



**DMD
CARE UK**

Best care for all

Patient information:

Respiratory Guidelines



Duchenne
UK

dmdcareuk.org

This guide is for older children or adults living with DMD and for parents or carers. It refers to 'you' throughout but this may apply to 'your child'.

It is also important to realise that many of the symptoms discussed are usually a feature of later stages of Duchenne muscular dystrophy, but onset of symptoms varies for everyone.

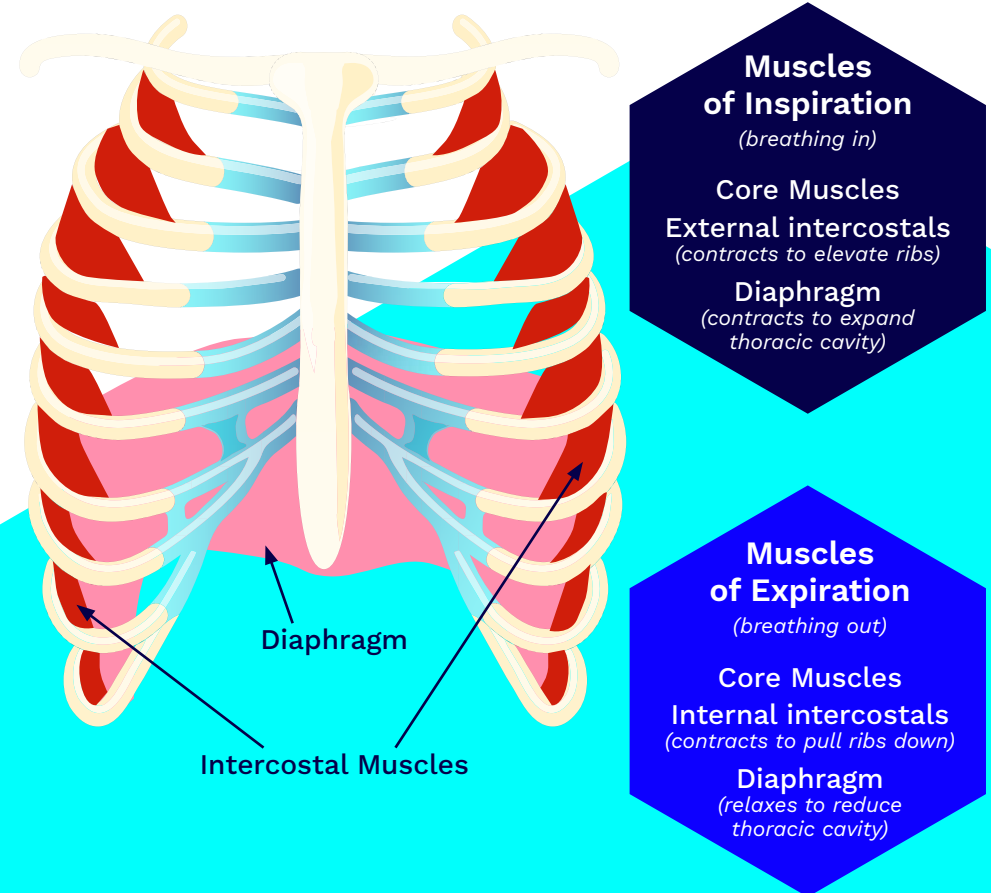
DMD Care UK has brought together experts in the management of neuromuscular diseases and respiratory conditions to agree the best standards of respiratory care in the UK. Respiratory care monitors and treats issues that might arise because of weakness in the breathing muscles for people with Duchenne muscular dystrophy (DMD). These clinical recommendations have been published in *Thorax* (2023, open access), and are based on the latest evidence, expert-opinions, and the patient perspective. They have been endorsed by the British Thoracic Society.

This booklet is designed to help DMD patients and parents/caregivers better understand these clinical recommendations for their respiratory care.

1. How the respiratory system is affected in DMD

In DMD, a lack of a protein called dystrophin means that all muscles weaken over time. This includes the respiratory muscles that control breathing and coughing.

One of these muscles is the diaphragm. The diaphragm is below the lungs and works to pull air into the lungs (fig 1). The lungs are not directly affected by DMD, but the muscles around them are.



2. When is breathing affected

The age when respiratory muscles are affected in DMD varies, but it is usually not until the late teens. Younger teenagers and children with DMD don't usually have respiratory symptoms. However, weakening respiratory muscles are part of the natural progression of the disease which varies for everyone. This means we need to look more closely at the breathing muscles in older teenagers and adults, especially when they become non-ambulant (not able to walk).

Early monitoring and ongoing management, including chest physiotherapy, helps to keep the respiratory system healthy for as long as possible. It also means that doctors can spot problems as early as possible and treat them.

3. How do we monitor respiratory function?

Monitoring the respiratory muscles involves the neuromuscular team checking:

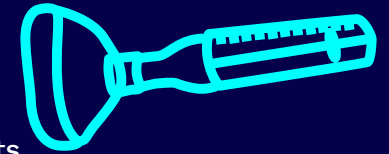
- How often you get chest infections
- How well you can cough up secretions (phlegm) produced in the lungs (peak cough flow) – see box 1
- Whether you have problems with swallowing
- For symptoms of breathing problems whilst sleeping (sleep studies)
- How well you are walking (ambulatory status)
- Lung function tests (pulmonary function tests or PFTs) – these include measuring forced vital capacity to measure how much air the lungs can breathe in and out – see box 2

More details about some of these tests are given in the boxes below. The tests will normally be carried out by your neuromuscular doctor or physiotherapist. You may also be referred to a respiratory specialist with experience in DMD for more assessments and/or treatments.

1

Peak Cough Flow (PCF)

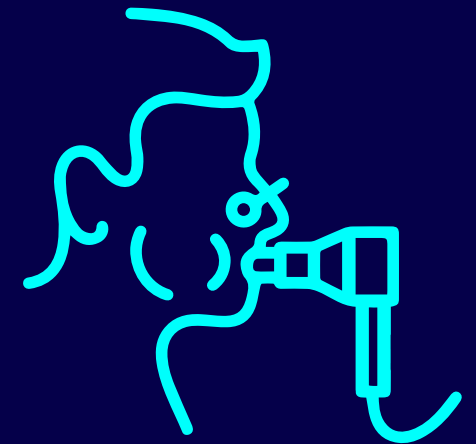
The ability to cough is important to keep airways clear, especially of phlegm. As the breathing muscles get weaker, the cough will also get weaker. Doctors and physiotherapists measure the strength of cough by checking the Peak Cough Flow (PCF). This is done asking you to 'cough' through a mouthpiece attached to a flow meter. The level of PCF will be compared to what is 'normal for you'. If it has gone down, you may be referred for more assessments.



2

Forced Vital Capacity (FVC)

This is a measure of the amount of air you can breathe out (exhale) as hard and quickly as possible after breathing in (inhaling) as much as you can. It is measured using a spirometer. It will be recorded in litres and as a '% of predicted value'. Predicted value is the amount that would be expected for your weight and height.



4. How often should respiratory function be checked in DMD?

Although it is unusual to see breathing problems until you are no longer walking independently, it is important to start routine respiratory (breathing) checks from around six years of age.

This is so that:

- You can get used to ‘doing’ the lung function tests so they give accurate information about your breathing and coughing later on
- You get a ‘baseline’ for respiratory function (to understand what is normal for you)
- Any problems can be spotted and treated as early as possible
- You know what to look out for as you grow

People with DMD who are ambulatory (able to walk) should have checks every 6-12 months. People who are non-ambulatory (not able to walk) should have checks at least every six months. You may need more frequent checks if you have any symptoms suggesting problems with your chest or breathing muscles – this includes frequent chest infections.

5. How can we delay respiratory problems?

The following steps can help keep the respiratory muscles working well for as long as possible:

- Follow the recommended care for the heart and bones: both heart muscle weakness and scoliosis (curved spine) can make respiratory muscles weaker

- Prevent respiratory illness by getting vaccines recommended by the government, including an annual flu vaccine, COVID-19 and pneumococcal vaccines as advised (see Box 3)
- Treat chest infections quickly with antibiotics
- Using high doses of corticosteroids has been shown to slow down changes in the respiratory muscles caused by abnormal dystrophin levels. These are prescribed to most children with DMD (see Box 3)

3

Important information about steroids

ADRENAL SUPPRESSION

Long-term steroids cause adrenal suppression and mean you are at risk of adrenal crisis during illness, injury or stress. This is a serious condition. It is particularly important to follow the advice on stress dosing in your own emergency care plan as outlined in the endocrine guidance:

<https://dmdcareuk.org/information-and-guidance>

VACCINES

Some vaccines (varicella and nasal influenza) are ‘live’ vaccines and contain inactivated viruses, whereas others are based on key viral proteins. When someone is taking immunosuppressive drugs like steroids, they may have a different response to live vaccines – potentially not developing the same immunity to the vaccine or experiencing symptoms of viral infection. We therefore recommend avoiding live vaccines when on higher doses of corticosteroids.

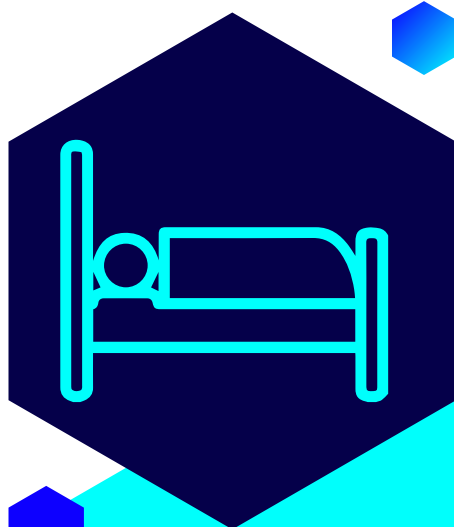
There is more specific advice about how to calculate whether it is safe to have a live vaccine in the government’s ‘green book’, and your GP and neuromuscular team should follow this guidance.

6. How does sleep affect respiratory function?

Changes in respiratory function are often seen at night when the breathing rate is typically lower and the 'body clock' is in sleep mode. The first signs of a decline in respiratory function can be very subtle and cause disrupted sleep (sleep disordered breathing) without you realising there is a problem, as everything seems fine during the day (see Box 4).

You should be monitored for signs and symptoms of sleep disordered breathing at all ages. Problems are more likely if people are overweight or have cushingoid features (a round face and puffy cheeks as a side-effect of steroid use).

If you notice any of the symptoms listed, report it at your next clinical appointment. Your neuromuscular team should refer you to a specialist respiratory service to assess these and advise you on further management. You will probably have a sleep study as part of this assessment.



Sleep Disordered Breathing

Symptoms of sleep disordered breathing that you may notice include:

- Excessive daytime sleepiness
- Poor concentration
- Disturbed sleep with frequent waking (possibly with a racing heart or breathlessness)
- Sleep that is not refreshing
- Stopping breathing for brief periods whilst sleeping, snoring or shallow, irregular breathing (sleep apnoea)
- Morning headaches (typically a throbbing headache that settles by mid-day)
- Poor appetite, especially in the morning
- Irritability

Note that some people may find it hard to report these symptoms due to **communication or learning difficulties**. However, you might notice changes in daily routine and/or behaviour.

It is a good idea to make a log of these issues if you notice them so that you can discuss with your neuromuscular team.

Sleep Studies

You may be referred for an overnight sleep study to check your breathing patterns through the night whilst you sleep normally. This can be at home or in hospital. Depending on the results, your specialist team will recommend any extra help or treatment needed to improve your breathing when you are asleep and reduce your symptoms. This might include night-time ventilation.

Sleep studies are important because they allow you and your doctors to make the best treatment decisions for you – if you are not breathing well at night, your ability to do the things you want to during the day can be affected.

7. Treatment and referrals

Your neuromuscular doctor should make a referral for you to a respiratory service experienced in management of DMD if there are:

- Signs of sleep-disordered breathing
- Frequent chest infections
- Abnormal or worsening respiratory function test results (for example a reduced FVC)

If you have frequent chest infections, it is important to check that this is not because you are accidentally swallowing food or liquid into your airway (aspiration). This usually causes coughing or choking but can sometimes happen ‘silently’ without you realising. If there are concerns about aspiration, you will be referred to a speech and language therapist for a more detailed assessment and advice on your swallowing.

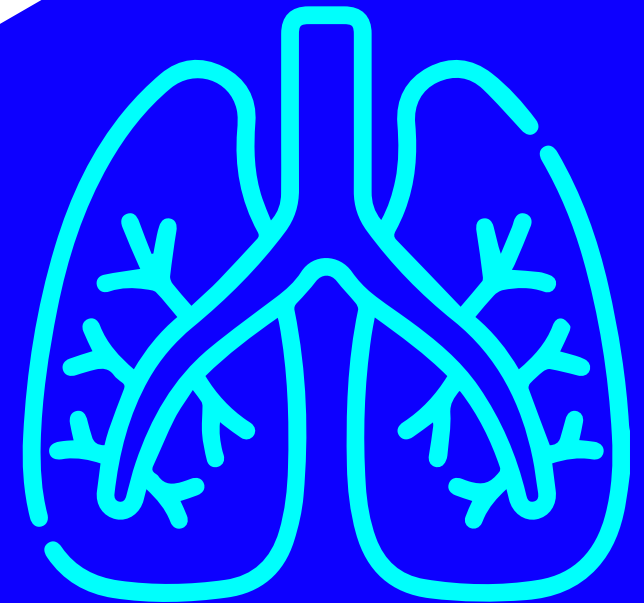
If you have a weak cough or difficulty clearing the phlegm (secretions) from your chest, you should be referred to a specialist respiratory physiotherapist who can advise on how to clear your airways more effectively. They may recommend you use specific equipment like a cough-assist machine.

8. Ventilation

As you get older and especially as walking becomes more difficult, you are likely to need some support with breathing and coughing. This will help you sleep better, keep your airways clear, keep your lungs healthy for longer and reduce the risks of infection. Ventilation can also improve your ability to keep active and do the things you want to do. There are many different forms of ventilatory support and you can ask your respiratory team to explain these in detail.

You may find information provided by Parent Project Muscular Dystrophy useful here too:

<https://www.parentprojectmd.org/care/care-guidelines/by-area/care-for-lung-muscles/assistive-devices-for-breathing/>



9. Warning signs and what to do in an emergency

Children and adults with reduced respiratory function can get worse very quickly during an infection. You should have an Emergency Health Care Plan outlining what to do if you become unwell with an infection or other illness and require treatment in hospital.

You can also download Duchenne UK's In Case of Emergency information to help you be prepared:
<https://dmdcareuk.org/emergency-support>

A person with DMD may not show expected signs during respiratory difficulties. They may not show laboured or more rapid breathing (e.g. sucking in of the throat and the chest wall) because of muscle weakness.

If there is an unplanned or emergency admission to hospital as a result of a chest infection or breathing issues, a child or adult with DMD should have an urgent assessment by a physiotherapist with experience in airway clearance.

If specialist-assessment of respiratory function shows any problems:

- Care should be led by a specialist respiratory team
- Medical teams should be cautious about using extra oxygen without careful monitoring as it can affect the body's drive to breathe
- Those who have significant breathing difficulties or abnormal gas exchange should be offered breathing support (ventilation). This will usually be delivered through a mask if possible and should be done in a critical care/specialist respiratory unit

If in doubt about the treatment being offered, ask the Emergency Department team to contact your neuromuscular specialist or respiratory consultant if you have one.

10. Other issues to consider if you have sudden changes in your breathing

People with DMD are at risk of a **heart muscle disease** called dilated cardiomyopathy. Cardiac function should always be reviewed in a patient with DMD developing respiratory symptoms. This is important whether these symptoms have come on gradually over time or more suddenly.

Be aware of the risk of fat embolism syndrome (FES) if there are sudden respiratory symptoms that get worse quickly, especially after a fall or stumble. FES happens when small droplets of fat get into the bloodstream after a bone fracture and can block blood flow. Symptoms can be misinterpreted as agitation or panic (see Box 5). FES is rare but it is a medical emergency.

5

Symptoms of Fat Embolism Syndrome (FES)

Symptoms below may be a sign of rare but dangerous FES and should not be ignored:

- Rapid less effective breathing
- Drowsiness
- Pinpoint rash
- Confusion
- Not 'seeming like yourself'

FES is rare and normally caused by a bone fracture releasing fat droplets into the bloodstream. In DMD it is still a risk even if a fracture is not obvious.

About DMD Care UK

DMD Care UK is a nationwide initiative to ensure every person living with Duchenne muscular dystrophy (DMD) in the UK has access to the best care.

This project is funded by Duchenne UK, Joining Jack and the Duchenne Research Fund. They work closely with the John Walton Muscular Dystrophy Research Centre in Newcastle and in collaboration with the North Star Network, funded by MDUK.

DMD Care UK has produced a series of information resources for UK DMD patients, families and other non-specialists on the recommended standards of care for DMD.

Find out more at dmdcareuk.org

Do you have questions or feedback about this booklet? Get in touch with support@duchenneuk.org



Notes

You may find it useful to jot down questions, thoughts or useful information here. We have included a few suggestions below. You can also find a glossary of terms here: <https://dmdcareuk.org/glossary>

Name and contact details of your neuromuscular specialist:

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Name and contact details of your respiratory specialist/s (this may include a respiratory physiotherapist):

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This booklet has been developed by DMD Care UK and reviewed by clinicians and the family focus group within the project. It is based on the DMD Care UK recommendations endorsed by the British Thoracic Society.

This booklet is for informational and educational purposes only. You should always discuss your medical care with your clinical team.