ABMGG Ctyogenetics and Genomics CertLink
Blueprint and Content Codes

Blueprint

ID Number	Exam Content Topic	Approximate Percentage
I	Normal structure and function	5%
III	Pathogenesis	25%
III	Application and interpretation	30%
IV	Laboratory management	20%
V	Nomenclature	10%
VI	Practice Guidelines	5%
VII	Emerging Applications	5%

Content

I. Normal structure and function
   a) Chromosome structure
      i) Banding (heterochromatic/euchromatin/ideograms)
   b) Mitosis, meiosis, cell cycle
      i) Mitosis
      ii) Meiosis I and II
         (1) Disjunction/segregation
         (2) Spermatogenesis/oogenesis/fertilization
            (a) Molar pregnancies, teratomas
   c) Chromosome/gene structure and function
      i) X-inactivation

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
   f) Imprinting/methylation
   g) Polymorphisms/normal variants
   h) LOH/AOH/LCSH/UPD
   i) Monitoring minimal residual disease
   j) Neoplasia
      i) Hematological conditions
         (1) Myeloid
         (2) Lymphoid
      ii) Solid tumors
         (1) Pediatric
         (2) Adult
   k) Sex chromosome abnormalities
   l) Other
      i) Chromosome instability
      ii) Chromothripsis

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) SNP arrays
vi) Nucleic acid isolation
b) Diagnosis of symptomatic individuals
c) Carrier testing
d) Screening
i) Preconception/prenatal (prenatal cfDNA)
e) Prenatal diagnosis
f) Identity testing
i) Maternal cell contamination of fetal samples
g) Preanalytic variables
h) Analytic variables
i) 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

VI. Practice Guidelines

VII. Emerging Applications