ABMGG Biochemical Genetics Blueprint and Content Codes

(Effective August 2023)

Exam Blueprint

ID Number	Exam Content Topic	Approximate Percentage
I	Amino acids	16%
II	Organic acids	15%
III	Cofactors	6%
IV	Carbohydrates	10%
V	Lipids	10%
VI	Lysosomes	11%
VII	Mitochondria	7%
VIII	Peroxisomes	3%
IX	Purines and pyrimidines	3%
X	Transport	3%
XI	Creatine	1%
XII	Neurotransmitters	1%
XIII	Metals	2%
XIV	Laboratory	8%
XV	Other disease category	3%

Biochemical Genetics

I. Amino acids
   a) PKU and hyperphenylalaninemia
   b) Tyrosinemias
   c) Glycine encephalopathy/hyperglycinemia
   d) MSUD and disorders of branched-chain amino acids
   e) Homocystinuria/disorders of sulfur amino acids and re-methylation
   f) Urea cycle defects
   g) Other/Serine

II. Organic acids
   a) Methylmalonic acidemia (excluding cobalamin defects) and propionic acidemia
   b) 3-MCC deficiency and other C5OH-related disorders
      i) 3-MCC deficiency
      ii) other C5OH-related disorders
   c) IVA and other C5-related disorders
   d) Glutaric acidemia type I
   e) Disorders of ketone body metabolism (HMG-CoA lyase, SCOT)
   f) Other (Glutathione synthetase deficiency, ECHS1, 3-methylglutaconic aciduria non-type 1)

III. Cofactors
   a) Cobalamin
   b) Biotin
   c) Molybdenum
   d) Folate
   e) Pyridoxine
   f) Biopterin
   g) Other (riboflavin, niacin, thiamine)

IV. Carbohydrates
   a) Glycogenoses (except GSD II [see LSD])
   b) Disorders of glycolysis
   c) Disorders of gluconeogenesis (including fructose-1,6-bisphosphatase deficiency)
   d) Disorders of Galactose metabolism
   e) Disorders of fructose metabolism
   f) Congenital disorders of glycosylation
   g) Other (ex: pentose phosphate)

V. Lipids
   a) Fatty acid oxidation disorders
      i) Short-chain
      ii) Medium-chain
      iii) Long-chain
      iv) Multiple acyl-CoA dehydrogenase deficiency (GA2)
   b) Hyperlipidemias
      i) Tangier disease
      ii) Other

c) Disorders of cholesterol metabolism
    i) Smith-Lemli-Opitz
    ii) Niemann-Pick C
    iii) Other (Bile Acid)

VI. Lysosomes
a) Mucopolysaccharidoses
    i) Type I
    ii) Type II
    iii) Type III
    iv) Type IV
    v) Type VI
    vi) Type VII
b) Sphingolipidoses
    i) Gaucher disease
    ii) Fabry disease
    iii) GM2 gangliosidoses (including Tay-Sachs disease and variants, and Sandhoff disease)
    iv) Krabbe disease
    v) Neimann Pick types A and B
    vi) GM1 gangliosidosis
    vii) Other
c) Multiple enzyme deficiencies
    i) Mucolipidoses (ML-II and ML-III)
    ii) Multiple sulfatase deficiency
    iii) Other
d) Other (Pompe and Cystinosis, NCL)

VII. Mitochondria
a) Mitochondrial myopathies, laboratory findings
b) Mitochondrial myopathies, Leigh syndrome
c) Mitochondrial myopathies, adult presentations
d) Disorders of electron transport chain function and assembly
e) Mitochondrial DNA mutation disorders
f) Disorders of mtDNA maintenance
g) Mitochondrial DNA depletion syndromes
h) Other

VIII. Peroxisomes
a) Biogenesis disorders: Zellweger syndrome
b) Single-enzyme disorders: X-linked adrenoleukodystrophy
c) Other (Refsum - infantile)

IX. Purines and pyrimidines
a) Purine disorders
    i) Lesch-Nyhan disease
    ii) Adenosine deaminase deficiency
    iii) Other
b) Pyrimidine disorders

i) UMP synthase deficiency
ii) Other

X. Transport
a) Dibasic aminoaciduria (includes cystinuria and lysinuric protein intolerance)
b) Other amino acid transport disorders
c) Glucose
d) Carnitine
e) Other

XI. Creatine

XII. Neurotransmitters
a) Succinic semialdehyde dehydrogenase (SSADH) deficiency
b) Disorders of dopamine synthesis
c) Other (GABA metabolism)

XIII. Metals
a) Copper
i) Wilson disease
ii) Menkes disease
iii) Other
b) Iron
c) Zinc
d) Other

XIV. Laboratory
a) QA/QC
b) Artifact
c) Techniques
d) Regulations

XV. Other disease category