

[Open Peer Review on Qeios](#)

Carnosinemia

National Cancer Institute

Source

National Cancer Institute. *Carnosinemia*. NCI Thesaurus. Code C125661.

A rare autosomal recessive metabolic disorder caused by mutation in CNDP1 gene. It is characterized by deficiency of carnosinase and manifests with severe mental defects and myoclonic seizures.