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Adenylosuccinate lyase deficiency

INSERM

Source

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

Adenylosuccinate lyase deficiency. ORPHA:46

Adenylosuccinate lyase deficiency (ADSL deficiency) is a disorder of purine metabolism characterized by intellectual disability, psychomotor delay and/or regression, seizures, and autistic features.