Pyruvate carboxylase deficiency, severe neonatal type

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Pyruvate carboxylase deficiency, severe neonatal type. ORPHA:353314

Severe neonatal pyruvate carboxylase (PC) deficiency (Type B) is a rare, extremely severe form of PC deficiency characterized by severe, early-onset metabolic acidosis, and a generally fatal outcome in early infancy.