# AMERICAN BOARD OF GENETIC COUNSELING (ABGC) BOARDS 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 is a characteristic sign of Cockayne Syndrome?**

A. Decreased fertility

B. Butterfly rash

C. Sensitivity to sunlight

D. Short stature

**2. What does 'malformation' typically indicate in the context of congenital defects?**

A. Issues caused by external forces

B. Genetic predisposition only

C. Failure of tissue formation

D. Injury after sufficient development

**3. In which disease is excess glycosaminoglycans a key feature?**

A. Homocystinuria

B. Hunter Syndrome

C. Maple Syrup Urine Disease

D. Homocystinuria

**4. What is an effective approach to alleviate a patient's worry about their genetic risk?**

A. Encouraging them to ignore their concerns

B. Discussing potential negative outcomes only

C. Talking through safety measures if their test results are negative

D. Providing a list of all possible mutations

**5. What is a premium in health insurance terms?**

A. Payment made only when needing medical services

B. Regular payment which keeps plan active

C. Amount users pay before insurance kicks in

D. A limit on insurance payments over a lifetime

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**6. What is the SMA carrier frequency for the general population?**

A. 1/40 to 1/60

B. 1/25

C. 1/50

D. 1/30

**7. Which test is the preferred initial diagnostic approach for Neurofibromatosis 2?**

A. MLPA for methylation

B. Sequencing followed by deletion/duplication analysis

C. Phenotypic assessment alone

D. Imaging studies

**8. An ultrasound finding of 2-3 syndactyly is linked to which genetic condition?**

A. Trisomy 21

B. Short femur

C. Syndrome of Laterality (SLO)

D. Thanatophoric dysplasia

**9. Which coping mechanism involves making specific plans for the future?**

A. Coping: confrontation

B. Planning

C. Coping: positive reapprasial

D. Self-controlling

**10. What is the design characteristic of a case-control study?**

A. It retrospectively compares patients with and without a disease

B. It follows a cohort forward in time

C. It uses random sampling of the general population

D. It includes only healthy individuals for analysis

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# Answers

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

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# Explanations

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## 1. What is a characteristic sign of Cockayne Syndrome?

A. Decreased fertility

B. Butterfly rash

C. Sensitivity to sunlight

D. Short stature

*Cockayne Syndrome is a genetic disorder characterized by the body's inability to adequately repair DNA damage, primarily affecting growth and development. A hallmark feature of Cockayne Syndrome is sensitivity to sunlight, also known as photosensitivity. Individuals with this condition often exhibit a range of symptoms upon exposure to sunlight, including skin damage, which underscores the necessity for protection from ultraviolet light. This sensitivity is particularly important in Cockayne Syndrome because it reflects the underlying cellular mechanisms of the disorder. The impaired DNA repair pathways not only affect the skin but also contribute to other systemic manifestations associated with the syndrome. In contrast, while other choices might describe aspects of genetic disorders broadly, they do not specifically relate to the hallmark symptoms of Cockayne Syndrome. For instance, decreased fertility, a butterfly rash, and short stature are associated with different conditions or syndromes, but none are defining features associated with Cockayne Syndrome. Understanding the specific manifestations of this syndrome, particularly photosensitivity, is crucial for diagnosis and management.*

## 2. What does 'malformation' typically indicate in the context of congenital defects?

A. Issues caused by external forces

B. Genetic predisposition only

C. Failure of tissue formation

D. Injury after sufficient development

*Malformation typically indicates a failure of tissue formation during the embryonic or fetal development stage. In the context of congenital defects, this term refers specifically to structural abnormalities that arise due to intrinsic factors affecting the formation and differentiation of tissues, organs, or body parts in the developing fetus. These malformations occur early in development and can result in noticeable changes in anatomy that can be detected at birth or later in life. This understanding distinguishes malformations from other types of congenital defects that may be caused by external factors, genetic predispositions, or injuries sustained after development is well underway. For example, issues caused by external forces would fall under the category of deformations or disruptions, not malformations. Similarly, while genetic predisposition can lead to various congenital conditions, malformations specifically derive from failures in the initial formation processes. Lastly, injuries sustained after sufficient development relate to acquired conditions rather than inherent developmental issues, which further clarifies the exclusivity of the term 'malformation.'*

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## 3. In which disease is excess glycosaminoglycans a key feature?

A. Homocystinuria

B. Hunter Syndrome

C. Maple Syrup Urine Disease

D. Homocystinuria

*Hunter syndrome, also known as mucopolysaccharidosis type II (MPS II), is characterized by the accumulation of glycosaminoglycans (GAGs) in the body due to a deficiency of the enzyme iduronate-2-sulfatase. This enzyme is responsible for breaking down specific GAGs, and its deficiency leads to the abnormal buildup of these molecules, resulting in various systemic and neurological symptoms. The presence of excess GAGs is central to the diagnosis of Hunter syndrome and affects multiple organ systems, causing a range of clinical manifestations, including skeletal abnormalities, cardiovascular issues, and developmental delays. Recognizing this accumulation is critical for both diagnosis and management of the disease. Although there are other metabolic disorders listed that involve amino acid or metabolic abnormalities, they do not feature the excess accumulation of glycosaminoglycans as a primary characteristic, making Hunter syndrome the correct answer.*

## 4. What is an effective approach to alleviate a patient's worry about their genetic risk?

A. Encouraging them to ignore their concerns

B. Discussing potential negative outcomes only

C. Talking through safety measures if their test results are negative

D. Providing a list of all possible mutations

*Talking through safety measures if their test results are negative is an effective approach to alleviate a patient's worry about their genetic risk because it aims to provide reassurance and a sense of control. This strategy helps patients to visualize a positive scenario and think about how they can take proactive steps to protect their health, even if they may be at risk. By focusing on what steps can be taken after receiving negative test results, it shifts the conversation to empowerment and reassurance, which can reduce anxiety and help the patient feel supported. Additionally, discussing safety measures reinforces the idea that they have options and tools at their disposal to manage their health, which can mitigate feelings of helplessness that often accompany worries regarding genetic risks. It also fosters an open dialogue where patients can express their concerns, ask questions, and feel like their worries are being taken seriously. In contrast, encouraging patients to ignore their concerns would likely increase anxiety, as ignoring worries does not resolve the underlying issues they face. Simply discussing potential negative outcomes could further heighten their fears without offering any constructive ways to cope. Providing a list of all possible mutations may overwhelm the patient with information, potentially increasing their anxiety rather than alleviating it, as they might fixate on rare or severe mutations without any context for action or management.*

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## 5. What is a premium in health insurance terms?

A. Payment made only when needing medical services

B. Regular payment which keeps plan active

C. Amount users pay before insurance kicks in

D. A limit on insurance payments over a lifetime

*In health insurance terms, a premium refers to the regular payment made by the policyholder to maintain an active health insurance plan. This payment is typically made monthly or annually and secures the policyholder's access to specified medical services and benefits covered under the insurance plan. Without the timely payment of premiums, the insurance coverage could lapse, leaving the individual without protection for healthcare costs. The concept of a premium is fundamental in understanding how insurance functions, as it represents the financial commitment required to access the coverage that insurance provides. Premiums are essential for the operation of insurance companies as they fund the claims that policyholders might submit, and they also contribute to the overall risk pool of the insurance system. Understanding this aspect is crucial for anyone navigating health insurance options. Being aware of the premium helps individuals budget for insurance costs and evaluate how they fit within their broader financial planning for healthcare needs.*

## 6. What is the SMA carrier frequency for the general population?

A. 1/40 to 1/60

B. 1/25

C. 1/50

D. 1/30

*SMA (Spinal Muscular Atrophy) is a genetic disorder caused by a deficiency of the survival motor neuron (SMN) protein, with carrier frequency providing insight into how prevalent the condition is within the general population. The carrier frequency for SMA in the general population is indeed estimated to be between 1 in 40 to 1 in 60 individuals. This range reflects the proportion of people who carry one copy of the mutated gene, while not expressing the disease themselves. Understanding that carrier frequencies can vary slightly based on different populations, the range of 1/40 to 1/60 is commonly cited in literature and clinical guidelines, making it an important figure for genetic counselors when assessing risk and providing information to individuals and families potentially affected by SMA. This data is crucial for carrier screening and counseling practices, emphasizing the need for awareness of genetic risks in family planning and prenatal settings.*

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## 7. Which test is the preferred initial diagnostic approach for Neurofibromatosis 2?

A. MLPA for methylation

B. Sequencing followed by deletion/duplication analysis

C. Phenotypic assessment alone

D. Imaging studies

*The preferred initial diagnostic approach for Neurofibromatosis 2 is sequencing followed by deletion/duplication analysis. Neurofibromatosis type 2 (NF2) is primarily caused by mutations in the NF2 gene, which encodes the protein merlin. Genetic testing that includes sequencing the NF2 gene allows for the identification of point mutations. Following sequencing, deletion/duplication analysis is essential to detect any larger genomic alterations, such as deletions or duplications, that may not be captured through standard sequencing methods. This comprehensive approach ensures that both small and large changes in the NF2 gene are identified, providing a definitive diagnosis. It is particularly important in genetic conditions like NF2 where both types of alterations can significantly contribute to the disease phenotype. While imaging studies may be helpful for evaluating symptoms related to tumor development associated with NF2, they do not provide a genetic diagnosis and are not the primary method recommended for initial diagnosis. Similarly, MLPA (Multiplex Ligation-dependent Probe Amplification) for methylation would be a follow-up tool rather than a first-line approach, and phenotypic assessment alone may miss the molecular underpinnings of the condition. Thus, starting with genetic sequencing followed by deletion/duplication analysis maximizes*

## 8. An ultrasound finding of 2-3 syndactyly is linked to which genetic condition?

A. Trisomy 21

B. Short femur

C. Syndrome of Laterality (SLO)

D. Thanatophoric dysplasia

*The ultrasound finding of 2-3 syndactyly is notably associated with the Syndrome of Laterality (SLO), which involves various forms of congenital malformations related to the body's laterality. In SLO, syndactyly, particularly between the second and third digits, is a characteristic finding. This condition results from disruptions in the normal development of the body's structure, leading to issues such as laterality defects and polydactyly, along with syndactyly. Other conditions mentioned do not typically link with this specific ultrasound finding. Trisomy 21, also known as Down syndrome, may present with other structural anomalies but does not commonly feature 2-3 syndactyly as a hallmark finding. Short femur does not specifically correlate with syndactyly and pertains more to measurements of skeletal development. Thanatophoric dysplasia is primarily characterized by severe skeletal dysplasia and limb shortening, but it does not typically present with syndactyly. Thus, the combination of features typical of the Syndrome of Laterality distinctly aligns with the ultrasound finding of 2-3 syndactyly, making it the correct choice in this context.*

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## 9. Which coping mechanism involves making specific plans for the future?

A. Coping: confrontation

B. Planning

C. Coping: positive reapprasial

D. Self-controlling

*The correct answer is planning because it directly refers to the process of making specific strategies or outlines for future actions. This coping mechanism is characterized by the active formulation of steps to address challenges or prepare for upcoming situations. It allows individuals to take control of uncertainty by thinking ahead and preparing for potential outcomes, which can help reduce anxiety and enhance a sense of agency. In this context, planning can involve anything from setting goals to anticipating future challenges and outlining the steps necessary to address them. This proactive approach empowers individuals to feel more equipped to handle what lies ahead, as it gives a sense of direction and purpose in the face of stressors. The other choices represent different coping strategies that do not specifically focus on planning. Confrontation involves actively addressing a problem or conflict but may not necessarily include forward-looking planning. Positive appraisal refers to reframing a situation in a more positive light, which can help shift one's emotional response but doesn't involve the formulation of concrete future plans. Self-controlling indicates a focus on managing one's own feelings and reactions, often by restraining impulses or emotions, rather than creating a structured plan for the future.*

## 10. What is the design characteristic of a case-control study?

A. It retrospectively compares patients with and without a disease

B. It follows a cohort forward in time

C. It uses random sampling of the general population

D. It includes only healthy individuals for analysis

*A case-control study is designed to retrospectively compare individuals who have a specific disease or condition (the cases) with individuals who do not have the disease (the controls). This design characteristic allows researchers to identify potential risk factors or associations that may contribute to the disease. By looking back in time at prior exposures or characteristics of both groups, researchers can gather insights into factors that may be linked to the onset of the disease. This type of study is particularly useful in understanding rare diseases as it allows for the examination of a smaller, more targeted group of individuals affected by the disease compared to the larger general population. The other options do not align with the characteristics of a case-control study. A cohort study, for example, follows a group of individuals over time, which is not retrospective. Random sampling of the general population is more aligned with cross-sectional study designs, while including only healthy individuals is not representative of a case-control study where both affected and unaffected individuals are compared to understand the disease's context. Thus, the key characteristic that defines a case-control study is its retrospective comparative approach between those with and without a disease.*

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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://abgcboards.examzify.com

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

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