ABMGG General Exam
Blueprint and Content Codes
(Effective August 2025)

Exam Blueprint

ID Number	Exam Content Topic	Approximate Percentage
I	Chromosomal basis of inheritance	5%
III	Chromosome and gene structure and function	6%
III	Population genetics	6%
IV	Cytogenetics	8%
V	Single gene inheritance	36%
VI	Gene environment interactions	6%
VII	Reproductive Genetics	6%
VIII	Cancer genetics	6%
IX	Population screening	4%
X	Genetic counseling	3%
XI	Genomics	14%

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) 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) Prenatal cell-free DNA screening
   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