ABMGG Molecular Genetics and Genomics CertLink
Blueprint and Content Codes

Blueprint

ID Number	Exam Content Topic	Approximate Percentage
I	Normal structure and function	5%
III	Pathogenesis	35%
III	Application and interpretation	35%
IV	Laboratory management	10%
V	Nomenclature	5%
VI	Practice Guidelines	5%
VII	Emerging Applications	5%

Content

I. Normal structure and function
   a) 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
II. Pathogenesis
   a) Mosaicism/pseudomosaicism
   b) Mitochondrial mutations/heteroplasmy
   c) Repeat expansion disorders
   d) Point mutations
   e) Imprinting/methylation
   f) Polymorphisms/normal variants
   g) Insertions/deletions/duplications
   h) LOH/AOH/LCSH/UPD
   i) Microsatellite instability
   j) Monitoring minimal residual disease
   k) Neoplasia
III. Applications and interpretation
   a) Core techniques, principles and practice
      i) Copy number detection
         (1) Chromosomal microarray
         (2) MLPA
      ii) Nucleic acid isolation
      iii) PCR
         (1) Qualitative
         (2) Quantitative
      iv) Allele discrimination
      v) Sequencing
         (1) Dideoxy sequencing
         (2) NGS
      vi) Methylation analysis
      vii) Nucleic acid hybridization
      viii) Targeted mutation analysis
      ix) 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) Molecular nomenclature

VI. Practice Guidelines

VII. Emerging Applications