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美国医生执照考试（一）—生化和遗传学 Biochemistry & Genetics
PRETEST USMLE STEP 1

Biochemistry and Genetics

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Biochemistry and Genetics

PreTest® Self-Assessment and Review

Notice

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Preface

This new edition of *Biochemistry and Genetics PreTest®: Self-Assessment and Review* is based in part on the earlier biochemistry editions prepared by Francis J. Chlapowski, Ph.D., Department of Biochemistry and Molecular Biology, University of Massachusetts Medical School. All questions are now in single-best-answer format and a large number are analogous to those of the United States Medical Licensing Examination (USMLE), Part I. Questions are updated to the most current editions of leading textbooks in medical biochemistry and medical genetics.

Introduction

Each *PreTest® Self-Assessment and Review* allows medical students to comprehensively and conveniently assess and review their knowledge of a particular basic science, in this instance biochemistry. The 500 questions parallel the format and degree of difficulty of the questions found in the United States Medical Licensing Examination (USMLE), Step 1. Appendix 1 lists the major subject areas of the biochemistry, genetics, and nutrition portions of the USMLE Step 1 content outline together with questions in this book that cover those areas. Practicing physicians who want to hone their skills before USMLE Step 3 or recertification may find this to be a good beginning in their review process.

Each question is accompanied by an answer, a paragraph explanation, and a specific page reference to an appropriate textbook. Over 20 reference figures have been added to help with review, and there are an additional 40 figures geared to specific questions. A bibliography listing sources can be found following the second appendix of this text, and a list of abbreviations used in the text follows this introduction. As listed in Appendix 2, over 100 clinical disorders or processes are discussed and related to biochemical and/or genetic mechanisms. For genetic disorders, a McKusick number is included that allows the reader to immediately access information about the disorder using the Online Mendelian Inheritance in Man Internet site (see the bibliography).

An effective way to use this PreTest® is to allow yourself one minute to answer each question in a given chapter. As you proceed, indicate your answer beside each question. By following this suggestion, you approximate the time limits imposed by the USMLE Step 1 examination. After you finish going through the questions in the section, spend as much time as you need verifying your answers and carefully reading the explanations provided. Pay special attention to the explanations for the questions you answered incorrectly—but read every explanation. The authors of this material have designed the explanations to reinforce and supplement the information tested by the questions. If you feel you need further information about the material covered, consult and study the text or online references indicated.

The High-Yield Facts in this book are provided to facilitate rapid review of biochemistry and genetics. It is anticipated that the reader will use the High-Yield Facts as a "memory jog" before proceeding through the questions.

Abbreviations

ACAT	acyl CoA–cholesterol acyl transferase
ACTH	adrenocorticotropic hormone
ADP	adenosine diphosphate
AMP	adenosine monophosphate
ATP	adenosine triphosphate
ATPase	adenosine triphosphatase
CAP	catabolite activator protein
CDP	cytidine diphosphate
CMP	cytidine monophosphate (cytidylic acid)
CoA	coenzyme A
cyclic AMP	adenosine 3′,5′-cyclic monophosphate (3′,5′-cyclic adenylic acid)
DHAP	dihydroxyacetone phosphate
DNA	deoxyribonucleic acid
DNP	2,4-dinitrophenol
DPG	diphosphoglycerate
dTMP	deoxythymidine monophosphate
dUMP	deoxyuridine monophosphate
EF	elongation factor
FAD (FADH)	flavin adenine dinucleotide (reduced form)
FMN	flavin mononucleotide
FSH	follicle-stimulating hormone
GDP	guanosine diphosphate
GMP	guanosine 5′-monophosphate (guanylic acid)
GTP	guanosine triphosphate
hCG	human chorionic gonadotropin
HDL	high-density lipoprotein
HGPRT	hypoxanthine-guanine phosphoribosyltransferase
HMG CoA	3-hydroxy-3-methylglutaryl coenzyme A
hnRNA	heterogeneous RNA of the nucleus
IDL	intermediate-density lipoprotein
IMP	inosine 5′-monophosphate (inosinic acid)
IP_3	inositol 1,4,5-triphosphate
LDH	lactate dehydrogenase
LDL	low-density lipoprotein
LH	luteinizing hormone

mRNA	messenger RNA
MSH	melanocyte-stimulating hormone
NAD (NADH)	nicotinamide adenine dinucleotide (reduced form)
NADP (NADPH)	nicotinamide adenine dinucleotide phosphate (reduced form)
PGH	pituitary growth hormone
P_i	inorganic orthophosphate
PP_i	inorganic pyrophosphate
PRPP	5-phosphoribosylpyrophosphate
RNA	ribonucleic acid
RQ	respiratory quotient
rRNA	ribosomal RNA
TMP	thymidine monophosphate
TPP	thymidine pyrophosphate
tRNA	transfer RNA
TSH	thyroid-stimulating hormone
TTP	thymidine triphosphate
UDP	uridine diphosphate
UMP	uridine monophosphate
UTP	uridine triphosphate
VLDL	very-low-density lipoprotein

Contents

High-Yield Facts in Biochemistry and Genetics

HORMONAL CONTROL OF METABOLISM

Metabolism is precisely regulated by hormones controlling the level of blood fuels and their delivery to tissues. The primary control hormones of metabolism are insulin and glucagon. Epinephrine has effects similar to those of glucagon, except that glucagon has a greater effect on the liver while epinephrine has a greater effect on muscle. Blood levels of glucose, amino acids, fatty acids, and ketone bodies are maintained by variations in the [insulin]/[glucagon] ratio. When blood sugar is high, the ratio increases and insulin signals the fed state, promoting anabolic activities. The ratio decreases as glucagon is released to direct catabolic activities when blood glucose falls between meals, during fasting, and during starvation. Epinephrine or norepinephrine is released during exercise to promote catabolism of glucose and fat that supports muscular activity. Under normal conditions, the very precise interplay between insulin and glucagon maintains homeostatic blood fuel levels at about: glucose, 4.5 m*M*; fatty acids, 0.5 m*M*; amino acids, 4.5 m*M*; ketone bodies, 0.02 m*M*. Blood levels of ketone bodies and fatty acids rise during fasting or during starvation, with blood glucose levels being maintained. However, during uncontrolled juvenile diabetes, blood glucose levels rise greatly. The lack of insulin in this disease otherwise mimics starvation. The activity of various pathways during different metabolic states is summarized in the following table.

ACTIVITY OF METABOLIC PATHWAYS

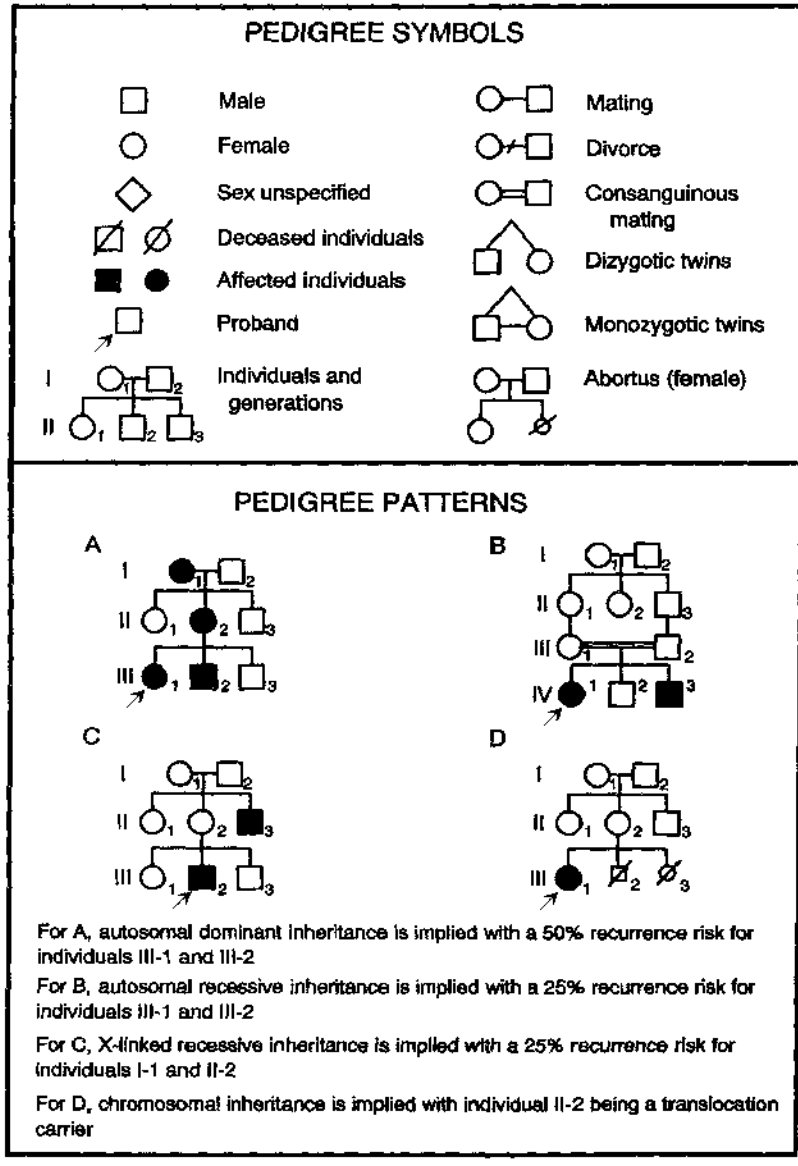
Pathway	Fed	Fasted	Diabetes
Glycogen synthesis	+	–	–
Glycolysis (liver)	+	–	–
Triacylglyceride synthesis	+	–	–
Fatty acid synthesis	+	–	–
Protein synthesis	+	–	–
Cholesterol synthesis	+	–	–
Glycogenolysis	–	+	+
Gluconeogenesis (liver)	–	+	+
Lipolysis	–	+	+
Fatty acid oxidation	–	+	+
Protein breakdown	–	+/–	+/–
Ketogenesis (liver)	–	+	+
Ketone body utilization (non-hepatic tissues)	–	+	+

KEY FACTS ABOUT INHERITANCE

- Human gametes have 23 chromosomes (haploid chromosome number $n = 23$), while most somatic cells have 46 chromosomes (diploid chromosome number $2n = 46$).
- Genes occupy sites on chromosomes (loci) and occur in alternative forms (alleles).
- Mendelian diseases exhibit autosomal dominant, autosomal recessive, or X-linked inheritance, while multifactorial diseases (e.g., cleft palate, diabetes mellitus, schizophrenia, hypertension) are determined by multiple genes plus the environment.
- Characteristics of autosomal dominant diseases include a vertical pedigree pattern, affliction of both males and females, variable expressivity (variable severity among affected individuals), frequent new mutations, and a 50% recurrence risk for offspring of affected individuals (see pedigree A on chart). *Corollary*: germ-line mosaicism may produce affected siblings with autosomal dominant disease when neither parent is affected.
- Characteristics of autosomal recessive diseases include a horizontal pedigree pattern, affliction of males and females, frequent consanguinity

(inbreeding), frequent carriers (heterozygotes without manifestations of disease), and a 25% recurrence risk for carrier parents (see pedigree B on chart). *Corollary*: normal siblings of individuals with autosomal recessive disease have a 2/3 chance of being carriers.

- Characteristics of X-linked recessive diseases include an oblique pedigree pattern, affliction of males only, frequent female carriers, and a 25% recurrence risk for carrier females (see pedigree C on chart). *Corollary*: Haldane's law predicts a 2/3 chance that the mother of an affected male with X-linked recessive disease is a carrier (and a 1/3 chance the affected male represents a new mutation).
- Ethnic correlations with Mendelian disorders include higher frequencies of cystic fibrosis in whites, sickle cell anemia in blacks, β-thalassemia in Italians and Greeks, α-thalassemia in Asians, and Tay-Sachs disease in Jews.
- Advanced maternal age is associated with higher risks for chromosomal disorders (e.g., Down's syndrome, trisomy 13), while advanced paternal age is associated with higher risks for new mutations (e.g., those producing achondroplasia or Marfan's syndrome).
- The Hardy-Weinberg law predicts allele frequencies in an idealized population according to the formula $p^2 + 2pq + q^2 = 1$. Applied to cystic fibrosis, the law predicts that homozygotes (q^2) have a frequency of 1 in 1600, predicting that carriers ($2pq$) have a frequency of 1 in 20.
- A karyotype is an ordered arrangement of chromosomes that is described by cytogenetic notation. A karyotype can be obtained from dividing cells (blood leukocytes, bone marrow, fibroblasts, amniocytes), but not from frozen or formalin-fixed cells.
- Cytogenetic notation includes the chromosome number (usually 46), description of the sex chromosomes (usually XX or XY), and indication of missing, extra, or rearranged chromosomes. Examples include 47,XY,+21 (male with Down's syndrome); 47,XX,+13 (female with trisomy 13); 45,X (female with monosomy X or Turner's syndrome), 46,XX,del(5p) (female with deletion of the chromosome 5 short arm)
- DNA diagnosis examines specific regions of genes for altered nucleotide sequences or deletions that affect gene expression and function; techniques include Southern blotting, gene amplification with the polymerase chain reaction (PCR), and mutant allele detection by



Pedigree symbols and pedigree patterns.

hybridization with allele-specific oligonucleotides (ASOs). Chromosome microdeletions encompass several genes and are detected by fluorescent in situ hybridization (FISH).

- Non-Mendelian inheritance mechanisms include mitochondrial inheritance (exhibiting maternal transmission), expansion of triplet repeats (exhibiting anticipation in pedigrees as in the fragile X syndrome), and genomic imprinting (exhibiting different phenotypes according to maternal or paternal origin of the aberrant genes).
- Prenatal diagnosis can include fetal ultrasound, maternal serum studies, or sampling of cells from the fetoplacental unit by chorionic villus sampling [CVS at 8 to 10 weeks, amniocentesis at 12 to 18 weeks, or percutaneous umbilical sampling (PUBS) from 16 weeks to term].