

ABMGG Clinical Genetics and Genomics Blueprint and Content Codes

(Effective August 2023)

Exam Blueprint

ID Number	Exam Content Topic	Approximate Percentage
I	Multiple malformations/Dysmorphology	17%
II	Intellectual Disability/Autism	10%
III	Connective Tissue Disorders	1%
IV	Skeletal Disorders/Dysplasias	1%
V	Hematologic Disorders	1%
VI	Cardiovascular Disorders	3%
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	1%
XVII	Complex Disorders (Common Adult Disorders)	1%
XVIII	Cancer	8%
XIX	Diagnostic techniques (utility and limitations)	12%
XX	Reproductive Genetics	8%
XXI	Inborn Errors	11%
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 (dystonias, 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) Non-pregnant
 - i) Genetic counseling
 - ii) Preconception Genetic Screening
 - iii) Pre-implantation Genetics Diagnosis
 - iv) Assisted Reproductive Techniques
 - b) Pregnant
 - 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) Other
 - j) Fatty acid oxidation disorders
- XXII. Ethical/Legal Principles
 - a) Direct to consumer testing
- XXIII. Population Genetics
- XXIV. Molecular mechanisms/genomics