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Nota di bibliografia	Includes bibliographical references at the end of each chapters and index.
Nota di contenuto	Coenzyme Q10 and Pyridoxal Phosphate Deficiency Is a Common Feature in Mucopolysaccharidosis Type III -- Pitfalls in Diagnosing Neuraminidase Deficiency: Psychosomatics and Normal Sialic Acid Excretion -- New Cases of DHTKD1 Mutations in Patients with 2-Ketoadipic Aciduria -- Screening Mucopolysaccharidosis Type IX in Patients with Juvenile Idiopathic Arthritis -- The Pathobiochemistry of Gastrointestinal Symptoms in a Patient with Niemann-Pick Type C Disease -- Improvement of Diffusion Tensor Imaging (DTI) Parameters with Decoppering Treatment in Wilson's Disease -- Vitamin E Improves Clinical Outcome of Patients Affected by Glycogen Storage Disease Type Ib -- Urine Beta2-Microglobulin Is an Early Marker of Renal Involvement in LPI -- The Spectrum of Krabbe Disease in Greece: Biochemical and Molecular Findings -- Exercise Intolerance and Myoglobinuria Associated with a Novel Maternally Inherited MT-ND1 Mutation --

	PNPO Deficiency and Cirrhosis: Expanding the Clinical Phenotype? -- Liver Fibrosis Associated with Iron Accumulation Due to Long-Term Heme-Arginate Treatment in Acute Intermittent Porphyria: A Case Series -- GM2-Gangliosidosis, AB Variant: Clinical, Ophthalmological, MRI, and Molecular Findings -- LC-MS/MS Analysis of Cerebrospinal Fluid Metabolites in the Pterin Biosynthetic Pathway -- Reduction of plasma globotriaosylsphingosine levels after switching from agalsidase alfa to agalsidase beta as enzyme replacement therapy for Fabry disease.
Sommario/riassunto	JIMD Reports publishes case and short research reports in the area of inherited metabolic disorders. Case reports highlight some unusual or previously unrecorded feature relevant to the disorder, or serve as an important reminder of clinical or biochemical features of a Mendelian disorder.