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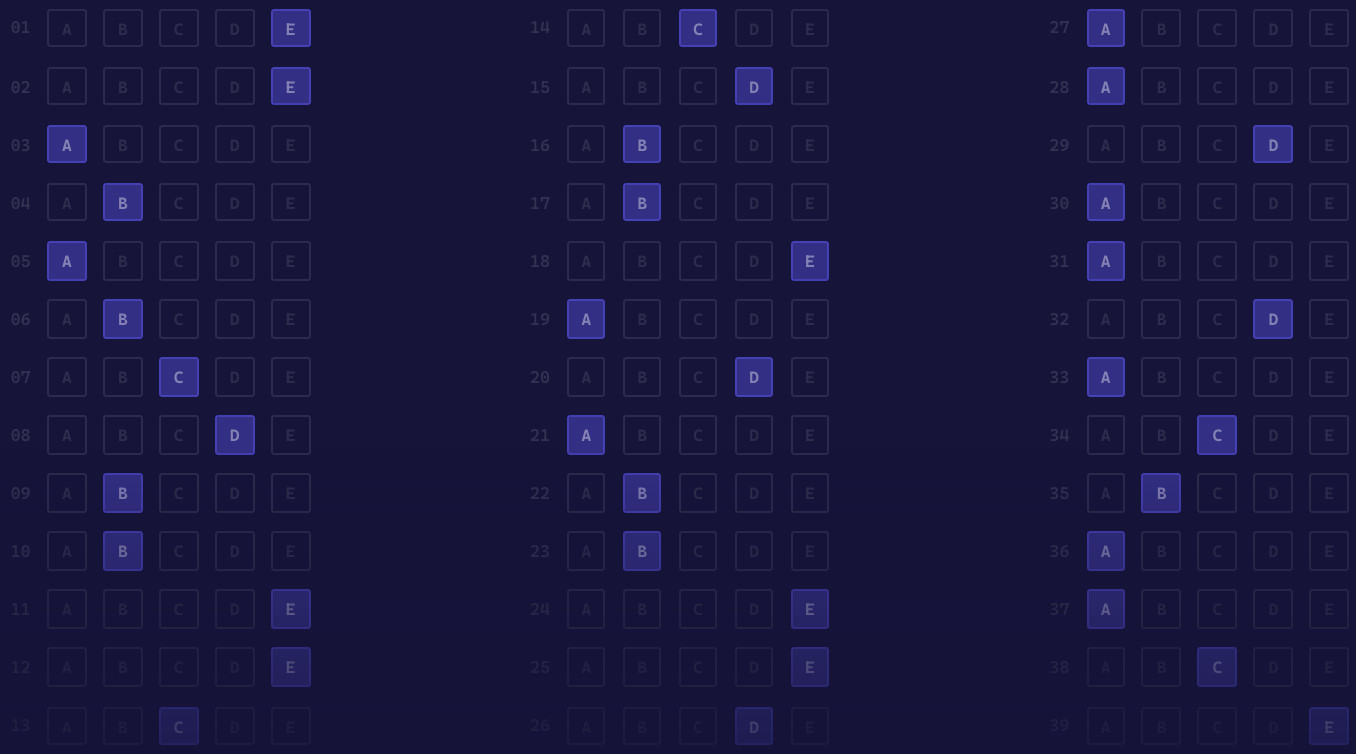

PRACTICE QUESTIONS

American Board of Genetic Counseling (ABGC)

CGC

Certified Genetic Counselor

EDITION	QUESTIONS	FORMAT
Demo	9	Limited Edition PDF

WEB www.mometrix.net

SUPPORT support@mometrix.net

Before you begin

What this is

9 practice questions for American Board of Genetic Counseling (ABGC) `CGC`, 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, `CGC`
- 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 32-year-old woman presents for genetic counseling regarding her family history of hereditary breast and ovarian cancer. Her mother was diagnosed with ovarian cancer at age 54, and her maternal aunt was diagnosed with breast cancer at age 47. Which of the following inheritance patterns and risk assessment considerations are most appropriate to include in the initial counseling session? (Choose two.)

A **Autosomal dominant inheritance is the most likely pattern given the maternal lineage pattern of breast and ovarian cancer across two generations.**

B An autosomal recessive pattern should be considered first because both mother and maternal aunt were affected in the same sibship.

C **The unaffected 35-year-old brother of the patient should be offered testing because male BRCA2 carriers have an elevated lifetime risk of breast and prostate cancer.**

D Mitochondrial inheritance should be excluded because ovarian cancer is a recognized feature of mitochondrial disorders.

E The patient's lifetime risk for breast cancer in the general population (approximately 12%) should be quoted without modification because no affected first-degree relatives are alive to test.

F Anticipation is a hallmark of hereditary breast and ovarian cancer syndrome and should be discussed when reviewing the pedigree.

ANSWER **A · C**

WHY Hereditary breast and ovarian cancer syndrome (most often associated with BRCA1/BRCA2) follows an autosomal dominant inheritance pattern with each affected generation showing multiple cancers and both sexes capable of carrying and being affected by pathogenic variants. Males who carry BRCA2 (and to a lesser extent BRCA1) pathogenic variants have an elevated lifetime risk of breast cancer and prostate cancer, so the patient's brother should be offered appropriate testing and counseling if a familial variant is identified. Autosomal recessive inheritance is incorrect because the pattern involves affected individuals in successive generations with both sexes at risk. Mitochondrial inheritance is incorrect because BRCA-associated cancers are not mitochondrial, and anticipation is not a feature of BRCA-associated hereditary cancer syndromes (it is seen in trinucleotide repeat disorders). Quoting general-population risk ignores the family history and is not appropriate counseling practice.

QUESTION 2 MULTIPLE CHOICE

Which of the following statements about Huntington disease are accurate and appropriate for inclusion in pretest genetic counseling? (Choose two.)

A **Huntington disease is caused by a CAG trinucleotide repeat expansion in the HTT gene on chromosome 4.**

B Huntington disease is caused by a point mutation in the HTT gene and shows a stable number of repeats across generations.

C **Anticipation, particularly with paternal transmission, may lead to earlier onset and more severe disease in subsequent generations.**

D Huntington disease demonstrates anticipation equally with maternal and paternal transmission.

E A predictive test is typically offered only to individuals who are 18 years of age or older because of the implications for minors.

F Presymptomatic testing in minors at 50% risk is routinely encouraged to allow early life planning.

ANSWER **A · C**

WHY Huntington disease is caused by a CAG trinucleotide repeat expansion in the HTT gene on chromosome 4, and the repeat is unstable and tends to expand during transmission, leading to anticipation (earlier age of onset in successive generations). Anticipation is more pronounced with paternal transmission because spermatogenesis is more prone to repeat expansion. Predictive testing in at-risk minors is generally not recommended by professional societies (e.g., ACMG, NSGC/Huntington Disease Society of America guidelines) because the result removes the minor's future autonomous choice and there is no preventive intervention during childhood; testing is therefore typically deferred until the individual can give informed consent as an adult.

QUESTION 3 SINGLE CHOICE

A 28-year-old pregnant patient at 10 weeks gestation asks about prenatal aneuploidy screening. Which test most directly analyzes placental DNA circulating in maternal blood and can screen for common trisomies as early as 9–10 weeks gestation?

A First-trimester combined screening (nuchal translucency plus serum PAPP-A and free beta-hCG)

B Cell-free DNA screening (non-invasive prenatal testing)

C Chorionic villus sampling

D Amniocentesis

ANSWER B

WHY Cell-free DNA screening (commonly called non-invasive prenatal testing or NIPT) analyzes fragments of placental DNA that circulate in maternal plasma and screens for common aneuploidies such as trisomy 21, trisomy 18, and trisomy 13. It can be performed from approximately 9–10 weeks gestation onward. First-trimester combined screening is a serum and ultrasound screen, not a DNA-based test. Chorionic villus sampling and amniocentesis are diagnostic (not screening) tests that sample placental tissue or amniotic fluid, respectively, and carry a procedure-related risk of pregnancy loss.

QUESTION 4 SINGLE CHOICE

A genetic counselor learns that an adult patient has chosen not to share a positive BRCA1 result with her adult sister who is also at 50% risk. The patient has decisional capacity and understands the implications. According to the ethical principle of autonomy and ABGC/NSGC practice standards, what is the most appropriate course of action?

A Immediately contact the sister because she is at risk and has a right to know.

B Respect the patient's decision and continue to encourage voluntary family communication, while exploring the patient's concerns and offering resources such as a family letter.

C Document the sister's contact information and forward the test result without the patient's consent.

D Refuse to see the patient again until she agrees to contact her sister.

ANSWER B

WHY Genetic counselors uphold patient autonomy and confidentiality. While they strongly encourage sharing of clinically actionable results with at-risk relatives, they do not override a competent adult patient's decision to withhold their own genetic information. Best practice is to explore the patient's reasons, address concerns, offer tools such as a family letter or facilitation letter, and document the discussion. Directly contacting relatives or forwarding results without consent would breach confidentiality and is not ethically or legally appropriate under standard U.S. genetic counseling practice.

QUESTION 5 SINGLE CHOICE

Which statement best characterizes the U.S. Recommended Uniform Screening Panel (RUSP) for newborns?

A It is a federal mandate that every newborn in the United States must be screened for every condition on the panel.

B **It is a set of recommendations developed by the Advisory Committee on Heritable Disorders in Newborns and Children; each state decides which conditions to include on its newborn screening panel.**

C It is a list of conditions screened only by private laboratories and is not used in state public health programs.

D It is restricted to metabolic disorders detectable by tandem mass spectrometry.

ANSWER B

WHY The Recommended Uniform Screening Panel (RUSP) is a list of disorders recommended by the Health Resources and Services Administration's Advisory Committee on Heritable Disorders in Newborns and Children. It is not a federal mandate; each state independently decides which conditions to include in its newborn screening program. The RUSP includes metabolic, endocrine, hematologic, immunologic, and other disorders, well beyond those detectable by tandem mass spectrometry, and is implemented through state public health newborn screening programs.

QUESTION 6 SINGLE CHOICE

Which hereditary cancer syndrome is most strongly associated with germline pathogenic variants in the TP53 gene and involves early-onset sarcomas, breast cancer, adrenocortical carcinoma, and brain tumors?

A Lynch syndrome

B **Li-Fraumeni syndrome**

C Cowden syndrome

D Familial adenomatous polyposis

ANSWER **B**

WHY Li-Fraumeni syndrome is caused by germline pathogenic variants in TP53 and is characterized by a wide spectrum of early-onset malignancies, including soft-tissue sarcomas, osteosarcomas, premenopausal breast cancer, adrenocortical carcinoma, and brain tumors, often in childhood or young adulthood. Lynch syndrome involves MLH1, MSH2, MSH6, PMS2, or EPCAM and predisposes primarily to colorectal, endometrial, and other cancers. Cowden syndrome involves PTEN and predisposes to breast, thyroid, and endometrial cancers. Familial adenomatous polyposis involves APC and predisposes to colon polyps and colorectal cancer.

QUESTION 7 SINGLE CHOICE

A couple presents for genetic counseling. The partner who is phenotypically normal has a balanced Robertsonian translocation between chromosomes 14 and 21. Which statement best describes the recurrence risk for this couple?

A All pregnancies will be unaffected because the translocation is balanced in the carrier.

B **There is an increased risk of liveborn offspring with Down syndrome and an increased risk of pregnancy loss due to unbalanced gametes.**

C Only male offspring will be affected because the translocation involves chromosome 21.

D The recurrence risk is identical to the general population risk because the carrier is phenotypically normal.

ANSWER B

WHY Carriers of a balanced Robertsonian translocation between chromosomes 14 and 21 are phenotypically normal but produce a proportion of unbalanced gametes (translocation Down syndrome, monosomy 21, or trisomy 21 with monosomy 14), resulting in an increased risk of having a child with Down syndrome and an increased risk of miscarriage. The risk does not depend on the sex of the offspring, and it is not the same as the general population risk.

QUESTION 8 SINGLE CHOICE

A patient is being started on the anticoagulant warfarin. Which pharmacogenomic association is most clinically established and relevant to dosing decisions?

A CYP2C19 genotype and clopidogrel activation

B SLCO1B1 genotype and simvastatin-induced myopathy

C CYP2C9 and VKORC1 genotypes and warfarin dosing

D HLA-B*57:01 and abacavir hypersensitivity

ANSWER C

WHY CYP2C9 (and to a lesser extent CYP2C19) and VKORC1 genotypes together account for a substantial proportion of the inter-individual variability in warfarin dose requirement, and dosing algorithms incorporating these genotypes are widely used in clinical practice. CYP2C19/clopidogrel is a well-known pharmacogenomic association but not relevant to warfarin. SLCO1B1/simvastatin is also an established association but not relevant to warfarin. HLA-B*57:01/abacavir is a strong pharmacogenomic association used in HIV care, not anticoagulation.

QUESTION 9 SINGLE CHOICE

A 6-month-old infant presents with failure to thrive, hepatosplenomegaly, cataracts, and developmental delay. Newborn screening was reportedly normal. Which condition is most likely on the differential?

A Phenylketonuria

B Classic galactosemia

C Cystic fibrosis

D Maple syrup urine disease

ANSWER B

WHY Classic galactosemia, caused by deficiency of galactose-1-phosphate uridyltransferase (GALT), presents in the neonatal period after the introduction of milk (lactose → glucose + galactose) with vomiting, failure to thrive, hepatomegaly, jaundice, cataracts, and E. coli sepsis risk. Some U.S. states do not include classic galactosemia on their newborn screening panel, and infants who are not screened or are breastfed before screening results return can present clinically as described. PKU does not cause cataracts or hepatosplenomegaly in this pattern. Cystic fibrosis typically presents with respiratory and pancreatic insufficiency rather than cataracts. MSUD presents with neurologic deterioration and a characteristic sweet-smelling urine, not cataracts.