

[Open Peer Review on Qeios](#)

Ornithine transcarbamylase deficiency

INSERM

Source

INSERM. (1999). *Orphanet: an online rare disease and orphan drug data base. Ornithine transcarbamylase deficiency. ORPHA:664*

Ornithine transcarbamylase deficiency (OTCD) is a disorder of urea cycle metabolism and ammonia detoxification (see this term) characterized by either a severe, neonatal-onset disease found almost exclusively in males, or later-onset (partial) forms of the disease. Both present with episodes of hyperammonemia that can be fatal and which can lead to neurological complications.