YOU HAVE A TEAM ON YOUR SIDE

To help provide the best care for your child, a team of physicians will be there to help:

- Your primary doctor, often a paediatric neurologist, will coordinate your child's care
- Various healthcare professionals may be needed at different stages
- They provide support to maximise quality of life and help maintain independence

When all those involved in a child's care work together, the challenges of Duchenne can be easier to deal with.

FURTHER INFORMATION AND SUPPORT

Receiving a diagnosis of Duchenne can be upsetting, but your healthcare team is there to help. They will be able to offer information and support as you need it, so remember to discuss your concerns with them. Contact with a support group or patient organisation may also be of help.

Whatever your situation, support is available to help you and your family plan for a positive future.

Here are some useful resources:

*Duchenne and You** *www.duchenneandyou.eu*

Insert local online resources

PTC THERAPEUTICS

*Duchenne and You is developed and funded by PTC Therapeutics.

This information given in this leaflet is not intended to replace medical advice. Always refer to your healthcare professional for any questions you may have about Duchenne.

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PTC THERAPEUTICS

Images used are of fictional patients

This leaflet is developed and funded by PTC Therapeutics, Ltd

DUCHENNE MUSCULAR DYSTROPHY

Duchenne muscular dystrophy (also called DMD or just Duchenne) is a muscle condition caused when the body cannot make a protein called dystrophin. This happens because of a change (mutation) in the gene for dystrophin.

Duchenne is a rare condition that mainly affects boys, with one in 3,600–6,000 being born with it. Around a third of all cases are due to a spontaneous mutation, which cannot be predicted or prevented.

A gene can be thought of as being an instruction guide on how to make a protein.

Because Duchenne can be caused by different types of gene changes, different treatments may be needed.

HOW LACK OF DYSTROPHIN LEADS TO THE SYMPTOMS OF DUCHENNE

Dystrophin acts as a shock absorber or stabiliser for muscles. Without dystrophin, muscle is more sensitive to damage and deteriorates over time. The injured muscles are gradually replaced with fat and scar tissue.

The lack of dystrophin and resulting loss of muscle lead to the symptoms of Duchenne.

WHAT TO EXPECT WITH DUCHENNE

The symptoms of Duchenne are different for each child and the rate at which symptoms worsen varies.

As time goes on, the symptoms of Duchenne will change:

- As the muscles get weaker, the ability to walk is slowly lost
- As the back muscles become weaker, the spine may begin to curve (called scoliosis)
- In late childhood to early adulthood, the upper body and arm muscles also weaken
- In late childhood to early adulthood, the upper body and arm muscles also weaken
- The heart and respiratory (or breathing) muscles also weaken over time, which can lead to complications

As Duchenne is a progressive disease, meaning the symptoms become more apparent over time, it is important to get your child the right treatments and management as soon as possible. This gives your child the best chance of maintaining their independence and quality of life for as long as possible.

HOW IS DUCHENNE TREATED

Even though Duchenne is not curable, management and treatment options are available:

- Physiotherapy to help maintain muscles and joints
- Medications to reduce symptoms of Duchenne
- Other therapies that are specific to the mutation causing Duchenne
- Potential enrollment into clinical trials for emerging therapies

An understanding of the specific mutation causing Duchnenne is important to make sure your child is on the right management pathway. This is done through genetic testing and should be completed as early as possible.