Physical exercise in adults with hereditary muscular disease

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Hereditary muscular diseases are a heterogeneous group of diseases that have previously been advised to avoid harsh physical activity due to fear that exercise would damage the already weakened muscles. Recent studies show that physical activity and exercise can have beneficial effects, but must be adapted individually based on diagnosis and level of function.

This article discusses hereditary myopathies and adult muscular dystrophies (Table 1) and builds on a discretionary selection of review articles and original articles as well as the authors’ clinical experience with the patient group. Common to these muscular diseases is that they mainly affect skeletal muscle, although other organs, especially the heart, may be affected. The inheritance pattern can be dominant, recessive or gender-bound. Most conditions are progressive. Some diagnoses progress quickly, others remain relatively stable over time. The course of the disease depends on the specific muscle disease, but even among those with the same genetic mutation, there are large individual differences. Much has been done in the diagnosis of muscular diseases, and large sack diagnosis such as limb girdle muscular dystrophy has now many genetically defined subgroups. A specific genetic diagnosis can provide knowledge of specific issues that are relevant to the particular variant of the disease. This is important in order to provide proper guidance and advice on exercise and physical activity.

**TABLE 1**

Examples of hereditary muscle diseases. AD = autosomal dominant,
We know from the normal population that lack of exercise and a sedentary lifestyle can lead to health problems and illness (3). Physical activity can to some extent prevent chronic disease and have a positive effect on, among other things, depression and pain (4). Recommendations for physical activity have been developed for the general population and for persons with failing health (5). But this is general advice that is not necessarily adapted to people with muscle disease (4). We know that lack of physical activity could lead to further loss of muscle mass, reduced walking distance and increased risk of overweight, fatigue and pain in people with muscle disease (4). Increasing the activity level must be adapted individually based on interests, wishes and functional level and contribute to an experience of coping. Physical activity need not be synonymous with organized exercise, but may include daily activities where fitness, strength, and mobility are challenged and adapted to frequency, duration and intensity (5).

Complications with significance for exercise

WEAKNESS, STIFFNESS AND CONTRACTURES
Muscle weakness is what is primarily associated with muscle disease with different distribution in each subgroup (1, 2). You often distinguish between proximal and distal muscle weakness. In addition, weakening of the back and abdominal muscles can be significant in some diagnoses (6). Muscle stiffness (myotonia) is typical, especially in dystrophy myotonica, but also in other muscle diseases (2). Contractures are common (7) and arise due to imbalance in muscle use around joints and reduced ability to use muscle power. It can also be a consequence of the dystrophic process as with Emery-Dreifuss muscle dystrophy and Ullrich’s congenital muscular dystrophy (1). Contractures can cause
secondary ailments and difficulty in using otherwise functioning muscles.

PAIN AND FATIGUE
Pain in patients with hereditary muscle diseases has been poorly studied (7), but is often described by the patients themselves. Pain may be due to secondary changes such as stiffness or inappropriate use of the body, but may also be a characteristic of the diagnosis as with facioscapulohumoral muscular dystrophy (8). Fatigue is another common challenge in muscle disease (7, 9). Here, exercise in the right amount and shape can have a positive effect (8, 10, 11).

ELEVATED CREATINE KINASE VALUES
Some muscular dystrophies and metabolic myopathies have an increased risk of rhabdomyolysis (2). Rhabdomyolysis is characterized by acute elevation of the muscle enzyme creatine kinase (CK) due to extensive muscle cell damage (12). Deposition of myoglobin in the kidneys by such acute injury is optionally treated with advanced alkaline diuresis (12). In healthy, it is suggested that a CK level > 5000 IU/l is indicative of treatment for rhabdomyolysis (12). Elevated CK values are common in muscle disease, however, it may be normal in slowly progressing myopathy and some muscle dystrophies (2). It is important to know that some patients with muscular dystrophy may have significantly higher CK values without symptoms of rhabdomyolysis. When exercising sensibly in muscle patients, CK will not normally be dangerous. Nevertheless, it is useful to know which variant of hereditary muscular disease the patient has to assess the risk of triggering rhabdomyolysis.

AFFECTION OF HEART MUSCLE
In some muscle diseases, affection of the heart muscle is common (13). This is the case, inter alia, for a large number of muscular dystrophies (Duchennes, Beckers and Emery-Dreifuss muscular dystrophies, dystrophy myotonics 1 and 2, limb girdle muscular dystrophy 1B, 2C-F, 2G and 2I) (13) and by some congenital
myopathy (14). Cardiac failure may result in cardiomyopathy or cardiac arrhythmias (14). For some muscle diseases such as limb girdle muscular dystrophy 1B (laminopathy), heart failure may be the first and sometimes only sign of muscle disease (13). The most common variant of limb girdle muscular dystrophy in Norway, type 2I, can cause dilated cardiomyopathy and conduction disorders (13). In case of infection or suspicion of affecting the heart muscle, it is therefore important to follow up with a cardiologist, regardless of whether the patient has symptoms (13, 14). In muscular diseases known to affect cardiac muscles, training is usually not dangerous, but advice on physical activity must be given in consultation with the cardiologist and preferably after the cardiac examination (13).

**Affection of Respiratory Muscles**

Some muscle diseases affect respiration, where the affection is mainly due to weakening of the respiratory muscles (15). This is the case with some metabolic myopathies and mitochondrial myopathies, in congenital myopathies and a number of muscular dystrophies (Duchennes, Beckers, Emery-Dreifuss and facioscapulohumoral muscular dystrophy, dystrophy myotonica 1 and limb-girdle muscle dystrophy 2A and 2I) (2). Endurance training, strength training and respiratory muscle training can be useful, and studies have shown improvement in, among other things, daily activities, quality of life and oxygen uptake (VO₂ max) (15). A probable effect of fitness and strength training has been shown in muscle disease and that cardiovascular changes due to exercise can be compared to that of healthy (15). Respiratory training in neuromuscular diseases is somewhat controversial and requires specific knowledge of pulmonary physiotherapy and individual muscle disease (15). In case of severe restrictive lung function, respiratory training is not safe, and it is important to have a good knowledge of different breathing aids (15).

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**Recommended physiotherapy and exercise**

PHYSIOTHERAPY

People with a hereditary muscle disease should be referred to physiotherapy for follow-up. Muscle diseases can cause improper use of the body as a result of muscle weakness, stiffness and pain. Physiotherapists have specialized expertise in assessing the muscle skeleton apparatus and can map the individual’s resources and challenges (7). They can do individual mapping of motion strategies and patterns and take steps to help the individual use their body more effectively in everyday life and during exercise. Mapping is also central to making recommendations on relevant exercises, aids, activity adaptation and total stress in everyday life.

Balance disorders are common in people with muscular disease (7). Balance training can provide better body awareness and balance confidence and reduce the risk of falls (7). Walking aids or other activity aids must be considered continuously (8). This often means that the desire for preserved activity level and independent mobility must be weighed against the risk of falls and injuries. Preventive work with exercise to maintain the movement and muscle length, and possibly the use of orthoses, is important. There is often a need for assessment of assistive technology as well as for sitting and lying positions. Immobilization after severe fractures often leads to accelerated muscle loss.

EXERCISE

People with muscular disease have previously been advised to avoid exercise because it was considered harmful to the weakened musculature (3), and one was uncertain as to whether the exercise could be effective. More recently, studies have shown that adapted physical activity can have a positive effect on people with muscle disease, e.g. by cardio fitness training or moderate strength training (3, 4, 10). No signs of exercise-induced muscle damage have been found in these patients assessed by CK measurements and muscle biopsy studies (8, 11). Therefore, the recommendations are no longer that one should avoid training, but that this must be adapted from diagnosis and function level (3, 4, 7, 10).
STRENGTH
The studies done on strength training are small, often with mixed intervention groups and often without control groups (10, 16). The results of the studies must therefore be interpreted with caution. The goal of the exercise is to maintain existing strength or reduce the progression of muscle weakness, not necessarily to strengthen the affected muscles (7). A possible increase in muscle strength is probably due to an effect on the muscles which is relatively unaffected by the muscular disease, but which may be deconditioned as a result of inactivity (10). Low intensity strength training (10-15 repetitions) may be beneficial for people who have enough muscle strength to move against gravity (4). There is no documented positive additional effects of heavy strength training compared to moderate exercise. Such training should therefore be avoided, as it can lead to overload (4). For the same reason, heavy eccentric strength training (9) is also not recommended. It is considered to be a frequent trigger for serious muscle damage also in muscle-healthy (12). Studies on limb girdle muscular dystrophy, facioscapulohumoral muscular dystrophy, dystrophy myotonica 1 and mitochondrial myopathies have shown that moderate strength training is safe and can have an effect on muscle strength and endurance, but results vary (3, 4, 9).

Although the studies point positively towards moderate strength training, there is currently no basis for making general recommendations for muscle diseases (10, 16). There are large individual differences in terms of how hard exercise is tolerated, both in different muscle diseases and among those with the same genetic variant.

ENDURANCE TRAINING
Moderate endurance training where the muscles are adequately supplied with oxygen, but one can easily breathe, can improve the cardiovascular function of people with muscle disease (4). Small endurance studies have been performed on limb girdle muscular dystrophy (9), dystrophy myotonica 1, metabolic myopathy (McArdle's disease) and mitochondrial myopathies where this has not done any harm and has had a moderate positive effect (3, 4).
Ergometer cycling studies have also been carried out on facioscapulohumoral muscular dystrophy which have shown that this is safe and that it can improve endurance (8, 11). Relieved walking training and pool training are other examples of training that some may enjoy and potentially improve endurance (3).

Summary

We recommend avoiding inactivity leading to increased decomposition and further reduction of muscle strength beyond the primary muscle disorder. This can in turn lead to increased fatigue, pain and reduction of movement and function. Adapted training and physical activity are generally safe, but must be adapted to the individual’s diagnosis, level of function and lifestyle.

Adapted physical activity that takes into account disease-specific issues is recommended, as it can facilitate daily activities and improve physical fitness. In order to get advice on the type of activity, it may be useful to get follow-up from the physiotherapist with neurological competence and insight into adapted training and dosing.
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