

Contents

Part One: Approach to Diagnosis	1
A Simple Tests in Urine and Blood	3
NENAD BLAU, MILAN E. BLASKOVICS, MARINUS DURAN	
B Amino Acid Analysis	11
VIVIAN E. SHIH	
C Organic Acid Analysis	27
GEORG F. HOFFMANN, PATRIK FEYH	
D Miscellaneous Analyses	45
MARINUS DURAN	
E Tandem Mass Spectrometry in Clinical Diagnosis	57
DAVID S. MILLINGTON	
F Proton NMR Spectroscopy of Body Fluids	77
RON WEVERS, UDO ENGELKE, SYTSKE MOOLENAAR	
Part Two: Disorders	87
1 Disorders of Phenylalanine and Tetrahydrobiopterin Metabolism ..	89
NENAD BLAU, LUISA BONAFÉ, MILAN E. BLASKOVICS	
2 Disorders of Neurotransmitter Metabolism	107
KEITH HYLAND	
3 Disorders of GABA, Glycine, Serine and Proline	123
JAAK JAEKEN, TOM DE KONING, JOHAN VAN HOVE	
4 Disorders of Tyrosine Degradation	141
ELISABETH HOLME	

- 5** Disorders of Histidine Metabolism 155
YASUHIRO KURODA, MICHINORI ITO
- 6** Disorders of Leucine Metabolism 165
K. MICHAEL GIBSON, ORLY N. ELPELEG, D. HOLMES MORTON,
REBECCA S. WAPPNER
- 7** Disorders of Valine-Isoleucine Metabolism 191
WILLIAM L. NYHAN, K. MICHAEL GIBSON
- 8** Various Organic Acidurias 215
CORNELIS JAKOBS, NANDA M. VERHOEVEN,
MARJO S. VAN DER KNAAP
- 9** Disorders of the γ -Glutamyl Cycle 233
AGNE LARSSON, ELLINOR RISTOFF
- 10** Disorders of Sulfur Amino Acids 243
FLEMMING SKOVBY
- 11** Inherited Hyperammonemias 261
CLAUDE BACHMANN
- 12** Disorders of Ornithine, Lysine and Tryptophan 277
HILDEGARD PRZYREMBEL
- 13** Defective Transcellular Transport of Amino Acids 301
SUSANNE SCHWEITZER-KRANTZ
- 14** Disorders of Mitochondrial Fatty Acid Oxidation
and Ketone Body Handling 309
MARINUS DURAN
- 15** Disorders of Carbohydrate and Glycogen Metabolism 335
THOMAS F. ROE, WON G. NG, PETER G. A. SMIT
- 16** Disorders of Glucose Transport 357
RENÉ SANTER
- 17** Disorders of Glycerol Metabolism 369
KATRINA M. DIPPLE, EDWARD R. B. McCABE
- 18** The Mucopolysaccharidoses 377
JERRY N. THOMPSON

19	Oligosaccharidoses and Related Disorders	399
	GENERO SO ANDRIA, GIANCARLO PARENTI	
20	Congenital Disorders of Glycosylation	411
	JAAK JAEKEN, GERT MATTHIJS	
21	Cystinosis	423
	ERIK HARMS	
22	Other Storage Disorders	431
	MICHAEL BECK	
23	Purine and Pyrimidine Disorders	445
	H. ANNE SIMMONDS, ALBERT H. VAN GENNIP	
24	Disorders of Creatine Metabolism	467
	SYLVIA STÖCKLER-İPSIROGLU, CARMEN STROMBERGER, CHIKE B. ITEM, ADOLF MÜHL	
25	Peroxisomal Disorders	481
	RONALD J. A. WANDERS, PETER G. BARTH, BWEE TIEN POLL-THE	
26	Hyperoxaluria	509
	CHRISTIAN VON SCHNAKENBURG, KAY LATTA	
27	Mitochondrial Energy Metabolism	519
	JAN SMEITINK, BERT VAN DEN HEUVEL, FRANS TRIJBELS, WIM RUITENBEEK, ROB SENGERS	
28	Genetic Dyslipoproteinemias	537
	DANIEL J. RADER	
29	Disorders of Steroid Synthesis and Metabolism	551
	ANNA BIASON-LAUBER	
30	Inborn Errors of Cholesterol Biosynthesis	573
	LISA E. KRATZ, RICHARD I. KELLEY	
31	The Porphyrias	593
	ELISABETH I. MINDER, XIAOYE SCHNEIDER-YIN	
32	Disorders of Bile Acid Synthesis	615
	PETER T. CLAYTON, HUGH A. LEMONDE	

- 33** Disorders of Copper, Zinc and Iron Metabolism 631
KURT BAERLOCHER, MARC SOLIOZ

- 34** Leukotrienes 659
ERTAN MAYATEPEK

- 35** Other Metabolic Disorders 669
ANDREA SUPERTI-FURGA

Part Three: Indices 675

Disorders Index 677

Signs and Symptoms Index 689

Tests Index 703