Pyruvate carboxylase deficiency, infantile type

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Pyruvate carboxylase deficiency, infantile type. ORPHA:353308

Infantile pyruvate carboxylase (PC) deficiency (Type A) is a rare, severe form of PC deficiency characterized by infantile-onset, mild to moderate lactic acidemia, and a generally severe course.