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Neurological conditions associated with aminoacylase 1 deficiency

INSERM

Source

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

Neurological conditions associated with aminoacylase 1 deficiency. ORPHA:137754

Aminoacylase 1 deficiency (ACY1D) is an inborn error of metabolism marked by a characteristic pattern of urinary N-acetyl amino acid excretion and neurologic symptoms.