ABMGG Clinical Genetics and Genomics Blueprint and Content Codes

(Effective August 2025)

Exam Blueprint

ID Number	Exam Content Topic	Approximate Percentage
I	Multiple malformations/Dysmorphology	16%
II	Intellectual Disability/Autism	10%
III	Connective Tissue Disorders	1%
IV	Skeletal Disorders/Dysplasias	1%
V	Hematologic Disorders	1%
VI	Cardiovascular Disorders	4%
VII	Hearing Loss Disorders	1%
VIII	Ophthalmologic disorders	3%
IX	Neurogenetic/Neuromuscular Disorders	3%
X	Neurodegenerative Disorders	1%
XI	Renal Disorders	1%
XII	Immunologic/Immunodeficiency Disorders	1%
XIII	Pulmonary Disorders	1%
XIV	Cutaneous Disorders	1%
XV	GI Disorders	1%
XVI	Pharmacogenomics	2%
XVII	Complex Disorders (Common Adult Disorders)	1%
XVIII	Cancer	9%
XIX	Diagnostic techniques (utility and limitations)	11%
XX	Reproductive Genetics	7%
XXI	Inborn Errors	10%
XXII	Ethical/Legal Principles	1%
XXIII	Population Genetics	1%
XXIV	Molecular mechanisms/genomics	8%

Content

I. Multiple malformations/Dysmorphology
   a) Chromosomal Disorders
      i) Aneuploidies
      ii) Microdeletion/microduplication syndromes/CNV
      iii) UPD disorders
      iv) Other
   b) Common dysmorphic syndromes
      i) Craniofacial
      ii) Short stature
      iii) Overgrowth
      iv) Other
   c) Congenital abnormalities
      i) Syndromic
      ii) Nonsyndromic
      iii) Multifactorial
      iv) Teratogenic
      v) Deformation/disruption
      vi) Other
   d) Disorders of sexual differentiation
II. Intellectual Disability/Autism
   a) Syndromic
   b) Nonsyndromic
   c) X-linked
      i) Fragile X
      ii) Other X-linked disorders
      iii) X-linked dominant
      iv) Skewed X-inactivation
   d) Other
III. Connective Tissue Disorders
IV. Skeletal Disorders/Dysplasias
   a) Lethal
   b) Non-lethal
   c) Other
V. Hematologic Disorders
   a) Hemoglobinopathies
   b) Thrombophilias
   c) Hemophilias
   d) Other
VI. Cardiovascular Disorders
   a) Cardiomyopathies
   b) Congenital
   c) Arrhythmias

d) Aortopathies
e) Other

VII. Hearing Loss Disorders
a) Syndromic
b) Nonsyndromic
c) Environmental
d) Other

VIII. Ophthalmologic disorders
a) Malformations
b) Retinitis pigmentosa
i) Syndromic
ii) Nonsyndromic
c) Optic atrophy disorders
d) Macular disorders
e) Other

IX. Neurogenetic/Neuromuscular Disorders
a) Brain malformations including microcephaly
b) Inherited neuropathies
c) Muscular dystrophies/myopathies
d) Leukodystrophies
e) Neurocutaneous/phakomatoses
f) Movement disorders (dystoniasm, etc.)
g) Epilepsy
h) Trinucleotide repeat disorders
i) Dementias
j) Other

X. Neurodegenerative Disorders
a) Pediatric onset
b) Adult onset
c) Other

XI. Renal Disorders
a) Syndromic
b) Nonsyndromic (including malformations)
c) Other

XII. Immunologic/Immunodeficiency Disorders
a) Primary
b) Syndromic
c) Other

XIII. Pulmonary Disorders
a) Cystic Fibrosis
b) Other

XIV. Cutaneous Disorders

XV. GI Disorders

XVI. Pharmacogenomics

XVII. Complex Disorders (Common Adult Disorders)
- a) Lipid disorders
- b) Multiple sclerosis
- c) Psychiatric disorders
- d) Other

XVIII. Cancer
- a) Adult Cancer
  - i) Syndromic
  - ii) Nonsyndromic
  - iii) Other
- b) Pediatric Cancer
  - i) Syndromic
  - ii) Nonsyndromic
  - iii) Other

XIX. Diagnostic techniques (utility and limitations)
- a) Arrays – Oligo vs. SNP
- b) Karyotype
- c) FISH/MLPA
- d) Single gene
- e) Gene panels
- f) Variant interpretation
  - i) Genomics/exome sequencing
- g) Biochemical testing
  - i) Amino acids
  - ii) Organic acids
  - iii) Acylcarnitine
- h) Urine GAGS, oligosaccharides
- i) Peroxisomal screening
- j) Transferrin electric focusing/glycans
- k) Enzyme testing
- l) Other

XX. Reproductive Genetics
- a) Preconception
  - i) Genetic counseling
  - ii) Preconception Genetic Screening
  - iii) Pre-implantation Genetic Testing (PGT)
  - iv) Assisted Reproductive Techniques
- b) Pregnancy
  - i) Genetic Screening Aneuploidy
  - ii) Teratogen Counseling
- c) Genetic Carrier screening
  - i) Expanded carrier screening
  - ii) Focused carrier screening

d) Prenatal diagnosis
    i) Indications
    ii) Procedures
    iii) Diagnostic techniques
e) Other

XXI. Inborn Errors
a) Lysosomal Storage Disorder
b) Mitochondrial disorders
c) Aminoacidopathies
d) Disorders of carbohydrates
e) Organic acidurias
f) Disorders of glycosylation
g) Peroxisomal disorders
h) Newborn screening
i) Fatty acid oxidation disorders
j) Other

XXII. Ethical/Legal Principles
a) Direct to consumer testing

XXIII. Population Genetics

XXIV. Molecular mechanisms/genomics