ABMGG Cytogenetics and Genomics
Blueprint and Content Codes
(Effective August 2019)

Exam Blueprint

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Cytogenetics and Genomics Content Codes

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

II. Pathogenesis
   A. Nondisjunction
      1) Meiosis I NDJ
      2) Meiosis II NDJ
      3) Mitotic NDJ
      4) Ploidy changes
   B. Unequal crossing over
   C. Abnormal segregation (adj-1, adj-2, 3:1, 4:0)
   D. Mosaicism/pseudomosaicism
   E. Structural aberrations (including translocations, inv, del, ins, dup, rings, mar dmin)
      1) Recombination
      2) Segregation
   F. Imprinting/methylation
   G. Polymorphisms/normal variants
   H. LOH/AOH/LCSH/UPD
   I. Other
      1) Chromosome instability
      2) Chromothripsis
   J. Neoplasia
   K. Sex chromosome abnormalities

III. Applications and Interpretation
   A. Core techniques, principles, and practice
      1) Cell culture/culture harvests
      2) Slidemaking/G-banding
      3) FISH
      4) Microscopy (brightfield and fluorescence)
5) Copy number detection
   a) Chromosomal microarray
   b) aCGH
   c) Exon-targeted aCGH
   d) SNP arrays
6) Nucleic acid isolation
7) Nucleic acid hybridization

B. Diagnosis of symptomatic individuals
C. Carrier testing
D. Screening
   1) Preconception/prenatal (including NIPT)
E. Prenatal diagnosis
F. Identity testing
   1) Maternal cell contamination of fetal samples
G. Preanalytic variables
H. Analytic variables
I. Postanalytic variables
   1) Interpretation of results
   2) Genotype:phenotype correlation
   3) Use of genomic databases
   4) Calculation of residual, recurrent risks
   5) Test limitations and recommendations
   6) Variant classification
   7) Variants of uncertain significance
   8) Surveillance/clinical follow up, additional testing
   9) Bioinformatics

IV. Laboratory Management
A. QA/QC
   1) Professional and regulatory guidelines
   2) Proficiency testing
   3) Instrument validation and calibration
   4) Assay validation
   5) Quality control/quality assurance
   6) Accuracy, precision, sensitivity, specificity
   7) Personnel competency assessment
B. Ethical/legal
   1) HIPAA, GINA
   2) Patient consent/assent
   3) Genetic counseling
C. Laboratory safety
   1) Chemical safety, fire safety
   2) Universal precautions, bloodborne pathogens

V. Nomenclature
A. Constitutional
   1) Numerical aberrations
      a) Aneusomies (autosomal and sex chromosomes)
      b) Ploidy changes
   2) Structural aberrations
   3) Mosaicism vs chimerism
B. Acquired (Somatic)
   1) Mainline/sideline/stemline
2) Use of “idem,” “cp,” “c”
3) Chimerism vs mosaicism
4) Numerical aberrations (aneusomies, ploidy)
5) Structural aberrations (including chromothripsis)
6) Mechanisms leading to gain of function/loss of function

C. FISH
   1) Metaphase vs interphase
   2) Probe designs (dual fusion, breakapart, enumeration, extra signal)

D. Chromosomal microarray
   1) Aneuploidy/polyploidy
   2) Copy number changes
   3) LOH/AOH/ROH/LCSH/UPD
   4) Complex