

# Contents

<i>Preface to the Third Edition</i>	<i>vii</i>
<i>Preface to the First Edition</i>	<i>ix</i>

## Section I: Carbohydrate Metabolism

CASE 1: Classical Galactosemia	1
CASE 2: Glucose-6-Phosphate Dehydrogenase Deficiency	4
CASE 3: Hereditary Fructose Intolerance	8
CASE 4: Lactose Intolerance	11
CASE 5: Sucrose Intolerance	15
CASE 6: Diabetes Mellitus	18
CASE 7: Diabetic Ketoacidosis	22
CASE 8: Correct Vacutainer for Blood Collection for Glucose Estimation	25
CASE 9: von Gierke Disease (Glycogen Storage Disorder Type Ia)	28
CASE 10: Cori Disease/Forbes Disease/Limit Dextrinosis (Glycogen Storage Disorder Type III)	31
CASE 11: McArdle Disease (Glycogen Storage Disorder Type V)	34
CASE 12: Pompe Disease (Glycogen Storage Disorder Type II)	38
CASE 13: Hurler Syndrome (Mucopolysaccharidosis Type I)	40
CASE 14: Hunter Syndrome (Mucopolysaccharidosis Type II)	44

## Section II: Lipid Metabolism

CASE 15: Zellweger Syndrome	47
CASE 16: Refsum Disease	50
CASE 17: Respiratory Distress Syndrome (RDS)	52
CASE 18: Carnitine Deficiency in Premature Baby	54
CASE 19: Type IIa Hyperlipoproteinemia (Familial Hypercholesterolemia)	56
CASE 20: Acute Pancreatitis in Hypertriglyceridemia	59
CASE 21: Type III Hyperlipoproteinemia (Remnant Removal Disease or Broad Beta Disease)	61
CASE 22: Xanthelasma	65
CASE 23: Tay-Sachs Disease	67
CASE 24: Niemann-Pick Disease	69
CASE 25: Jamaican Vomiting Sickness	71
CASE 26: Sudden Infant Death Syndrome (SIDS)	73

### Section III: Amino Acid Metabolism

CASE 27: Urea Cycle Disorder	75
CASE 28: Phenylketonuria (PKU)	78
CASE 29: Albinism	81
CASE 30: Vitiligo/Leukoderma	83
CASE 31: Pheochromocytoma	85
CASE 32: Parkinson's Disease	88
CASE 33: Tyrosinemia Type I (Tyrosinosis)	91
CASE 34: Tyrosinemia Type II (Richner-Hanhart Syndrome/ Oculocutaneous Involvement)	93
CASE 35: Alkaptonuria	96
CASE 36: Hartnup's Disease	98
CASE 37: B <sub>6</sub> Pellagra	101
CASE 38: Carcinoid Syndrome	104
CASE 39: Classical or Typical Homocystinuria	107
CASE 40: Maple Syrup Urine Disease (MSUD)	110
CASE 41: Isovaleric Acidemia	113

### Section IV: Enzymes

CASE 42: Myocardial Infarction and Enzyme Marker	115
CASE 43: Acute Pancreatitis	118
CASE 44: Ethanol used as Therapeutic Agent in Methanol Poisoning	121

### Section V: Vitamins

CASE 45: Scurvy in Adult	123
CASE 46: Scurvy in Child	126
CASE 47: Vitamin B <sub>1</sub> Deficiency (Wernicke-Korsakoff Psychosis)	128
CASE 48: Wet Beriberi (Shoshin Beriberi)	131
CASE 49: Ariboflavinosis (Vitamin B <sub>2</sub> Deficiency)	133
CASE 50: Pellagra (Niacin Deficiency)	135
CASE 51: B <sub>6</sub> Deficiency in Isoniazid (INH) Treatment	138
CASE 52: Biotin Deficiency in Raw Egg Consumption	140
CASE 53: Vitamin B <sub>12</sub> Deficiency in Vegans	142
CASE 54: Vitamin B <sub>12</sub> Deficiency Presenting as Neurological Manifestation	144
CASE 55: Vitamin B <sub>12</sub> Deficiency Presenting as Knuckle Hyperpigmentation	146
CASE 56: Folate Trap	148

CASE 57: Rickets	150
CASE 58: Osteomalacia Case 1	154
CASE 59: Osteomalacia Case 2	156
CASE 60: Vitamin D Toxicity	160
CASE 61: Vitamin A Deficiency	162
CASE 62: Vitamin A Toxicity	164
CASE 63: Vitamin K Deficiency in Newborn	166

## Section VI: Mineral Metabolism

CASE 64: Iron Deficiency Anemia	169
CASE 65: Hemosiderosis, Bronze Diabetes, Bantu Siderosis	171
CASE 66: Hypocalcemic Tetany	174
CASE 67: Hypocalcemia in a Case of Respiratory Alkalosis	177
CASE 68: Dental Fluorosis	179
CASE 69: Keshan Disease/Endemic Cardiomyopathy	181
CASE 70: Indian Childhood Cirrhosis	183
CASE 71: Wilson Disease	185
CASE 72: Menke Kinky Steely Hair Syndrome	187
CASE 73: Acrodermatitis Enteropathica	189
CASE 74: Iodine Deficiency Multinodular Goiter (Hypothyroidism)	191

## Section VII: Nutrition

CASE 75: Obesity	193
CASE 76: Marasmus	196
CASE 77: Kwashiorkor	197

## Section VIII: Heme Metabolism

CASE 78: Lead Toxicity: Plumboporphyria and Lead-induced Anemia	201
CASE 79: Porphyria Cutanea Tarda (PCT)	203
CASE 80: Acute Intermittent Porphyria (AIP)	206
CASE 81: Prehepatic Jaundice	208
CASE 82: Rh Incompatibility(Erythroblastosis Fetalis) Presenting as Kernicterus	210
CASE 83: Hepatic Jaundice	212
CASE 84: Posthepatic/Obstructive Jaundice	215
CASE 85: Pathological Jaundice in Newborn (Dubin-Johnson Syndrome)	218

## Section IX: Hemoglobinopathy

CASE 86: Sickle Cell Disease	221
CASE 87: Thalassemia Major	225

## Section X: Collagenopathy

CASE 88: Marfan Syndrome (MFS)	227
CASE 89: Osteogenesis Imperfecta (Brittle Bone Diseases)	231
CASE 90: Ehlers-Danlos Syndrome	233

## Section XI: Nucleotide Metabolism

CASE 91: Gout	237
CASE 92: Lesch-Nyhan Syndrome	241
CASE 93: Methotrexate and Antineoplastic Drug	243
CASE 94: 5-Fluorouracil and its Mechanism of Action	245

## Section XII: Miscellaneous

CASE 95: Amyl Nitrite as an Antidote of Cyanide Poisoning	247
CASE 96: Cholera Toxicity	250
CASE 97: Multiple Myeloma	252
CASE 98: Nephrotic Syndrome	257
CASE 99: Inclusion Cell Disease (I Cell Disease or Mucopolipidosis II)	260
CASE 100: Leber Hereditary Optic Neuropathy (LHON)/ Congenital Amaurosis	262
CASE 101: Cystic Fibrosis	265
CASE 102: Orotic Aciduria	268
CASE 103: Fatty Liver in an Alcoholic Patient	272
<i>Index</i>	275