## Exam Blueprint

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<th>ID Number</th>
<th>Exam Topic Content</th>
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<tr>
<td>I</td>
<td>Normal structure and function</td>
<td>13%</td>
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<td>II</td>
<td>Pathogenesis</td>
<td>20%</td>
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<tr>
<td>III</td>
<td>Applications and interpretation</td>
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<td>IV</td>
<td>Laboratory management</td>
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<td>V</td>
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Laboratory Genetics and Genomics Content Codes

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

II. Pathogenesis
   A. Nondisjunction
      a. Meiosis I NDJ
      b. Meiosis II NDJ
      c. Mitotic NDJ
      d. 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)
      a. Recombination
      b. Segregation
   F. Mitochondrial mutations/heteroplasmy
   G. Triplet expansion disorders
   H. Point mutations
   I. Imprinting/methylation
   J. Polymorphisms/normal variants
   K. Insertions/deletions/duplications
   L. LOH/AOH/LCSH/UPD
   M. Microsatellite instability
   N. Monitoring minimal residual disease
   O. Other
      a. Chromosome instability
      b. Chromothripsis

III. Applications and interpretation
A. Core techniques, principles and practice
   a. Cell culture/culture harvests
   b. Slidemaking/G-banding
   c. FISH
   d. Microscopy (brightfield and fluorescence)
   e. Copy number detection
      1. Chromosomal microarray
         a. aCGH
         b. Exon-targeted aCGH
         c. SNP arrays
      2. MLPA
   f. Nucleic acid isolation
   g. PCR
      1. Qualitative
      2. Quantitative
   h. Allele discrimination
   i. Sequencing
      1. Dideoxy sequencing
      2. NGS
   j. Methylation analysis
   k. Nucleic acid hybridization
   l. Targeted mutation analysis
   m. Mutation scanning methods
   n. SNP analysis (molecular)
B. Diagnosis of symptomatic individuals
C. Predictive testing
   a. Hereditary disease (presymptomatic)
   b. Pharmacogenetic
D. Carrier testing
E. Screening
   a. Population based
F. Preconception/prenatal (including NIPT)
G. Prenatal diagnosis
   a. Identity testing
   b. Maternal cell contamination of fetal samples
H. Paternity testing
I. Preanalytic variables
J. Analytic variables
K. Postanalytic variables
   a. Interpretation of results
   b. Genotype:phenotype correlation
   c. Use of genomic databases
   d. Calculation of residual, recurrent risks
   e. Test limitations and recommendations
   f. Variant classification
   g. Variants of uncertain significance
   h. Surveillance/clinical follow up, additional testing
   i. Bioinformatics
IV. Laboratory management
   A. QA/QC
      a. Professional and regulatory guidelines
      b. Proficiency testing
      c. Instrument validation and calibration
      d. Assay validation
      e. Quality control/quality assurance
      f. Accuracy, precision, sensitivity, specificity
      g. Personnel competency assessment
   B. Ethical/legal
      a. HIPAA, GINA
      b. Patient consent/assent
      c. Genetic counseling
   C. Lab safety
      a. Chemical safety, fire safety
      b. Universal precautions, bloodborne pathogens

V. Nomenclature
   A. Constitutional
      a. Numerical aberrations
      b. Aneusomies (autosomal and sex chromosomes)
      c. Ploidy changes
      d. Structural aberrations
      e. Mosaicism vs. chimerism
   B. Acquired (Somatic)
      a. Mainline/sideline/stemline
      b. Use of "idem", "cp", "c"
      c. Chimerism vs. mosaicism
      d. Numerical aberrations (aneusomies, ploidy)
      e. Structural aberrations (including chromothripsis)
      f. Mechanisms leading to gain of function/loss of function
   C. FISH
      a. Metaphase vs. interphase
      b. Probe designs (dual fusion, breakapart, enumeration, extra signal)
   D. Chromosomal microarray
      a. Aneuploidy/polyploidy
      b. Copy number changes
      c. LOH/AOH/ROH/LCSH/UPD
      d. Complex
   E. Molecular nomenclature