1.1 Diagnosis

Some children will have a diagnosis of their muscle-wasting condition when they begin their school life. Others will still be having investigations and tests, and some will only begin to show signs and symptoms of the condition when they are already at school.

It may be that school staff will be the first to recognise a problem, perhaps during PE or a games lesson. Should this happen, the specific concerns need to be discussed sensitively with parents and the school medical officer, who may suggest referring the child to a paediatric consultant. Do remember, however, that many children have co-ordination and movement difficulties. This does not necessarily relate to a muscle-wasting condition.

Obviously the time around diagnosis will be extremely stressful for all family members. Chapter 4 deals with this in more detail. Once a child has been diagnosed with a muscle-wasting condition, it is vital to:

- find out accurate information about the specific condition and how it is likely to progress (you can visit Muscular Dystrophy UK’s website, or call the helpline on 0800 652 6352 to find out more)
- discuss the child’s condition and related needs with his or her family, while being aware that they may find it difficult to share information, especially about how the condition may progress
- gain practical information and advice from specialist advisory teachers, health professionals and your local neuromuscular care advisor.

1.2 Neuromuscular conditions

More than 70,000 children and adults in the UK have a muscle-wasting condition (please note that we use the umbrella term ‘muscle-wasting conditions’ to refer to muscular dystrophy and related neuromuscular conditions). A further 350,000 people are affected indirectly as family, friends or carers.

Muscle-wasting conditions cause muscles to weaken and waste over time, leading to increasing disability. These rare and very rare genetic conditions may affect not only the muscles in the limbs, but also those of the heart and lungs, sometimes significantly shortening life-expectancy.

Genetic conditions result from alterations in the genetic make-up of an individual. They may be caused by defects in single genes or whole chromosomes, parts of which may be lost, duplicated, misplaced or replaced. Genetic disorders can be caused by defects in one or more genes. (Contact Muscular Dystrophy UK to find out more.)

In these conditions, a gene may fail to produce one of the proteins needed for normal muscle function. While the conditions are generally inherited, in some cases, they occur ‘out of the blue’ with no family history of the condition.

Muscular Dystrophy UK also supports families living with conditions that affect nerves, including spinal muscular atrophy (SMA) and the hereditary sensory neuropathies, which cause muscle weakness. The junction between the nerve and muscle (neuromuscular junction) is affected in the myasthenias.
Below is a brief summary of some of the more frequently encountered conditions. For more in-depth information on a specific condition please read the Muscular Dystrophy UK factsheets available to download from www.musculardystrophyuk.org or call 0800 652 6352.

**Duchenne muscular dystrophy**

Duchenne muscular dystrophy is one of the more common conditions; it is a life-shortening muscle-wasting condition caused by the lack of a vital muscle protein called dystrophin. The condition causes muscles to weaken and waste over time, leading to increasing and severe disability.

Duchenne muscular dystrophy currently affects around 2,500 people in the UK and around 100 babies are born in the UK with the condition each year. It is caused by genetic mutations on the ‘X’ chromosome, meaning almost all of those affected are male. These mutations prevent the body from producing the vital protein, dystrophin, which is needed to build and repair muscle.

Most children with Duchenne muscular dystrophy are diagnosed by the age of five. Most will use a wheelchair by the age of 12, and will face life-threatening health problems by their late teens as the muscles of the heart and lungs weaken. Few of those born with the condition currently live to see their 30th birthday; with medical advances, however, life-expectancy is improving.

The right specialist healthcare and equipment, including access to heart and respiratory experts and regular physiotherapy, can make a significant difference to both quality and length of the life for people with Duchenne muscular dystrophy.

While the condition is severely disabling, many of those living with Duchenne muscular dystrophy lead full and active lives. There are authors, designers, campaigners and Paralympians with Duchenne muscular dystrophy.

**Learning abilities in Duchenne muscular dystrophy**

Some children with Duchenne muscular dystrophy may also have learning difficulties or find some aspects of learning more difficult. These are rarely severe and are not progressive. The main difficulties are usually language and communication skills.

The protein dystrophin is absent from muscle tissue and it is now known that part of the protein is also missing from the brain. The contribution made by dystrophin to the brain is not fully understood, although it is thought to be involved with selective cognitive processes.

Most children with Duchenne muscular dystrophy do not have any difficulties with their ability to learn, however, it is thought that all do have some cognitive involvement. It is generally believed that a little over two-thirds of those who are diagnosed with Duchenne muscular dystrophy have an IQ score one point lower than their peer group, which means that they score within the average range. However, about 19 percent will have a more significant difference with a score in the 70s, and some will fall into the high average group.

It is important to stress that there is much variability in skill, so accurate assessment is required to inform appropriate management.

Studies indicate that the main difficulties experienced by these children are verbal skills and reading skills, limited verbal memory, reduced attention and poor social and interpersonal behaviour skills. These are the ‘hidden problems’ of Duchenne muscular dystrophy and can sometimes be overlooked when the primary concerns seem to focus on physical skills and access.
It has been suggested that about half of all children with Duchenne muscular dystrophy are thought to have a form of developmental dyslexia. This could be, in particular, a difficulty with phonological processing, which can mean a reading age two years behind their class group. These children can struggle to learn when presented with too many verbal instructions or non-contextual aural information, which is not accompanied by kinaesthetic or visual reinforcement. They may, therefore, find it difficult to repeat back verbal information presented only once.

There can be a reduction in digit span, which is a measure of short-term memory. It involves testing the number of digits a person can remember and recall in a correct sequence after hearing or seeing them. A child needs to recall approximately six digits in order to utilise phonics. If this is not possible, the strategies employed in the classroom need to focus on memory development. Phonological skills underpin reading.

The social interaction difficulties can manifest as the child’s being less compliant or not noticing social subtleties.

These difficulties are thought to be a direct result of compromised verbal skills and can be a great source of stress to caregivers. These children and young people can be skilled in other areas, for example rote memory, which does not require mental manipulation. Long-term memory is effective and intact so these skills need to be nurtured in educational and clinical settings. Some children with Duchenne muscular dystrophy can appear to have difficulties processing sensory information from the environment around them. In these cases, the children would require support from an occupational therapist.

All involved in education and care must be mindful that the difficulties experienced by children with Duchenne muscular dystrophy can be far-reaching and go beyond physical difficulties. Consequently, it is essential to have accurate assessment for the most effective and accessible teaching techniques to be employed to deliver the national curriculum. Any assessment needs to cover the child’s development, academic skills, verbal abilities, working memory and attention span.

Suggested strategies for use in the classroom include:

- **assessment – developmental level, academic milestones, sensory and aural processing** (this information may need to come from different people who know and support the child, for example parent/carer, OT, SENCO and neuromuscular care advisor)
- simplifying instructions
- breaking aural information and instructions into chunks appropriate to the results of the aural processing assessment
- introducing a sensory programme, if this is deemed to be a barrier to learning
- frequently checking the child’s understanding of information
- using mixed media presentations, with emphasis on visual and kinaesthetic learning
- introducing games and activities that help the child to practise memory improving strategies
- using phonological awareness as with the dyslexic population
- using time-limited activities to promote success, followed by an activity of choice.

Each strategy should be employed in a way that does not alienate the child who is already aware that he is different from his peers. In addition, parents need to be supported through the process of assessment so they can carry through the suggested management techniques at home.

**Communication needs**

Some boys with Duchenne muscular
dystrophy may have delayed acquisition of language skills, and therefore delayed expressive language. This can have a significant impact on their social life, affecting their ability to build and maintain friendships.

Pupils may have difficulties with processing and retaining complex information delivered verbally by an adult. This can mean that they are unable to carry out a full set of instructions and can look like they are distracted or just being difficult. For example, when a parent asks a young child to get ready to go to the park by going to the toilet, washing their hands and getting their coat and shoes, they may only follow one instruction. This is because the volume of information is likely to be too much for the child. To those who do not understand the hidden problems of Duchenne muscular dystrophy, this can be frustrating.

Some parents report that their child is ‘very clever’ because they have a keen knowledge of certain topics of interest. On closer assessment, it is often the case that the child has an extensive vocabulary related to this, but it may mask the difficulties listed previously. In such cases, the agenda is set by the child and it is harder to engage him in other learning tasks such as asking questions or engaging with activities not closely related to the topic of interest. The child’s knowledge of topic-specific vocabulary can affect how the teacher assesses the child’s ability to function in class and may therefore affect expectations.

Becker muscular dystrophy
Becker muscular dystrophy is a genetic muscle-wasting condition, which causes muscles to weaken and waste over time leading to increasing disability. It is caused when the body’s ability to produce a fully-functioning version of a vital muscle protein called dystrophin is compromised.

Over 2,400 people in the UK are thought to have Becker muscular dystrophy. Most people with the condition are diagnosed by the time they reach their 20s and indeed, some are severely affected from childhood. However, others do not know they are affected until well into adult life.

Becker muscular dystrophy almost always affects boys and men. It is not uncommon for several members of a family across generations to be affected. The severity of the condition varies. People with Becker muscular dystrophy may struggle with sport as children, with muscle weakness becoming more pronounced in their teens or 20s, causing difficulty in walking quickly, running and climbing stairs. In most cases they will continue to be able to walk until their 40s or 50s, but for some the decline in mobility will be much faster.

Spinal muscular atrophy
Spinal muscular atrophy (SMA) is a rare inherited neuromuscular condition, of which there are several distinct types. The condition may affect crawling and walking ability, arm, hand, head and neck movement, breathing and swallowing. SMA does not affect a child’s intellectual development.

There are four main types of SMA, and they vary in severity:

SMA Type 1
The symptoms of SMA Type 1 appear within the first few months of life, sometimes before birth. It is the most severe form of SMA. Children are never able to sit unaided and rarely survive their second birthday. However survival rates are increasing and with full ventilator support there are a small number of children with SMA Type 1 attending school.

SMA Type 2
The symptoms of SMA Type 2 usually appear between the ages of seven and 18 months. The condition is severely physically disabling, with children never able to stand unaided. Though this is a serious inherited
neuromuscular condition that may shorten life-expectancy, improvements in care standards mean that the majority of people can live long, fulfilling and productive lives.

SMA Type 3
The symptoms of SMA Type 3 appear after 18 months of age. Children are able to stand and walk, though will experience reduced walking ability over time. It is a less disabling condition. Life-expectancy for children diagnosed with SMA Type 3 is normal and most people can live long productive lives.

SMA Type 4
The symptoms of SMA Type 4 appear in adulthood. It is also known as Adult Onset SMA and is not life-threatening.

Myotonic and congenital myotonic dystrophy
Myotonic dystrophy is a genetic condition, which causes muscles to weaken, stiffen and waste over time, leading to increasing disability.

Myotonic dystrophy affects approximately 9,500 people in the UK. The age at which symptoms appear can vary from birth to old age. The condition is progressive and the earlier symptoms appear, the more severe they will become.

Muscle stiffness or ‘myotonia’ is a characteristic symptom, especially in the hands. People may also experience muscle weakness in the face, jaw and neck, often resulting in speech and swallowing difficulties. Heart problems, digestive problems, and cataracts may also be caused by myotonic dystrophy.

High-risk complications for people with myotonic dystrophy include an irregular heart rhythm and chest infections caused by weakened chest muscles.

Congenital (from birth) myotonic dystrophy
Children with congenital myotonic dystrophy tend to reach physical and learning milestones later than their peers.

Often children with congenital myotonic dystrophy have facial weakness and a lack of facial expression. This does not mean that the child is unresponsive; it is simply that he or she may be unable to make the usual range of facial movements.

The severity of the condition varies considerably from child to child. A few affected children need to use a wheelchair. Significant features, particularly in congenital myotonic dystrophy, are learning difficulties (which can be severe), tiredness, lethargy and cataracts at an unusually early age.

Congenital muscular dystrophy
Congenital (from birth) muscular dystrophy refers to a number of genetic muscle-wasting conditions which take effect from infancy, causing muscles to weaken and waste over time and leading to increasing disability.

It is thought that around 500 people in the UK have congenital muscular dystrophy, the main forms being Ullrich congenital muscular dystrophy and merosin-deficient congenital muscular dystrophy.

Congenital muscular dystrophy is caused by mutations in genes affecting the production and repair of muscle. Mutations causing the condition have been discovered across at least 19 genes and there are likely to be many more. Owing to the complexity of the condition and the number of genes affected, some people will never know which type they have.

The types fit into two groups: the first cause weakness in all the muscles in the body, but do not affect the intellect; the second cause muscle weakness, along with learning difficulties, and sometimes seizures.
The severity of congenital muscular dystrophy varies greatly between types and individuals. Some children affected will walk, but sometimes this can be delayed until five years of age or older. Some children who have achieved independent walking may lose this ability later on because, as they grow heavier, the muscles are unable to cope with a greater strain. Other children may never be able to walk.

Facioscapulohumeral muscular dystrophy
Facioscapulohumeral muscular dystrophy (FSHD) is a genetic muscle-wasting condition that particularly affects the muscles of the limbs, shoulders and face. It is thought to be the third most common form of muscular dystrophy, affecting over 1,300 people in the UK and at least 140,000 worldwide.

FSHD is caused by a genetic mutation that causes the production of a toxic protein in the muscle, which kills the muscle cells. Often several generations of a family may be affected by FSHD, which can be diagnosed at any age and varies widely between individuals, even within the same family. The earlier muscle weakness appears, the more severe it is likely to become.

Approximately 10 to 20 percent of people with the condition eventually require a wheelchair. The muscles of the eyes and mouth can also be involved, affecting facial expression, and the ability to smile.

Limb girdle muscular dystrophy
Limb girdle muscular dystrophy is a large group of conditions that cause weakness of the large muscles at the top of arms and legs, attached to the shoulder and pelvic ‘girdles’. In some people, the muscles of the heart and lungs are also affected, leading to life-threatening health problems. Around 1,400 people in the UK have one of the many types of limb girdle muscular dystrophy.

A very wide range of genetic mutations, all affecting the production and repair of muscles, cause limb girdle muscular dystrophy. Owing to this complexity, approximately 25 percent of all people with limb girdle muscular dystrophy do not receive a precise genetic diagnosis.

Some people with milder forms will never become seriously affected, while others may struggle to lift their arms above their heads, or lose the ability to walk. The muscles of the legs may deteriorate faster than those of the upper body, resulting in frequent falls, difficulty in running and climbing stairs, and rising from the floor. Usually, the earlier the symptoms become apparent, the more severe the condition will be.

Congenital myopathies
This is a group of inherited muscle conditions that are present from birth.

Congenital myopathies include central core disease, congenital myopathy with fibre-type disproportion, multi (mini) core disease, myotubular (centronuclear) myopathy and nemaline myopathy.

Congenital myopathies are conditions where changes in the muscle cells make them less able to contract. All these forms of congenital muscular dystrophy lead to muscle weakness and a decrease of muscle tone in early childhood. Later in life they are sometimes associated with delayed motor development and speech and learning difficulties.

About 1,000 people in the UK have a form of congenital myopathy.

Charcot-Marie-Tooth disease
Charcot-Marie-Tooth disease (CMT) is a progressive, inherited condition which affects nerves controlling movement of the hands and lower legs. It affects around 23,000 people in the UK, an equal spread of male and female.
Also commonly referred to as hereditary motor and sensory neuropathy (HMSN), CMT can involve loss of feeling in the hands and feet (the ‘sensory’ component). The term ‘neuropathy’ refers to the peripheral nerves, which connect the spinal cord to the muscles, joints and skin, carrying messages in both directions, and which do not function normally.

The first symptoms of CMT include difficulties picking up the feet and very high foot arches. These are usually apparent from childhood. Those affected may have problems with balance and walking longer distances, bone abnormalities, loss of reflexes and weakness of the hands. It is unusual for people to lose the ability to walk, however some will need to use walking aids as they grow older.

Owing to loss of sensation in the hands and feet, people with CMT need to be very careful to prevent and detect injuries to these areas. This is particularly important when planning PE activities, as the foot or ankle can be damaged without the pupil’s being aware. Fine motor skills may also be affected as the pupil may not receive sensory feedback from the equipment. This could result in difficulties with handwriting, manipulating science or technology equipment, distinguishing temperature, sharpness or pain.

Myasthenia gravis
Myasthenia gravis is an autoimmune condition. It is caused when the body’s immune system attacks the junctions responsible for transferring messages from the nerves to the muscles. It causes weakness and fatigue in the muscles of the limbs, face, eyes and throat, and also in the respiratory muscles vital for breathing.

Myasthenia gravis is thought to affect between 5,000 and 10,000 people in the UK – about 15 in every 100,000 individuals. It can develop at any age from childhood to extreme old age. Most of those affected are female, although men are more likely than women to develop it in later life.

The impact of the condition is very unpredictable and can vary dramatically day-to-day and between individuals. Frequently, the muscles of the eyes and face are affected, causing double vision, drooping of the eyelids (the child looks as if he or she is sleepy) and making it difficult for the child to smile. The arms and legs may be weak and, in severe cases, there can be problems with breathing muscles.

Myasthenia gravis can also cause speech disturbances, such as slurred speech, which can cause difficulty in a classroom setting and when socialising with peers. Swallowing difficulties are also common in myasthenia gravis. Swallowing is a complex process involving around 50 pairs of muscles. These muscles can become fatigued, particularly towards the end of a meal or when food has required a lot of chewing. It is important that a child with swallowing difficulties has an assessment with a speech and language therapist, so that recommendations can be put in place to help manage the symptoms.

Please note that even after successful treatment of juvenile myasthenia gravis, pupils can be left with significant neuromuscular fatigue. This may make it difficult when returning to school once treatment is finished and symptoms have improved. The fatigue is unpredictable; it can present a day or two after a period of physical exercise or emotional stress and can be very debilitating.

Congenital myasthenic syndrome
This condition, which is very rare, can be difficult to distinguish from myasthenia gravis. It is a genetic condition in which one of the proteins at the neuromuscular junction functions abnormally. The symptoms and signs are similar to myasthenia gravis. The first evidence of the condition is at birth or shortly afterwards. In some cases the
child experiences potentially fatal episodes of breathing failure. Drugs can help the weakness and it may improve spontaneously as the child gets older. Again, speech and swallowing difficulties can be common.

**Mitochondrial myopathy**
Mitochondrial myopathies are a group of conditions that particularly affect muscle, but may also affect every other part of the body, including the brain and the eye.

Mitochondrial myopathy is caused by genetic faults, which affect the function of mitochondria (the parts of muscle cells that generate the energy required for a muscle to contract). The causes and results of mitochondrial myopathy vary but most lead to some form of muscle weakness, and some affect specific parts of the body.

About 3,500 people in the UK have mitochondrial myopathy. The impact of mitochondrial myopathy varies dramatically from person to person. Many of those diagnosed are unaware that there is a family history of the condition.

The most common symptom is a combination of mild weakness of the arms and legs, droopy eyelids and difficulty moving the eyes. Some people only have the weakness in their arms and legs and find this gets worse if they exert themselves. In others it is more severe. For example, some babies with the condition may have difficulties with swallowing and feeding, which can affect their life-expectancy.

**Metabolic myopathies**
Also known as metabolic muscle conditions, these are conditions that interfere with the way muscles provide energy.

Metabolic myopathies, which include McArdle’s disease and Pompe disease (also known as acid maltase deficiency, or glycogen storage disease type II) are caused by mutations in the genes involved the production of energy in skeletal muscles. The mutations generally block the chemical reactions that take place during energy production, so the muscle cells cannot work properly. In some cases, this can lead to episodes of muscle damage, causing acute kidney failure. In others there is progressive muscle weakness.

Metabolic myopathies that affect young children tend to be the most severe and can, in some cases, be fatal. Those with a later onset tend to have less severe symptoms, and in very mild cases changes in diet and lifestyle can ease symptoms. About 700 people in the UK have a form of metabolic myopathy.

**Neuromuscular conditions (non-specified)**
Despite medical and technological advantages, a number of people will not have a definitive diagnosis of their muscle-wasting condition.

Not having a diagnosis can be difficult for these children and their families. However, they should keep in close contact with their care advisor or specialist nurse to find out what research is taking place or if there are any clinical trials. It is important for school to understand that the pupil will have specific difficulties which need to be managed to optimise safety and to enable the pupil to achieve their academic potential.

**Information resources**
Muscular Dystrophy UK has alert cards and Information Standard-accredited factsheets on a number of muscle-wasting conditions – including those without a specific diagnosis. The alert cards include information on the vital and specific issues that affect children with these conditions. They also outline key recommendations and precautions that non-specialist clinicians would need to know in times of worsening health.
1.3 Key issues for the conditions

- Mobility and physical activity
- Falling/losing balance
- Fatigue
- Upper limb function
- Personal care and comfort
- Moving and handling
- Seating, standing, comfort and positioning
- Temperature control
- Eating and swallowing
- Going to the toilet
- Self-image and emotional wellbeing
- Communication needs

The above issues are discussed in Chapter 5: Health Issues.