

MUSCULAR DYSTROPHIES & SPINAL MUSCULAR ATROPHY 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.

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!

Questions

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- 1. In myotonic dystrophy type 1, non-skeletal complications requiring monitoring most include which?**
 - A. Cardiac conduction defects and endocrine issues
 - B. Pulmonary hypertension
 - C. Hepatic failure
 - D. Kidney stones
- 2. Which of the following is a clinical red flag for SMA?**
 - A. Hypotonia and lack of head control
 - B. Hypertonia and brisk reflexes
 - C. Hyperflexibility with joint laxity
 - D. Normal reflexes with strong respiratory drive
- 3. How do exon-skipping therapies for DMD work mechanistically?**
 - A. They bind dystrophin pre-mRNA to modulate splicing and skip exons to restore the reading frame and produce functional dystrophin
 - B. They replace dystrophin with a synthetic protein
 - C. They inhibit dystrophin production
 - D. They deliver dystrophin protein directly
- 4. What genetic abnormality causes facioscapulohumeral muscular dystrophy type 1 (FSHD1)?**
 - A. Expansion of D4Z4 macrosatellite repeats on chromosome 4q35
 - B. Duplication of the D4Z4 region on 4q35
 - C. Contraction of D4Z4 macrosatellite repeats on chromosome 4q35 leading to misexpression of the DUX4 gene
 - D. Deletion of the DUX4 gene
- 5. In SMA, disease severity is determined by what?**
 - A. Gender
 - B. Age of Onset
 - C. Number of SMN2 Gene Copies
 - D. SMN1 Gene Dosage

- 6. What is the hallmark molecular basis of SMA?**
- A. Loss of SMN1 with SMN2 acting as a modifier; low SMN protein leads to motor neuron loss.
 - B. Overexpression of SMN1 causing motor neuron loss.
 - C. Mutation in dystrophin gene is SMA cause.
 - D. Mitochondrial dysfunction is SMA cause.
- 7. How early on should power mobility be introduced for sitters with SMA?**
- A. Very late, after age 6
 - B. Very early on, before age 2
 - C. After formal gait training begins
 - D. Only when walking is achieved
- 8. What is SMA type 4, and when does it typically present?**
- A. Adult-onset SMA with mild proximal weakness after age 30
 - B. Infant with severe proximal weakness
 - C. Teen with distal weakness
 - D. Late childhood with respiratory failure
- 9. A CM patient should primarily aim to...**
- A. Energy Conservation and Activity Modification
 - B. Speed Training
 - C. Sprint Intervals
 - D. Free Weight Maximal Lifts
- 10. Which biomarker is typically markedly elevated in Duchenne muscular dystrophy and aids in diagnosis?**
- A. Hemoglobin A1c
 - B. Alkaline phosphatase
 - C. Creatine kinase
 - D. Thyroid-stimulating hormone

Answers

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

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Explanations

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1. In myotonic dystrophy type 1, non-skeletal complications requiring monitoring most include which?

A. Cardiac conduction defects and endocrine issues

B. Pulmonary hypertension

C. Hepatic failure

D. Kidney stones

Myotonic dystrophy type 1 affects many organ systems beyond muscles, but the non-skeletal problems that most often require regular monitoring are problems with the heart's electrical conduction and with endocrine/metabolic regulation. Cardiac conduction defects arise when the heart's electrical pathways weaken or become blocked, which can progress and cause dangerous arrhythmias or heart block even in people with mild weakness. Because of this, baseline and periodic ECGs, sometimes extended rhythm monitoring, and assessment of conduction disease are essential; some patients may need a pacemaker if blocks advance. Endocrine issues are also common in DM1—insulin resistance and diabetes can develop, and thyroid or gonadal function can be affected. Regular screening for glucose metabolism and thyroid and gonadal function helps catch and manage these problems early. Pulmonary or hepatic problems can occur in DM1, but they are not as consistently or as strongly emphasized for routine monitoring as cardiac conduction and endocrine issues. Kidney stones are not a typical feature of DM1.

2. Which of the following is a clinical red flag for SMA?

A. Hypotonia and lack of head control

B. Hypertonia and brisk reflexes

C. Hyperflexibility with joint laxity

D. Normal reflexes with strong respiratory drive

The main idea here is that spinal muscular atrophy causes loss of anterior horn cells leading to low muscle tone and proximal weakness, so signs reproduce as a floppy, weak infant with poor head control. When the neck and trunk muscles can't support the head, it reflects early axial weakness from motor neuron loss, which is a classic red flag for SMA and prompts neuro evaluation. This pattern helps distinguish SMA from other issues. Hypertonia with brisk reflexes suggests upper motor neuron involvement (like cerebral palsy), not SMA. Hyperflexibility with joint laxity isn't typical of SMA, which presents with weakness rather than excessive joint looseness. Normal reflexes with strong respiratory drive don't fit SMA well, because SMA often involves reduced reflexes and progressive respiratory muscle weakness. So, hypotonia with lack of head control best signals SMA in infants.

3. How do exon-skipping therapies for DMD work mechanistically?

- A. They bind dystrophin pre-mRNA to modulate splicing and skip exons to restore the reading frame and produce functional dystrophin
- B. They replace dystrophin with a synthetic protein
- C. They inhibit dystrophin production
- D. They deliver dystrophin protein directly

Exon-skipping therapies work by using antisense molecules that bind to dystrophin pre-mRNA and mask specific splice sites or regulatory elements. This modulates how the cell's splicing machinery chooses which exons to include, so the targeted exon is skipped during mRNA processing. If skipping that exon restores the reading frame, the resulting mRNA codes for a dystrophin protein that is shorter but still functional enough to anchor the muscle membrane to the extracellular matrix. In effect, mutations that previously disrupted the reading frame can become in-frame, producing a Becker-like dystrophin and improving muscle stability and function. This approach does not replace dystrophin with a synthetic protein, nor does it inhibit its production or deliver the protein directly; it leverages the body's own gene to generate a usable, shorter dystrophin.

4. What genetic abnormality causes facioscapulohumeral muscular dystrophy type 1 (FSHD1)?

- A. Expansion of D4Z4 macrosatellite repeats on chromosome 4q35
- B. Duplication of the D4Z4 region on 4q35
- C. Contraction of D4Z4 macrosatellite repeats on chromosome 4q35 leading to misexpression of the DUX4 gene
- D. Deletion of the DUX4 gene

FSHD1 arises when the D4Z4 macrosatellite repeats on chromosome 4q35 are abnormally shortened. In healthy individuals there are roughly 11 to 100 repeats; in FSHD1 the repeats contract to about 1–10 units. This shortening changes the chromatin to a more open state, allowing the DUX4 gene, located within those repeats, to be inappropriately expressed in skeletal muscle. For DUX4 expression to produce a stable transcript, the chromosome also needs a permissive 4qA haplotype that provides a polyadenylation signal; without that signal, the transcript may be unstable. The misexpression of DUX4 in muscle drives the pathogenic process seen in FSHD1. Expanding or duplicating the repeat, or deleting DUX4, would not produce the same disease mechanism, and deletion of DUX4 would remove the toxic transcript rather than cause disease.

5. In SMA, disease severity is determined by what?

- A. Gender
- B. Age of Onset
- C. Number of SMN2 Gene Copies
- D. SMN1 Gene Dosage

In spinal muscular atrophy, how severe the disease is mainly depends on how many SMN2 gene copies a person has. SMN1 loss causes the condition, but SMN2 can partly compensate by producing functional SMN protein; the more SMN2 copies present, the more functional protein can be made, which tends to lessen weakness, delay onset, and produce a milder overall course. Gender does not influence severity, and while age of onset often correlates with SMN2 copy number, the primary determinant is the amount of SMN2 available to supplement SMN1 loss. SMN1 dosage explains the presence of disease, but the variation in severity across individuals is driven by SMN2 copy number.

6. What is the hallmark molecular basis of SMA?

- A. Loss of SMN1 with SMN2 acting as a modifier; low SMN protein leads to motor neuron loss.**
- B. Overexpression of SMN1 causing motor neuron loss.
- C. Mutation in dystrophin gene is SMA cause.
- D. Mitochondrial dysfunction is SMA cause.

Spinal muscular atrophy is driven by a deficiency of SMN protein due to loss or damaging mutations of the SMN1 gene. The SMN2 gene can partially compensate, but it mostly produces a nonfunctional form, so it only modestly rescues SMN levels. With SMN1 gone or severely reduced, low SMN protein leads to motor neuron degeneration. That's why the hallmark is SMN1 loss with SMN2 acting as a modifier, not overexpression of SMN1, dystrophin mutations, or mitochondrial dysfunction.

7. How early on should power mobility be introduced for sitters with SMA?

- A. Very late, after age 6
- B. Very early on, before age 2**
- C. After formal gait training begins
- D. Only when walking is achieved

Introducing powered mobility very early, before age 2, allows a child with SMA to explore their environment and participate in daily activities during crucial years of development. When a sitter can move independently in a powered chair, they gain opportunity for environmental interaction, social engagement, and motivation to engage in play and caregiver routines. This early access supports cognitive and social growth and helps families integrate mobility into everyday life, rather than waiting for a future milestone that may or may not occur. In SMA, where muscle weakness progresses and walking may never be achieved for many children, providing mobility early ensures the child isn't robbed of independent exploration and learning opportunities simply because walking isn't part of their trajectory. Delaying mobility overlooks these formative windows, and using gait training as a prerequisite or waiting until walking is achieved excludes non-ambulatory children from crucial participation and development.

8. What is SMA type 4, and when does it typically present?

- A. Adult-onset SMA with mild proximal weakness after age 30
- B. Infant with severe proximal weakness
- C. Teen with distal weakness
- D. Late childhood with respiratory failure**

SMA type 4 is the adult-onset form, characterized by onset after about 30 years of age with relatively mild proximal muscle weakness. Respiratory involvement is not a defining feature and, when it occurs, tends to be less prominent than in earlier-onset forms. So the best description is someone who develops symptoms in adulthood with mild proximal weakness, not in childhood. The other scenarios don't fit SMA type 4 well: infant onset with severe proximal weakness is classic for the early forms (SMA type 1); teen with distal weakness is not typical for classic SMA patterns, which are more proximal; late childhood with respiratory failure points to earlier-onset SMA with more significant respiratory involvement.

9. A CM patient should primarily aim to...

A. Energy Conservation and Activity Modification

B. Speed Training

C. Sprint Intervals

D. Free Weight Maximal Lifts

The focus here is on preserving function by reducing energy expenditure. In muscular dystrophy, weakness comes with increased fatigue and a limited ability to sustain activity. Prioritizing energy conservation and activity modification helps a person stay independent longer, complete essential tasks, and participate in daily life without overexerting fragile muscles. This approach worked into daily practice by pacing activities, planning rest breaks, using assistive devices or task modifications, and choosing lower-effort ways to accomplish goals. High-demand options like sprint work, maximal lifts, or rapid movements require a lot of energy and place more stress on weakened muscles and joints, which can lead to overuse injuries or faster fatigue. They don't support sustaining function over time the way energy-conserving strategies do.

10. Which biomarker is typically markedly elevated in Duchenne muscular dystrophy and aids in diagnosis?

A. Hemoglobin A1c

B. Alkaline phosphatase

C. Creatine kinase

D. Thyroid-stimulating hormone

In Duchenne muscular dystrophy, the lack of dystrophin makes muscle membranes fragile, so when muscle fibers are damaged their contents spill into the blood. Creatine kinase is a large enzyme abundant in skeletal muscle, and its levels rise dramatically when muscle injury occurs. Because the muscle breakdown in Duchenne is ongoing, the blood CK level is typically markedly elevated—often many times above the normal upper limit—making it a valuable clue that prompts genetic testing for dystrophin mutations. This marker is useful for screening and monitoring, though it isn't specific to Duchenne and must be confirmed with dystrophin gene testing. Other systemic biomarkers listed are not related to muscle disease: hemoglobin A1c reflects glucose control, alkaline phosphatase relates to bone/liver processes, and thyroid-stimulating hormone relates to thyroid function.

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://mdspinalmuscularatrophy.examzify.com>

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

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