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	Uniparental Isodisomy of Chromosome 1 -- Severe Neonatal Manifestations of Infantile Liver Failure Syndrome Type 1 Caused by Cytosolic Leucine-tRNA Synthetase Deficiency -- Enzyme Replacement Therapy in Pregnant Women with Fabry Disease: A Case Series -- Survival of a Male Infant with a Familial Xp11.4 Deletion Causing Ornithine Transcarbamylase Deficiency -- The Unique Spectrum of Mutations in Patients with Hereditary Tyrosinemia Type 1 in Different Regions of the Russian Federation -- Elevated Lyso-Gb3 Suggests the R118C GLA Mutation Is a Pathological Fabry Variant -- Glycogen Storage Disease Type IV: A Rare Cause for Neuromuscular Disorders or Often Missed? -- A Hemizygous Deletion Within the PGK1 Gene in Males with PGK1 Deficiency.
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.