

[Open Peer Review on Qeios](#)

Homocystinuria due to methylene tetrahydrofolate reductase deficiency

INSERM

Source

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

Homocystinuria due to methylene tetrahydrofolate reductase deficiency. ORPHA:395

Homocystinuria due to methylene tetrahydrofolate reductase (MTHFR) deficiency is a metabolic disorder characterised by neurological manifestations.