

[Open Peer Review on Qeios](#)

MFSD8 wt Allele

National Cancer Institute

Source

National Cancer Institute. *MFSD8 wt Allele*. NCI Thesaurus. Code C157081.

Human MFSD8 wild-type allele is located in the vicinity of 4q28.2 and is approximately 48 kb in length. This allele, which encodes major facilitator superfamily domain-containing protein 8, plays a role in lysosome activity. Missense mutations in the gene are associated with both macular dystrophy with central cone involvement and late infantile-onset neuronal ceroid lipofuscinosis (vLINCL; neuronal ceroid lipofuscinosis type 7; CLN7 Batten disease).