

ABMGG General Exam
Blueprint and Content Codes
(Effective August 2023)

Exam Blueprint

ID Number	Exam Content Topic	Approximate Percentage
I	Chromosomal basis of inheritance	6%
III	Chromosome and gene structure and function	6%
III	Population genetics	7%
IV	Cytogenetics	8%
V	Single gene inheritance	36%
VI	Gene environment interactions	6%
VII	Reproductive Genetics	6%
VIII	Cancer genetics	6%
IX	Population screening	5%
X	Genetic counseling	3%
XI	Genomics	13%

Content

- I. Chromosomal basis of inheritance
 - a) Cell cycle
 - b) Mitosis
 - c) Meiosis
 - d) Oogenesis
 - e) Spermatogenesis
 - f) Other
- II. Chromosome and gene structure and function
 - a) DNA structure
 - b) Gene structure and organization
 - c) Transcription and translation
 - d) Gene regulation
 - e) Chromosome structure
 - f) Epigenetics
 - g) Non-coding RNA
 - h) Other
- III. Population genetics
 - a) Genetic drift and genetic flow
 - b) Founder effect
 - c) Hardy-Weinberg equilibrium
 - d) Consanguinity, genetic isolates and ethnic predilection
 - e) Fitness and selection
 - f) Other
- IV. Cytogenetics
 - a) Cytogenetic techniques
 - i) Cell culture
 - ii) Microscopy/karyotyping
 - iii) FISH
 - iv) Chromosomal microarrays
 - v) Special Techniques (e.g. breakage, SCE)
 - vi) Other
 - b) Chromosome abnormalities
 - i) Aneuploidy
 - ii) Structural rearrangements
 - iii) Mosaicism/chimerism
 - iv) Deletions/Duplications
 - v) Other
- V. Single gene inheritance
 - a) General principles
 - i) Mutation classes
 - ii) Mutation effects
 - iii) Genetic heterogeneity

- iv) Variable expression
 - v) Penetrance
 - vi) Pleiotropy
 - vii) Lyonization
 - viii) Other
 - b) Patterns of mendelian inheritance
 - i) Autosomal dominant
 - ii) Autosomal recessive
 - iii) X-linked
 - (1) X-inactivation
 - (2) X-linked recessive inheritance
 - (3) X-linked dominant inheritance
 - iv) Other
 - c) Atypical inheritance
 - i) Mitochondrial inheritance
 - ii) Parent of origin effects on inheritance (genomic imprinting)
 - iii) Uniparental disomy
 - iv) Dynamic mutations: trinucleotide repeat expansions
 - v) Mosaicism
 - (1) Germline
 - (2) Somatic
 - vi) Other
 - d) Single gene disorders
 - i) Connective tissue disorders
 - ii) Skeletal dysplasias
 - iii) Cardiac disorders
 - iv) Hematologic disorders
 - v) Neurogenetic disorders
 - vi) Neurocutaneous disorders
 - vii) Ophthalmic genetic disorders
 - viii) Deafness
 - ix) Metabolic disease
 - (1) Amino acid disorders
 - (2) Organic acid disorders
 - (3) Fatty acid oxidation disorders
 - (4) Sugar (or carbohydrate) metabolism disorders
 - (5) Lipid metabolism disorders
 - (6) Lysosomal storage disorders
 - (7) Mitochondrial disorders
 - (8) Peroxisomal disorders
 - x) Other
- VI. Gene environment interactions
- a) Multifactorial inheritance
 - i) Estimates of heritability

- ii) Risk threshold model
 - iii) Factors influencing degree of risk
 - b) Common complex disease
 - i) Gene associations
 - ii) HLA associations
 - iii) Pharmacogenetics
 - c) Teratology
 - i) Principles of teratology
 - ii) Common teratogens
 - d) Other
- VII. Reproductive Genetics
 - a) Carrier screening
 - i) Targeted (ethnic) screening
 - ii) Expanded carrier screening
 - b) Non-invasive prenatal screening
 - i) Maternal serum analyte screening
 - ii) Non-invasive aneuploidy screening by massively parallel sequencing
 - iii) Ultrasound
 - c) Invasive prenatal testing
 - i) Chorionic villus sampling
 - ii) Amniocentesis
 - d) Preimplantation genetic diagnosis
 - i) Aneuploidy screening
 - ii) Single gene disorder testing
 - e) Other
- VIII. Cancer genetics
 - a) Genetic basis of cancer
 - i) Oncogenes
 - ii) Tumor suppressor genes
 - b) Hereditary cancer predisposition
 - i) Autosomal dominant cancer predisposition syndromes
 - ii) Autosomal recessive cancer predisposition syndromes
 - c) Acquired abnormalities
 - i) Overgrowth syndromes
 - ii) Somatic genetic changes
 - d) Other
- IX. Population screening
 - a) Newborn screening
 - i) Blood spot screening
 - ii) Other screening (point of care, etc.)
 - b) Heterozygote screening
 - c) Other
- X. Genetic counseling
 - a) Psychosocial issues

- b) Legal issues
 - i) Confidentiality
- c) Ethical issues
 - i) Autonomy, beneficence, non-maleficence, justice
 - ii) Genetic testing of minors
 - iii) Human subjects protection
- d) Other

XI. Genomics

- a) General/targeted molecular testing
 - i) PCR and related techniques
 - ii) Gene sequencing - Sanger
 - iii) Gene sequencing - NGS panel testing
- b) Genomics
 - i) Whole exome sequencing
 - ii) Whole genome sequencing
 - iii) Chromosomal microarray
 - iv) Variant interpretation - constitutional
 - v) Variant interpretation - somatic
- c) Other
 - i) RNA-based analysis
 - ii) Genetic identity
 - iii) Methylation testing