UNLOCK NDM

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What you need to know about Non-Dystrophic Myotonia (NDM)

Information for patients

Booklet produced by

LUPIN HEALTHCARE

Contents

Welcome

Welcome to this non-dystrophic myotonia information booklet. If you are looking through this booklet, it is likely that you (or someone you know) have a condition called **Non-Dystrophic Myotonia (NDM).**

You may have many questions about your condition, such as ‘how will my NDM continue to affect me?’ and ‘how will my healthcare team and I manage my NDM?’

This booklet will help you understand NDM, its symptoms and management options. It is not intended to replace a thorough discussion with your specialist doctor (neurologist). The information provided in this booklet will also give you a better insight into discussions you will have with your healthcare team.

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What is non-dystrophic myotonia?

NDM – what is it and how does it affect me?

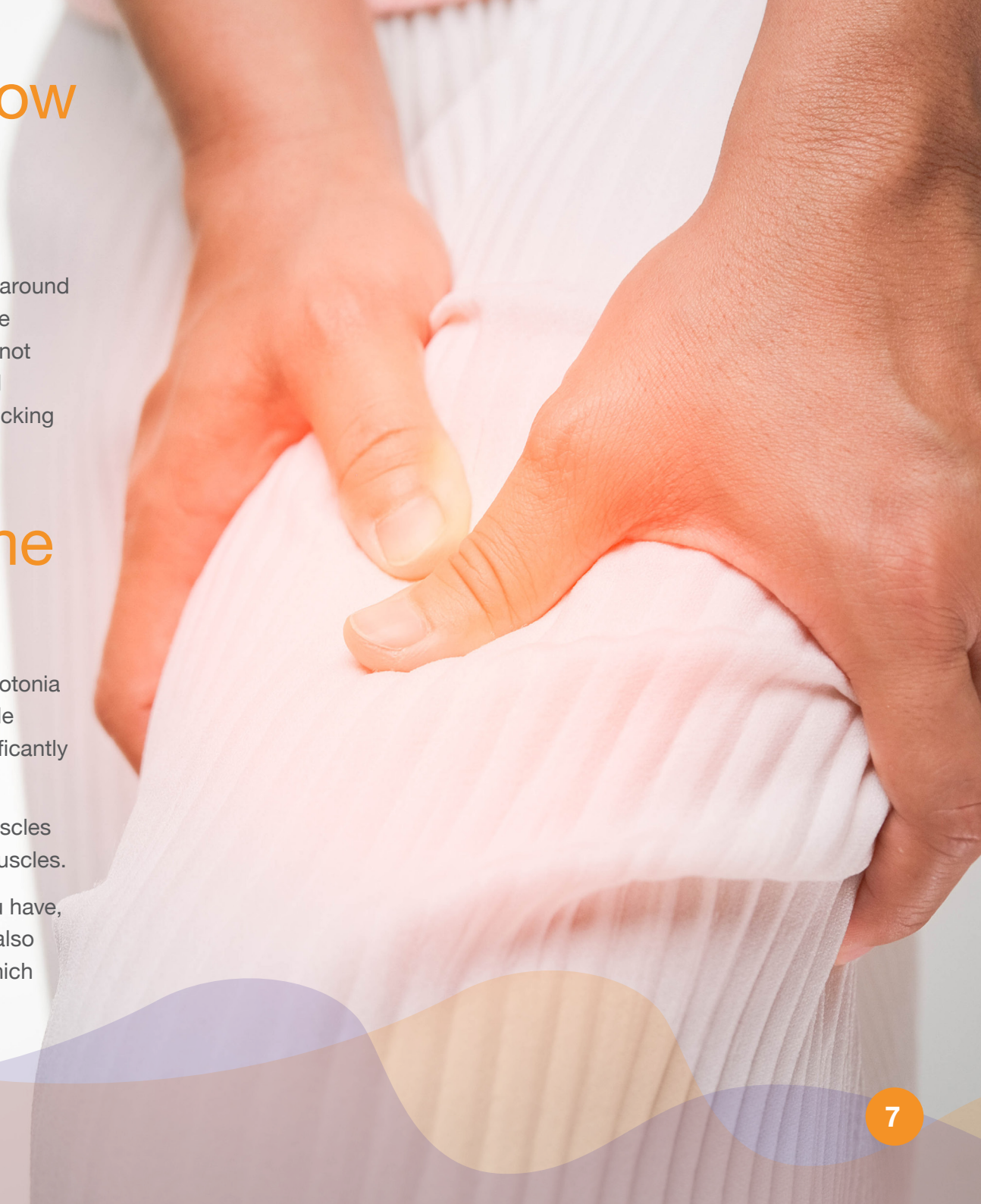
NDM is a group of rare conditions that affects your muscles. Only around 1 in 100,000 people worldwide have these conditions, but there are countries where NDM is more common. In all NDMs, muscles are not able to relax immediately after they have been used – this is called myotonia. You may experience myotonia as stiffness, cramps or locking of your muscles during everyday tasks and movements.

Is my myotonia the same as everyone else's?

The simple answer is 'no'. Your myotonia will be unique to you. Myotonia symptoms may range from subtle effects on muscles that have little impact on the ability to move, to more severe symptoms that significantly affect daily activities.

There are several different forms of NDM. Some mainly involve muscles of the face, tongue and hands, while others involve leg and arm muscles.

Even though your doctor will have told you which type of NDM you have, the range of symptoms you experience will be unique to you. It is also likely that your symptoms will vary from time to time in terms of which muscles are affected and by how much.



How might my NDM affect my daily life?

Depending on your symptoms, NDM may make aspects of your daily life challenging. Some individuals find that their symptoms increase as they get older *(Figure 1).*

Despite this, many individuals develop strategies to help them cope with the daily impact of NDM and lead happy and fulfilling lives. Over time, these strategies may need to change though, and eventually may not be enough.

NDM can impact everyday life

Negative emotions

- Feeling restricted
- Tired all the time
- Worries due to not knowing what's coming
- Pain and cramping

Social anxiety

- Unable to release handshake
- Difficulty eating
- Problems speaking

Lack of independance

- Driving difficulties
- Needing help with tasks
- Locking during movement

Issues with daily living

- Challenges dressing and limited (manual) skills
- Stiffness
- Problems climbing stairs

Reduced activity

- Driving difficulties
- Work duties can be challenging
- Difficulty crossing streets

Figure 1.

Why does Non-Dystrophic Myotonia occur?

Muscles require messages from the brain conducted via nerves in order to work. When a message from the brain reaches the muscles, microscopic channels in the muscle cell wall – called sodium and chloride ion channels – work to turn this message into a very brief electrical signal in the muscle cell. It is this signal that causes the muscle to work (tighten; contract). When the signal ends the muscle relaxes.

In NDM, changes in the sodium or chloride ion channels caused by a gene defect mean that, instead of a single signal, each message from the nerve causes repeated signals in the muscle cells. These repeated signals cause the muscles to remain tensed up and it can take much longer for them to relax ***(Figure 2)***. This can lead to difficulties in moving around and may even affect your balance.

You may experience your NDM in a variety of ways, such as stiffness, cramps, difficulty releasing a fist, problems with chewing and swallowing, difficulty navigating stairs or standing up from a sitting position, difficulty opening the eyelids or a range of other similar problems. Many people with NDM also experience pain.

Other factors such as exercise, cold weather, certain foods, shock, stress and periods of rest and inactivity may make your NDM worse. Your pattern of experiences will be unique to you and will probably differ from day to day. This is normal with NDM.

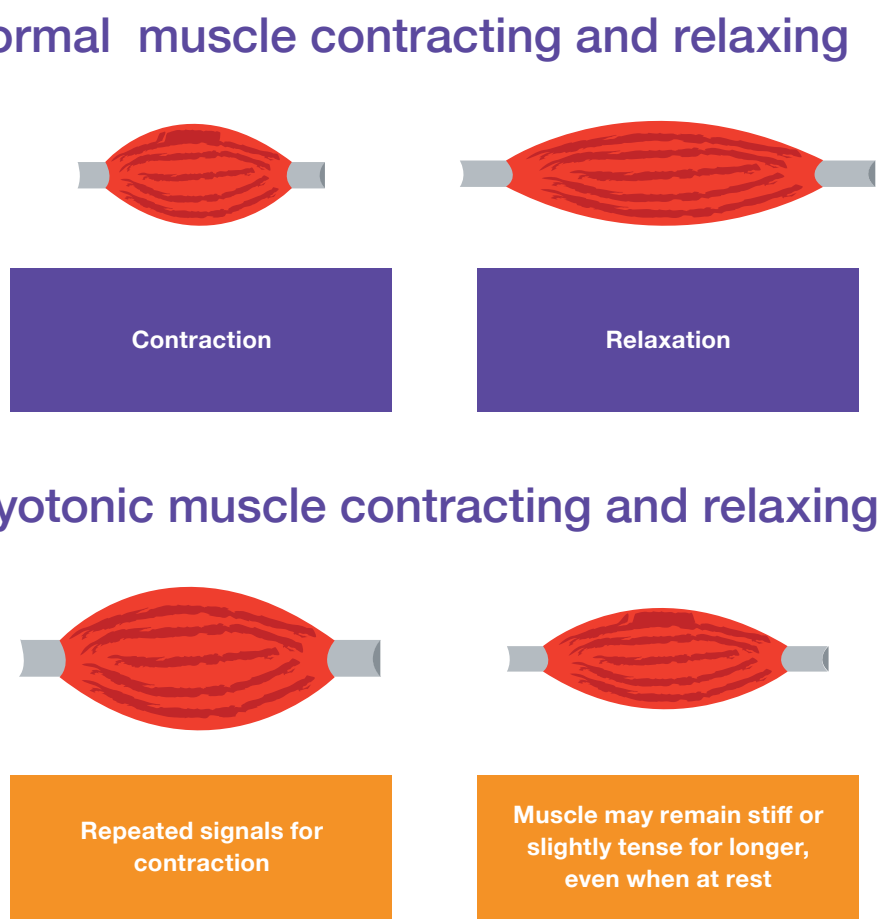


Figure 2.

In addition to delays in muscle relaxation, NDM can also be associated with an increase in muscle size.

What are the different types of NDM?

There are 4 main types of NDM:

- **Recessive myotonia congenita**
- **Dominant myotonia congenita**
- **Paramyotonia congenita**
- **Hyperkalemic periodic paralysis (hyperPP)**

The types of NDM reflect different problems in the genes that tell the body how to make the sodium or chloride ion channels that control the muscle's ability to work and then relax.

Another difference between types of NDM is how warming up affects muscles before any activity.

Some patients with recessive myotonia congenita and dominant myotonia congenita find warming up can reduce their symptoms, but some patients with paramyotonia congenita often find that muscle warm-up can make their symptoms worse.

Non-dystrophic myotonia (NDM)	Includes Becker myotonia, Thomsen myotonia, paramyotonia congenita (von Eulenberg) and other rare sodium channel defects
Myotonia congenita	Includes Becker and Thomsen myotonias
Recessive myotonia congenita	Also called Becker myotonia congenita, Becker disease, generalised myotonia, recessive generalised myotonia or autosomal recessive myotonia congenita
Dominant myotonia congenita	Also called Thomsen myotonia congenita or autosomal dominant myotonia congenita
Paramyotonia congenita	Also called von Eulenburg's disease, paralysis periodica paramyotonia, paramyotonia congenita of von Eulenburg, PMC
Potassium-aggrevated myotonia	Also known as sodium channel myotonia (SCM) or simple myotonia
Hyperkalemic periodic paralysis	Also known as HyperPP

Which type of NDM do I have?

Your doctor will review all of the information they have gathered about your symptoms and tests performed during your visits, and will be able to tell you which form of NDM you have. However management of your symptoms will probably be similar to that for all types of NDM. The aim will be to reduce how your symptoms impact your daily life as much as possible.

If you have been diagnosed with paramyotonia congenita or dominant myotonia congenita, you are much more likely to experience worse symptoms in your arms or hands. In contrast, if you have recessive myotonia congenita, your leg muscles are more likely to be the ones in which the myotonia is worse. However you can experience these symptoms anywhere in your body regardless of which type of NDM you have *(Figure 3).*

Types of NDM

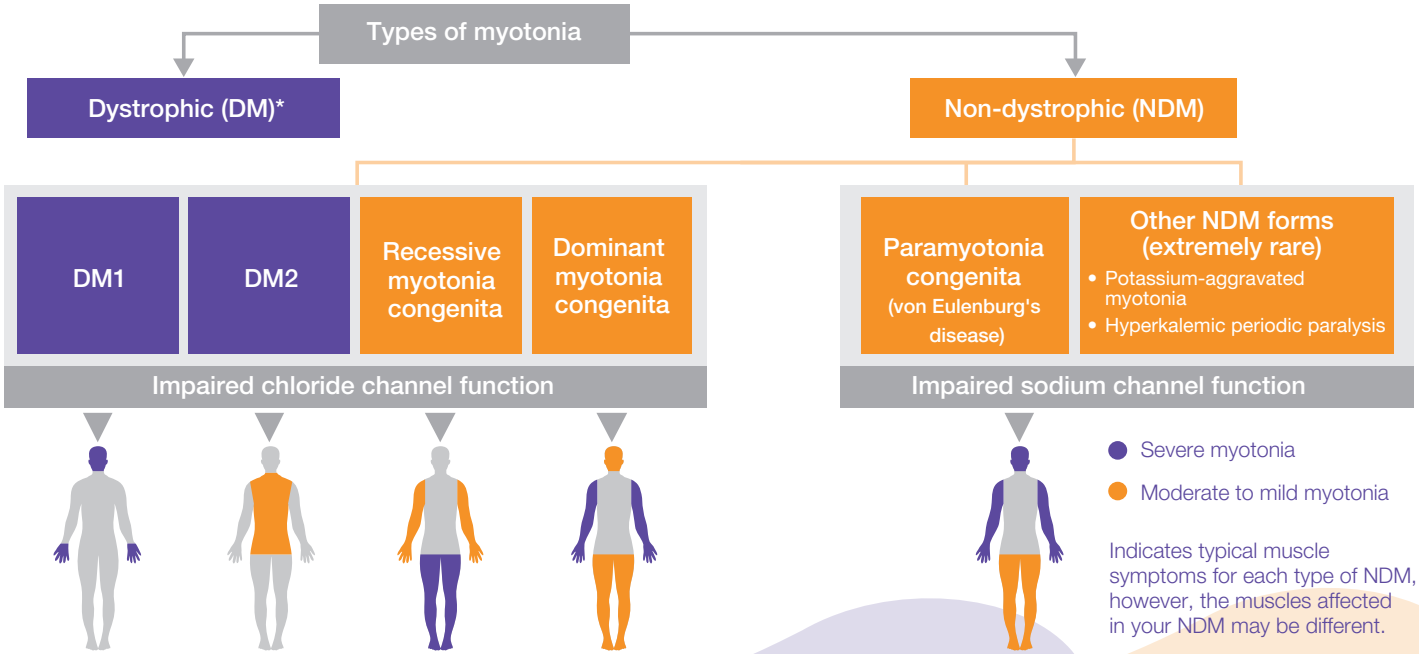


Figure 3.

Indicates typical muscle symptoms for each type of NDM, however, the muscles affected in your NDM may be different.

*Dystrophic myotonia type 1 (DM1) and type 2 (DM2) are a separate group of disorders that are not discussed in this booklet.

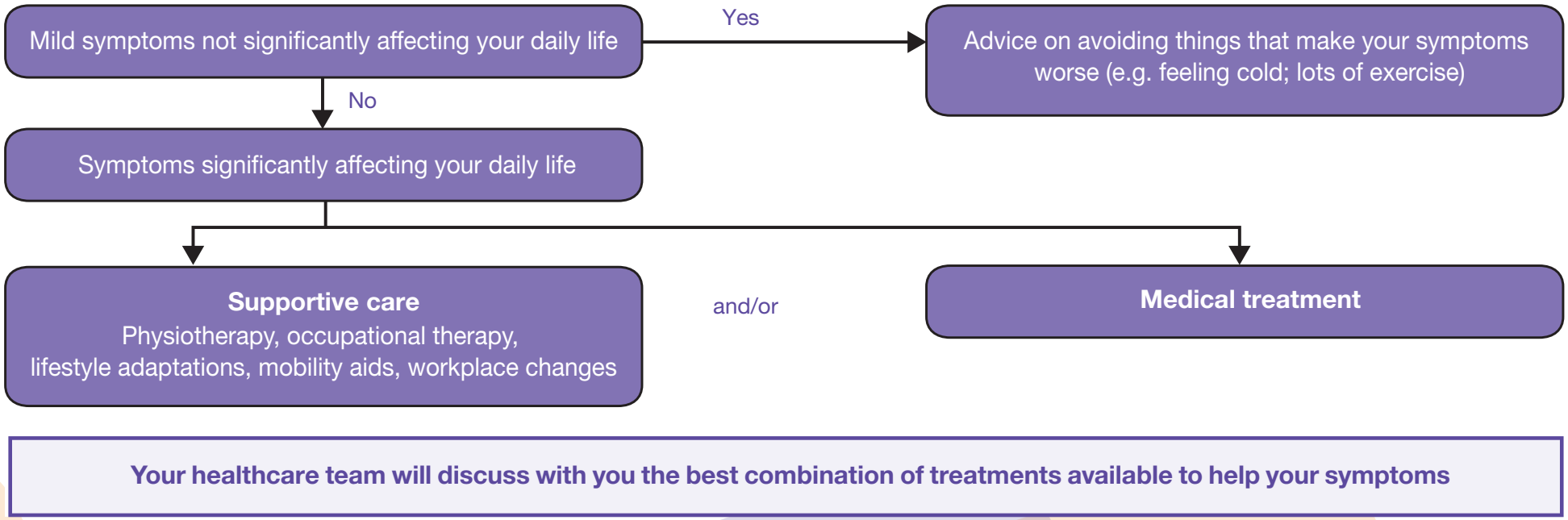
How will my healthcare team and I manage my NDM?

The main goal you will have is to manage your NDM symptoms so that you may live an easier life without having to find ways to work around them.

Depending on how much your symptoms affect your daily life, your healthcare team may suggest physiotherapy, lifestyle adaptations or drug treatment. In some cases, a combination of these options will have the best chance of positively affecting your daily life ***(Figure 4)***. You should discuss with your healthcare team what management options will help you to have the best possible relief from your daily symptoms.

NDM treatment pathway

Figure 4.



NDM: answering other questions you might have

Will NDM reduce how long I live?

No, there is no evidence that people with NDM have, on average, a shorter lifespan than people without the condition.

Will my NDM get worse over time?

NDM is generally not considered to get worse over time. However, in one study of people with NDM, around 6 out of 10 indicated that the severity of their NDM symptoms increased over time.

What symptoms may I experience?

You may experience myotonia in a variety of ways, including stiffness, cramps, difficulty releasing a fist, problems with chewing and swallowing, difficulty navigating stairs, difficulty opening the eyelids and/or standing from sitting. While pain is not always mentioned as a symptom of NDM, many people with NDM experience pain. Depending on the type of NDM you have, you may be more likely to experience symptoms in your arms or hands (dominant myotonia congenita or paramyotonia congenita/ von Eulenburg's), or in your legs (recessive myotonia congenita).

How common is it? Why me?

NDM is a rare disease that is passed down in families through their genes. The condition is caused by errors in the genes that build chloride or sodium ion channels for muscle cells. These channels control the muscle working and then relaxing. NDM caused by defects in the chloride ion channel affects around 1 in 25,000 people (recessive myotonia congenita) to 1 in 400,000 people (dominant myotonia congenita). NDM caused by sodium ion channel defects (eg. paramyotonia congenita/von Eulenburg's) is less common, with around 1 in 250,000 people affected.

Will my children get NDM?

NDM is caused by a range of different errors in a person's genes. These errors can be passed down between generations. The risks of passing on the NDM faulty gene are higher for some forms of NDM than for others. If you are planning to have children, you should discuss the risks with your healthcare team.

NDM: answering other questions you might have

Is there anything I can do to cure it?

Unfortunately, while there are no treatments that will cure your NDM, talk to your doctor about managing your symptoms. You should discuss available treatment options with your healthcare team to find the treatment, or combination of treatments, that will allow you to have the best possible mobility and improve your day-to-day quality of life.

What are coping mechanisms? What works for others?

Everyone experiences their NDM in a unique way, depending on the severity of their symptoms and which parts of their body are most affected. You may have developed a number of ways of coping with your NDM. In some cases, parents with NDM teach their children how to 'hide' their symptoms. It is important to remember that what works for one person may not work for another. Coping mechanisms can range from avoiding key triggers for particular symptoms (e.g. cold, stress, exercise, certain foods such as bananas and potatoes) to lifestyle adaptations that might reduce the daily impact of your symptoms.

Can I work? Can I drive?

Your ability to work will depend on the type of job you have and how severe your NDM symptoms are. Your ability to drive may also be affected by your NDM symptoms and their impact on your movements. In both cases, you should discuss these issues with your healthcare team.

Where can I go to talk about it? Are there others like me?

NDM is a rare disease, but there are other people affected like you are. The internet and social media will likely have some patient forums that you might find useful. Some web resources are listed later in this booklet.

If I'm anxious or depressed, what can I do?

NDM can have a great impact on your overall well-being because of the frustrating nature of the limitations it may put on your everyday life or cause anxiety about displaying symptoms of NDM in public. If you feel anxious or depressed about your NDM, please discuss this with your healthcare team.

My NDM diary

Since NDM is unpredictable, your symptoms may be better or worse than normal when you visit your doctor. It is helpful for your healthcare team to have as much information as possible about your daily experiences with symptoms in order to optimise your care.

You might find it helpful to use these pages to record days when your symptoms are particularly bad, when new symptoms occur or if you have any questions you would like to ask your healthcare team at your next visit.

Please see the first diary entry below as an example.

Date:	Description:
31 June	Stiff legs worse than usual

Notes:

Made getting up out of seat difficult and missed my stop on the bus.

NDM experience:

Everything feels worse today.
MUST TELL DOCTOR!

Date:	Description:

Notes:

NDM experience:

Date:	Description:

Notes:

NDM experience:

Date:	Description:

Notes:

NDM experience:

Date:	Description:

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NDM experience:

Support and where to go for information

Hospital Contact Details

Hospital Name:
Address:
Postcode:
Tel:
Email:
Name of specialist doctor / nurse

Web Resources

Unlock NDM UK website: **https://www.unlockndm.uk/**

Muscular Dystrophy UK: **https://www.musculardystrophyuk.org/**

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www.unlockndm.uk

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