ABMGG Laboratory Genetics and Genomics
Blueprint and Content Codes

(Effective August 2023)

Exam Blueprint

ID Number	Exam Content Topic	Approximate Percentage
I	Normal structure and function	10%
III	Pathogenesis	20%
III	Application and interpretation	53%
IV	Laboratory management	7%
V	Nomenclature	10%

Content

I. Normal structure and function
   a) Chromosome structure
      i) Genome organization/chromatin packaging
      ii) Nucleolar organizing regions
      iii) Banding (heterochromatic/euchromatin/ideograms)
   b) Mitosis, meiosis, cell cycle
      i) Cell cycle
      ii) Mitosis
      iii) Meiosis I and II
         (1) Pachytene pairing
         (2) Disjunction/segregation
         (3) Spermatogenesis/oogenesis/fertilization
            (a) Molar pregnancies, teratomas
   c) Chromosome/gene structure and function
      i) RNA and DNA structures
      ii) Elements/features of genes and RNA
      iii) Transcription/translation/regulatory elements
      iv) RNA processing
      v) X-inactivation
      vi) Gene mapping/linkage

II. Pathogenesis
   a) Nondisjunction
      i) Meoisis I NDJ
      ii) Meoisis II NDJ
      iii) Mitotic NDJ
      iv) Ploidy changes
   b) Unequal crossing over
   c) Abnormal segregation (adj-1, adj-2, 3:1, 4:0)
   d) Mosaicism/ pseudomosaicism
   e) Structural aberrations (incl. translocations, inv, del, ins, dup, rings, mar, dmin)
      i) Recombination
      ii) Segregation
   f) Mitochondrial mutations/heteroplasmy
   g) Triplet expansion disorders
   h) Single Nucleotide Variants
   i) Imprinting/methylation
   j) Normal variants
   k) Insertions/deletions/duplications
   l) LOH/AOH/LCSH/UPD
   m) Microsatellite instability
   n) Monitoring minimal residual disease
   o) Other

i) Chromosome instability
ii) Chromothripsis
iii) Nonsense mediated decay

III. Applications and interpretation

a) Core techniques, principles and practice
i) Cell culture/culture harvests
ii) Slidemaking/G-banding
iii) FISH
iv) Microscopy (brightfield and fluorescence)
v) Copy number detection
(1) Chromosomal microarray
(a) aCGH
(b) Exon-targeted aCGH
(c) SNP arrays
(2) MLPA
vi) Nucleic acid isolation
vii) PCR
(1) Qualitative
(2) Quantitative
viii) Allele discrimination
ix) Sequencing
(1) Dideoxy sequencing
(2) NGS
(a) Somatic NGS
(b) Germline NGS
x) Methylation analysis
xi) Nucleic acid hybridization
xii) Targeted mutation analysis
xiii) Mutation scanning methods
xiv) SNP analysis (molecular)

b) Diagnosis of symptomatic individuals

c) Predictive testing
i) Hereditary disease (presymptomatic)
ii) Pharmacogenetic

d) Carrier testing

e) Screening
i) Population based
ii) Preconception/prenatal (including NIPT)

f) Prenatal diagnosis

g) Identity testing
i) Maternal cell contamination of fetal samples
ii) Paternity testing

h) Preanalytic variables

i) Analytic variables

- j) Postanalytic variables
  - i) Interpretation of results
  - ii) Genotype:phenotype correlation
  - iii) Use of genomic databases
  - iv) Calculation of residual, recurrent risks
  - v) Test limitations and recommendations
  - vi) Variant classification
  - vii) Variants of uncertain significance
  - viii) Surveillance/clinical follow up, additional testing
  - ix) Bioinformatics

IV. Laboratory management

- a) QA/QC
  - i) Professional and regulatory guidelines
  - ii) Proficiency testing
  - iii) Instrument validation and calibration
  - iv) Assay validation
  - v) Quality control/quality assurance
  - vi) Accuracy, precision, sensitivity, specificity
  - vii) Personnel competency assessment
- b) Ethical/legal
  - i) HIPAA, GINA
  - ii) Patient consent/assent
  - iii) Genetic counseling
- c) Lab safety
  - i) Chemical safety, fire safety
  - ii) Universal precautions, bloodborne pathogens

V. Nomenclature

- a) Constitutional
  - i) Numerical aberrations
    - (1) Aneusomies (autosomal and sex chromosomes)
    - (2) Ploidy changes
  - ii) Structural aberrations
  - iii) Mosaicism vs. chimerism
- b) Acquired (Somatic)
  - i) Mainline/sideline/stemline
  - ii) Use of "idem", "cp", "c"
  - iii) Chimerism vs. mosaicism
  - iv) Numerical aberrations (aneusomies, ploidy)
  - v) Structural aberrations (including chromothripsis)
  - vi) Mechanisms leading to gain of function/loss of function
- c) FISH
  - i) Metaphase vs. interphase
  - ii) Probe designs (dual fusion, breakapart, enumeration, extra signal)
- d) Chromosomal microarray

i) Aneuploidy/polyploidy
ii) Copy number changes
iii) LOH/AOH/ROH/LCSH/UPD
iv) Complex

e) Molecular nomenclature