Oxoglutaric aciduria

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Oxoglutaric aciduria. ORPHA:31

A rare, genetic, inborn error of metabolism disorder characterized by neonatal-onset of developmental delay, hypotonia, hepatomegaly, lactic acidemia, increased creatine kinase levels, elevated alpha-ketoglutaric acid in urine, and a decreased plasma beta-hydroxybutyrate-to-acetoacetate ratio. Pyruvate dehydrogenase deficiency can be associated, leading to hypoglycemia and neurologic anomalies, including seizures.