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

Study Guide

Everything you need from our exam experts!

This is a sample study guide. To access the full version with hundreds of 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. Don't worry about getting everything right, 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, and take breaks to retain information better.

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.

7. Use Other Tools

Pair this guide with other Examzify tools like flashcards, and digital practice tests to strengthen your preparation across formats.

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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 the carrier frequency of Tay Sachs disease in the Ashkenazi Jewish population?**

   A. 1 in 20

   B. 1 in 30

   C. 1 in 40

   D. 1 in 10

2. **What is considered a teratogenic factor that can impact fetal development?**

   A. Exercise

   B. Alcohol

   C. Hydration

   D. Weight maintenance

3. **Which condition does not typically associate with a finding of thick nuchal translucency?**

   A. Trisomy 21

   B. Congenital heart defects

   C. Skeletal dysplasia

   D. Turner syndrome

4. **What is common among disorders related to FGFR2?**

   A. Midface retrusion

   B. Absent limbs

   C. Aplasia of middle phalanges

   D. Preaxial polydactyly

5. **Which enzyme deficiency is primarily responsible for Methylmalonic Acidemia?**

   A. Methylmalonyl CoA mutase

   B. Propionyl-CoA carboxylase

   C. Ornithine transcarbamylase

   D. Alpha-galactosidase A

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6. What term describes not providing full care or a breach of contract?

A. Abandonment

B. Negligence

C. Battery

D. Rights

7. Which inheritance pattern is primarily seen in Coffin-Lowry Syndrome?

A. Autosomal Dominant

B. Autosomal Recessive

C. X-Linked Recessive

D. Mitochondrial

8. What does a false positive indicate in a test result?

A. Test indicates positive disease status in an unaffected individual

B. Test indicates negative disease status in an affected individual

C. Test indicates positive disease status in an affected individual

D. Test indicates negative disease status in an unaffected individual

9. What are the primary neuropathological findings in Alzheimer's Disease?

A. Neurofibrillary tangles and amyloid plaques

B. White matter hyperintensities

C. Chromosomal deletions

D. Ischemic strokes

10. Which inheritance pattern is characteristic of Orofaciodigital Syndrome?

A. Autosomal dominant

B. Autosomal recessive

C. X-linked dominant

D. Y-linked

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Answers

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

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Explanations

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1. What is the carrier frequency of Tay Sachs disease in the Ashkenazi Jewish population?

A. 1 in 20

B. 1 in 30

C. 1 in 40

D. 1 in 10

The carrier frequency of Tay Sachs disease in the Ashkenazi Jewish population is approximately 1 in 30. This higher carrier frequency is a result of a population bottleneck effect and certain historical factors that have increased the prevalence of specific genetic disorders among Ashkenazi Jews. Tay Sachs disease is an autosomal recessive disorder caused by a deficiency in the enzyme hexosaminidase A. People who are carriers of the disease usually do not show symptoms, but if two carriers have a child, there is a 25% chance that their child will be affected by the disorder. Understanding the carrier frequency helps in genetic counseling, particularly for individuals from high-risk populations such as the Ashkenazi Jewish community. It allows genetic counselors to provide accurate risk assessments and facilitate appropriate testing and reproductive options for couples who may be carriers.

2. What is considered a teratogenic factor that can impact fetal development?

A. Exercise

B. Alcohol

C. Hydration

D. Weight maintenance

Alcohol is recognized as a teratogenic factor that can adversely affect fetal development. Teratogens are substances or environmental factors that can cause developmental abnormalities in an embryo or fetus. Maternal alcohol consumption during pregnancy can lead to a range of physical, behavioral, and cognitive issues in the child, collectively known as Fetal Alcohol Spectrum Disorders (FASDs). These disorders can vary in severity, but they often include growth deficiencies, facial anomalies, and neurodevelopmental problems. In contrast, exercise, hydration, and weight maintenance, while important aspects of a healthy pregnancy, are not classified as teratogenic factors. Moderate exercise is generally beneficial for pregnant individuals, aiding in overall well-being and potentially improving outcomes. Adequate hydration and maintaining a healthy weight are also essential for supporting both maternal health and fetal development, but they do not exhibit teratogenic effects. Understanding the distinction between beneficial factors and teratogens is crucial in prenatal care and counseling.

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3. Which condition does not typically associate with a finding of thick nuchal translucency?

A. Trisomy 21

B. Congenital heart defects

C. Skeletal dysplasia

D. Turner syndrome

Thick nuchal translucency is a measurement taken during the first trimester of pregnancy, particularly during an ultrasound, and is often used as a marker for certain chromosomal and structural abnormalities. A finding of increased nuchal translucency is commonly linked with conditions such as Trisomy 21 (Down syndrome), congenital heart defects, and Turner syndrome. Skeletal dysplasia, on the other hand, primarily refers to a group of disorders characterized by abnormal bone growth and development. These conditions typically do not present with thick nuchal translucency as a routine marker because they have different etiological underpinnings and associated ultrasound findings. While some skeletal dysplasias can be detected through ultrasound, they are often associated with other specific signs rather than increased nuchal translucency. Therefore, it is more accurate to state that skeletal dysplasia does not typically associate with this particular ultrasound finding, making it the correct answer.

4. What is common among disorders related to FGFR2?

A. Midface retrusion

B. Absent limbs

C. Aplasia of middle phalanges

D. Preaxial polydactyly

Midface retrusion is a common characteristic observed in disorders associated with FGFR2 (Fibroblast Growth Factor Receptor 2), such as Crouzon syndrome and Apert syndrome. FGFR2 plays a crucial role in cranial development, and mutations in this gene can lead to craniosynostosis, where the sutures of the skull fuse prematurely. This has a direct impact on the shape of the skull and facial features, often resulting in midface hypoplasia or retrusion. Conditions linked to FGFR2 mutations typically manifest in craniofacial abnormalities alongside potential limb or digit anomalies, although midface retrusion remains a hallmark feature. The other choices do not prominently feature in this context: absent limbs is typically associated with different genetic pathways, aplasia of middle phalanges is more often related to limb malformations not specifically tied to FGFR2, and preaxial polydactyly is associated with the SHH pathway rather than FGFR2. Thus, midface retrusion stands out as the characteristic feature most commonly recognized in FGFR2-related disorders.

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5. Which enzyme deficiency is primarily responsible for Methylmalonic Acidemia?

A. Methylmalonyl CoA mutase

B. Propionyl-CoA carboxylase

C. Ornithine transcarbamylase

D. Alpha-galactosidase A

Methylmalonic Acidemia is primarily caused by a deficiency in Methylmalonyl CoA mutase, which is an enzyme critical for the metabolism of certain amino acids and fatty acids. This enzyme converts methylmalonyl-CoA to succinyl-CoA, a necessary step in the pathway that leads to the energy production from these nutrients. When there is a deficiency in Methylmalonyl CoA mutase, it leads to a buildup of methylmalonic acid in the bloodstream, which can cause a range of metabolic disturbances. While other enzymes mentioned are associated with different metabolic disorders, they do not play a role in the specific pathology of Methylmalonic Acidemia. Propionyl-CoA carboxylase is involved in the metabolism of fatty acids and amino acids but is not the direct cause of this condition. Ornithine transcarbamylase is involved in the urea cycle and causes a different metabolic disorder. Alpha-galactosidase A is related to Fabry disease, which is unrelated to the metabolism impacted in Methylmalonic Acidemia. Thus, understanding the role of Methylmalonyl CoA mutase in the metabolic pathway is crucial to recognizing why its deficiency

6. What term describes not providing full care or a breach of contract?

A. Abandonment

B. Negligence

C. Battery

D. Rights

The correct term that describes the failure to provide full care or a breach of contract is abandonment. In the context of healthcare and genetic counseling, abandonment refers to the situation where a healthcare provider, including a genetic counselor, discontinues care for a patient without proper notification or without ensuring that the patient has a suitable substitute provider. This can occur when a counselor stops meeting with a patient or fails to provide necessary follow-up, leaving the patient without adequate support or guidance, which constitutes a breach of the duty of care. In contrast, negligence specifically refers to the failure to exercise the standard of care that a reasonably prudent professional would in a similar situation, which can result in harm to the patient. While negligence can involve abandonment, the term directly associated with the concept of halting care or failing to fulfill a professional obligation is abandonment. Battery involves unlawful physical contact, which is unrelated to the scenario of not providing care. Rights refer to the entitlements individuals have in the context of legal or ethical standards, rather than describing a specific failure in care provision.

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7. Which inheritance pattern is primarily seen in Coffin-Lowry Syndrome?

A. Autosomal Dominant

B. Autosomal Recessive

C. X-Linked Recessive

D. Mitochondrial

Coffin-Lowry Syndrome is primarily associated with an X-linked inheritance pattern, which is indicated by the correct answer. This means that the gene responsible for the condition is located on the X chromosome. Males, who have only one X chromosome, are more severely affected by this syndrome, as they will express the condition if they inherit the mutated gene. Females, having two X chromosomes, may be carriers and show milder symptoms due to the potential presence of a normal allele on their second X chromosome. Understanding the specific inheritance pattern of Coffin-Lowry Syndrome helps in genetic counseling, as it informs families about the risks of passing the condition to offspring. In particular, it is important to know that the affected fathers will pass the condition to all of their daughters, who will then become carriers, while their sons will not be affected. This knowledge allows for better family planning and risk assessment for future generations.

8. What does a false positive indicate in a test result?

A. Test indicates positive disease status in an unaffected individual

B. Test indicates negative disease status in an affected individual

C. Test indicates positive disease status in an affected individual

D. Test indicates negative disease status in an unaffected individual

A false positive indicates that a test suggests the presence of a disease or condition in an individual who does not actually have it. This situation arises when the test produces a positive result despite the absence of the condition in the individual being tested. Therefore, a false positive is characterized by the incorrect interpretation of the test result as indicating positive disease status, leading to potential anxiety, further unnecessary testing, or treatment for a condition that is not present. In the context of this question, option A accurately describes the scenario of a false positive. It is important to understand the implications of such errors in diagnostic testing, as they impact not only clinical decisions but also patient care and outcomes. On the other hand, the other options do not capture the essence of a false positive; they either describe true negative findings or misinterpretations of affected individuals, which do not pertain to the definition of a false positive.

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9. What are the primary neuropathological findings in Alzheimer's Disease?

A. Neurofibrillary tangles and amyloid plaques

B. White matter hyperintensities

C. Chromosomal deletions

D. Ischemic strokes

Alzheimer's Disease is characterized by distinct neuropathological changes that primarily include neurofibrillary tangles and amyloid plaques. Neurofibrillary tangles are composed of hyperphosphorylated tau protein and are indicative of intracellular disruptions in neuronal function. These tangles disrupt communication between neurons, leading to cell death. Amyloid plaques, on the other hand, are formed by the accumulation of beta-amyloid peptides outside the neurons. The buildup of these plaques can lead to inflammation and further neuronal damage, contributing to the cognitive decline seen in individuals with Alzheimer's Disease. Together, these two findings—neurofibrillary tangles and amyloid plaques—are considered hallmark features of the disease and are critical components of its pathology. Other options such as white matter hyperintensities, chromosomal deletions, and ischemic strokes do not represent the primary neuropathological features of Alzheimer's. While white matter hyperintensities can be found in older adults and may be associated with vascular dementia, they are not specific to Alzheimer's Disease. Chromosomal deletions can lead to various genetic disorders but are not directly related to the neuropathology of Alzheimer's. Ischemic strokes primarily involve blood flow disruption and can cause localized

10. Which inheritance pattern is characteristic of Orofaciodigital Syndrome?

A. Autosomal dominant

B. Autosomal recessive

C. X-linked dominant

D. Y-linked

Orofaciodigital Syndrome is primarily associated with an X-linked dominant inheritance pattern. This means that the gene mutation responsible for the syndrome is located on the X chromosome, and it affects both males and females, although males are typically more severely affected. In X-linked dominant disorders, an affected male will pass the trait to all of his daughters but none of his sons, while an affected female has a 50% chance of passing the mutation to each child, regardless of their sex. The characteristic features of Orofaciodigital Syndrome include abnormalities of the face, oral cavity, and digits, and its inheritance pattern is significant in genetic counseling as it affects familial risk assessments and implications for affected families. Understanding this inheritance pattern helps genetics professionals provide crucial information regarding recurrence risks and the implications for family planning for those involved. The other inheritance patterns listed are not consistent with the characteristics of Orofaciodigital Syndrome. For example, autosomal dominant disorders would present differently in terms of male-to-male transmission and risks passed to offspring, and autosomal recessive conditions require two copies of the mutated gene for manifestation, which does not apply in this syndrome. Y-linked inheritance affects traits only in males and is not relevant in the context of Orofaciodigital Syndrome

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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:

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

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