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Urocanic aciduria

INSERM

Source

INSERM. (1999). Orphanet: an online rare disease and orphan drug data base. Urocanic aciduria. ORPHA:210128

Encephalopathy due to urocanase deficiency is an extremely rare histidine metabolism disorder characterized by urocanic aciduria and other variable manifestations including intellectual deficit and intermittent ataxia in the 4 cases reported to date.