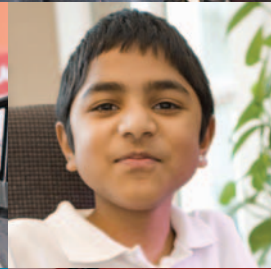




# Muscle disease: the impact

Incidence and Prevalence of  
Neuromuscular Conditions in the UK



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# Foreword

**This report, compiled by the Muscular Dystrophy Campaign, is the first document that comprehensively reveals the number of children and adults who are affected by genetic or acquired neuromuscular conditions in the UK.**

Epidemiological studies like this are the cornerstone methodology of public health research. They provide evidence and guidance for the planning and provision of accurate health care services and are particularly relevant in the field of neuromuscular conditions, as these conditions are very rare and require complex diagnostic approaches and multi-disciplinary care. The chronic conditions can affect newborns, children and adults and the clinical picture can vary significantly between conditions.

Up until now, precise and up to date information on patient numbers for those with genetic and acquired neuromuscular conditions has been sparse. Compiling the report was a challenging exercise because only a small number of epidemiological studies about the frequency of neuromuscular diseases in the UK have been published. As long as data that underline the need for improved clinical care are missing, services for patients with neuromuscular conditions will remain extremely underfunded and limited – knowledge of patient numbers for these diagnostically and clinically challenging conditions is of vital importance for NHS and local authority commissioners so that they can accurately plan provisions of multidisciplinary health and social care services.

At the moment there are no effective treatments for the majority of the neuromuscular conditions listed in this report. Increased life expectancy and

improved quality of life for many affected children and adults are due solely to better care and clinical management that is delivered by a multi-disciplinary approach. **As clinicians we urge policy makers, parliamentarians and health care providers to use the data in this timely and seminal report to plan vastly improved access to vital health care services that will change the lives of 1000s of people in the UK.**



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# Executive summary

Muscular dystrophies and related neuromuscular conditions are regarded as rare and, in many cases, even ultra-rare conditions for which there are currently no known cures and few effective treatments. The prevalence data showing the numbers of individuals living with these conditions are based on existing studies and clinical records and are published here for the first time in the UK. The EU suggests a rare disease is one with a prevalence of less than 1 in 2,000, although this is not binding. In the UK, a condition is considered to be a rare disease when the prevalence is less than 1 in 50,000.

The publication of the data by the Muscular Dystrophy Campaign serves several important purposes. It indicates to researchers and clinicians the numbers of individuals who need clinical care and support and also suggests the patient population from which participants in clinical trials can be found. Further, the report is a valuable tool for NHS and local authority commissioners and service leads who are responsible for the provision of health and social care to this important group of children and adults.

Individuals living with one of more than 60 neuromuscular conditions supported by the Muscular Dystrophy Campaign require specialist care and treatment from dedicated neuromuscular clinicians. With such rare conditions, the necessary knowledge and expertise usually lies with neuromuscular experts who provide leadership and guidance to clinical colleagues in secondary and primary care. Experience in several areas of the UK shows that a managed clinical network provides effective co-ordination and development of services and leads to optimal care of patients and families.

The most important part of this report will be found in the Results section where the data

show that at least 70,000 people in the UK are living with a neuromuscular condition. The data also show the groups of conditions that fall within the broad category of neuromuscular conditions. Patient numbers in the UK for each group of conditions range from some 23,000 people living with one of the hereditary neuropathies to as few as 10 people living with some forms of lipid storage myopathies. Further information about each group of conditions can be found in the section *Neuromuscular conditions – a summary*.

Bringing together the data from a number of sources has underlined the need for epidemiological studies to be undertaken in the UK to provide further robust, comprehensive data on the incidence and prevalence of these often devastating conditions. It is suggested that nationally important epidemiological studies should be commissioned by the National Institute for Health Research or the Medical Research Council.

## The need

The Muscular Dystrophy Campaign funds research and provides support for over 60 muscle conditions some of which are extraordinarily rare with as few as 10 known individuals in the UK. Incredible advances have been made in the field of neuromuscular research, particularly in the last 20 years. However, for many of the conditions there is still little information available on the exact numbers of affected people.

The rarity of the conditions and the absence of reliable epidemiological data are two of the reasons why the needs of children and adults with muscle disease are often overlooked by the NHS. In several reports prepared by the Muscular Dystrophy Campaign and leading clinicians in the field, it has become evident that specialist health services in England, Scotland, Wales and

Northern Ireland are extremely patchy and a 'postcode lottery' applies. Patients and families living in those regions with the poorest services sometimes refer to a 'no code lottery' in terms of inadequate care and support services. Nearly all of the conditions are of a chronic and progressive nature and healthcare providers often have little understanding that a multidisciplinary team is essential to improve people's survival, their independence and quality of life.

There are currently no efficient treatments or cures available for most of these conditions. Research, however, advances fast and a number of clinical trials to test the clinical benefit of promising technology are under way (details of current clinical trials are given on the Muscular Dystrophy Campaign's website at [www.muscular-dystrophy.org/research/clinical\\_trials](http://www.muscular-dystrophy.org/research/clinical_trials)). Scientists and clinicians are beginning to express cautious optimism that treatments

might become a reality in the near future. This makes the need for a reliable source of patient numbers even more urgent, as it will give NHS commissioners important information with which they can allocate funds efficiently to maximise patient benefit.

For this study we reviewed the relevant international literature, in particular a recent report published by Norwood *et al.* (2009) on the prevalence of genetic muscle disease in Northern England. The data from the literature, together with estimates on patients numbers from leading clinicians were used in the compilation of this comprehensive report on all neuromuscular conditions supported by the Muscular Dystrophy Campaign. This is the first report published on patient numbers for both inherited and acquired neuromuscular conditions in the whole of the UK. It is an important resource for policy makers, healthcare providers and healthcare professionals.

**Table 1** Number of neuromuscular patients living in each NHS region or country in the UK

| Country              | Population        | Individuals living with a neuromuscular condition |
|----------------------|-------------------|---|
| Scotland             | 5,062,000         | 5,984   |
| Wales                | 2,903,000         | 3,432   |
| Northern Ireland     | 1,685,000         | 1,992   |
| English Regions      | Population        | Individuals living with a neuromuscular condition |
| North East           | 2,623,000         | 3,101   |
| North West           | 6,887,000         | 8,141   |
| Yorkshire and Humber | 5,140,000         | 6,076   |
| East Midlands        | 4,333,000         | 5,122   |
| West Midlands        | 5,359,000         | 6,235   |
| South Central        | 4,034,000         | 4,769   |
| South West           | 5,130,000         | 6,064   |
| South East Coast     | 4,237,000         | 5,008   |
| East of England      | 5,607,000         | 6,628   |
| London               | 7,512,000         | 8,880   |
| <b>UK Total</b>      | <b>60,512,000</b> | <b>71,432</b>                                     |

Based on this review we estimate that at least 70,000 children and adults in the UK have a genetic or acquired neuromuscular condition supported by the Muscular Dystrophy Campaign. The estimated numbers for each NHS region are set out in the table above.

### The method

Several available resources were used to ensure that the data presented here are as accurate and reliable as possible. First we examined scientific journal articles obtained via the Internet, in particular through access to MEDLINE – a literature database of life sciences and biomedical information. Prevalence data for many genetic muscle diseases were based on a recently published study by the Newcastle MRC Centre for Neuromuscular Disease for the Northern region of England (Norwood *et al.* 2009). Secondly, we were advised by scientists and clinicians working in the field of neuromuscular conditions, including the specialist neuromuscular centres in London, Newcastle, Oxford and Oswestry and thirdly, where available, we used information from Muscular Dystrophy Campaign factsheets – written by expert clinicians – about specific conditions. We also took information from a number of reputable websites such as the website of the National Institute of Health, USA, and where possible we have referenced the source.

While every effort has been made to exhaust all available information on patient numbers, several issues should be noted in order to understand the full value (and limitations) of this review. In the UK very few scientific epidemiological studies about neuromuscular conditions have been published to date. Where studies have been carried out, they often give incidence and prevalence data for specific populations or ethnic groups for which an unusually high or low occurrence of

a disease might appear. In some cases epidemiological studies have been conducted several years if not decades ago and patient numbers might have changed because of scientific advances, such as genetic testing techniques or a general increase in life expectancy.

It should also be noted that conditions are sometimes grouped differently based either on their clinical picture (phenotype), or the underlying genetic defect (genotype).

However, this is the first comprehensive report on patient numbers for both genetic and acquired neuromuscular conditions in the UK and the first time that data from several different resources have been combined. As such, patient numbers presented here will be as reliable and accurate as the current state of epidemiological research allows.

**The data about the numbers of affected individuals in the UK have been gathered from a variety of sources, including scientific papers, and have been approved by leading clinicians and scientists.**

## The results

An overview of the different groups of neuromuscular conditions is provided in table 2.

**Table 2** Conditions and numbers – prevalence in the UK

| Group of conditions                      | Patient number in UK | Percentage of total (%) |
|--|----------------------|-------------------------|
| Muscular dystrophies                     | 8,000-10,000         | 12.6                    |
| Myotonic disorders                       | 9,500                | 13.3                    |
| Congenital myopathies                    | 1,000                | 1.4                     |
| Distal myopathies                        | 300                  | 0.4                     |
| Mitochondrial myopathies                 | 3,500                | 4.9                     |
| Metabolic myopathies                     | 700                  | 1.0                     |
| Periodic paralysis                       | 900                  | 1.3                     |
| Myositis                                 | 5,000-6,000          | 7.7                     |
| Spinal muscular atrophies (SMA)          | 1,200                | 1.7                     |
| Hereditary neuropathies                  | 23,000               | 32.0                    |
| Inflammatory and autoimmune neuropathies | 6,400                | 8.9                     |
| Disorders of the neuromuscular junction  | 10,500               | 14.7                    |
| Myositis ossificans progressiva (MOP)    | 60                   | 0.1                     |
| <b>Total number (prevalence)</b>         | <b>70,060-73,060</b> | <b>100</b>              |

## The study shows:

- There are at least 70,000 children and adults living in the UK with one of the 60 genetic or acquired neuromuscular conditions supported by the Muscular Dystrophy Campaign.
- The severity of the muscle conditions supported by the Muscular Dystrophy Campaign is highly variable – for the most severe disorders children can die at birth or in their first year of life while the mildest forms only slightly affect elderly people and have no life limiting impact.
- About 70% of all people with conditions supported by the Muscular Dystrophy Campaign are affected by muscle diseases that are inherited and about 30% by acquired muscle diseases.
- The number of patients for a given disease varies from 23,000 (hereditary neuropathies) to as few as 10 (some forms of lipid storage myopathies) or one (rare congenital muscular dystrophy variants such as MDC1D).
- Some 13% of the estimated 70,000 people affected by muscle disease have a form of muscular dystrophy.
- There is a need for governmental research bodies to commission national epidemiological studies which will reliably define the number of people living with a neuromuscular condition in the UK.

## Conclusion

This report represents the first publication of patient numbers for genetic and acquired neuromuscular conditions in the UK. There has been a pressing need to bring the data together in one document to establish a reliable source of reference that is both available and accessible. The data in this report are essential in enabling healthcare commissioners and managers to plan and budget specialist services effectively and to ensure that affected individuals and families receive the necessary *multi-disciplinary* health care at both specialist centres and their local areas.

In recent years, promising technology that has been developed in the laboratory is now being brought forward into clinical trials. The current focus is on translational research to speed up the bench-to-bedside transfer of these therapeutic approaches. As most of the conditions are relatively rare, knowledge of accurate patient numbers is particularly important for the logistics of clinical trials. This is especially significant as more “mutation-specific” treatments are in development – such as for Duchenne muscular dystrophy – which will only be appropriate for a proportion of the total number of individuals affected by the condition.

The authors have identified a lack of reliable data and up-to-date epidemiological studies as we often had to rely on personal communication with expert clinicians who deduced estimates according to the number of patients currently treated in their clinics. Our experiences strengthen the need for the research community to carry out epidemiological studies that reflect the current situation and we call for a national patient registry for neuromuscular conditions to be introduced in the UK.

# Neuromuscular conditions – a summary

## Muscular dystrophies

The term 'muscular dystrophy' defines a group of genetic muscle diseases that present with progressive skeletal muscle weakness and wasting due to the degeneration of muscle cells that can include the heart. The severity of muscular dystrophy can be hugely variable and symptoms can be apparent at birth or in the first months of life, as observed for congenital muscular dystrophy, or the symptoms can be very mild and are first noticed later in life between the ages of 40 and 50, in the case of oculopharyngeal muscular dystrophy.

There are more than 35 different clinically described conditions that can be categorised into seven groups, including:

- Duchenne muscular dystrophy
- Becker muscular dystrophy
- limb girdle muscular dystrophy
- congenital muscular dystrophy
- facioscapulohumeral muscular dystrophy
- oculopharyngeal muscular dystrophy
- Emery-Dreifuss muscular dystrophy

For most of the muscular dystrophies the genetic defect has been described – it generally involves a defect in a protein that plays a vital role in muscle cell function or repair. However, for a small number of individuals the genetic defect has yet to be found, but as research advances the categorisation of muscular dystrophies becomes increasingly complex. For example there are now 19 different genes known to cause limb girdle muscular dystrophy.

An estimated 8,000 to 10,000 people in the UK have a form of muscular dystrophy<sup>1,2</sup>. This number does not include myotonic dystrophy which can be found under "myotonic disorders".

## Myotonic disorders

These are inherited conditions characterised by myotonia which describes a certain muscle stiffness that makes it difficult to relax the muscles after they have been contracted.

The most prevalent form is myotonic dystrophy characterised by muscle weakness and myotonia that particularly affects hands and ankles. It also affects other organs and is associated with cataracts, disturbance of heart rhythm and, in children, learning difficulties. The condition shows enormous clinical variability ranging from the presence of severe symptoms present at birth to the development of cataracts as the only symptoms in middle age. The genetic defects have been described as unstable mutations that tend to get worse when passed from generation to generation.

This group also includes myotonia congenita and paramyotonia congenita. Patients with these conditions also experience significant muscle stiffness because of an inability of muscle relaxation. This can range in severity from severe neonatal myotonia causing respiratory compromise through to adult onset muscle stiffness with limitation of daily activities.

An estimated 9,500 people living in the UK have a form of a myotonic disorder<sup>3,10,17</sup>.

## Congenital myopathies

Congenital myopathies are a group of inherited muscle diseases that are present from birth. They are characterised by changes in the muscle cells that lead to a reduced contractile ability of the muscle. Four conditions generally account for this group:

- nemaline myopathy
- central core disease
- centronuclear myopathy
- multiminicore disease

All four conditions lead to muscle weakness and a decrease of muscle tone in early childhood. They are also sometimes associated with delayed motor development later in life and speech and learning difficulties.

An estimated 1,000 people in the UK have a form of congenital myopathy<sup>4,5,6,7</sup>.

### **Distal myopathies**

Distal myopathy describes a group of inherited disorders that primarily affect distal muscles – those in the lower arms or legs, hands and feet. Although muscle weakness is generally first observed in distal muscles, with time other muscle groups will also be affected. The distal myopathies are caused by defects in a variety of genes and not all affected individuals can be accurately diagnosed.

An estimated 300 people in the UK have a form of distal myopathy<sup>1</sup>.

### **Mitochondrial myopathies**

‘Mitochondrial myopathies’ is a collective term for a group of diseases that particularly affect muscle, but which may also affect every other part of the body, including the brain and the eye. The clinical features may be fairly mild with predominantly ophthalmoplegia (weakness of the eye muscles) or severe with progressive muscle weakness leading to respiratory failure.

Mitochondrial myopathies are often caused by a defect in the mitochondrial genome that is maternally transmitted. Mitochondria are the “powerhouses” of the cells that convert the food we eat into energy. Due to the presence of multiple copies and a mixture of mutated and unaffected mitochondria in the muscle cell, genetic counselling, pre-implantation genetic diagnosis and chorionic villus biopsy all have limitations in providing accurate genetic advice. In addition, only limited treatment is available.

An estimated 3,500 people living in the UK have a form of mitochondrial myopathy. An additional 6,000 people are also considered “at risk”: These are children and first degree relatives who might develop the condition during their lives<sup>8</sup>.

### **Metabolic myopathies**

Metabolic myopathies are caused by mutations in genes involved in the energy metabolism in skeletal muscles. The defects generally block the chemical reactions involved in the provision of energy and so the muscle cells cannot work properly. In some cases this can lead to episodes of muscle damage, causing acute kidney failure and requiring admission to intensive care, and in others there is progressive muscle weakness.

Metabolic myopathies that affect young children tend to be the most severe and can be fatal. Those with a later onset tend to be less severe, and in very mild cases changes in diet and lifestyle can alleviate symptoms.

An estimated 700 people in the UK have a form of metabolic disorder of muscle<sup>9</sup>.

### **Periodic paralysis (channelopathies)**

Periodic paralysis describes a group of rare genetic conditions characterised by intermittent (periodic) attacks of muscle weakness (paralysis). The attacks can be the result of stress, excitement, physical activity, heat or cold. The muscle weakness may be confined to a small group of muscles or be more generalised, in which case the whole body is unable to move (paralysis). It is very rare for the breathing, speaking or swallowing muscles or the heart to be involved. The duration of an attack varies from minutes to days depending upon the type of periodic paralysis and at the end of an attack the muscle strength returns. Individuals can

experience slow, progressive muscle weakness over time.

An estimated 900 people in the UK have a form of periodic paralysis<sup>3,10,17</sup>.

### **Myositis**

Myositis is a general term used to describe a number of inflammatory myopathies including:

- dermatomyositis
- inclusion body myositis
- juvenile forms of myositis
- polymyositis

Inflammatory myopathies are considered to be autoimmune diseases meaning that the body's own immune system attacks the muscle. All forms are characterised by degeneration of muscle, that causes progressive weakness. Symptoms include difficulty walking and climbing stairs, falling, and weakness of the arms. Swallowing and breathing may be affected. Inflammatory myopathies are potentially life-threatening, especially dermatomyositis which can involve the heart and the lungs. It can also be associated with cancer in up to 20% of patients. Treatments are available for some forms of myositis and about half of people treated recover so that, for example, they can return to employment.

An estimated 5,000 to 6,000 people in the UK have a form of myositis<sup>11,3</sup>.

### **Spinal muscular atrophy**

Spinal muscular atrophies (SMA) are a group of genetic conditions characterised by the loss of nerve cells (motor neurons) in the spinal cord. This leads to progressive muscle weakness and wasting, particularly in the trunk, upper arms and thighs. Respiratory problems can also develop. The various types of SMA differ considerably in age of onset and severity. The most severe type, SMA type I, represents the

most common single genetic cause of death in infancy. The mildest form, SMA type III, generally manifests when children are between five and ten years old and many individuals keep their ability to walk decades after the symptoms are first observed.

An estimated 1,200 people in the UK have a form of spinal muscular atrophy<sup>12,9</sup>.

### **Hereditary neuropathies**

Hereditary neuropathies describe a group of inherited conditions that affect the peripheral nervous system. The group includes Charcot-Marie-Tooth disease (CMT), one of the most common inherited neuromuscular disorders that affects both the sensory and motor nerves (hereditary motor and sensory neuropathy). CMT is characterised by muscle weakness below the knees and in the hands. Individuals may experience motor problems as the motor nerves from the spinal cord to the muscles are damaged. It also causes numbness and sensory problems as the sensory nerves travelling from the skin to the spinal cord are damaged. There are several forms of CMT disease with differing inheritance patterns. More than 30 causative genes have been described to date.

An estimated 23,000 people in the UK have a form of Charcot-Marie-Tooth disease<sup>14</sup>.

### **Inflammatory and autoimmune neuropathies**

Inflammatory and autoimmune neuropathies are a group of peripheral neuropathies that arise when the immune system attacks the peripheral nerves, disrupting the signal to the muscles. The group includes:

- acute inflammatory demyelinating polyneuropathy, also called Guillain Barré syndrome
- chronic inflammatory demyelinating peripheral neuropathy (CIDP).

Guillain Barré syndrome is characterised by the rapid development of weakness and numbness caused by widespread inflammation that is first observed in the legs and then in the arms and face. In severe cases swallowing and breathing are involved, so individuals need artificial ventilation and feeding. The exact cause is not known, but in most cases individuals had a prior viral or bacterial infection. Individuals often make a full recovery.

Chronic inflammatory demyelinating peripheral neuropathy has similar characteristics to Guillain Barré syndrome and is often regarded as the chronic counterpart. It is more common in younger people and affects more women than men.

An estimated 6,400 people in the UK have a form of inflammatory and autoimmune neuropathy<sup>14</sup>.

### **Disorders of the neuromuscular junction**

Disorders of the neuromuscular junction are caused when the electric signal from the nerve to the muscle is disrupted in some way resulting in muscle weakness and fatigue. Two groups fall into this category:

- myasthenia gravis
- congenital myasthenic syndromes

Myasthenia gravis is a chronic, autoimmune disease that causes muscle weakness and excessive muscle fatigue. It can affect individuals of all ages. The disease can vary in severity and distribution of weakness between individuals, and in any one patient the symptoms fluctuate with relapses and remissions. Myasthenia gravis can resolve spontaneously, but for most patients the condition persists for life. It can be life threatening, but 90% of patients become symptom-free with modern treatments.

Congenital myasthenic syndromes are a group of inherited conditions caused by mutations in the genes that play an important role in the transmission of the signal from the nerve to the muscle. The symptoms are similar to myasthenia gravis although it should be noted that treatments for myasthenia gravis are not useful for the congenital form.

An estimated 10,500 people in the UK have a form of myasthenic disorder<sup>15,3</sup>.

### **Myositis ossificans progressiva (MOP)**

Myositis ossificans progressiva (MOP), also known as fibrodysplasia ossificans progressiva, is a condition caused by bone forming within muscles. The bone tissue is generated as a result of faulty repair processes of damaged muscle tissue. The bone formation is usually first noticed in early childhood as a series of hard lumps in the neck or along the spine. The lumps, which may be tender, gradually shrink in size as the affected muscles are replaced by bone. As this process progresses it will lock joints in place and will make movement difficult or impossible.

This is a very rare condition. An estimated 60 people in the UK have this condition<sup>16,4</sup>.

# Appendix 2

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# Appendix 3

## About the Muscular Dystrophy Campaign

The Muscular Dystrophy Campaign has a very proud history. We were set up in 1959 by a scientist, a doctor and a family to help other families and children living with muscular dystrophy. Our main aim was to raise money to fund research to find the causes of and cures for muscular dystrophy.

Since then, diagnosis of the different forms of muscular dystrophy and related muscle diseases has improved hugely and we now provide support for over 60 different muscle-wasting conditions. Many key research developments during the last 50 years have been funded by us, including vital pre-clinical research for the exon skipping trial that is testing an exciting new technology to treat Duchenne muscular dystrophy.

Since 1959 we have invested £100 million in pioneering research and the provision of care and support.

We are dedicated to improving the lives of all babies, children and adults affected by muscle disease.

## Our priorities today:

- **We fund world-class research to find treatments and cures.**
- **We provide free practical and emotional support.**
- **We campaign to raise awareness, improve care and support and bring about change**
- **We award grants towards the cost of specialist equipment.**
- **We continue to lead the fight against muscle disease today.**



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