



Making breathing easier

Breathing difficulties can affect some individuals with neuromuscular disorders. Simple measures can be taken to reduce these problems, and in many situations it is possible to provide excellent control of symptoms.

Breathing problems include an increased susceptibility to chest infections, difficulty coughing and clearing phlegm, breathlessness, and under-breathing (known as hypoventilation) particularly during sleep. This factsheet provides background information on how families can help in the management of respiratory problems and identify symptoms which require investigation.

Who develops breathing problems, and at what age?

Respiratory muscle weakness is relatively common in most neuromuscular conditions and is inevitable in the late stages of Duchenne muscular dystrophy. The age at which respiratory problems develop varies enormously in different conditions. Breathing difficulties can arise shortly after birth in children with severe Type 1 spinal muscular atrophy and during the first few months of life in some other myopathies. Individuals with congenital muscular dystrophy or Type 2 spinal muscular atrophy may run into breathing problems in childhood. Young men with Duchenne muscular dystrophy tend to develop symptoms of nocturnal hypoventilation aged 15-20 years, and respiratory muscle involvement in conditions such as limb girdle muscular dystrophy and acid maltase deficiency may not occur until adulthood.

Respiratory muscle strength can be very variable within and between neuromuscular disorders, therefore investigations and treatment must be tailored to the individual. Long-term management of breathing problems should always be medically supervised.

Recent research (*Neuromuscular Disorders* November 2002) shown that non-invasive nocturnal ventilation can have a significant effect on extending the life span of children with Duchenne muscular dystrophy (DMD). Overall, better co-ordinated care has probably improved the chances of survival to 25 years from 0% in the 60s to 4% in the 70s and 12% in the 80s. Nocturnal ventilation has increased this possibility to 53% in the 1990s, and these figures are continuing to improve all the time.

How do breathing problems arise?

When we breathe, certain muscles act as bellows to expand our lungs; these are called inspiratory muscles. They cause oxygen to be drawn into the lungs. The most important inspiratory muscle is the diaphragm. Weak inspiratory muscles reduce lung volume.

This factsheet is under review, due for updating later in 2017. If you have any queries, please contact us.

Version: 2. Published: August 2007. Updated: April 2008. Date of review: November 2011 /
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Breathing out the waste gas (carbon dioxide) from the lungs is known as expiration. This is usually passive and does not require particularly strong muscles, but coughing does require effective contraction of these expiratory muscles and normal functioning of the upper airway (bulbar) muscles. Scoliosis (a curved spine) reduces lung volume even more, and causes the respiratory muscles to contract inefficiently due to the asymmetrical shape of the chest.

After a period of breathing at low lung volumes, the chest wall tends to become stiff and less compliant making it more difficult for the respiratory muscles to expand and draw enough oxygen into the lungs. Children and adults with low lung capacity are prone to chest infections and these are slow to clear because coughing is ineffective. If the swallowing muscles are weak, food may go down the wrong way into the lungs leading to recurrent infections.

During sleep, inspiratory and upper airway muscles normally relax and each breath becomes smaller so the oxygen level in the body goes down. If these muscles are already very weak then the oxygen level becomes even lower, this is known as under ventilation or hypoventilation. In mild cases, hypoventilation does not cause any symptoms and is only noticeable in rapid eye movement (REM) sleep when we often dream. However, if hypoventilation at night progresses it can lead to low oxygen and high carbon dioxide levels during the day.

If upper airway muscles are particularly weak, short episodes of upper airway obstruction may occur during sleep; this is known as obstructive sleep apnoea. Often hypoventilation and obstructive sleep apnoea coexist.

Spotting the symptoms and acting on them

Symptoms of nocturnal hypoventilation include morning headaches, lethargy, breathlessness, disturbed sleep, sweating at night and poor appetite. Erratic noisy breathing during sleep may be observed. In young children, failure to thrive or gain weight is not uncommon. These symptoms should be reported to your doctor.

Breathing problems can also be picked up during routine clinic visits. Vital capacity and overall respiratory muscle strength can be measured by simple blowing tests. Once vital lung capacity is less than 60% predicted and respiratory muscle strength falls below 30% of normal, hypoventilation becomes a possibility and regular checks should be made. Hypoventilation is unlikely if vital capacity is over 60% of normal, other than at the time of a severe chest infection. If problems with breathing during sleep are suspected a 'sleep study' will usually be carried out. Here oxygen and carbon dioxide levels (and sometimes the pattern of breathing) are monitored overnight using small probes attached painlessly to the surface of the body. The sleep study may show a variety of findings. The most common of these is a fall in oxygen level and rise in carbon dioxide particularly during REM sleep due to under ventilation. In others short episodes of stopping breathing (apnoeas) due to either obstruction of a floppy upper airway (obstructive sleep apnoea) or lack of breathing effort (central sleep apnoea) are seen.

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Measures to help avoid breathing problems

Healthy eating

It is important to eat a sensible, balanced diet. Obesity should be avoided as it impedes breathing and increases the work of the respiratory muscles. It may also increase a tendency to obstructive sleep apnoea. Remember that it is always easier to prevent obesity than to lose weight! Constipation leading to abdominal distension is not only uncomfortable but reduces diaphragm movement. It is best dealt with by a good fibre intake, although sometimes mild laxatives are needed.

Influenza and pneumococcal vaccine

The influenza (flu) and pneumococcal vaccines are recommended for individuals with breathing problems. These vaccines are not 100% effective, but will significantly reduce the risk of infection. The influenza vaccine is given once a year, usually in October. The pneumococcus bacterium is one of the most common causes of bacterial pneumonia. The pneumococcal vaccine lasts about five to eight years and in some people does not need to be repeated as protection remains long term. Both vaccines can be given by your GP and are very safe, however individuals who are allergic to eggs should not receive them.

Breathing exercises and physiotherapy

Deep breathing helps to fully inflate the lungs and puts the lungs, respiratory muscles and chest wall through a good range of movement. Assisted coughing or 'huffing' is especially beneficial in clearing secretions at the time of a chest infection. Huffing is achieved by taking a few deep breaths and then forcing the air out as rapidly as possible with the mouth open. Phlegm is shifted from deep within the lungs to the main airways and then can be expectorated more easily. A family member or friend can help expand the chest in this region by placing their hands over the lower rib cage. Your physiotherapist will advise on breathing exercises and other forms of treatment if appropriate, such as postural drainage and chest clapping if secretions are excessive. Cough assist devices such as the In-Exsufflator may be of value in some individuals with a weak cough (see below).

Confidence about breathing can be boosted by breathing control advice and activities such as singing and playing the musical instruments like the recorder. Some respiratory muscle training devices have been developed, but evidence to support respiratory muscle training is not conclusive.

Posture and curvature of the spine

Good posture is essential to allow the rib cage to expand optimally. It requires attention in the sitting, lying and standing positions. For wheelchair users appropriate special seating not only improves posture and comfort but may prevent the development of skeletal deformity. Around half of boys with Duchenne muscular dystrophy develop a significant curvature of the spine (scoliosis) and a scoliosis is relatively common in other neuromuscular disorders which arise before adolescence. Surgical correction of the scoliosis is used in some individuals to stabilise the spine and prevent further loss of lung

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volume. However spinal surgery does not prevent further loss of lung capacity when it is caused by progressive muscle weakness.

Treatments

Steroids therapy in Duchenne muscular dystrophy may help preserve respiratory function. See the factsheet called *Steroids and Duchenne muscular dystrophy (DMD) – some questions and answers*.

Chest infections

Prompt use of an antibiotic is recommended if phlegm is discoloured or copious and the individual feels unwell and/or feverish. Flu and colds are often followed by a bacterial infection so that an antibiotic may be required if symptoms don't clear up. Huffing and chest physiotherapy should be started as soon as possible.

If these measures are not beneficial then further medical help should be sought. Children and adults with a tendency to wheeze may be prescribed a bronchodilator drug such as salbutamol (ventolin) which can be delivered by inhaler, spacer device or nebuliser.

A reserve course of antibiotics at home is useful to provide peace of mind and treat infections swiftly.

Ventilation

Breathing can be assisted by artificial ventilation if simpler medical measures fail to maintain carbon dioxide and oxygen levels, for example during a chest infection or for nocturnal hypoventilation. Nearly always breathing can be supported using a non-invasive system. Ventilation via tracheostomy is only required in individuals with severe swallowing problems, or inability to use non-invasive ventilation. A recent study suggests the best time to start non-invasive ventilation is when the child or adult develops symptomatic nocturnal hypoventilation. Without non-invasive ventilation individuals with nocturnal hypoventilation are likely to develop respiratory failure in the day during the next 1-2 years.

Non-invasive ventilation

This system consists of a closely fitting facemask joined to a portable positive pressure ventilator which augments the patient's breathing, thereby increasing oxygen and reducing carbon dioxide levels. A range of ventilators can be used such as the BiPAP (Respironics), VPAP (ResMed), Breas PV 403, Nippy (B&D Electromedical). Many of these are portable and all are easy to operate. There is also an increasing variety of masks and interfaces. In most individuals ventilation is initially needed only at night. If respiratory muscle weakness progresses then use during the day may be required. Most patients are able to quickly acclimatise to non-invasive ventilation and find that their sleep quality improves. Non-invasive ventilation can produce excellent control of symptoms in adults and children, sometimes over long periods of time. Evidence suggests that non-invasive ventilation may also reduce the frequency of chest infections and decrease hospital admissions. However, if the swallowing muscles are very weak, non-invasive ventilation cannot prevent aspiration and in this situation another form of ventilation (tracheostomy ventilation) that will protect the airway can be considered. Ventilatory options should always be discussed

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on an individual basis, taking into full account the wishes of the patient and family, and the clinical circumstances.

Cough assistance

Physiotherapy can be very helpful in assisting cough and clearing secretions. The efficiency of coughing can be assessed by a cough peak flow measurement. Values in teenagers and adults below 270/minute suggest some weakness of cough and if values are less than 160/minute clearance of phlegm can be a major problem.

In individuals who are already using non-invasive ventilation, physiotherapy when resting on the ventilator can improve the effectiveness of a physiotherapy session and make it less tiring for the child or adult.

Use of an ambu bag can help the individual to take bigger breaths. If these measures are insufficient a cough in-exsufflator device may be valuable in adults and children with frequent chest infections and a poor cough. The cough in-exsufflator provides a large breath in (via mask or mouthpiece) and then applies a negative pressure (suction) with expiration so that secretions are sucked away. The cough in-exsufflator can be used in adults and children (even in children below 1 year old) but there is little experience with these in tiny babies. There is good evidence that the combination of non-invasive ventilation and the cough in-exsufflator can reduce the need for tracheostomy ventilation in Duchenne muscular dystrophy. Devices such as the Percussionaire or 'Vest' aim to vibrate secretions loose but have not been fully evaluated yet.

Other forms of ventilatory support

'Continuous positive airway pressure' (CPAP) is a mask and compressor system which is a form of non-invasive respiratory support but delivers a constant pressure rather than ventilatory assistance. This treatment is helpful in obstructive sleep apnoea as the pressure acts to hold the upper airway open, but is usually not sufficient to deal with nocturnal hypoventilation, or in individuals with respiratory muscle weakness where non-invasive ventilation is almost always preferable. Negative pressure devices such as the tank ventilator, cuirass, and other devices such as the rocking bed were used in the past to treat breathing problems caused by neuromuscular disorders, but have now been mainly superseded by non-invasive positive pressure ventilation. However, negative pressure ventilation is still available for individuals who do not adapt to mask ventilation, and newer negative pressure models such as the Hayek Oscillator may have a role in very young children and some adults. Ventilation via a mouth-piece is a variant of non-invasive ventilation that can be used at night or as a breathing aid during the day.

There is much that can be done now to control respiratory problems in neuromuscular disorders. Although a decline in respiratory capacity is inevitable in some conditions, symptom control produced by treatment such as non-invasive ventilation has been shown to improve quality of life. Children and adolescents using nocturnal non-invasive ventilation are able to return to school or higher education and adults are able to return to their usual daily activities.

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Centres providing respiratory support

There are an increasing number of hospitals which carry out sleep studies and provide respiratory support for patients with neuromuscular disorders. The centres with major interest:

Royal Brompton Hospital, London
St.Thomas' / Guy's Hospital, London
Great Ormond Street Hospital
Hammersmith Hospital, London
Fazackerley Hospital, Liverpool
City General Hospital, Stoke
Glenfield Hospital, Leicester
Papworth Hospital, Cambridgeshire
Newcastle General Hospital
Wythenshawe Hospital, Manchester
St. James Hospital, Leeds
Radcliffe Infirmary, Oxford
Sheffield Hospital
Hospital for Sick Children, Glasgow
Queens Medical Centre, Nottingham

If you have feedback about this factsheet please email info@muscular dystrophyuk.org.

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Here for you

The friendly staff in the care and support team at the Muscular Dystrophy UK's London office are available on **0800 652 6352** or info@muscular dystrophyuk.org from 8.30am to 6pm Monday to Friday to offer free information and emotional support.

If they can't help you, they are more than happy to signpost you to specialist services close to you, or to other people who can help.

www.muscular dystrophyuk.org

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