Open Peer Review on Qeios

Pyruvate carboxylase deficiency

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Pyruvate carboxylase deficiency. ORPHA:3008

Pyruvate carboxylase (PC) deficiency is a rare neurometabolic disorder characterized by metabolic acidosis, failure to thrive, developmental delay, and recurrent seizures at an early age in severely affected patients.