MUSCULAR DYSTROPHY (MD) PRACTICE TEST (SAMPLE)

STUDY GUIDE



SAMPLE STUDY GUIDE WITH LIMITED QUESTIONS

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Introduction

Preparing for a certification exam can feel overwhelming, but with the right tools, it becomes an opportunity to build confidence, sharpen your skills, and move one step closer to your goals. At Examzify, we believe that effective exam preparation isn't just about memorization, it's about understanding the material, identifying knowledge gaps, and building the test-taking strategies that lead to success.

This guide was designed to help you do exactly that.

Whether you're preparing for a licensing exam, professional certification, or entry-level qualification, this book offers structured practice to reinforce key concepts. You'll find a wide range of multiple-choice questions, each followed by clear explanations to help you understand not just the right answer, but why it's correct.

The content in this guide is based on real-world exam objectives and aligned with the types of questions and topics commonly found on official tests. It's ideal for learners who want to:

- Practice answering questions under realistic conditions,
- Improve accuracy and speed,
- Review explanations to strengthen weak areas, and
- Approach the exam with greater confidence.

We recommend using this book not as a stand-alone study tool, but alongside other resources like flashcards, textbooks, or hands-on training. For best results, we recommend working through each question, reflecting on the explanation provided, and revisiting the topics that challenge you most.

Remember: successful test preparation isn't about getting every question right the first time, it's about learning from your mistakes and improving over time. Stay focused, trust the process, and know that every page you turn brings you closer to success.

Let's begin.

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How to Use This Guide

This guide is designed to help you study more effectively and approach your exam with confidence. Whether you're reviewing for the first time or doing a final refresh, here's how to get the most out of your Examzify study guide:

1. Start with a Diagnostic Review

Skim through the questions to get a sense of what you know and what you need to focus on. Your goal is to identify knowledge gaps early.

2. Study in Short, Focused Sessions

Break your study time into manageable blocks (e.g. 30 – 45 minutes). Review a handful of questions, reflect on the explanations.

3. Learn from the Explanations

After answering a question, always read the explanation, even if you got it right. It reinforces key points, corrects misunderstandings, and teaches subtle distinctions between similar answers.

4. Track Your Progress

Use bookmarks or notes (if reading digitally) to mark difficult questions. Revisit these regularly and track improvements over time.

5. Simulate the Real Exam

Once you're comfortable, try taking a full set of questions without pausing. Set a timer and simulate test-day conditions to build confidence and time management skills.

6. Repeat and Review

Don't just study once, repetition builds retention. Re-attempt questions after a few days and revisit explanations to reinforce learning. Pair this guide with other Examzify tools like flashcards, and digital practice tests to strengthen your preparation across formats.

There's no single right way to study, but consistent, thoughtful effort always wins. Use this guide flexibly, adapt the tips above to fit your pace and learning style. You've got this!

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Questions

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1. What genetic mechanism causes DM1?

A. Point mutation in the DMPK gene

B. Deletion in the DMPK gene

C. Duplication of the DMPK gene

D. CTG trinucleotide repeat expansion in the DMPK gene

2. If initial deletion/duplication analysis is negative, what is the next step in molecular testing for DMD?

A. Repeat deletion/duplication analysis

B. Sequencing of the DMD gene

C. Muscle biopsy

D. Karyotyping

3. What is a key clinical takeaway for OT management in muscular dystrophy?

A. Keep the person doing what they can for as long as possible

B. Push for rapid progression to full independence

C. Delay all intervention until symptoms are severe

D. Limit therapy to passive activities

4. LGMD2I is caused by mutations in which gene?

A. FKRP

B. DMD

C. LAMA2

D. COL6A1

5. Weakness in DMD follows which pattern?

A. Proximal to distal (hips shoulders distal muscles)

B. Distal to proximal

C. Facial to limb

D. Trunk to limbs

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6. Calf pseudohypertrophy in Duchenne muscular dystrophy is primarily due to replacement of muscle by which tissues?

A. Edema and inflammatory infiltrates within the muscle

B. Adipose tissue infiltrating muscle but not fibrous tissue

C. Fibrous tissue replacing muscle without fat

D. Fat and fibrous tissue replacing muscle

7. To support participation across the lifespan, OT should ?

A. Tailor tasks and environments to changing abilities

B. Use a fixed plan for all ages

C. Focus only on hospital-based care

D. Remove community participation

8. In MD, cognition is typically what?

A. Education impact

B. Cognition in MD

C. Psychosocial impact

D. ADL impact in MD

9. What is the typical inheritance pattern for FSHD1?

A. Autosomal recessive

B. Autosomal dominant (with a permissive 4qA haplotype)

C. X-linked recessive

D. Mitochondrial inheritance

10. Distal weakness pattern involving face, hands, and forearms is characteristic of which dystrophy?

A. Myotonic Dystrophy

B. Becker Muscular Dystrophy

C. Limb-Girdle Muscular Dystrophy

D. Duchenne Muscular Dystrophy

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Answers

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1. D
2. B
3. A
4. A
5. A
6. D
7. A
8. B
9. B
10. A

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Explanations

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1. What genetic mechanism causes DM1?

A. Point mutation in the DMPK gene

B. Deletion in the DMPK gene

C. Duplication of the DMPK gene

D. CTG trinucleotide repeat expansion in the DMPK gene

DM1 is caused by a CTG trinucleotide repeat expansion in the DMPK gene. When the number of CTG repeats becomes large, the transcribed RNA contains long CUG repeats that act as a toxic RNA, sequestering splicing factors and causing mis-splicing of many transcripts. This RNA gain-of-function effect leads to the muscle weakness, myotonia, and multisystem features seen in DM1 and explains inheritance with anticipation. In contrast, a point mutation would change a single amino acid, a deletion would remove part of the gene, and a duplication would alter gene dosage—none of which produce the toxic RNA–driven mis-splicing characteristic of DM1.

2. If initial deletion/duplication analysis is negative, what is the next step in molecular testing for DMD?

A. Repeat deletion/duplication analysis

B. Sequencing of the DMD gene

C. Muscle biopsy

D. Karyotyping

When the initial screening for large deletions or duplications in the DMD gene comes back negative, the next step is sequencing the DMD gene. This is because many patients carry small-scale mutations—such as point mutations or small insertions/deletions—that do not change copy number and, therefore, aren't detected by CNV analysis. Sequencing examines all coding exons and essential splice sites, and is typically done with next-generation sequencing (often with confirmation by another method if needed). If sequencing also finds nothing, further testing like RNA analysis to detect deep intronic or regulatory variants may be considered. Repeating the deletion/duplication test wouldn't reveal small variants; muscle biopsy provides tissue pathology rather than mutation information; and karyotyping looks for whole-chromosome problems, not gene-level changes.

3. What is a key clinical takeaway for OT management in muscular dystrophy?

A. Keep the person doing what they can for as long as possible

B. Push for rapid progression to full independence

C. Delay all intervention until symptoms are severe

D. Limit therapy to passive activities

Preserving function and independence by enabling ongoing participation is the goal of occupational therapy in muscular dystrophy. Because MD causes progressive weakness, the OT focuses on helping the person stay as active as possible for as long as feasible, using strategies like pacing, energy conservation, activity modification, and joint protection. Providing or teaching adaptive equipment, ergonomic techniques, and simple home modifications supports engagement in meaningful daily activities while minimizing fatigue and risk of injury. This approach helps maintain range of motion, prevents contractures, and preserves autonomy in everyday tasks, which are all essential for quality of life as the disease progresses. Pushing for rapid progression to full independence isn't realistic or safe given the gradual decline in strength, and it can lead to overexertion or injury. Delaying all intervention until symptoms are severe misses opportunities to slow functional loss and complicates later rehabilitation. Limiting therapy to passive activities neglects the need to actively participate, build skills, and strategize around remaining abilities, which undermines long-term independence.

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4. LGMD2I is caused by mutations in which gene?

A. FKRP

B. DMD

C. LAMA2

D. COL6A1

Mutations in FKRP cause LGMD type 2I. FKRP encodes fukutin-related protein, which is involved in the glycosylation of alpha-dystroglycan. Proper glycosylation of alpha-dystroglycan is essential for linking the muscle cell membrane to the extracellular matrix; when FKRP is defective, this connection weakens, leading to the proximal muscle weakness seen in LGMD2I. This form is typically inherited in an autosomal recessive pattern. The other genes listed are linked to different muscular dystrophies: DMD encodes dystrophin (Duchenne/Becker muscular dystrophy), LAMA2 encodes merosin (alpha2-laminin) associated with MDC1A, and COL6A1 encodes collagen VI (Ullrich/Bethlem myopathy).

5. Weakness in DMD follows which pattern?

A. Proximal to distal (hips shoulders distal muscles)

B. Distal to proximal

C. Facial to limb

D. Trunk to limbs

Duchenne muscular dystrophy weakens proximal muscles first, especially the hip and pelvic girdle, so individuals often have trouble rising from the floor (Gowers' sign). As the disease progresses, the weakness extends to the proximal muscles of the shoulders and upper arms, with distal muscles affected later. This proximal-to-distal pattern is characteristic, whereas distal-to-proximal patterns or facial-to-limb or trunk-to-limb sequences are seen in other conditions. Therefore, the weakness in DMD follows a proximal-to-distal progression.

6. Calf pseudohypertrophy in Duchenne muscular dystrophy is primarily due to replacement of muscle by which tissues?

A. Edema and inflammatory infiltrates within the muscle

B. Adipose tissue infiltrating muscle but not fibrous tissue

C. Fibrous tissue replacing muscle without fat

D. Fat and fibrous tissue replacing muscle

Calf pseudohypertrophy in Duchenne muscular dystrophy happens because degenerating muscle fibers are progressively replaced with noncontractile tissue. In DMD, the lack of dystrophin leads to repeated cycles of muscle fiber death, and the muscle is gradually replaced not just by fat but also by fibrous connective tissue. This combination of fat and fibrous tissue expands the calf region, making it look larger while the muscle's strength is actually decreased. Edema and inflammatory infiltrates describe an active, acute process, which isn't the chronic replacement pattern seen in established pseudohypertrophy. Fat infiltration alone doesn't capture the full, stiff, nonfunctional replacement that fibrous tissue adds, and fibrosis plus fat together best explain the enlargement with functional loss.

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7. To support participation across the lifespan, OT should ?

A. Tailor tasks and environments to changing abilities

B. Use a fixed plan for all ages

C. Focus only on hospital-based care

D. Remove community participation

Adapting activities and environments to the person's changing abilities is central to OT across the lifespan. Occupational therapy aims to help people participate in meaningful activities by adjusting tasks, supports, and the surroundings as abilities shift with aging, development, or health changes. For example, someone might benefit from simpler task steps, assistive devices, or a modified home layout to maintain independence and safety at home, at school, work, or in the community. This flexible, person-centered approach keeps participation realistic and relevant as needs evolve. Choosing a fixed plan for all ages ignores how abilities and contexts change, making it hard to stay effective over time. Focusing only on hospital-based care misses opportunities to support participation in everyday life like home routines or community activities. Removing community participation would directly undermine the goal of helping people stay engaged in meaningful roles across their lives.

8. In MD, cognition is typically what?

A. Education impact

B. Cognition in MD

C. Psychosocial impact

D. ADL impact in MD

In muscular dystrophy, thinking and problem-solving abilities are usually preserved. The disease targets skeletal muscles and causes weakness and degeneration, but the brain isn't affected in the same direct way, so overall intellectual function tends to be normal. Some individuals, particularly with Duchenne-type changes, may have learning or attention challenges, but these are not a defining feature of the condition and occur in a minority. So the statement that cognition in MD is typically preserved best captures the common pattern. Other aspects like education, psychosocial well-being, or daily living activities can be impacted by physical limitations or chronic illness, but they don't reflect changes in cognitive function itself.

9. What is the typical inheritance pattern for FSHD1?

A. Autosomal recessive

B. Autosomal dominant (with a permissive 4qA haplotype)

C. X-linked recessive

D. Mitochondrial inheritance

FSHD1 follows an autosomal dominant pattern, meaning a single pathogenic allele can cause the disease in a family. What makes it specific is that the disease only manifests when the D4Z4 repeat contraction on chromosome 4q35 occurs on a chromosome with the permissive 4qA haplotype. That 4qA haplotype provides the necessary polyadenylation signal for DUX4, so the misexpressed DUX4 gene leads to muscle weakness. If the contraction sits on a non-permissive 4qB haplotype, DUX4 isn't properly expressed and the disease doesn't occur. So the inheritance is autosomal dominant with a haplotype requirement, though penetrance and age of onset can vary.

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10. Distal weakness pattern involving face, hands, and forearms is characteristic of which dystrophy?

A. Myotonic Dystrophy

B. Becker Muscular Dystrophy

C. Limb-Girdle Muscular Dystrophy

D. Duchenne Muscular Dystrophy

Distal weakness with facial involvement is a hallmark of myotonic dystrophy. This condition features myotonia (delayed relaxation after contraction) and weakness that often starts in distal muscles of the hands and forearms, while also affecting facial muscles, producing ptosis and facial weakness. By contrast, Duchenne/Becker and limb-girdle muscular dystrophies mostly affect proximal muscles—the shoulder/hip girdles—early on, with less emphasis on the face and distal hand/forearm muscles. So the described pattern best fits myotonic dystrophy.

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Next Steps

Congratulations on reaching the final section of this guide. You've taken a meaningful step toward passing your certification exam and advancing your career.

As you continue preparing, remember that consistent practice, review, and self-reflection are key to success. Make time to revisit difficult topics, simulate exam conditions, and track your progress along the way.

If you need help, have suggestions, or want to share feedback, we'd love to hear from you. Reach out to our team at hello@examzify.com.

Or visit your dedicated course page for more study tools and resources:

https://musculardystrophy.examzify.com

We wish you the very best on your exam journey. You've got this!

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