

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.

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. What is the hallmark diagnostic feature of the hypermobile type of Ehlers Danlos Syndrome?**
 - A. Skin hyperextensibility
 - B. Joint hypermobility
 - C. Heart murmurs
 - D. Bone fragility
- 2. Who benefits from the military insurance known as Tricare?**
 - A. Only retirees
 - B. All US citizens
 - C. Active and retired military personnel and their families
 - D. Individuals with chronic illnesses
- 3. Which condition is associated with heart and arm anomalies detected by ultrasound?**
 - A. Beckwith Wiedemann
 - B. Meckel Gruber
 - C. Stickler
 - D. Holt Oram
- 4. Which condition is characterized by high levels of inhibin A?**
 - A. Neural tube defects
 - B. Down syndrome
 - C. Turner syndrome
 - D. Edwards syndrome
- 5. What is the carrier frequency for PKU in the general population?**
 - A. 1/40
 - B. 1/50
 - C. 1/30
 - D. 1/60

- 6. Which gene mutation is most commonly associated with Ehlers Danlos Syndrome Classic type?**
- A. COL1A1
 - B. COL3A1
 - C. COL5A1/2
 - D. ACTA2
- 7. In terms of recurrence risk, what does a genetic counseling professional imply when assessing conditions?**
- A. That all conditions have equal risk
 - B. Recurrence risk is not influenced by family history
 - C. More affected relatives indicate higher risk
 - D. Risk is irrelevant to the patient's family medical history
- 8. What kind of partnerships might one anticipate with clinics listed in GeneClinics?**
- A. Partnerships with financial institutions
 - B. Affiliations with national health services
 - C. Collaboration with insurance companies
 - D. Links to educational institutions only
- 9. Which syndrome is associated with TWIST1?**
- A. Saethre-Chotzen Syndrome
 - B. Muenke Syndrome
 - C. Apert Syndrome
 - D. Carpenter Syndrome
- 10. What type of dietary management is often required in the treatment of Maple Syrup Urine Disease?**
- A. High protein diet
 - B. Leucine restriction
 - C. Folate supplementation
 - D. Low carbohydrate diet

Answers

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

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Explanations

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1. What is the hallmark diagnostic feature of the hypermobile type of Ehlers Danlos Syndrome?

- A. Skin hyperextensibility
- B. Joint hypermobility**
- C. Heart murmurs
- D. Bone fragility

The hallmark diagnostic feature of hypermobile Ehlers-Danlos Syndrome (hEDS) is joint hypermobility, which is characterized by an increased range of motion in joints beyond what is considered normal. This feature is a key criterion for diagnosing hEDS, as it can lead to various symptoms, including pain and increased susceptibility to joint dislocations and injuries. Joint hypermobility in hEDS is often assessed through the Beighton score, which evaluates a combination of movements in various joints. Unlike other types of Ehlers-Danlos Syndrome, such as the classical type, hEDS lacks the marked skin hyperextensibility, fragility, or additional systemic features; thus, joint hypermobility stands out as the primary diagnostic indicator for this specific type. Other features like skin hyperextensibility, heart murmurs, and bone fragility may be relevant to different types of Ehlers-Danlos Syndrome but are not central to the diagnosis of hEDS. For instance, skin hyperextensibility is more characteristic of the classical type, and heart murmurs may indicate vascular complications associated with another type, such as the vascular type of EDS. Bone fragility is also more associated with osteogenesis imperfecta rather than h

2. Who benefits from the military insurance known as Tricare?

- A. Only retirees
- B. All US citizens
- C. Active and retired military personnel and their families**
- D. Individuals with chronic illnesses

Tricare is a comprehensive health insurance program specifically designed for military personnel in the United States. Its coverage extends not just to active-duty service members but also to retired military members and their immediate family members. This inclusivity is essential because it aims to support those who have sacrificed for their country by providing them with necessary healthcare services. The program addresses unique healthcare needs and offers various plans tailored to different situations, ensuring ongoing medical support for both active and retired personnel, as well as their dependents. This focus on families acknowledges the sacrifices made by service members and provides them with the necessary resources for maintaining their health and well-being during and after their service. The other options do not accurately represent the scope of who benefits from Tricare. For example, it does not extend universally to all US citizens, nor is it specifically designed for individuals with chronic illnesses; rather, it is primarily targeted at military families. Therefore, recognizing this specific target demographic highlights why this particular answer is indeed the most appropriate.

3. Which condition is associated with heart and arm anomalies detected by ultrasound?

- A. Beckwith Wiedemann
- B. Meckel Gruber
- C. Stickler
- D. Holt Oram**

The condition associated with heart and arm anomalies detected by ultrasound is Holt-Oram syndrome. Holt-Oram syndrome is a genetic disorder characterized by congenital heart defects and upper limb malformations, which can include issues such as absent or poorly formed arms. The heart defects often involve the septum or other structural anomalies. In prenatal diagnosis, ultrasound is a critical tool for detecting these types of anomalies. The presence of both cardiac defects and limb anomalies in a developing fetus would point towards Holt-Oram syndrome, allowing for further genetic counseling and management considerations. Other conditions, while they may present with various anomalies, do not specifically manifest both heart and arm anomalies in the combination characteristic of Holt-Oram syndrome. For instance, Beckwith-Wiedemann syndrome primarily involves overgrowth and organomegaly, Meckel-Gruber syndrome is associated with renal cysts and neural tube defects, and Stickler syndrome is characterized by ocular, auditory, and skeletal abnormalities, but not typically with the specific combination of heart and arm anomalies that define Holt-Oram. Thus, Holt-Oram syndrome is the most appropriate answer for the described condition in the question.

4. Which condition is characterized by high levels of inhibin A?

- A. Neural tube defects
- B. Down syndrome**
- C. Turner syndrome
- D. Edwards syndrome

High levels of inhibin A are associated with Down syndrome, also known as Trisomy 21. Inhibin A is a protein produced by the placenta and the ovaries, and its levels can be measured during prenatal screening tests. Elevated levels of inhibin A, particularly when considered alongside other markers such as maternal serum alpha-fetoprotein (MSAFP) and estriol, can indicate an increased risk for chromosomal abnormalities, specifically Down syndrome. In the context of prenatal screening, the presence of high levels of inhibin A is part of the combined first-trimester screening that can help assess the likelihood of Down syndrome in a fetus. This marker is specifically valuable as it reflects placental function and abnormalities present in conditions like Down syndrome, making it a crucial factor in prenatal risk assessment. Other conditions listed, like neural tube defects, Turner syndrome, and Edwards syndrome, do not typically present with high levels of inhibin A. Instead, they are diagnosed based on different markers and ultrasound findings. Neural tube defects often show elevated MSAFP rather than inhibin A. Turner syndrome generally involves low levels of various markers, and Edwards syndrome has distinct findings that differ from those associated with Down syndrome.

5. What is the carrier frequency for PKU in the general population?

- A. 1/40
- B. 1/50**
- C. 1/30
- D. 1/60

The carrier frequency for phenylketonuria (PKU) in the general population is approximately 1 in 50 individuals. This statistic reflects the prevalence of carriers, meaning that these individuals possess one copy of the mutated gene associated with PKU but typically do not express the disorder themselves since PKU is autosomal recessive. Understanding why this carrier frequency is significant involves recognizing its implications for genetic counseling and population screening. Carriers have a 25% chance of having an affected child with another carrier, emphasizing the importance of genetic testing for couples planning to conceive, especially if they belong to groups known to have higher frequencies of PKU. The other options do not accurately reflect the established carrier prevalence for PKU in the general population, and therefore do not serve as correct answers. Accurate knowledge of such statistics is crucial for genetic counselors when assessing risk and guiding families in decision-making regarding genetic testing and family planning.

6. Which gene mutation is most commonly associated with Ehlers Danlos Syndrome Classic type?

- A. COL1A1
- B. COL3A1
- C. COL5A1/2**
- D. ACTA2

The most commonly associated gene mutations with Ehlers-Danlos Syndrome (EDS) Classic type are in the COL5A1 and COL5A2 genes. EDS Classic type primarily affects the synthesis of type V collagen, which is crucial for the structural integrity of connective tissues. Type V collagen is a component of the extracellular matrix and plays a significant role in the regulation of collagen fibril organization. Mutations in COL5A1 and COL5A2 lead to the characteristic features of Classic type EDS, including skin hyperextensibility, joint hypermobility, and a tendency to bruise easily. These gene mutations impair collagen structure and function, resulting in weakened connective tissues. In comparison, mutations in COL1A1 and COL3A1 can cause other forms of EDS and connective tissue disorders, but they are not primarily responsible for EDS Classic type. Similarly, while ACTA2 mutations are associated with vascular-related disorders, they are not the primary contributors to the Classic type of EDS. Thus, the correct association with EDS Classic type lies with the mutations in COL5A1 and COL5A2 genes.

7. In terms of recurrence risk, what does a genetic counseling professional imply when assessing conditions?

- A. That all conditions have equal risk
- B. Recurrence risk is not influenced by family history
- C. More affected relatives indicate higher risk**
- D. Risk is irrelevant to the patient's family medical history

When assessing the recurrence risk of genetic conditions, a genetic counseling professional implies that more affected relatives within a family can suggest a higher risk of the condition occurring in future generations. This is rooted in the understanding of inheritance patterns; many genetic conditions are influenced by the family structure and how conditions are transmitted across generations. In families where multiple members are affected by the same condition, it is often indicative of a genetic component that may increase the likelihood of the condition appearing in other relatives or future offspring. Therefore, counselors consider the number and relation of affected individuals to provide an estimate of risk that is tailored to each family's unique genetic landscape. Understanding how genetic conditions cluster within families allows counselors to guide families on potential risks and the implications for family planning or monitoring. This context is crucial for families to make informed decisions about their health and the health of potential future generations.

8. What kind of partnerships might one anticipate with clinics listed in GeneClinics?

- A. Partnerships with financial institutions
- B. Affiliations with national health services**
- C. Collaboration with insurance companies
- D. Links to educational institutions only

The choice highlighting affiliations with national health services is the most relevant and accurate when considering the types of partnerships that might be found with clinics listed in GeneClinics. These clinics typically focus on genetic counseling and testing, which is often integrated with broader health services provided by national health systems. Such affiliations can enhance access to genetic services and ensure that they are part of the public healthcare framework. Approaching the question from the aspect of clinics and how they function within the healthcare system helps delineate why the other options don't align as well with the context of GeneClinics. While partnerships with financial institutions, collaboration with insurance companies, and links to educational institutions may exist in some capacity, they do not fundamentally capture the essence of the primary relationships that GeneClinics would likely prioritize, which center around providing seamless healthcare access, aligning with the goals of national health services. These services are essential in promoting and integrating genetic counseling into general health practices, thereby improving overall patient care and accessibility to genetic information.

9. Which syndrome is associated with TWIST1?

A. Saethre-Chotzen Syndrome

B. Muenke Syndrome

C. Apert Syndrome

D. Carpenter Syndrome

Saethre-Chotzen syndrome is indeed associated with mutations in the TWIST1 gene. This genetic condition is characterized by craniosynostosis, which is the premature fusion of certain skull bones, leading to a misshapen head. Additional features may include facial asymmetry, dental anomalies, and hearing loss, which are attributable to abnormal bone development during embryogenesis. The TWIST1 gene plays a crucial role in regulating the differentiation of mesodermal tissues, which include bone and cartilage. Mutations in this gene can disrupt normal developmental processes, specifically affecting craniofacial structures. This connection between TWIST1 and Saethre-Chotzen syndrome highlights the importance of genetic factors in craniosynostosis syndromes. In contrast, the other conditions listed, such as Muenke syndrome, Apert syndrome, and Carpenter syndrome, are associated with different genes and pathways, emphasizing the specificity of genetic syndromes to certain genes involved in development.

10. What type of dietary management is often required in the treatment of Maple Syrup Urine Disease?

A. High protein diet

B. Leucine restriction

C. Folate supplementation

D. Low carbohydrate diet

In the treatment of Maple Syrup Urine Disease (MSUD), the primary dietary management strategy involves restricting the intake of specific branched-chain amino acids, particularly leucine. This condition arises from a deficiency in the branched-chain alpha-keto acid dehydrogenase complex, leading to the accumulation of leucine, isoleucine, and valine in the body. High levels of these amino acids, particularly leucine, can be toxic and cause serious neurological issues. Restricting leucine in the diet helps prevent these toxic accumulations. In individuals with MSUD, a carefully monitored diet is essential to maintain amino acid balance, ensuring they receive adequate nutrition while avoiding the buildup of harmful substances. Patients often consume a low-protein diet that is supplemented with specially designed formulas that provide the necessary nutrients without the harmful levels of branched-chain amino acids. Other choices are less relevant in the context of dietary management specifically for MSUD. For instance, a high protein diet could exacerbate the condition by increasing the levels of branched-chain amino acids in the body. Folate supplementation is not a targeted treatment for MSUD and does not address the core issue of amino acid management. A low carbohydrate diet might be beneficial for other metabolic disorders,

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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