

ABMGG Biochemical Genetics
Blueprint and Content Codes
(Effective August 2025)

Exam Blueprint

ID Number	Exam Content Topic	Approximate Percentage
I	Amino acids	15%
II	Organic acids	12%
III	Cofactors	7%
IV	Carbohydrates	10%
V	Lipids	13%
VI	Lysosomes	12%
VII	Mitochondria	7%
VIII	Peroxisomes	3%
IX	Purines and pyrimidines	3%
X	Transport	3%
XI	Creatine	1%
XII	Neurotransmitters	2%
XIII	Metals	3%
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 (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) Mucopolysaccharidoses (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 (MELAS)

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