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Nota di contenuto	PART 1: ORGANIC ACIDEMIAS. Introduction to the organic acidemias. Propionic academia. Methylmalonic academia. Cobalamin C, D, F, G diseases; methylmalonic aciduria and homocystinuria. The methylmalonic aciduria of deficiency of AcylCoA synthetase (ACSF3). Multiple carboxylase deficiency/holocarboxylase synthetase deficiency. Multiple carboxylase deficiency/biotinidase deficiency. Isovaleric academia. Glutaric aciduria (type I). 3-MethylcrotonylCoA carboxylase deficiency/3-methylcrotonylglycinuria. D-2-hydroxyglutaric aciduria. L-2-hydroxyglutaric aciduria. 4-Hydroxybutyric aciduria. PART 2: DISORDERS OF AMINO ACID METABOLOISM. Alkaptonuria. Phenylketonuria. Hyperphenylalaninemia and defective metabolism of tetrahydrobiopterin. Biogenic amines. Homocystinuria. Maple syrup urine disease (branched-chain oxoaciduria). Branched chain keto acid dehydrogenase kinase (BCKDK) deficiency. Oculocutaneous tyrosinemia/tyrosine aminotransferase deficiency. Hepatorenal tyrosinemia/fumarylacetoacetate hydrolase deficiency. Nonketotic hyperglycinemia. Serine deficiencies. PART 3: HYPERAMMONEMIA AND DISORDERS OF THE UREA CYCLE. Introduction to hyperammonemia and disorders of the urea cycle. Ornithine transcarbamylase deficiency. Carbamylphosphate synthetase deficiency. Citrullinemia type I. Argininosuccinic aciduria. Argininemia. Hyperornithinemia, hyperammonemia, homocitrullinuria syndrome. Lysinuric protein

intolerance. Glutamine synthetase deficiency. PART 4: DISORDERS OF FATTY ACID OXIDATION. Introduction to disorders of fatty acid oxidation. Carnitine transporter deficiency. Carnitine-ayclcarnitine translocase deficiency. Carnitine palmitoyl transferase I deficiency. Carnitine palmitoyl transferase II deficiency, lethal neonatal. Medium-chain acyl CoA dehydrogenase deficiency. Very long-chain acyl-CoA dehydrogenase (VLCAD) deficiency. Long chain L-3-hydroxyacl CoA dehydrogenase deficiency -- (trifunctional protein) deficiency. Short-chain acyl CoA dehydrogenase (SCAD) deficiency. Short-chain 3-hydroxyacylCoA dehydrogenase (SCHAD) deficiency. Short/branched-chain acyl-CoA dehydrogenase (2-methylbutyrylCoA dehydrogenase) deficiency. Multiple acyl CoA dehydrogenase deficiency/glutaric aciduria, type II ethylmalonic-adipic aciduria. 3-Hydroxy-3-methylglutarylCoA lyase deficiency. PART 5: THE LACTIC ACIDEMIAS AND MITOCHONDRIAL DISEASE. Introduction to the lactic acidemias. Pyruvate carboxylase deficiency. Fructose-1,6-diphosphatase deficiency. Deficiency of the pyruvate dehydrogenase complex. Mitochondrial encephalomyelopathy, lactic acidosis, and stroke-like episodes (MELAS). Myoclonic epilepsy and ragged red fiber (MERRF) disease. Neurodegeneration, ataxia and retinitis pigmentosa (NARP). Kearns-Sayre syndrome. Pearson syndrome. The mitochondrial DNA depletion syndromes: mitochondrial DNA polymerase deficiency. PART 6: DISORDERS OF CARBOHYDRATE METABOLISM. Galactosemia. Glycogen storage disease: introduction. Glycogenosis type I -- von Gierke disease. Glycogenosis type II/Pompe/lysosomal [alpha]-glucosidase deficiency. Glycogenosis type III/amylo-1,6-glucosidase (debrancher) deficiency. PART 7: PEROXISOMAL DISORDERS. Adrenoleukodystrophy. Neonatal adrenoleukodystrophy/disorders of peroxisomal biogenesis. PART 8: DISORDERS OF PURINE AND PYRIDINE METABOLISM. Introduction to the disorders of purine and pyrimidine deficiencies. Lesch-Nyhan disease and variants. Adenine phosphoribosyltransferase (APRT) deficiency. Phosphoribosylpyrophosphate synthetase and its abnormalities. Adenosine deaminase deficiency. Adenosine kinase deficiency. Purine nucleoside phosphorylase deficiency. Adenylosuccinate lyase deficiency. Santhinuria, xanthine oxidase deficiency. Orotic aciduria. Molybdenum cofactor deficiency. PART 9: MUCOPOLYSACCHARIDOSES. Introduction to mucopolysaccharidoses. Hurler disease/mucopolysaccharidosis type IH (MPSIH)/[alpha]-L-iduronidase deficiency. Scheie and Hurler-Scheie diseases/mucopolysaccharidosis IS and IHS/[alpha]-iduronidase deficiency. Hunter disease/mucopolysaccharidosis type II/iduronate sulfatase deficiency. Sanfilippo disease/mucopolysaccharidosis type III. Morquio syndrome/mucopolysaccharidosis type IV/keratan sulfaturia. Maroteaux-Lamy disease/mucopolysaccharidosis VI/N-acetylgalactosamine-4-sulfatase deficiency. Sly disease/[beta]-glucuronidase deficiency/mucopolysaccharidosis VII. PART 10: MUCOLIPIDOSIS. Mucolipidosis II and III/ (I-cell and pseudo-Hurler polydystrophy N-acetyl-glucosaminyl-l-phosphotransferase deficiency. PART 11: DISORDERS OF CHOLESTEROL AND NEUTRAL LIPID METABOLISM. Familial hypercholesterolemia. Mevalonic aciduria. Lipoprotein lipase deficiency/type I hyperlipoproteinemia. PART 12: LIPID STORAGE DISORDERS. Fabry disease. Tay-Sachs disease/hexosaminidase A deficiency. Sandhoff disease/GM2 gangliosidosis/deficiency of Hex A and Hex B subunit deficiency. Gaucher disease. Niemann-Pick disease. Niemann-Pick type C disease/cholesterol-processing abnormality. Krabbe disease/galactosylceramide lipidosis/globoid cell leukodystrophy.

Lysosomal acid lipase deficiency: Wolman disease/cholesteryl ester storage disease. Fucosidosis. [alpha]-Mannosidosis ([beta]-Mannosidosis). Galactosialidosis. Metachromatic leukodystrophy. Multiple sulfatase deficiency. PART 13: MISCELLANEOUS. Disorders of vitamin B6 metabolism. PMM2-CDG (Congenital disorders of glycosylation, type Ia). Ethylmalonic encephalopathy. Disorders of creatine metabolism. GLUT1 deficiency. Hypophosphatasia. NBAS/RALF deficiency. [alpha]1-Antitrypsin deficiency.

Sommario/riassunto

In a field where even experts may find that years have elapsed since they last encountered a child with a given disorder, it is essential for the clinician to have a comprehensive source of practical and highly illustrated information covering the whole spectrum of metabolic disease to refer to. The content is divided into sections of related disorders, including disorders of amino acid metabolism, lipid storage disorders, and mitochondrial diseases for ease of reference, with an introductory outline where appropriate summarizing the biochemical features and general management issues. Within the sections, each chapter deals with an individual disease, opening with a useful summary of major phenotypic expression including clear and helpful biochemical pathways, identifying for the reader exactly where the defect occurs. Throughout the book, plentiful photographs, often showing extremely rare disorders, are an invaluable aid to diagnosis. Key Features Fully updated to incorporate all new developments in the field Brand new chapters cover methylmalonic aciduria of ACSF3 deficiency, branched chain keto acid dehydrogenase deficiency, serine deficiencies, purine nucleoside phosphorylase deficiency, antiquitin deficiency, and others Excellent and detailed clinical descriptions, with numerous valuable hints and suggestions for management Helpful explanatory algorithms and decision trees, and high-quality illustrative material including biochemical pathways and an unrivaled photographic collection, which enhance clinical applicability The fourth edition of this highly regarded book, authored by two of the foremost authorities in pediatric metabolic medicine, continues to provide incomparable insight into the problems associated with metabolic diseases and remains invaluable to pediatricians, geneticists, and general clinicians worldwide.