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37	A	B	C	D	E
38	A	B	C	D	E
39	A	B	C	D	E

PRACTICE QUESTIONS

American Board of Medical Genetics And Genomics (ABMGG)

MG-GE

General Examination

EDITION

Demo

QUESTIONS

9

FORMAT

Limited Edition PDF

Before you begin

What this is

9 practice questions for American Board of Medical Genetics And Genomics (ABMGG) **MG-GE**, 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, **MG-GE**
- the question number
- the email address on your order

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

QUESTION 1

SINGLE CHOICE

A male with classic hemophilia A (X-linked recessive) marries a woman whose brother is affected and whose mother is an obligate carrier. What is the probability that their first-born son will be affected?

- ☐ A 0
- ☐ B $1/8$
- ☐ C $1/4$
- ☒ D $1/2$

ANSWER D

WHY The wife's mother is an obligate carrier (heterozygous, $X^H X^h$). The wife therefore has a $1/2$ chance of being a carrier. If she is a carrier, half of her sons will be affected; if she is not a carrier, none will be. Combined probability = $1/2 \times 1/2 = 1/4$. The husband being affected does not influence a son's status because he passes his Y to sons.

QUESTION 2

SINGLE CHOICE

In a three-generation pedigree, an autosomal dominant trait shows male-to-male transmission, every affected individual has an affected parent, and unaffected individuals do not transmit the trait. The most likely mode of inheritance is:

- ☐ A X-linked dominant
- ☐ B Mitochondrial
- ☒ C Autosomal dominant
- ☐ D Autosomal recessive with pseudodominance

ANSWER C

WHY Male-to-male transmission excludes X-linked and mitochondrial inheritance. Vertical transmission with affected individuals in every generation and the absence of skipping is characteristic of autosomal dominant inheritance.

QUESTION 3

SINGLE CHOICE

A newborn with hypotonia, upslanting palpebral fissures, epicanthal folds, a single transverse palmar crease, and a heart murmur has a karyotype most likely to show:

- ☐ A 45,X
- ☐ B 47,XX,+18
- ☒ C 47,XY,+21
- ☐ D 47,XXY

ANSWER C

WHY The described constellation of features is classic for Down syndrome (Trisomy 21). The other options correspond to Turner syndrome (45,X), Edwards syndrome (47,+18), and Klinefelter syndrome (47,XXY), which have different clinical pictures.

QUESTION 4

SINGLE CHOICE

Which DNA repair mechanism is primarily responsible for removing cyclobutane pyrimidine dimers created by ultraviolet light exposure?

- ☐ A Mismatch repair (MMR)
- ☒ B Nucleotide excision repair (NER)
- ☐ C Base excision repair (BER)
- ☐ D Homologous recombination repair (HR)

ANSWER B

WHY UV radiation produces cyclobutane pyrimidine dimers and 6-4 photoproducts, which distort the DNA helix and are removed by nucleotide excision repair. Defects in NER cause xeroderma pigmentosum. Mismatch repair corrects replication errors, base excision repair removes damaged individual bases (e.g., via glycosylases), and homologous recombination repairs double-strand breaks.

QUESTION 5

SINGLE CHOICE

A laboratory wants to simultaneously assess gene expression levels of thousands of transcripts in a tumor sample. The most appropriate high-throughput method is:

- ☐ A Sanger sequencing
- ☒ B RNA sequencing (RNA-seq)
- ☐ C Southern blot
- ☐ D Allele-specific PCR

ANSWER B

WHY RNA-seq provides genome-wide quantitative measurement of transcript abundance. Sanger sequencing determines the nucleotide sequence of individual amplicons. Southern blot detects specific DNA fragments. Allele-specific PCR interrogates a single known variant.

QUESTION 6

SINGLE CHOICE

Deficiency of phenylalanine hydroxylase most directly causes which biochemical finding in untreated patients?

- ☒ A Decreased tyrosine and elevated phenylalanine
- ☐ B Elevated tyrosine and decreased phenylalanine
- ☐ C Decreased homogentisic acid
- ☐ D Elevated branched-chain amino acids

ANSWER A

WHY Phenylalanine hydroxylase converts phenylalanine to tyrosine. Deficiency (classic phenylketonuria) leads to accumulation of phenylalanine and its keto acids, with reduced tyrosine, which becomes a conditionally essential amino acid. Elevated branched-chain amino acids occur in maple syrup urine disease, and elevated homogentisic acid occurs in alkaptonuria.

QUESTION 7

SINGLE CHOICE

In a population in Hardy-Weinberg equilibrium, the carrier frequency ($2pq$) for an autosomal recessive condition with disease frequency $1/10,000$ is approximately:

- ☒ A 1/50
- ☐ B 1/100
- ☐ C 1/200
- ☐ D 1/10,000

ANSWER A

WHY If $q^2 = 1/10,000$, then $q = 1/100$ and $p \approx 1$. The carrier frequency $2pq \approx 2(1)(1/100) = 1/50$. This illustrates that carriers of rare recessive alleles are far more common than affected individuals.

QUESTION 8

SINGLE CHOICE

A family presents with early-onset colorectal cancer, endometrial cancer, and numerous colonic adenomas in multiple relatives. Germline testing most likely reveals a pathogenic variant in:

- ☐ A RB1
- ☒ B MSH2 (Lynch syndrome)
- ☐ C APC (Familial Adenomatous Polyposis)
- ☐ D TP53

ANSWER B

WHY Lynch syndrome (hereditary nonpolyposis colorectal cancer) is caused by germline mutations in DNA mismatch repair genes (MLH1, MSH2, MSH6, PMS2) or EPCAM, predisposing to colorectal, endometrial, ovarian, gastric, and other cancers without the hundreds of polyps seen in FAP (APC). RB1 causes retinoblastoma, and TP53 causes Li-Fraumeni syndrome.

QUESTION 9

MULTIPLE CHOICE

Which of the following are recognized core components of informed consent in genetic testing? (Choose three.)

- ☒ **A** Description of the condition being tested, including its variability and natural history
- ☒ **B** Discussion of risks and benefits of testing, including implications for family members
- ☐ **C** Guarantee that results will be used only for the stated clinical purpose
- ☒ **D** Explanation of alternatives to testing, including the option to decline

ANSWER A • B • D

WHY Core elements of informed consent include disclosure of the test's purpose and the condition (including variability, natural history, and treatability), risks/benefits/limitations, implications for family members, alternatives including the option to decline, confidentiality protections, and options for future use of the specimen. A guarantee of restricted use is not standard—appropriate uses (such as de-identified research with consent) are generally discussed rather than guaranteed.