

Open Peer Review on Qeios

## Citrullinemia

National Cancer Institute

## Source

National Cancer Institute. <u>Citrullinemia</u>. NCI Thesaurus. Code C84639.

A rare autosomal recessive inherited disorder caused by mutations in the ASS1 and SLC25A13 genes. It is characterized by a defective urea cycle, resulting in the accumulation of ammonia and other toxic substances in the blood.

Qeios ID: YUZRQ6 · https://doi.org/10.32388/YUZRQ6