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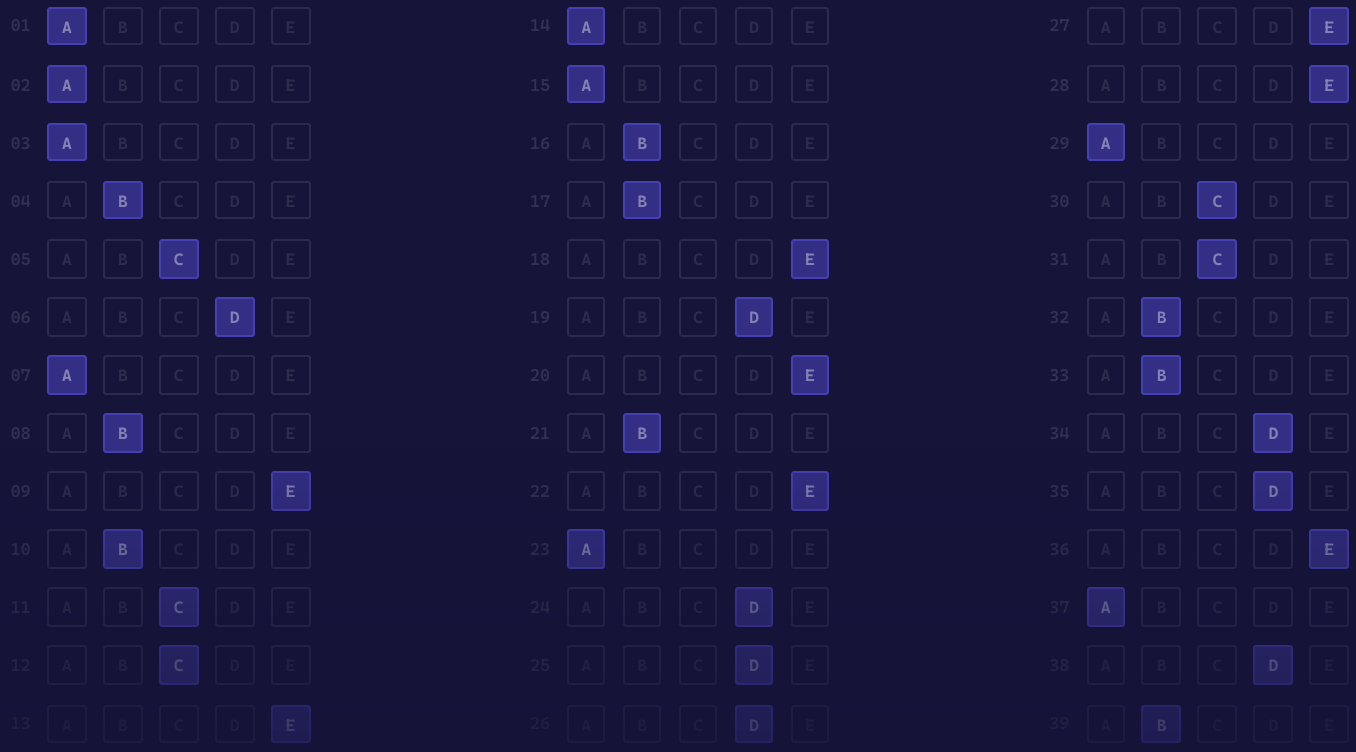

PRACTICE QUESTIONS

American Board of Medical Genetics And Genomics (ABMGG)

MG–CY

Clinical Cytogenetics & Genomics Exam

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 Medical Genetics And Genomics (ABMGG) `MG-CY`, 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-CY`
- the question number
- the email address on your order

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

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QUESTION 1 SINGLE CHOICE

A peripheral blood sample is referred for chromosome analysis. The referring clinician suspects a diagnosis most commonly associated with a deletion on the short arm of chromosome 5. Which banding technique is the laboratory most likely to use as a first-line method to detect this deletion?

A **G-banding (Giemsa banding) at approximately 400–550 band resolution**

B Q-banding using quinacrine mustard staining

C T-banding to highlight terminal chromosome regions

D R-banding (reverse banding) using acridine orange

ANSWER **A**

WHY G-banding (Giemsa staining following trypsin treatment) is the standard first-line banding technique used in clinical cytogenetics laboratories. It produces a reproducible pattern of light and dark bands and, at a typical 400–550 band level of resolution, is sufficient to detect deletions such as the 5p deletion seen in Cri du Chat syndrome. Q-banding requires fluorescence microscopy and is rarely used in routine practice. T-banding and R-banding are specialized techniques used for specific purposes rather than routine karyotyping.

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QUESTION 2 SINGLE CHOICE

An amniocyte karyotype reveals 47,XX,+21 in all 15 cells examined from two independent cultures. Which interpretation is most accurate?

A Mosaic Down syndrome

B Constitutional (non-mosaic) Trisomy 21

C Trisomy 21 due to an unbalanced Robertsonian translocation

D Maternal cell contamination artifact

ANSWER B

WHY When an extra chromosome 21 is present in every cell analyzed from two independent cultures, this represents a constitutional (non-mosaic) Trisomy 21, the most common cause of Down syndrome. Mosaicism would require two or more distinct cell lines. A Robertsonian translocation would be written as a derivative chromosome, such as der(14;21)(q10;q10), not as simple +21. Maternal cell contamination (MCC) is suspected when the result is inconsistent with the fetal phenotype or when only female cells are seen, and is typically excluded by comparing maternal and fetal genotypes.

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QUESTION 3 SINGLE CHOICE

A 3-year-old child presents with global developmental delay, hypotonia, and facial dysmorphism. A previous high-resolution G-banded karyotype was reported as normal. Which test is most appropriate as the next-tier investigation?

A Repeat G-banded karyotype at higher band resolution

B **Chromosomal microarray analysis (CMA)**

C Standard whole exome sequencing

D Mitochondrial DNA sequencing

ANSWER **B**

WHY When a high-resolution karyotype is normal in a child with unexplained developmental delay and dysmorphic features, chromosomal microarray analysis (CMA) is the recommended next-tier test. CMA detects copy number variants (microdeletions and microduplications) that are below the resolution of light microscopy. Repeating the karyotype at higher resolution is unlikely to add yield. Whole exome sequencing is typically used when CMA is non-diagnostic, not as a first follow-up. Mitochondrial DNA sequencing is reserved for specific clinical indications such as suspected mitochondrial disease.

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QUESTION 4 SINGLE CHOICE

A patient with a normal male karyotype is found by chromosomal microarray to have a 1.5 Mb deletion on the X chromosome encompassing the DMD gene. Which statement best explains why this deletion is pathogenic in this male patient?

A X-linked deletions in males are always pathogenic because there is no second allele

B Males are hemizygous for the X chromosome, so loss of a single copy of DMD results in loss of function

C The deletion disrupts an autosomal recessive locus

D Males inherit two X chromosomes, one of which is inactivated

ANSWER B

WHY Males normally have a single X chromosome (46,XY) and are therefore hemizygous for X-linked genes such as DMD. Loss of a single functional copy of DMD on the X chromosome results in loss of dystrophin expression, which is the basis of Duchenne muscular dystrophy in this patient. The statement that 'X-linked deletions in males are always pathogenic' is too broad; some X-linked deletions may involve genes with variable expressivity or be compensated. DMD is not autosomal recessive, and males do not inherit two X chromosomes.

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QUESTION 5 SINGLE CHOICE

Which cytogenetic abnormality most commonly results from breakage and reunion at the centromeres of two acrocentric chromosomes with loss of the short arms?

A Reciprocal translocation

B Pericentric inversion

C Robertsonian translocation

D Isochromosome formation

ANSWER C

WHY Robertsonian translocations result from breakage at or near the centromeres of two acrocentric chromosomes (13, 14, 15, 21, 22) with fusion of the long arms and loss of the short arms, which contain ribosomal RNA genes that are present in multiple copies elsewhere. Reciprocal translocations involve exchange of segments between non-homologous chromosomes. Pericentric inversions include the centromere. Isochromosomes arise from misdivision through the centromere, producing a chromosome with two identical arms.

QUESTION 6 SINGLE CHOICE

A bone marrow karyotype from a patient with newly diagnosed chronic myeloid leukemia (CML) is most likely to demonstrate which of the following abnormalities?

A t(9;22)(q34;q11.2)

B t(8;21)(q22;q22)

C t(15;17)(q22;q12)

D inv(16)(p13.1q22)

ANSWER A

WHY The Philadelphia chromosome, der(22)t(9;22)(q34;q11.2), producing the BCR::ABL1 fusion, is the defining cytogenetic abnormality of chronic myeloid leukemia and is detected in approximately 95% of cases. t(8;21) and inv(16) are associated with core-binding factor acute myeloid leukemia (AML), while t(15;17) is characteristic of acute promyelocytic leukemia (APL/AML-M3).

QUESTION 7 SINGLE CHOICE

Fluorescence in situ hybridization (FISH) is performed on a metaphase spread using a locus-specific probe for the BCR gene on chromosome 22q11.2 and a second probe for the ABL1 gene on chromosome 9q34. In a normal cell, what hybridization pattern is expected?

A Two red signals only

B Two green signals only

C **Two red and two green signals that are not adjacent**

D One fused yellow signal and one separate red and one separate green signal

ANSWER C

WHY In a normal nucleus or metaphase, the BCR probe labels two chromosome 22 homologs (two red signals) and the ABL1 probe labels two chromosome 9 homologs (two green signals). Because BCR and ABL1 are on different chromosomes, the signals appear as separate red and green pairs, not adjacent to one another. A fused (yellow) signal representing the BCR::ABL1 fusion would only be expected if the Philadelphia chromosome translocation is present.

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QUESTION 8 MULTIPLE CHOICE

A peripheral blood karyotype from a 6-month-old infant with asymmetric growth shows two cell lines: 45,X in 8 cells and 46,XY in 12 cells, from a total of 20 cells analyzed. Which of the following statements accurately apply to this result? (Choose two.)

A **The result meets the ISCN/standard criterion to be reported as mosaic 45,X/46,XY**

B The presence of a 46,XY cell line excludes Turner syndrome in this patient

C **Mosaicism must be considered when interpreting the patient's phenotype**

D A minimum of 20 cells must show the same abnormality to call true mosaicism

ANSWER **A · C**

WHY Standard cytogenetic practice (per ACMG and ISCN guidelines) requires two or more cells with the same abnormality to call true mosaicism, and this case meets that criterion (8 cells with 45,X and 12 cells with 46,XY). Therefore the result should be reported as mosaic 45,X/46,XY. Mosaicism must always be considered when interpreting the patient's phenotype because the proportion and tissue distribution of each cell line influences clinical features. The presence of a 46,XY cell line does NOT exclude Turner syndrome, since the 45,X line can produce features of Turner syndrome depending on tissue distribution. The '20 cells' rule is incorrect; the standard threshold is observation of an abnormality in two or more cells.

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QUESTION 9 MULTIPLE CHOICE

Which of the following are appropriate clinical indications for chromosomal microarray analysis (CMA) rather than a standard karyotype? (Choose two.)

A **Evaluation of a child with autism spectrum disorder and intellectual disability**

B Confirmation of a clinically suspected trisomy 21 in a newborn

C **A fetus with multiple anomalies on prenatal ultrasound and a normal karyotype**

D Detection of low-level mosaicism below 10%

ANSWER **A · C**

WHY CMA is the recommended first-tier test for children with unexplained developmental delay, intellectual disability, autism spectrum disorder, and multiple congenital anomalies because it detects submicroscopic copy number variants that are not visible by karyotype. A fetus with multiple anomalies and a normal karyotype is also an appropriate indication, since CMA may reveal pathogenic microdeletions or microduplications. Confirming a clinically suspected trisomy 21 is more appropriately done by rapid FISH or karyotype, both of which detect the aneuploidy reliably. Detecting low-level mosaicism below 10% is a known limitation of CMA; standard karyotype or FISH is preferred for mosaicism assessment.

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