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# X-Linked Creatine Transporter Deficiency

National Cancer Institute

## Source

National Cancer Institute. *X-Linked Creatine Transporter Deficiency*. NCI Thesaurus. Code C125665.

A rare X-linked inherited disorder. It is caused by mutations in the SLC6A8 gene resulting in the absence of a compound needed to transport creatine into cells. It manifests with intellectual disability, seizures, short stature, and midface hypoplasia.