

American Board of Genetic Counseling (ABGC) Boards Practice Test (Sample)

Study Guide



Everything you need from our exam experts!

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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. Which is a primary characteristic of right ventricular hypertrophy?**
 - A. Increased size of right ventricle**
 - B. Abnormal valve closure**
 - C. Reduced lung capacity**
 - D. Increased left ventricular output**

- 2. What percentage of individuals with Down Syndrome typically have ultrasound findings?**
 - A. 25%**
 - B. 50%**
 - C. 75%**
 - D. 90%**

- 3. What is the significance of high hCG levels?**
 - A. Indicates a normal pregnancy**
 - B. Associated with T18**
 - C. Associated with T21**
 - D. Indicates maternal health issues**

- 4. What is the carrier frequency of HBOC in the general population?**
 - A. 1 in 10**
 - B. 1 in 400**
 - C. 1 in 30**
 - D. 1 in 20**

- 5. How should follow-up diagnostics be handled if a screening test result is positive?**
 - A. Immediate referral to a specialist**
 - B. Based on family history assessment**
 - C. Confirmatory testing should be conducted**
 - D. Leave it to patient preference**

- 6. What is the carrier frequency for Beta Thalassemia in Mediterranean populations?**
- A. 1 in 10**
 - B. 1 in 20**
 - C. 1 in 30**
 - D. 1 in 40**
- 7. Engagement in counselees reflects what type of involvement?**
- A. Cognitive and emotional involvement**
 - B. Financial involvement**
 - C. Physical involvement**
 - D. Social involvement**
- 8. Which ultrasound finding is characteristic of Thanatophoric dysplasia type 1?**
- A. Molar tooth sign**
 - B. Telephone receiver femur**
 - C. Rhabdomyoma**
 - D. Interrupted aortic arch**
- 9. Which genetic mutation is primarily associated with Progeria?**
- A. MECP2**
 - B. GNAS**
 - C. LMNA**
 - D. RBM8A**
- 10. 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**

Answers

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

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Explanations

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1. Which is a primary characteristic of right ventricular hypertrophy?

A. Increased size of right ventricle

B. Abnormal valve closure

C. Reduced lung capacity

D. Increased left ventricular output

A primary characteristic of right ventricular hypertrophy is indeed the increased size of the right ventricle. This condition generally arises as a response to increased workload on the right side of the heart, commonly due to conditions such as pulmonary hypertension or chronic respiratory disorders that place stress on the right ventricle. As the ventricle works harder to pump blood into the pulmonary arteries, the muscle fibers adaptively thicken, resulting in hypertrophy, which is the enlargement of the heart muscle. The other options are related to various aspects of cardiac function or physiology but do not define the primary characteristic of right ventricular hypertrophy. Abnormal valve closure may occur with various cardiac conditions, but it is not a defining characteristic of right ventricular hypertrophy itself. Reduced lung capacity can be associated with conditions leading to right ventricular hypertrophy, such as chronic obstructive pulmonary disease (COPD), but it does not directly characterize the hypertrophy itself. Increased left ventricular output is typically not a feature of right ventricular hypertrophy and instead may refer to conditions affecting the left heart, making it unrelated to the primary defining characteristic of right ventricular hypertrophy.

2. What percentage of individuals with Down Syndrome typically have ultrasound findings?

A. 25%

B. 50%

C. 75%

D. 90%

Individuals with Down Syndrome often present with characteristic findings detectable via prenatal ultrasound. Studies indicate that approximately 50% of fetuses with Down Syndrome will exhibit one or more ultrasound markers. This can include structural abnormalities such as increased nuchal translucency, heart defects, and other physical anomalies that are more prevalent in this population. Recognizing that not all individuals with Down Syndrome will show detectable abnormalities on ultrasound is crucial; hence, while there is a significant presence of ultrasound findings in these cases, it does not reach the higher percentages that other options suggest. This understanding helps genetic counselors provide accurate information regarding the likelihood of ultrasound findings in pregnancies at risk for chromosomal abnormalities.

3. What is the significance of high hCG levels?

- A. Indicates a normal pregnancy
- B. Associated with T18
- C. Associated with T21**
- D. Indicates maternal health issues

High levels of human chorionic gonadotropin (hCG) are most notably associated with trisomy 21 (Down syndrome). In pregnancy, hCG is produced by the placenta shortly after implantation, and its levels can provide important insights into fetal health and development. In the context of screening for fetal anomalies, elevated hCG levels can be a marker for an increased risk of Down syndrome. This condition is characterized by the presence of an extra copy of chromosome 21, leading to the distinct phenotypic and developmental features associated with it. Therefore, when hCG levels are abnormally high, it raises the suspicion for trisomy 21, prompting further diagnostic testing such as non-invasive prenatal testing (NIPT) or amniocentesis. While high hCG can also be present in normal pregnancies and certain maternal health conditions, its role as an indicator for trisomy 21 is specifically significant in the context of genetic counseling and prenatal screening. This awareness is crucial for healthcare providers in evaluating and managing pregnancies.

4. What is the carrier frequency of HBOC in the general population?

- A. 1 in 10
- B. 1 in 400**
- C. 1 in 30
- D. 1 in 20

The carrier frequency of hereditary breast and ovarian cancer (HBOC) syndrome in the general population, particularly relating to mutations in the BRCA1 and BRCA2 genes, is approximately 1 in 400. This figure reflects the distribution of these specific genetic mutations that significantly increase the risk of developing breast and ovarian cancers. Understanding this frequency is crucial for genetic counseling and risk assessment, as it helps healthcare providers identify individuals at risk for HBOC. The identification of such carriers is important for recommending appropriate surveillance and preventive strategies, such as enhanced screening or prophylactic surgeries. In contrast, the other frequencies listed in the choices suggest either a significantly higher prevalence that is not supported by current epidemiological data or do not align with the established figures for HBOC. For instance, a carrier frequency of 1 in 30 is more characteristic of Ashkenazi Jewish populations, where specific mutations are more prevalent, rather than the general population frequency. The accuracy of the 1 in 400 statistic is employed in broader risk assessments and public health discussions related to genetic testing and cancer predisposition.

5. How should follow-up diagnostics be handled if a screening test result is positive?

- A. Immediate referral to a specialist**
- B. Based on family history assessment**
- C. Confirmatory testing should be conducted**
- D. Leave it to patient preference**

When a screening test result is positive, confirmatory testing is essential to accurately determine whether the condition is truly present. Screening tests are designed to identify individuals who may have a particular condition, but they are not definitive. This means that a positive result can occur in individuals who do not actually have the condition, leading to false positives. Confirmatory tests provide a more reliable assessment of the individual's health status and are necessary for making appropriate clinical decisions and guiding further care. Following a positive screening result, it is crucial to conduct confirmatory testing to ensure that the follow-up care is based on accurate information. This process may involve specific diagnostic procedures, additional testing, or clinical evaluations tailored to the suspected condition. Other potential options like referral to a specialist or considering family history might be appropriate in certain contexts, but they do not take precedence over the need for definitive diagnostic evaluation. Leaving the decision to patient preference could lead to a delay in necessary evaluations, which may prove harmful, particularly in conditions where timely intervention is critical. Therefore, confirmatory testing is the best course of action following a positive screening test result.

6. What is the carrier frequency for Beta Thalassemia in Mediterranean populations?

- A. 1 in 10**
- B. 1 in 20**
- C. 1 in 30**
- D. 1 in 40**

The carrier frequency for Beta Thalassemia in Mediterranean populations is indeed approximately 1 in 20. This relatively high carrier frequency can be attributed to the historical prevalence of malaria in these regions. Individuals who are carriers of the Beta Thalassemia trait often have a selective advantage in malaria-endemic areas, which has led to an increased frequency of the mutation within the population over generations. Understanding carrier frequencies is essential for genetic counseling, as it informs healthcare providers and patients about the risk of offspring being affected by genetic conditions like Beta Thalassemia. For populations in the Mediterranean, the prevalence means that it is important for individuals considering family planning to be aware of their carrier status and that of their partners to make informed reproductive choices.

7. Engagement in counselees reflects what type of involvement?

- A. Cognitive and emotional involvement**
- B. Financial involvement**
- C. Physical involvement**
- D. Social involvement**

The correct answer, which emphasizes cognitive and emotional involvement, highlights the dual aspects of engagement that are critical in the context of genetic counseling. Cognitive involvement refers to the mental processes of understanding, processing, and integrating information regarding genetic information, risks, and implications for health. Emotional involvement signifies the feelings and emotional responses that counselees may experience during the counseling process, which can significantly affect their decision-making and ability to cope with genetic information. In genetic counseling, it is essential to foster both cognitive and emotional engagement because individuals need to both comprehend the complex information presented to them and address the emotional impacts that may arise from learning about genetic risks or conditions. This comprehensive engagement allows for more personalized and effective counseling, facilitating better decisions and outcomes for counselees. While financial, physical, and social involvement may play roles in certain contexts, they do not encapsulate the comprehensive and deep-rooted engagement that cognitive and emotional involvement collectively provide in the realm of genetic counseling.

8. Which ultrasound finding is characteristic of Thanatophoric dysplasia type 1?

- A. Molar tooth sign**
- B. Telephone receiver femur**
- C. Rhabdomyoma**
- D. Interrupted aortic arch**

The characteristic ultrasound finding associated with Thanatophoric dysplasia type 1 is the "telephone receiver femur." This term describes the appearance of the femur, which is typically short and curved, resembling a telephone receiver. Thanatophoric dysplasia is a severe skeletal dysplasia, and the abnormalities in bone structure, particularly the femur, are a key diagnostic feature. In this condition, the femoral curvature is pronounced, leading to a distinctive diagnostic imaging appearance. The shortening of the long bones, particularly seen in the femur, is one of the primary indicators that clinicians look for when investigating skeletal dysplasias. This imaging feature, along with other clinical findings, helps confirm the diagnosis of Thanatophoric dysplasia type 1. The other options refer to conditions or findings not primarily associated with Thanatophoric dysplasia. For instance, the molar tooth sign is linked with Joubert syndrome, rhabdomyoma is a cardiac tumor seen in conditions like tuberous sclerosis, and interrupted aortic arch is a congenital heart defect. Each of these has its own specific characteristics and associated conditions, making them unrelated to Thanatophoric dysplasia type 1.

9. Which genetic mutation is primarily associated with Progeria?

- A. MECP2**
- B. GNAS**
- C. LMNA**
- D. RBM8A**

Progeria, specifically Hutchinson-Gilford Progeria Syndrome (HGPS), is primarily associated with mutations in the LMNA gene. This gene encodes for lamin A, a protein that is crucial for maintaining the structural integrity of the nuclear envelope. The specific mutation linked to Progeria is a single nucleotide mutation that results in the production of an abnormal form of lamin A known as progerin. This abnormal protein leads to severe developmental and physical changes that mimic aspects of aging at an accelerated rate. The association of LMNA with Progeria highlights the significance of nuclear stability and elasticity in normal cellular function and development. Therefore, individuals with Progeria exhibit symptoms that relate to both premature aging and cellular dysfunction, which are primarily driven by the defective lamin A protein that arises from mutations in this gene. In contrast, mutations in MECP2 are primarily associated with Rett syndrome, mutations in GNAS are linked to various disorders including Albright hereditary osteodystrophy, and mutations in RBM8A are connected to a different set of genetic disorders. These genes do not play a role in the pathology of Progeria, further underscoring the specificity of LMNA mutations to this condition.

10. 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.

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!