



Becker Muscular Dystrophy



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This guide aims to provide information and advice about Becker muscular dystrophy (BMD) but is not intended to replace the services of your healthcare provider. If you are an adult who has BMD you should consult your healthcare provider about all matters relating to your health. Similarly, if you are a parent or carer of someone with BMD, you should consult the person's healthcare provider about all matters relating to their health, involving them in discussions too if age appropriate.

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1. What is Becker muscular dystrophy?

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Becker muscular dystrophy (BMD) is a hereditary condition that causes progressive muscle weakness and wasting, particularly in muscles located in the hips, pelvic region, thighs, and shoulders. In addition to these skeletal muscles, the heart muscle may also be affected over time. BMD is classified as one of the nine primary forms of muscular dystrophy, a broader category of genetic disorders characterized by the gradual breakdown and weakening of voluntary muscles.

This specific type of muscular dystrophy is part of a subgroup known as dystrophinopathies, which are caused by mutations in the gene responsible for producing dystrophin - a protein crucial for muscle function. Dystrophinopathies include not only Becker muscular dystrophy but also Duchenne muscular dystrophy (DMD), as well as an intermediate form that shares features of both conditions. While DMD and BMD are related, they differ significantly in terms of onset, severity, and progression.



BMD was first identified and described in the 1950s by Dr. Peter Emil Becker, a German physician, who recognized it as a less severe variant of DMD. Unlike Duchenne muscular dystrophy, which typically presents in early childhood and progresses rapidly, Becker muscular dystrophy often appears during adolescence or early adulthood and advances more slowly. Individuals with BMD tend to retain muscle strength and mobility longer than those with DMD, and the degree of muscle impairment can vary widely from person to person.

Despite the milder progression of skeletal muscle involvement in BMD, the heart muscle (particularly the cardiac muscle) can be affected in ways similar to DMD. Cardiomyopathy, or disease of the heart muscle, is a common complication and may require ongoing cardiac monitoring and treatment.

In summary, Becker muscular dystrophy is a genetic, progressive neuromuscular disorder that primarily impacts voluntary muscles and the heart. While it shares genetic origins and similarities with Duchenne muscular dystrophy, it typically presents later and progresses at a slower rate, offering affected individuals a wider range of physical abilities over time.

2. Symptoms of Becker muscular dystrophy

The content of this section is from source 4 in the list of references.

The symptoms associated with Becker muscular dystrophy can differ greatly from one individual to another. While some individuals may only exhibit mild physical limitations, others may face more substantial impairments. BMD can be diagnosed at any point from early childhood through adulthood.

2.1. Muscle Weakness

Muscle weakness in BMD usually begins in the muscles of the pelvic area and lower limbs. This early weakness may make it challenging for affected individuals to engage in physical activities like running or participating in sports. One of the earliest indicators can be muscle cramps triggered by physical exertion. In some rare instances, children may notice dark red or brown-colored urine following exercise. This could be a sign of rhabdomyolysis, a condition involving the breakdown of muscle tissue, which requires immediate medical evaluation and intervention.

As the disorder progresses - typically during the teenage years or early adulthood - the muscle weakness becomes more pronounced. Everyday tasks such as walking long distances, climbing stairs, or rising from a seated or lying position may become increasingly difficult. Over time, weakness often spreads to the upper body, including the shoulders and arms. Activities such as lifting heavy items or reaching overhead can become problematic.

In middle age, usually during the 40s or 50s, mobility can become significantly impaired. Individuals may begin to experience frequent falls and difficulty walking. In some cases, the use of a wheelchair may become necessary. In more rapidly progressing cases of BMD, mobility aids might be needed earlier in life.

2.2. Cardiac Involvement

Heart complications are a known feature of BMD and typically present in the form of cardiomyopathy, a disease that weakens the heart's ability to pump blood effectively. Although these heart issues can occur at any age, they may initially be silent, showing no clear symptoms. In fact, for some, cardiac problems may be the first noticeable manifestation of BMD.

Signs that may point to heart involvement include breathlessness (especially during activity), irregular or rapid heartbeat (palpitations), dizziness, or sensations of chest discomfort or tightness. Any of these symptoms should be taken seriously and promptly discussed with healthcare providers, as early detection and treatment are crucial.

2.3. Breathing Difficulties

As BMD progresses, especially in individuals who use wheelchairs or are in the later stages of the condition, respiratory muscles may also become weakened. This can lead to diminished lung capacity and a reduced ability to clear mucus from the airways due to a weakened cough. As a result, the risk of developing chest infections increases, sometimes requiring antibiotics or other treatments.

Some individuals may also develop breathing-related issues during sleep. Conditions such as sleep apnea and nocturnal hypoventilation (shallow or slowed breathing at night) can lead to poor sleep quality and daytime fatigue. These problems may require specialized respiratory support, including non-invasive ventilation during sleep.

2.4. Neurodevelopmental and Behavioral Aspects

In addition to its physical effects, BMD may also influence brain function. This is because dystrophin, the protein that is deficient or abnormal in BMD, is also present in certain areas of the brain. As a result, individuals with BMD may have an increased risk of cognitive or behavioral challenges.

Commonly reported conditions include learning disabilities, autism spectrum disorder (ASD), attention deficit hyperactivity disorder (ADHD), obsessive-compulsive disorder (OCD), and anxiety disorders. Early identification of these difficulties is vital to ensure appropriate interventions and support systems are in place. For children, this might include educational support, tailored learning plans, and therapeutic services. Adults may benefit from accommodations in the workplace and access to mental health services. In some cases, consultation with neurodevelopmental or psychiatric specialists may be needed, and medication might be part of the treatment plan.

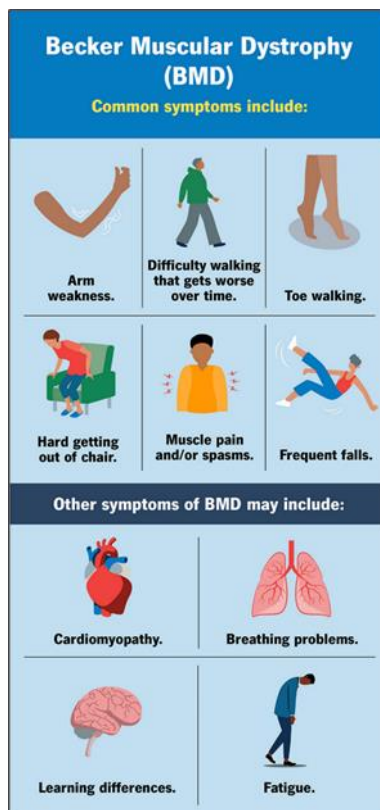


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3. How Becker muscular dystrophy is caused

3.1. Genetic changes

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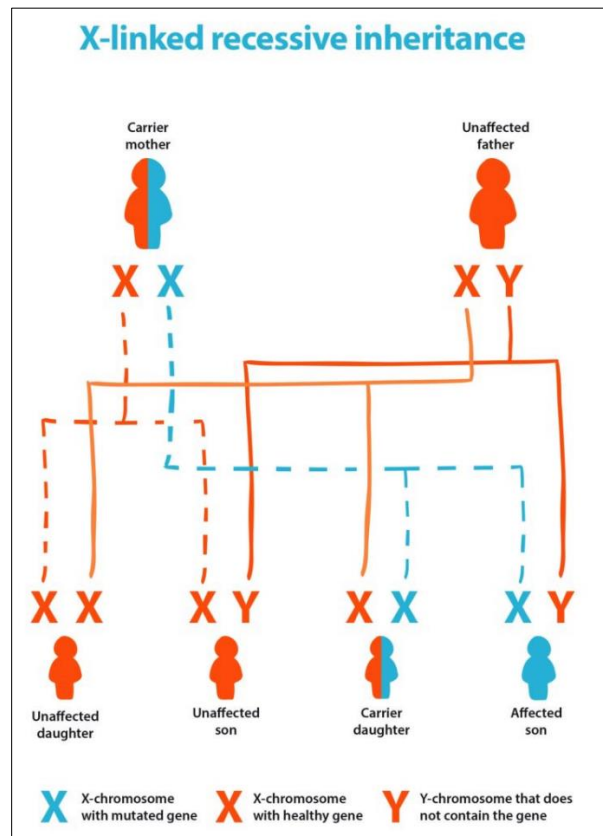
BMD results from a mutation in the gene responsible for producing dystrophin, a protein essential for maintaining muscle integrity. Although individuals with BMD do produce dystrophin, the amount or

quality is insufficient, leading to gradual muscle weakening as muscle fibers become damaged over time.

Duchenne muscular dystrophy (DMD) is a more severe form of the condition, caused by a total absence of dystrophin. DMD typically appears in early childhood and progresses rapidly, often resulting in loss of movement and serious medical complications. In contrast, BMD tends to have a slower progression and milder symptoms because some functional dystrophin is still present.

3.2. Inheritance

BMD follows an X-linked recessive inheritance pattern, meaning the faulty gene is located on the X chromosome. Since males have only one X chromosome, they are more susceptible to developing the condition. Females, with two X chromosomes, are less likely to show symptoms because they typically have one functioning copy of the gene to compensate.



4. The differences between Duchenne and Becker muscular dystrophy

Duchenne muscular dystrophy (DMD) and Becker muscular dystrophy (BMD) are both inherited in an X-linked recessive pattern and result from mutations in the gene responsible for producing dystrophin, a

protein vital for muscle function. These conditions lead to progressive muscle weakening and damage. Although they are related, DMD is more severe, begins earlier, and advances more quickly than BMD.

Key Differences Between DMD and BMD are:

Duchenne Muscular Dystrophy (DMD):

- Age of Onset: Typically begins before the age of six.
- Severity: Considered the most severe form of dystrophinopathy, leading to rapid muscle deterioration.
- Disease Course: Weakness advances quickly, often resulting in loss of the ability to walk during early adolescence.
- Dystrophin Levels: The dystrophin protein is nearly or completely absent.
- Life Expectancy: Individuals often live into their mid-to-late 20s, though advancements in care may extend lifespan.

Becker Muscular Dystrophy (BMD):

- Age of Onset: Usually appears later, between ages 8 and 12.
- Severity: Milder than DMD, with a slower and less aggressive progression.
- Disease Course: Muscle weakness develops more gradually, and walking ability may be preserved well into adulthood.
- Dystrophin Levels: Reduced levels of functional dystrophin are present.
- Life Expectancy: Generally longer than in DMD, although complications - particularly related to heart function - can still arise.

5. Manifesting carriers

The content of this section is from source 4 in the list of references.

Men affected by Becker muscular dystrophy (BMD) cannot transmit the altered gene to their sons, since males pass on a Y chromosome to male offspring. However, all of their daughters will inherit the affected X chromosome, making them carriers. Most female carriers do not show signs of the condition, but a minority may develop symptoms such as muscle weakness or heart-related issues later in life. When symptoms do appear, these individuals are referred to as manifesting carriers of BMD.

6. Can Becker muscular dystrophy be prevented?

The content of this section is from source 1 in the list of references.

Since Becker muscular dystrophy (BMD) is an inherited condition, there's nothing you can do to prevent it. If you have BMD, or if you're concerned about the risk of passing on BMD or other genetic conditions before trying to have a biological child, talk to your healthcare provider about genetic counseling.

7. Getting a diagnosis for Becker muscular dystrophy

The content of this section is from source 4 in the list of references.

If symptoms of muscle weakness or wasting are observed, a general practitioner (GP) can refer the individual to a neurologist for further evaluation. Neurologists are medical specialists who focus on disorders affecting the muscles and nervous system. To make a diagnosis, the neurologist may use several tools, including a physical exam, blood tests to check creatine kinase (CK) levels, and genetic testing to examine the dystrophin gene. In certain cases, a muscle biopsy may be performed, which involves removing a small sample of muscle tissue to assess whether dystrophin is present. Occasionally, BMD is identified during an evaluation for heart issues, even when no other muscle symptoms are present.

8. Condition management

The content of this section is from source 4 in the list of references.

A multidisciplinary approach plays a vital role not only in managing Becker muscular dystrophy and its symptoms but also in supporting overall health and wellbeing. Because BMD can vary significantly between individuals - starting at different ages and affecting people in different ways - care must be tailored to each person's needs.

8.1. Access to a Healthcare Team

People living with BMD benefit from being cared for by a team of healthcare professionals with different specialties. A neurologist often serves as the primary coordinator of care. If you're not currently seeing a neurologist or a relevant specialist, it's advisable to speak with your GP about arranging a referral.

8.2. Heart Health

Cardiac complications are a common feature of BMD, and they may not always cause noticeable symptoms. Routine check-ups with a cardiologist are essential for early detection and management. Typical heart evaluations include:

- **Electrocardiogram (ECG):** Measures the heart's electrical activity
- **Echocardiogram (ECHO):** Assesses heart structure and performance
- **Holter Monitor:** Tracks heart rhythms over a longer duration

Early use of medications such as ACE inhibitors and beta-blockers can help protect heart function and reduce the risk of serious issues like cardiomyopathy or heart failure. It's important to report symptoms like chest discomfort, dizziness, palpitations, or breathlessness to your medical team promptly.

8.3. Breathing Support

As muscle weakness progresses, respiratory issues may develop. Lung function may be assessed through tests such as forced vital capacity (FVC). Sleep studies might also be recommended to evaluate nighttime breathing. If needed, treatments like breathing exercises, cough assist devices, and non-invasive ventilation (NIV) may be used to maintain respiratory health.

8.4. Spine Monitoring

BMD can sometimes lead to scoliosis, a sideways curvature of the spine caused by uneven muscle strength. Regular monitoring is important. In some cases, spinal bracing can slow progression. Staying mobile and physically active may help reduce the risk. Surgery might be necessary for more severe curvature.

8.5. Physical Activity and Physiotherapy

Remaining active can help preserve muscle strength and joint flexibility. A physiotherapist (a specialist in movement and rehabilitation) can develop an individualized exercise plan.

Key recommendations include:

- Regular stretching to reduce stiffness
- Strengthening exercises to delay muscle weakening
- Aerobic activities such as walking, cycling, or swimming to support cardiovascular and respiratory health

It's important to balance activity with adequate recovery time. Some mild muscle soreness is expected, but persistent or intense pain should be reported.

8.6. Steroid Treatment

While corticosteroids have shown benefits in slowing the progression of Duchenne muscular dystrophy, they are not commonly prescribed for BMD. In more severe BMD cases, they may be considered, but due to potential side effects, their use should be carefully evaluated by a specialist.

9. Anesthesia safety

The content of this section is from source 4 in the list of references.

Anaesthesia is used to prevent pain during surgical procedures, but individuals with Becker muscular dystrophy (BMD) face an increased risk of complications when undergoing general anaesthesia. These risks are often linked to underlying heart or breathing issues, as well as potential negative reactions to specific anaesthetic agents. It is essential that the anaesthetist and surgical team are fully informed about the person's BMD diagnosis and any existing cardiac or respiratory concerns. A thorough pre-operative evaluation and careful post-operative monitoring are strongly recommended. Because each person with BMD has unique needs, anaesthetic plans should be tailored individually to ensure the highest level of safety.

Certain anaesthetic agents must be avoided, including:

- Suxamethonium (succinylcholine)
- Inhalational anaesthetics
- Neuromuscular blocking agents

These drugs can trigger acute rhabdomyolysis, a life-threatening condition involving rapid breakdown of muscle tissue. Signs of rhabdomyolysis include severe muscle pain and urine that appears dark red or brown. If these symptoms occur, immediate hospital admission and treatment are critical.

10. Becker Emergency Information

The content of this section is from source 2 in the list of references.

10.1. Cardiac Management

- Individuals with Becker muscular dystrophy (BMD) are at risk of developing cardiomyopathy, even if their muscle symptoms are mild. Regular cardiac evaluations are essential for all patients, regardless of severity.
- Early treatment with medications such as ACE inhibitors and beta-blockers can help delay the progression of heart muscle disease.
- Heart rhythm disturbances should be considered in anyone experiencing palpitations, dizziness, or near-fainting episodes. These symptoms warrant further investigation through electrocardiograms (ECG), 24-hour Holter monitoring, or similar diagnostic tests.

10.2. Respiratory Support

- Chronic breathing insufficiency in BMD may not present with obvious signs. Subtle symptoms can include morning headaches, poor-quality sleep, daytime fatigue, reduced appetite, weight loss, or excessive daytime sleepiness. Respiratory issues should also be considered if a chest infection occurs.
- If oxygen therapy is needed during a respiratory crisis, it must be used cautiously, with close monitoring for signs of acute respiratory failure. An arterial blood gas test is essential to evaluate oxygen, carbon dioxide, and bicarbonate levels. Non-invasive ventilation (NIV), with carefully regulated oxygen, may be necessary.
- Techniques such as assisted coughing, chest physiotherapy, and breath-stacking using an AMBU bag can help clear secretions from the lungs. A mechanical cough assist device may also be beneficial. These methods should only be performed by qualified professionals.

10.3. Anaesthetic Guidelines

- Only intravenous general anaesthetics should be used; suxamethonium (succinylcholine) must be avoided.
- Inhaled anaesthetic agents are not recommended due to the risk of complications.
- Local anaesthetics and nitrous oxide are considered safe, including for minor dental treatments.

10.4. Additional Recommendations and Safety Measures

- Vaccinations should be kept current. However, live vaccines should be avoided if the individual is on corticosteroid therapy.

- Wearing a medical alert bracelet is strongly advised.

Note: Blood tests may show elevated AST and ALT liver enzymes, which is common in BMD due to muscle breakdown. This should not automatically lead to liver-specific investigations unless other signs point to liver disease.

Please contact your local MDFSA office for a pocket-size emergency card.

11. Exercising with a muscle wasting condition

The content of this section is from source 4 in the list of references.

Engaging in physical activity offers numerous health benefits. However, for individuals with muscle-wasting conditions, it's important to choose appropriate strengthening exercises with care. Before beginning any exercise program - especially if you are still being diagnosed or undergoing heart-related tests - it's advisable to consult with your doctor or physiotherapist to ensure the activities are safe and suitable for your condition.

11.1. General health benefits of keeping active and exercising

Although researchers have not yet determined the ideal type or amount of exercise for individuals with muscle-wasting conditions, there is general agreement that avoiding physical activity can be harmful, and that staying active in any safe and manageable way is beneficial to overall health.

Benefits of Staying Physically Active:

- Lowers the risk of developing other health conditions such as heart disease and type 2 diabetes
- Enhances heart and lung function, which supports stamina and endurance
- Helps keep unaffected muscles strong, reducing weakness not directly related to the muscle condition
- Supports the ability to carry out daily activities and tasks
- Prevents additional weakness caused by lack of use
- Can help reduce feelings of fatigue
- Promotes stronger bones and helps maintain bone density
- Aids in weight management
- Boosts mental wellbeing and mood
- Supports stress management
- Encourages better quality sleep
- May assist in pain reduction
- Helps to preserve or improve joint flexibility and range of motion

11.2. Tips to help you get active

- If you haven't been active for a while, begin slowly and increase your activity level gradually. Everyday tasks such as cleaning the house or walking to your destination also count as forms of exercise.

- Set clear goals to stay motivated. For instance, aim not to sit still for more than 30 minutes at a time, or try to fit in physical activity at least three times per week.
- Whenever possible, exercise with others even if it's online. Sharing your fitness plans with friends or family can help you stay committed.
- Keep a log of your physical activity, including what you did and when, so you can track your progress over time.
- Using step counters or activity-tracking apps on your phone can also be a helpful way to monitor how much you move each day.

11.3. Planning Your Physical Activity

Try to schedule your activities or workouts on a weekly basis. Vary your routine to allow different muscle groups time to rest and recover. For instance, if one session involves a lot of walking (aerobic exercise using your legs), consider switching to strengthening exercises that target areas like your arms, legs, or core in your next session.

Always begin with a warm-up to prepare your body and end with a cool-down to help it recover.

11.4. What to Expect from Your Body

During aerobic exercises such as walking, you should feel mildly out of breath but still able to speak comfortably. Your heart rate will increase, and you might sweat slightly.

When starting new strength-based activities, some muscle soreness is normal, but this should subside within 48 hours. As you continue exercising regularly, you'll likely notice less discomfort afterward.

11.5. Safety Tips and Precautions

- Avoid pushing yourself to the point of exhaustion. Take breaks when needed.
- Space out your physical tasks throughout the day or week, considering other demands on your energy. Frequent, shorter sessions are often more effective.
- Your activity should not leave you overly fatigued the next day.
- Try not to overdo exercises that involve lengthening muscles under tension (eccentric movements), such as repeated squats.
- It's important to distinguish between muscle tiredness and weakness. Tired muscles generally recover with rest. Be mindful of your balance when fatigued, as your risk of falling increases.
- Exercise should not cause pain.
- Maintain proper posture and alignment during exercise to protect your joints.

In rare cases, individuals with muscle-wasting conditions may notice a dark color in their urine - similar to black tea or cola - after physical activity. If this happens, go to the emergency department, as it could be a sign of myoglobinuria, a potentially serious condition.

11.6. What type of activity or exercise can I do?

The content of this section is from source 4 in the list of references.

The most suitable type of physical activity or exercise can vary greatly between individuals. It's important to choose something that feels safe, is enjoyable, achievable, and fits into your daily schedule. What works best for you will depend on your personal needs, preferences, and physical condition. For instance, if balance is a concern, a stationary bike that's easy to mount and dismount might be a better option.

There are three main categories of physical activity:

- Aerobic
- Strength-based
- Stretching

1) Aerobic Activity

Aerobic exercises are those that raise your heart rate and breathing, involving large muscle groups and lasting for a few continuous minutes.

Examples include:

- Walking
- Swimming
- Cycling
- Moving your wheelchair
- Doing household chores
- Gardening

These activities support heart and lung health, improve circulation, and contribute to better overall fitness. Even modest increases in daily physical activity can reduce the risk of chronic illnesses and improve your ability to handle everyday tasks.

Before beginning any aerobic program, consult your physiotherapist, neurologist, or cardiologist to determine what's suitable for your condition.

How Often Should You Do Aerobic Activity?

Start by noting what kind of activity you can manage and how long you can do it comfortably. Gradually work on increasing either the duration or the frequency.

On days when fatigue is worse, you might not be able to do much - that's okay. This is a normal part of the process, and you can continue when you feel better. It's important to schedule rest periods to allow your muscles time to recover and prevent excessive tiredness. Try to spread your activity across the week to suit your routine.

How Intense Should It Be?

Aim for a moderate level of effort. This means:

- Feeling warmer and perhaps sweating lightly
- Breathing more deeply, though you should still be able to hold a conversation comfortably

Evaluate how you feel at the start of each session to ensure you're working within your limits. This will help you set a safe and realistic baseline.

If you plan longer or more intense sessions, be sure to include a 3–5 minute warm-up to gradually raise your body temperature and a 5–10 minute cool-down to bring your heart rate down and prevent stiffness afterward. Your therapist can guide you on warm-up and cool-down routines that suit your needs.

2) Strengthening exercises

Strengthening activities typically focus on the main muscle groups and may include movements using your own body weight, resistance bands, light weights, or exercises that work against gravity.

It's important to target muscles in your arms, legs, abdomen, and back - commonly referred to as the "core." Building core strength can improve your posture and balance, lowering your risk of falls and making everyday tasks like transferring in and out of a wheelchair easier.

Try to avoid overloading your muscles with repetitive movements that involve lengthening the muscle under tension - known as eccentric exercises (such as repeated squats). These place greater strain on the muscles, increasing the risk of soreness or possible muscle damage.

Since everyone's needs are different, speak with your physiotherapist to identify which muscle groups to focus on and the best way to strengthen them.

Rather than trying to increase muscle size or maximum strength, the goal should be to improve muscular endurance. Endurance allows you to perform tasks for longer before becoming fatigued, for example, being able to climb more stairs without needing a break.

Some muscles affected by a muscle-wasting condition may not be able to regain strength. However, it's still important to preserve existing muscle function for as long as possible.

How Often Should You Do Strengthening Exercises?

Aim to include strength-focused exercises in your routine twice a week. Avoid doing them on back-to-back days, as muscles need time to recover.

During a single session, vary the muscle groups you work. For instance, after doing an exercise for your arms, switch to one for your legs.

If you notice increased muscle weakness, fatigue, or soreness that interferes with your daily routine, reduce your activity or consult your physiotherapist for guidance.

How Intense Should Strengthening Activities Be?

Low to moderate resistance exercises are generally safe for individuals with muscle-wasting conditions.

- Avoid heavy lifting, as it may strain delicate muscles and increase stress on joints and ligaments.
- Maintain good posture and alignment during all exercises to protect your back and neck.

- Focus on increasing repetitions instead of resistance.
- Stop the activity if you experience excessive shaking or jerky movements.

It's normal to feel mild soreness when beginning new strengthening exercises, but this should resolve within 48 hours.

3) Stretching exercises

Including stretching in your regular activity or exercise routine is highly recommended. It's best to perform stretches when your muscles and joints are warm, such as after exercising or following a bath or shower, when your body is more relaxed and flexible.

Maintaining good flexibility and joint range is essential for performing daily tasks. For instance, having enough mobility in your shoulders can make actions like getting dressed or washing much easier.

In certain rare neuromuscular conditions, joint or spinal stiffness (contractures) can occur. In such cases, the focus of stretching is to maintain your current level of flexibility rather than trying to increase it.

Be cautious not to overstretch muscles that are weak or joints that are already highly flexible. If you have areas that feel particularly tight or restricted, you might benefit from a personalized stretching plan. A physiotherapist can guide you on which areas to target and how to stretch safely.

Gentle movement practices such as yoga or Pilates can also support flexibility and can be modified to suit your specific physical needs.

If you're unable to stretch independently, a family member or caregiver can assist you with passive stretches.

How Often and How Long Should I Stretch?

Stretching is most beneficial when done consistently. Try incorporating it into your daily routine, for example, stretching your calves while you brush your teeth.

Aim to hold each stretch for about 30 to 40 seconds. Movements should be slow and steady. Avoid bouncing or jerking during the stretch.

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