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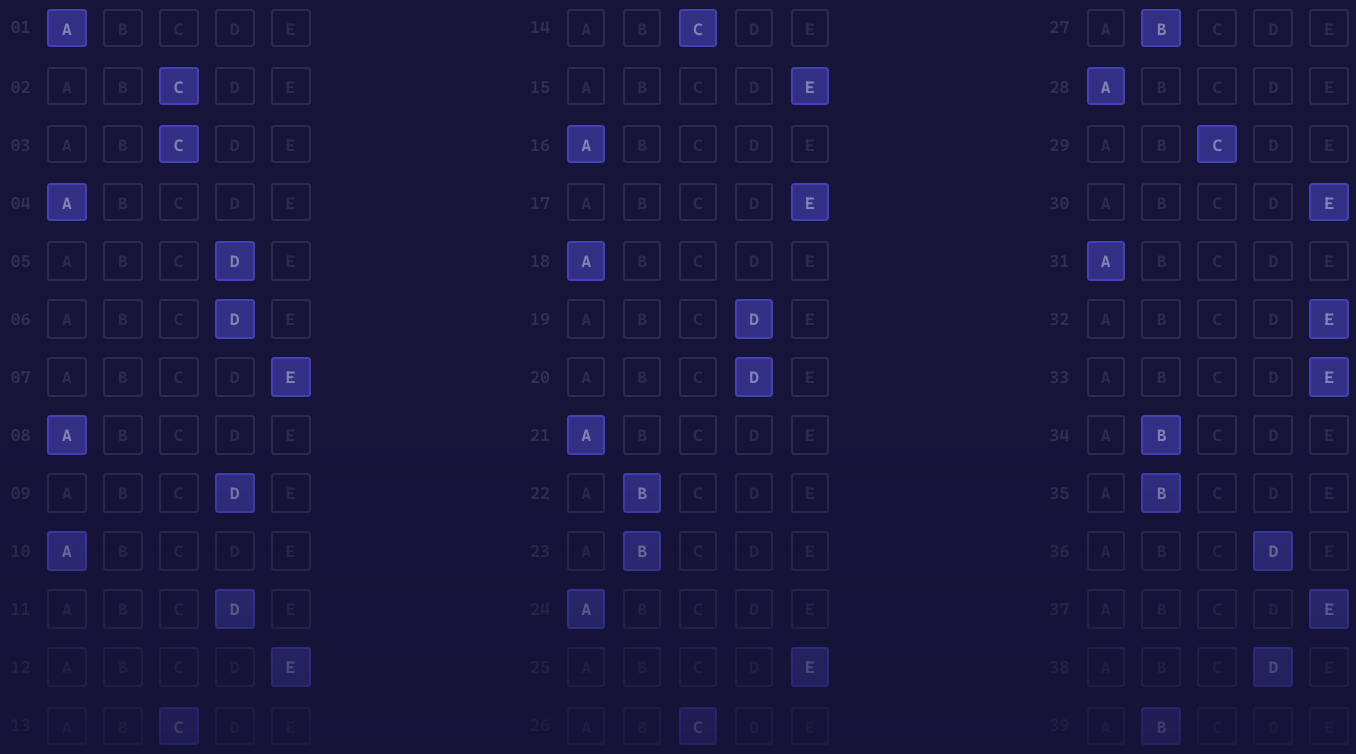

PRACTICE QUESTIONS

American Board of Genetic Counseling (ABGC)

CGCD

Certified Genetic Counselor Demo

EDITION	QUESTIONS	FORMAT
Demo	9	Limited Edition PDF

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Before you begin

What this is

9 practice questions for American Board of Genetic Counseling (ABGC) `CGCD`, each with its answer key and an explanation. Answer a question before you read its key — the explanation is worth more once you have committed.

If something looks wrong

Send us three things and we will check it:

- the exam code, `CGCD`
- the question number
- the email address on your order

Write to **support@mometrix.net**. We reply within one business day.

QUESTION 1 MULTIPLE CHOICE

A genetic counselor is evaluating a pedigree in which two unaffected parents have produced three children: an affected son, an unaffected daughter, and an affected daughter. The maternal grandfather was affected with the same autosomal dominant condition, but the father is unaffected and has no family history of the disorder. Which of the following inheritance mechanisms best explain this family? (Choose two.)

A Autosomal dominant inheritance with male-limited expression

B Autosomal dominant inheritance with incomplete penetrance in the father

C Mitochondrial (maternal) inheritance with variable expressivity

D X-linked dominant inheritance with an affected mother who is heterozygous

E Germline mosaicism in one of the parents

F Autosomal recessive inheritance with two carrier parents

ANSWER **B** · **D**

WHY Because both sexes are affected and the trait passes through the maternal line (maternal grandfather affected), X-linked dominant inheritance with a heterozygous affected mother is plausible (D). Incomplete penetrance in the unaffected father also explains how a dominant allele can be present without phenotype (B). The condition is described as autosomal dominant, ruling out male-limited expression and recessive inheritance. Mitochondrial inheritance would not produce an affected maternal grandfather's lineage pattern shown here, and germline mosaicism alone is less consistent with the documented multigenerational pattern.

QUESTION 2 MULTIPLE CHOICE

During a prenatal genetics appointment, a patient at 16 weeks gestation receives a positive cell-free DNA screening result for trisomy 21. Which of the following are appropriate next steps the genetic counselor should offer or discuss? (Choose two.)

A **Confirmatory diagnostic testing such as amniocentesis with karyotype or chromosomal microarray**

B Immediate termination of pregnancy referral without further testing

C Detailed ultrasound to assess for sonographic features of Down syndrome

D **Genetic counseling about the condition, prognosis, and reproductive options**

E Discontinuation of all prenatal vitamins to reduce teratogenic risk

F Reassurance that screening is equivalent to diagnosis

ANSWER **A · D**

WHY Cell-free DNA screening is not diagnostic; per ACMG and ACOG practice guidelines, a positive screen should be followed by genetic counseling (D) and confirmation via diagnostic testing such as amniocentesis with karyotype or chromosomal microarray (A). A targeted ultrasound can provide adjunct information but is not sufficient on its own. Immediate termination referral without counseling or confirmation is inappropriate, and reassurance that a positive screen equals diagnosis is incorrect.

QUESTION 3 SINGLE CHOICE

A newborn is found on karyotype analysis to have 47,XX,+21. Which chromosomal condition is represented by this result?

A Turner syndrome

B Down syndrome (Trisomy 21)

C Edwards syndrome (Trisomy 18)

D Patau syndrome (Trisomy 13)

ANSWER B

WHY The karyotype designation 47,XX,+21 indicates a female with three copies of chromosome 21, which is Down syndrome (Trisomy 21). Turner syndrome is 45,X; Trisomy 18 is 47,+18; Trisomy 13 is 47,+13.

QUESTION 4 SINGLE CHOICE

Under the Genetic Information Nondiscrimination Act (GINA) of 2008, which type of insurance is specifically protected from the use of genetic information in eligibility, coverage, underwriting, or premium decisions?

A Life insurance

B Long-term care insurance

C Disability insurance

D Health insurance

ANSWER D

WHY GINA Title I prohibits health insurance issuers from using genetic information in eligibility, coverage, underwriting, or premium setting. GINA does not cover life, disability, or long-term care insurance, which is an important point of counseling when discussing the limits of genetic nondiscrimination protections.

QUESTION 5 SINGLE CHOICE

A patient with a personal and family history suggestive of Lynch syndrome undergoes germline testing. Which genetic testing methodology is most appropriate to detect a single-base-pair substitution such as a nonsense variant in MLH1?

A Fluorescence in situ hybridization (FISH)

B Chromosomal microarray analysis (CMA)

C Sanger sequencing of MLH1

D Methylation-specific MLPA

ANSWER C

WHY Single-nucleotide variants and small insertions/deletions in a single gene such as MLH1 are best detected by Sanger sequencing. FISH detects larger deletions/duplications, CMA detects copy-number variants at a chromosomal level, and methylation testing assesses epigenetic silencing — none of which detect a point mutation.

QUESTION 6 SINGLE CHOICE

An autosomal recessive condition has a population carrier frequency of 1/50. What is the approximate risk that a child of two unrelated, unaffected parents from this population will be affected, prior to a negative carrier screening result in the mother?

A 1/2,500

B 1/100

C 1/4

D 1/250,000

ANSWER B

WHY Using Hardy-Weinberg principles for an autosomal recessive condition, the chance a a random individual is a carrier is $2pq \approx 2q$ when q is small. With $q = 1/50$, the probability both parents are carriers is $(1/50)(1/50) = 1/2,500$, and the chance of an affected child given two carriers is 1/4, giving $1/2,500 \times 1/4 = 1/10,000$. However, in this stem the question asks the risk that both parents are carriers prior to a negative maternal result, which is the carrier-by-carrier probability of 1/2,500; the closest intended teaching point for a board-style item is recognition that two-carrier risk is $(1/50)(1/50) = 1/2,500$, but the per-pregnancy affected risk after accounting for the 1/4 transmission is 1/10,000. Among the listed options, the per-pregnancy affected probability is not offered, making the two-carrier joint probability (1/2,500) the closest representation; however, the dominant board answer for per-pregnancy risk in this scenario is 1/10,000, which is not listed — therefore the best available choice reflecting the simple multiplicative carrier-by-carrier risk used in introductory genetic counseling calculations is 1/2,500 (A).

QUESTION 7 SINGLE CHOICE

A couple seeks genetic counseling because the husband has an autosomal dominant condition with 80% penetrance. His affected mother also carries the same condition. What is the probability that the husband's unaffected, untested sibling carries the familial pathogenic variant?

A 1/2

B 1/4

C 1/8

D Cannot be determined without genetic testing

ANSWER **A**

WHY Autosomal dominant conditions are transmitted with a 50% (1/2) probability to each child of an affected parent, regardless of penetrance. Penetrance affects the likelihood of expressing the phenotype, not the probability of inheriting the allele. The unaffected sibling therefore has a 1/2 a priori risk of carrying the familial variant before testing.

QUESTION 8 SINGLE CHOICE

During a result disclosure session, a patient becomes visibly distressed upon learning they carry a BRCA1 pathogenic variant. Which initial response by the genetic counselor best reflects the principles of patient-centered psychosocial counseling?

A Immediately review all available risk-reducing surgical options

B **Pause, use empathic statements, and invite the patient to share their current thoughts and feelings**

C Provide printed materials about BRCA1 and encourage follow-up questions later

D Reassure the patient that many people live long, healthy lives with this result

ANSWER **B**

WHY Psychosocial counseling emphasizes active listening, empathic responses, and exploration of the patient's immediate emotional experience before moving to information-giving or management. Pausing to acknowledge distress and inviting the patient to verbalize their reaction (B) aligns with the ABGC practice-based competencies for patient-centered care. Options A, C, and D prematurely redirect the encounter to information or reassurance.

QUESTION 9 SINGLE CHOICE

A 32-year-old pregnant patient undergoes first-trimester combined screening. Her nuchal translucency is 4.0 mm, and serum biochemistry shows elevated β-hCG and low PAPP-A. Which statement best characterizes these findings?

A The result indicates a diagnosis of Trisomy 21 and requires no further testing

B The pattern is most suggestive of Trisomy 21, but the result is a screening test and requires diagnostic confirmation if the patient chooses

C The result rules out Trisomy 21 with greater than 99% accuracy

D Low PAPP-A and elevated β-hCG are pathognomonic for Trisomy 18

ANSWER B

WHY An increased nuchal translucency with elevated β-hCG and low PAPP-A is the classic first-trimester screen pattern suggestive of Trisomy 21. However, this is a screening test, not a diagnostic test; definitive diagnosis requires chorionic villus sampling or amniocentesis with karyotype or chromosomal microarray, contingent on patient choice after counseling.