

01	☒ A	☐ B	☐ C	☐ D	☐ E
02	☐ A	☒ B	☐ C	☐ D	☐ E
03	☒ A	☐ B	☐ C	☐ D	☐ E
04	☐ A	☒ B	☐ C	☐ D	☐ E
05	☐ A	☐ B	☐ C	☐ D	☒ E
06	☐ A	☐ B	☐ C	☐ D	☒ E
07	☐ A	☐ B	☐ C	☐ D	☒ E
08	☐ A	☐ B	☒ C	☐ D	☐ E
09	☐ A	☐ B	☒ C	☐ D	☐ E
10	☐ A	☒ B	☐ C	☐ D	☐ E
11	☐ A	☐ B	☒ C	☐ D	☐ E
12	☐ A	☐ B	☐ C	☐ D	☒ E
13	☐ A	☒ B	☐ C	☐ D	☐ E

14	☐ A	☐ B	☒ C	☐ D	☐ E
15	☐ A	☐ B	☐ C	☒ D	☐ E
16	☐ A	☐ B	☒ C	☐ D	☐ E
17	☒ A	☐ B	☐ C	☐ D	☐ E
18	☐ A	☐ B	☒ C	☐ D	☐ E
19	☐ A	☐ B	☐ C	☐ D	☒ E
20	☐ A	☐ B	☐ C	☐ D	☒ E
21	☒ A	☐ B	☐ C	☐ D	☐ E
22	☐ A	☐ B	☐ C	☒ D	☐ E
23	☐ A	☐ B	☒ C	☐ D	☐ E
24	☐ A	☐ B	☐ C	☒ D	☐ E
25	☐ A	☐ B	☐ C	☐ D	☒ E
26	☒ A	☐ B	☐ C	☐ D	☐ E

27	☐ A	☒ B	☐ C	☐ D	☐ E
28	☒ A	☐ B	☐ C	☐ D	☐ E
29	☐ A	☐ B	☒ C	☐ D	☐ E
30	☐ A	☐ B	☐ C	☐ D	☒ E
31	☐ A	☒ B	☐ C	☐ D	☐ E
32	☒ A	☐ B	☐ C	☐ D	☐ E
33	☐ A	☐ B	☒ C	☐ D	☐ E
34	☐ A	☐ B	☐ C	☐ D	☒ E
35	☐ A	☐ B	☒ C	☐ D	☐ E
36	☐ A	☒ B	☐ C	☐ D	☐ E
37	☐ A	☒ B	☐ C	☐ D	☐ E
38	☒ A	☐ B	☐ C	☐ D	☐ E
39	☒ A	☐ B	☐ C	☐ D	☐ E

PRACTICE QUESTIONS

Canadian Alliance of Medical Laboratory Professionals Regulators (CAMLPR)

BCG

Clinical Genetics Bundle (Cytogenetics, Molecular Diag)

EDITION

Demo

QUESTIONS

9

FORMAT

Limited Edition PDF

Before you begin

What this is

9 practice questions for Canadian Alliance of Medical Laboratory Professionals Regulators (CAMLPR) BCG, 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, BCG
- 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 prenatal specimen is received for aneuploidy screening and rapid turnaround is required. Which molecular method is most appropriate for detecting trisomy 21, trisomy 18, and trisomy 13 within 24–48 hours?

- ☐ A Karyotype analysis on cultured amniocytes
- ☒ B Fluorescence in situ hybridization (FISH) on interphase nuclei
- ☐ C Chromosomal microarray (CMA) using SNP probes
- ☐ D Sanger sequencing of the BRCA1 gene

ANSWER B

WHY Interphase FISH (commonly on uncultured amniocytes or chorionic villi) detects the most common trisomies (13, 18, 21 and sex chromosome aneuploidies) rapidly without requiring cell culture. Karyotype provides a whole-genome view but takes longer; CMA can detect microdeletions/duplications but has a longer turnaround and is not the first-line rapid screen for common trisomies. BRCA1 sequencing is unrelated to aneuploidy detection.

QUESTION 2

SINGLE CHOICE

A chromosomal microarray on a pediatric patient shows a 2.5 Mb deletion on chromosome 15q11.2–q13, which includes the SNRPN locus. Which genetic disorder is most consistent with this finding?

- ☐ A Williams syndrome (7q11.23 deletion)
- ☒ B Prader-Willi / Angelman syndrome region (15q11–q13)
- ☐ C Cri-du-chat syndrome (5p deletion)
- ☐ D 22q11.2 deletion syndrome

ANSWER B

WHY The 15q11.2–q13 region containing SNRPN is the imprinted region responsible for Prader-Willi syndrome (paternal deletion) and Angelman syndrome (maternal deletion). Williams syndrome involves ELN at 7q11.23, Cri-du-chat involves 5p, and 22q11.2 deletion (DiGeorge/velocardiofacial) is a separate locus.

QUESTION 3

SINGLE CHOICE

A patient with suspected cystic fibrosis is found to carry the F508del mutation on one allele by standard CFTR panel testing. Which molecular technique is most appropriate to detect the mutation on the second allele if the panel covers only the most common CFTR variants?

- ☒ **A** Sanger sequencing of the entire CFTR gene
- ☐ **B** Karyotype analysis
- ☐ **C** Fluorescence in situ hybridization (FISH) with a CFTR probe
- ☐ **D** Serum protein electrophoresis

ANSWER A

WHY Mutation panels cover only the most common CFTR variants. To identify rare or novel mutations on the second allele, full gene Sanger sequencing (or comprehensive NGS panel) of CFTR is the appropriate molecular follow-up. Karyotype, FISH, and protein electrophoresis do not detect point mutations in CFTR.

QUESTION 4

SINGLE CHOICE

A peripheral blood karyotype reveals 46,XX,t(9;22)(q34;q11). Which clinical condition is this translocation classically associated with?

- ☐ **A** Burkitt lymphoma
- ☒ **B** Chronic myeloid leukemia (CML)
- ☐ **C** Acute promyelocytic leukemia (APL)
- ☐ **D** Down syndrome

ANSWER B

WHY The t(9;22)(q34;q11) translocation produces the BCR-ABL1 fusion (Philadelphia chromosome), the hallmark of chronic myeloid leukemia. Burkitt lymphoma is associated with t(8;14); APL with t(15;17); Down syndrome is trisomy 21.

QUESTION 5 SINGLE CHOICE

Which of the following is the principal purpose of including a no-template control (NTC) in every PCR run?

- ☐ A To quantify the amount of DNA in patient samples
- ☒ B To verify that reagents are not contaminated with template DNA
- ☐ C To calibrate the thermal cycler block temperature
- ☐ D To assess primer annealing efficiency

ANSWER B

WHY The NTC contains all PCR reagents except the template. Amplification in the NTC indicates reagent contamination, which would invalidate the run. Quantification, instrument calibration, and primer efficiency are assessed by other means (standards, calibration, and optimization curves, respectively).

QUESTION 6 SINGLE CHOICE

In a real-time PCR assay using TaqMan chemistry, what is the role of the fluorescently labeled probe?

- ☐ A It acts as a primer for DNA synthesis
- ☐ B It binds the double-stranded PCR product and fluoresces upon intercalation
- ☒ C It emits fluorescence only when cleaved by the 5'→3' exonuclease activity of Taq polymerase during elongation
- ☐ D It labels the RNA template for reverse transcription

ANSWER C

WHY A TaqMan probe carries a 5' reporter fluorophore and a 3' quencher; during extension, the 5'→3' exonuclease activity of Taq polymerase cleaves the probe, separating reporter from quencher and producing fluorescence. It is not a primer, is not an intercalator (that describes SYBR Green), and reverse transcription does not depend on the probe.

QUESTION 7

SINGLE CHOICE

Next-generation sequencing (NGS) of a tumor sample reveals a point variant listed as 'NM_004333.4:c.1799T>A (p.Val600Glu)' in the BRAF gene. What is the most appropriate clinical interpretation?

- ☐ A Benign polymorphism with no clinical significance
- ☒ B The BRAF V600E mutation, commonly found in melanoma and other cancers, often actionable by BRAF inhibitors
- ☐ C A germline mutation diagnostic of familial adenomatous polyposis
- ☐ D A balanced translocation involving BRAF

ANSWER B

WHY BRAF p.Val600Glu (V600E) is a well-known activating mutation in melanoma, colorectal cancer, and others, and patients may benefit from BRAF inhibitor therapy (e.g., vemurafenib). It is oncogenic, not benign; it is not a familial polyposis variant (APC); point mutations are not balanced translocations.

QUESTION 8

SINGLE CHOICE

What is the standard number of metaphase chromosome spreads analyzed from at least two cultured cell types before declaring a normal karyotype according to standard cytogenetic reporting conventions?

- ☐ A 5 metaphase cells from one culture
- ☒ B 20 metaphase cells (15 from one culture, 5 from another)
- ☐ C 50 metaphase cells from a single culture
- ☐ D All metaphase cells observed

ANSWER B

WHY Per standard cytogenetic practice (ISCN-aligned reporting), a minimum of 20 metaphase cells are analyzed for a constitutional peripheral blood karyotype: 15 from one culture and 5 from a second independent culture, to rule out cell-culture artifacts. Five cells is insufficient, 50 is not required for a normal constitutional result, and analyzing every cell observed is impractical.

QUESTION 9

MULTIPLE CHOICE

A peripheral blood karyotype from an adult patient with chronic myelogenous leukemia (CML) demonstrates the Philadelphia chromosome. Which two cytogenetic findings classically define this rearrangement? (Choose two.)

- ☐ A Deletion of the long arm of chromosome 5
- ☐ B Reciprocal translocation between chromosomes 9 and 22
- ☒ C **t(9;22)(q34;q11)**
- ☐ D Isochromosome formation of the X chromosome
- ☒ E **Presence of the BCR-ABL1 fusion gene**

ANSWER C - E

WHY The Philadelphia chromosome is the product of a reciprocal translocation between chromosomes 9 and 22, written as t(9;22)(q34;q11), which juxtaposes the BCR gene on 22q11 with the ABL1 gene on 9q34 to create the BCR-ABL1 fusion. The other options describe unrelated cytogenetic findings not associated with the Philadelphia chromosome.