

ABMGG Clinical Genetics and Genomics CertLink Blueprint and Content Codes

Blueprint

ID Number	Exam Content Topic	Approximate Percentage
I	Multiple malformations/Dysmorphology	8%
II	Intellectual Disability/Autism	5%
III	Connective Tissue Disorders	3%
IV	Skeletal Disorders/Dysplasias	3%
V	Hematologic Disorders	3%
VI	Cardiovascular Disorders	3%
VII	Hearing Loss Disorders	3%
VIII	Ophthalmologic disorders	3%
IX	Neurogenetic/Neuromuscular Disorders	3%
X	Neurodegenerative Disorders	3%
XI	Renal Disorders	3%
XII	Immunologic/Immunodeficiency Disorders	3%
XIII	Pulmonary Disorders	3%
XIV	Cutaneous Disorders	3%
XV	GI Disorders	3%
XVI	Pharmacogenomics	2%
XVII	Complex Disorders (Common Adult Disorders)	2%
XVIII	Cancer	8%
XIX	Diagnostic techniques (utility and limitations)	8%
XX	Reproductive Genetics	8%
XXI	Inborn Errors	8%
XXII	Ethical/Legal Principles	2%
XXIII	Population Genetics	2%
XXIV	Molecular mechanisms/genomics	2%
XXV	Practice Guidelines	3%
XXVI	Emerging Applications	3%

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
- III. Connective Tissue Disorders
- IV. Skeletal Disorders/Dysplasias
 - a) Lethal
 - b) Non-lethal
- 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
- XI. Renal Disorders
 - a) Syndromic
 - b) Nonsyndromic (including malformations)
 - c) Other
- XII. Immunologic/Immunodeficiency Disorders
 - a) Primary
 - b) Syndromic
- XIII. Pulmonary Disorders
- XIV. Cutaneous Disorders
- XV. GI Disorders
- XVI. Pharmacogenomics
- XVII. Complex Disorders (Common Adult Disorders)
 - a) Lipid disorders
 - b) Psychiatric disorders
 - c) 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) 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 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

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
- XXV. Practice Guidelines
- XXVI. Emerging Applications