

[Open Peer Review on Qeios](#)

Atypical hypotonia-cystinuria syndrome

INSERM

Source

INSERM. (1999). *Orphanet: an online rare disease and orphan drug data base. Atypical hypotonia-cystinuria syndrome. ORPHA:238523*

Atypical hypotonia-cystinuria syndrome is a form of hypotonia-cystinuria type 1 syndrome characterized by mild to moderate intellectual disability in addition to classic hypotonia-cystinuria syndrome phenotype (cystinuria type 1, generalised hypotonia, poor feeding, growth retardation, and minor facial dysmorphism).