



## Respiratory System

### Symptoms

#### Respiratory muscle weakness

People with myotonic dystrophy commonly have significant breathing problems that can lead to respiratory failure or require mechanical ventilation in severe cases. These issues may result from muscle weakness (diaphragm, abdominal, and intercostals muscles) and myotonia of respiratory muscles, which lead to poor breathing force and results in low blood oxygen/elevated carbon dioxide levels.

#### Aspiration

Breathing of foreign material, including food and drink, saliva, nasal secretions, and stomach fluids, into the lungs (aspiration) can result from abnormal swallowing. Without adequate diaphragm, abdomen and chest wall coughing strength to remove the foreign material, the inhaled acidic material can cause chemical injury and inflammation in the lungs and bronchial tubes. The injured lungs are then susceptible to infections that can lead to respiratory distress.

#### Sleep apnea

Insufficient airflow due to sleep apnea (periods of absent airflow due to narrow airways and interrupted breathing) can result in dangerously low levels of oxygen and high levels of carbon dioxide in the blood. In mild cases, apnea can cause disrupted sleep, excessive fatigue, and morning headaches. In severe cases, apnea can cause high blood pressure, cardiac arrhythmias, and heart attack.

The respiratory issues seen with myotonic dystrophy vary depending on the form of the disease.

### Patterns of Respiratory System Problems

Congenital DM1
<p>Prenatal</p> <ul style="list-style-type: none"> <li>• Failure of cerebral respiratory control, which may result in fetal distress</li> <li>• Pulmonary immaturity, which may be further complicated by premature birth</li> </ul>
<p>Newborn</p> <ul style="list-style-type: none"> <li>• Respiratory insufficiency due to a combination of weak diaphragm and intercostals muscles, pulmonary immaturity, and failure of cerebral respiratory control. Severe cases may require mechanical ventilation for extended periods. Respiratory issues are the principal cause of death in newborns with congenital myotonic dystrophy DM1.</li> <li>• Weak facial and esophagus muscles that may lead to sucking and swallowing problems, which can allow fluids to enter the lungs and result in chemical injury to the respiratory passages, chronic lung inflammation, and aspiration pneumonia.</li> </ul>
<p>Childhood/Adolescence</p> <ul style="list-style-type: none"> <li>• Weakness of the diaphragm, abdomen and chest wall muscles affecting the ability to cough, resulting in chronic lung infections, chronic bronchitis and bronchiectasis (abnormal stretching and enlarging of the bronchial tubes, which remain chronically infected)</li> <li>• Chronic upper airway infections, which potentially can lead to hearing loss at a young age</li> <li>• Weak facial and esophagus muscles that may lead to swallowing problems, which can allow fluids to enter the lungs and result in chemical injury to the respiratory passages, chronic lung inflammation, and aspiration pneumonia</li> </ul>



<p><b>Adulthood</b></p> <ul style="list-style-type: none"><li>• Weakness of the diaphragm, abdomen and chest wall muscles affecting the ability to cough, resulting in chronic lung infections, chronic bronchitis and bronchiectasis (abnormal stretching and enlarging of the bronchial tubes, which remain chronically infected)</li><li>• Weak facial and esophagus muscles that may lead to sucking and swallowing problems, which can allow fluids to enter the lungs and result in chemical injury to the respiratory passages, chronic lung inflammation, and aspiration pneumonia</li><li>• Weakness and myotonia of the diaphragm and other respiratory muscles, leading to insufficient exchange of oxygen and carbon dioxide in the lungs (hypoventilation)</li><li>• Sleep apnea, which can result in dangerously low levels of oxygen and high levels of carbon dioxide in the blood. In mild cases, apnea can cause disrupted sleep, excessive fatigue, and morning headaches. In severe cases, apnea can cause high blood pressure, cardiac arrhythmias, and heart attack.</li><li>• Severe respiratory failure is also seen in some individuals with myotonic dystrophy, particularly late in life. These pulmonary problems are one of the main causes of mortality in adults with the congenital form of myotonic dystrophy DM1.</li></ul>
<p><b>Childhood Onset DM1</b></p>
<p><b>Childhood/Adolescence</b></p> <ul style="list-style-type: none"><li>• Weakness of the diaphragm, abdomen, and chest wall muscles affecting the ability to cough, resulting in chronic lung infections, chronic bronchitis and bronchiectasis</li><li>• Chronic upper airway infections, which potentially can lead to hearing loss at a young age</li><li>• Weak facial and esophagus muscles leading to swallowing problems, which can result in fluids entering the lungs that cause chemical injury to the respiratory passages, chronic lung inflammation, and aspiration pneumonia</li></ul>
<p><b>Adulthood</b></p> <ul style="list-style-type: none"><li>• Weakness of the diaphragm, abdomen and chest wall muscles affecting the ability to cough, resulting in chronic lung infections, chronic bronchitis, and bronchiectasis</li><li>• Weak esophagus muscles and swallowing problems, which can allow fluids to enter the lungs and result in chemical injury to the respiratory passages, chronic lung inflammation, and aspiration pneumonia</li><li>• Weakness and myotonia of the diaphragm and other respiratory muscles, leading to insufficient exchange of oxygen and carbon dioxide in the lungs (hypoventilation)</li><li>• Sleep apnea which can result in dangerously low levels of oxygen and high levels of carbon dioxide in the blood. In mild cases, apnea can cause disrupted sleep, excessive fatigue, and morning headaches. In severe cases, apnea can cause high blood pressure, cardiac arrhythmias, and heart attack.</li><li>• Severe respiratory failure, particularly late in life. Pulmonary problems are one of the main causes of mortality for individuals with childhood onset myotonic dystrophy DM1</li></ul>
<p><b>Adult Onset DM1</b></p>
<ul style="list-style-type: none"><li>• Weakness of the diaphragm, abdomen and chest wall muscles affecting the ability to cough, resulting in chronic lung infections</li><li>• Weak esophagus muscles and swallowing problems, which can allow fluids to enter the lungs and result in chemical injury to the respiratory passages, chronic lung inflammation, and aspiration pneumonia</li><li>• Weakness and myotonia of the diaphragm and other respiratory muscles, leading to insufficient exchange of oxygen and carbon dioxide in the lungs (hypoventilation)</li><li>• Sleep apnea, which can result in dangerously low levels of oxygen and high levels of carbon dioxide in the blood. In mild cases, apnea can cause disrupted sleep, excessive fatigue, and morning headaches. In severe cases, apnea can cause high blood pressure, cardiac arrhythmias, and heart attack.</li><li>• Severe respiratory failure, particularly late in life. Pulmonary problems are one of the main causes of mortality for individuals with adult onset myotonic dystrophy DM1.</li></ul>
<p><b>DM2</b></p>
<ul style="list-style-type: none"><li>• Respiratory complications are uncommon.</li></ul>



## Diagnosis

### Clinical observation of gas exchange

- Measurement of respiration rate and work of breathing; comfort level; tachypnea
- Assessment of chest wall motion; abdominal muscle recruitment
- Observation for evidence of diaphragmatic paralysis
- Monitoring of breath sounds using a stethoscope (auscultation) to evaluate air entry into the lung base

### Observation for pneumonia

Weakened breathing muscles put patients at risk for lung infections so careful monitoring for signs of pneumonia is important.

### Inquiry about sleep disturbances

Symptoms such as nocturnal restlessness, unexplained awakenings, loud snoring punctuated by occasional awakening and gasping for breath may suggest the presence of a sleep-related respiratory disorder. Further study with a polysomnographic evaluation is recommended when symptoms are present.

### Pulmonary function tests

These measures are used as a predictive measure of respiratory failure susceptibility and likely need for mechanical ventilation, and include:

- FVC (forced vital capacity). The total amount of air that can be forcibly blown out after full inspiration, measured in liters
- FEV1 (Forced Expiratory Volume in 1 Second). The amount of air that can be forcibly blown out in one second, measured in liters
- Maximal inspiration force. Ability to force air into the lungs
- Gas diffusion studies
- Arterial blood gases
- Carbon monoxide diffusing capacity (also called transfer factor, or TF)

### Imaging studies

- Chest radiography to detect recurrent or chronic infections
- High-resolution computed tomography (HRCT) scans to look for lung abnormalities (e.g. pulmonary fibrosis, bronchiectasis, parenchymal scarring, pleural thickening) in patients with respiratory weakness with or without hypogammaglobulinemia. HRCT scans are considered to be more sensitive than chest radiography for helping detect the silent or asymptomatic structural changes of airways and lung parenchyma that sometimes occur.



## Treatment

### Nocturnal mechanical ventilation

Noninvasive positive pressure ventilation or bilevel positive airway pressure ventilation may relieve chronic hypoventilation-related symptoms and sleep apnea-hypopnea. In later stages, patients may become symptomatic from alveolar hypoventilation even with the use of nocturnal support as muscle weakness progresses; full-time ventilation may be required.

### Manual and assisted coughing and/or cough assist device

In patients demonstrated to have difficulty clearing airway secretions, regular use of manual assisted coughing and/or a cough assist device may help to reduce the risk of pneumonia.

### Incentive spirometry

Use of breathing exercise such as incentive spirometry may also help to clear mucus from the lungs and increase the amount of oxygen that gets deep into the lungs. Treatment for pneumonia follows standard clinical practice.

### Continuous endotracheal mechanical ventilatory support

Infants with congenital myotonic dystrophy DM1 often require this level of support.

### Nasal continuous positive airway pressure (N-CPAP)

This can facilitate weaning infants from ventilation and minimize morbidity and mortality associated with prolonged (>4 weeks) intubation.

### Gastrostomy tube

Because feeding difficulties are common for children with congenital myotonic dystrophy DM1 with an increased risk for aspiration, individuals may benefit from feeding evaluation and gastrostomy tube insertion for airway protection and enteral feeding in early life.