# ABMGG Laboratory Genetics and Genomics
## Blueprint and Content Codes
*(Effective August 2023)*

### Exam Blueprint

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<th>Exam Content Topic</th>
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<td>Normal structure and function</td>
<td>10%</td>
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<td>III</td>
<td>Pathogenesis</td>
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<tr>
<td>III</td>
<td>Application and interpretation</td>
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</table>
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) Meiosis I NDJ
      ii) Meiosis 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