genes, are organised into chromosomes.

The instructions in our chromosomes, including our genes and gene regulators, are hereditary. In other words, they are passed down from parents to children.

But genes can also spontaneously mutate (which means change) inside an embryo during pregnancy. A mutation in a gene can change the instructions it gives. This sometimes leads to a protein being made wrongly, or not at all.

> The human body is made **up of trillions of cells.**

Spontaneous mutation (changes within the child)

In one third of DMD cases, the mistake on the dystrophin gene happened spontaneously at the beginning of pregnancy.

The dystrophin gene is one of the largest genes in the body. And the larger the gene, the greater the chance that a mistake will happen. This is why DMD has a high rate of spontaneous mutation.

The genetic change occurs very early in the development of the fertilised egg. As a result, the change in the dystrophin gene is found in all the cells of the body.

Inherited (passed down from the parents)

In two thirds of DMD cases, the genetic change is passed down from the mother to the child.

These women are called 'carriers' because they carry the DMD gene but they do not have the condition themselves. Some carriers have symptoms of DMD. (You'll find more information about carriers in a few pages).

Every person has two sets of chromosomes which contain their genes.

One set of chromosomes is inherited from their father and the other set from their mother. The chromosomes are copied from the parents' cells into the child's cells when the egg is fertilised.

Every person also has two sex chromosomes, which determine if they are male or female. Females have two X chromosomes and males have one X chromosome and one Y chromosome.

The dystrophin gene is located on the X chromosome.

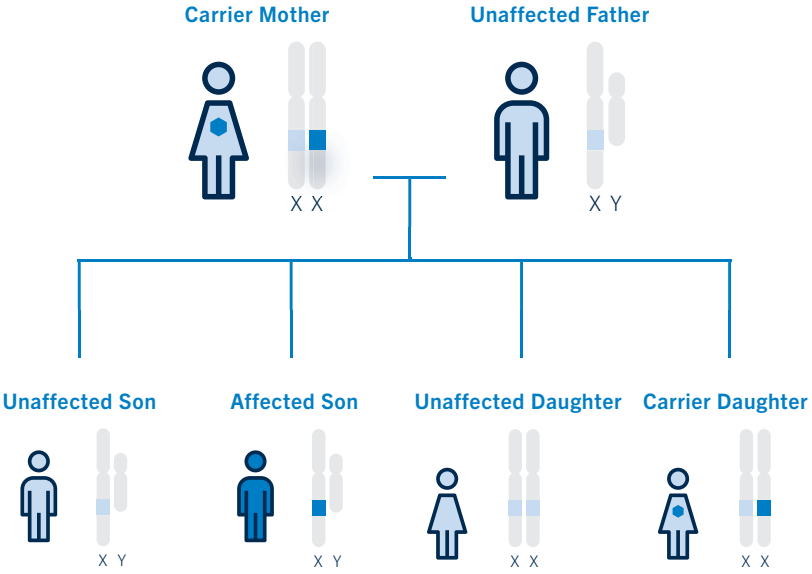
Males have one X chromosome (which they inherit from their mother) and one Y chromosome (which they inherit from their father).

If their X chromosome contains a faulty dystrophin gene, they will have DMD. This is why boys can only inherit DMD from their mothers and not their fathers, because they inherit their only X chromosome from their mother.

Females have two X chromosomes; one inherited from each parent.

If one of their X chromosomes has a faulty dystrophin gene, the healthy dystrophin gene on the other X chromosome usually compensates for it.

X-LINKED RECESSIVE, CARRIER MOTHER



So, if a woman who is a carrier is pregnant, there is a one in four chance that the baby will be:

- An unaffected son who has inherited his mother's healthy X chromosome and, therefore, doesn't have DMD
- An affected son who has inherited his mother's faulty X chromosome and, therefore, has DMD
- An unaffected daughter who has inherited her mother's healthy X chromosome and, therefore, is not a DMD carrier
- A carrier daughter who has inherited her mother's faulty X chromosome and, therefore, is a DMD carrier

If a man with DMD has children with a woman who is not a carrier, none of their sons will have DMD but all their daughters will be carriers.

Why has this happened?

No one is to blame. No one did anything wrong to cause your child to be born with DMD.

There's nothing you could have done during your pregnancy to prevent your child from developing DMD.

Genes are very complicated and sometimes they go wrong.

Females with DMD or who are manifesting carriers

DMD affects only 1 in 50 million girls (compared to 1 in 3,500 boys).

A girl only develops DMD if both of her X chromosomes have faults in their dystrophin genes.

This can only happen if one of the following extremely rare events takes place:

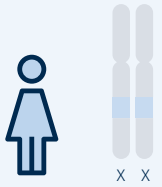
- The girl inherits a faulty dystrophin gene on one of her X chromosomes from her mother and also develops a spontaneous mutation in her other X chromosome
- The girl's healthy dystrophin gene on one X chromosome becomes 'switched off' by a process called X-linked inactivation because she has inherited a faulty dystrophin gene on her other X chromosome
- The girl's father is a man with DMD and her mother is a woman who carried a defective DMD gene ('a carrier')
- The girl develops spontaneous mutations in the DMD gene on both of her X chromosomes

Manifesting carriers

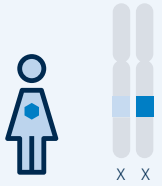
Some female carriers of a faulty dystrophin gene have very mild symptoms of DMD, including fatigue, mild muscle weakness and cramping.

These girls and women are called 'manifesting carriers' because they produce reduced amounts of dystrophin.

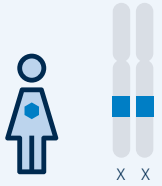
A girl only develops DMD if both of her X chromosomes have faults in their dystrophin genes.



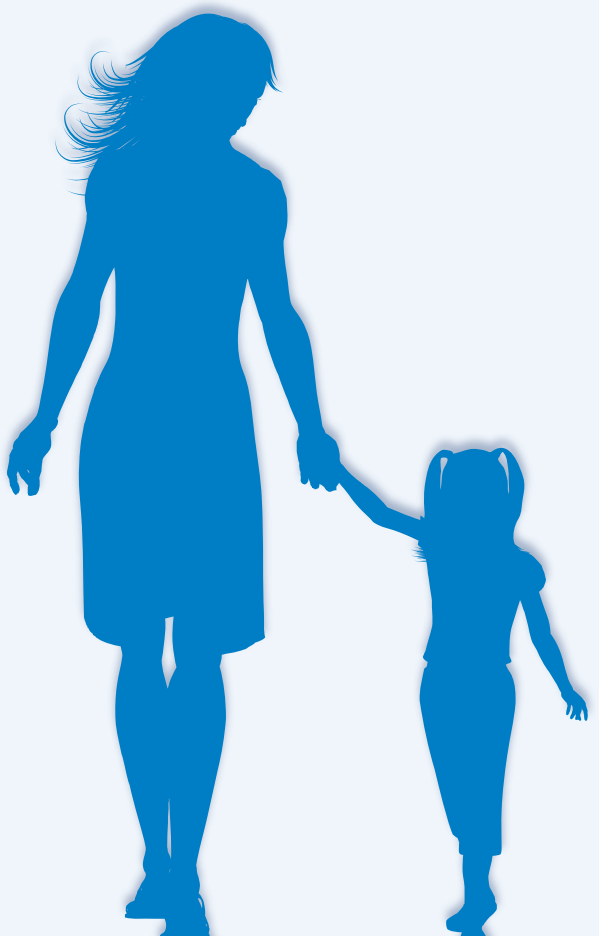
Unaffected Girl



Carrier Girl



Affected Girl



Where to get help for your daughter or yourself

If your daughter has DMD or you are a woman who is a manifesting carrier, please contact Duchenne UK via our website: **www.duchenneuk.org**

We are here to help everyone affected by DMD – girls and women, as well as boys and men.

You should also talk to your GP who can help you to manage any symptoms.

Support from other DMD carriers

If you are a carrier and would like to speak with someone else who is a carrier, please contact us at:

support@duchenneuk.org

Parents and Siblings

If your child has inherited DMD through their genes, other members of your family may also be affected.

Parents who are carriers of DMD

If you are a carrier, you may be at increased risk of heart disease.

We recommend that you ask your child's neuromuscular doctor for advice about heart check-ups. You should also let your GP know that you need heart check-ups.

Germline Mosaicism

A small number of women who do not test positive as carriers for DMD may still be able to pass a faulty DMD gene to their children. This is called germline mosaicism.

It is caused by a mutation in the DMD gene in the cells of some of the mother's eggs. But the mutation is not in the DMD gene in the rest of the mother's cells.

If one of the affected eggs is fertilised, the resulting baby will have DMD if it's a boy or be a carrier if it's a girl.

Doctors estimate that around 15% of mothers of boys with DMD have passed a faulty DMD gene to their children in this way – even though the mothers do not appear to be carriers.

Mothers who are pregnant

If you're pregnant when you receive your child's diagnosis, you'll need to speak to your neuromuscular consultant about your pregnancy and genetic testing.

This is a difficult and distressing situation.So, you should also ask your consultant or GP to refer you and your partner to counselling.

Siblings

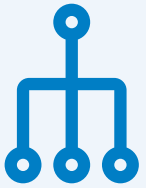
Your whole family will be affected when you get the diagnosis of DMD.

As well as dealing with your own emotions, you will need to cope with the feelings of your other children.

Siblings can often feel guilty for not being ill. They may feel jealous of the time you spend with the child who has DMD and their hospital commitments.

So, it's important to involve your other children as much as possible in the conversations and make time for them too.

You will need to cope with the feelings of your other children.



Siblings can often feel guilty for not being ill.

Ensure you meet with a doctor or researcher for genetic counselling.

Make a list of questions before your appointment.

Genetic counselling

Genetic counselling can give you and your family the information you need to plan for the future. It will help you understand how DMD can be passed on through families.

After your child has been diagnosed, you should ensure you meet with a doctor or researcher for genetic counselling.

It will help you to make informed choices about whether to have another baby, as there's a significant chance that they will either have DMD or be a carrier.

Genetic counselling also gives your extended family the opportunity to think through whether they should have genetic testing.

You may find you can get the most out of your counselling if you make a list of questions before your appointment. Then, you can make sure you cover everything you want to talk about.

Helping your child with DMD

The best thing you can do is make sure your GP refers your child to a multi-disciplinary clinic that specialises in neuromuscular conditions, such as a North Star Centre.

Early treatment and management strategies will give your child a much better chance of preserving their muscle strength and movement.

In this folder, you'll find information about the other things you should do for your child in the first year after diagnosis, including physiotherapy, diet, exercise and medical treatments.

Parents, siblings, extended family and schools need to work with doctors and healthcare professionals to ensure that each child with DMD thrives.

By learning as much about DMD as you can, you'll have the knowledge you need to give your child the best possible care.

> You are not alone - **there's a whole community of other parents who have been where you are now, ready to welcome you with open arms.**

Patient Information Days

Duchenne UK hosts free Patient Information Days for parents and caregivers. You can find out about current medical research, clinical trials and talk with DMD doctors.

Events are also a great opportunity to meet other families who have DMD. Meeting people who are in the same situation as you can be very useful because you can support each other.

You can hear about our events by signing up to our newsletter on our website: **www.duchenneuk.org** or by following us on Facebook or Twitter.

Hope for the future

At the time of diagnosis, you will be faced with the ‘worst case scenario’. Although it is useful to be prepared, it is important to remember that there is hope for the future.

With improved care standards, children with DMD are now living longer than ever before - and having productive, fulfilling lives into their 30s and 40s.

Many families also draw hope from research. It is moving forward at a fast pace with many promising treatments now being tested in clinical trials.

The Duchenne journey may seem overwhelming and challenging at the moment. And it’s true that life as you knew it will never be the same. But things do get better.

Take the time to enjoy activities as a family. Live each day as it comes.

Focus on what your child can do. Encourage your child to develop interests and talents and make friends. Promote your child’s independence. These are important life skills for their future.

You will meet some amazing people in the DMD community, who will give you support and advise you. You will discover an amazing strength within yourself. And you will become your child’s greatest advocate.

> Take the time to enjoy activities as a family. **Live each day as it comes.**

You will meet some amazing people in other DMD families, who will give you support and advise you.

You will discover an amazing strength within yourself. **And you will become your child’s greatest advocate.**

Introduction to Duchenne UK

Duchenne UK is a lean, ambitious and highly-focused charity with a clear vision - to fund and accelerate treatments and a cure for DMD.

We are already making great progress by focusing on three main activities:

£10 million
is committed to medical research

1 Funding research and trials

We fund medical research and clinical trials which increase the scientific understanding of DMD, and develop and test treatments.

We work closely with doctors, scientists, hospitals, pharmaceutical companies and regulators to get treatments approved as quickly as possible. Our aim is to get treatments to patients as fast as we can.

We actively seek out and invest in projects and possible therapies across the globe that could benefit this generation of patients.

2 Accelerating drug development

We actively invest in every stage of drug development to improve and speed-up the drug development process.

We work with healthcare providers, the pharmaceutical industry and governments in the UK and globally to achieve this aim.

We also run the DMD Hub, **www.dmdhub.org**, which supports pharmaceutical companies and hospitals running clinical trials. The Hub also provides patients and their families with information about trials so they can take part in them.

Great advances
have been made

Drive and ambition
comes from our deep understanding

3 Community Engagement and Support

The children and adults with DMD and their families are at the heart of everything we do.

Our drive and ambition comes from our deep understanding of the pain and the challenges that families face. So, we work closely with the Duchenne community and collaborate with other charities to improve the lives of people with DMD.

Why we have produced this folder for newly-diagnosed parents

Duchenne UK is a parent-led charity.

As parents of children with DMD, we all remember the shock, confusion and pain that comes with diagnosis.

Quite simply, we want to help other parents who are going through the same thing that we went through. And we want to offer hope.

We want to give you the information and the support that we would like to have had when we received the diagnosis for our children.

We do hope this folder will be helpful to you. And we hope that we will meet you in person one day soon.

With warmest wishes,

The Parents of Duchenne UK


WORLD'S
STRONGEST
BOYS