MFSD8 gene

major facilitator superfamily domain containing 8

Normal Function

The *MFSD8* gene provides instructions for making a protein whose function is unknown. The MFSD8 protein is embedded in the membrane of cell compartments called lysosomes, which digest and recycle different types of molecules. It is one of a large group of related proteins called the major facilitator superfamily of secondary active transporter proteins. Proteins in this family move certain molecules within a cell or in and out of cells. While it is likely that the MFSD8 protein transports molecules across the lysosomal membrane, the specific molecules it moves have not been identified.

Health Conditions Related to Genetic Changes

CLN7 disease

At least 30 mutations in the *MFSD8* gene have been found to cause CLN7 disease. This condition typically starts in early childhood with the loss of previously acquired skills (developmental regression), recurrent seizures (epilepsy), muscle twitches (myoclonus), difficulty coordinating movements (ataxia), speech impairment, and vision loss. Mental functioning and motor skills (such as sitting and walking) decline with age. Individuals with CLN7 disease typically do not survive past their teens.

MFSD8 gene mutations that cause CLN7 disease likely lead to the production of a protein with altered structure or function. One *MFSD8* gene mutation is responsible for almost all cases of CLN7 disease in the Roma population of the Czech Republic. This mutation replaces the protein building block (amino acid) threonine with the amino acid lysine at position 294 in the MFSD8 protein (written as T294K). A variety of other mutations cause the condition in other populations.

It is unclear how an altered MFSD8 protein leads to the severe neurological features of CLN7 disease. CLN7 disease is characterized by the accumulation of proteins and other substances in lysosomes. These accumulations occur in cells throughout the body; however, nerve cells seem to be particularly vulnerable to their effects. These accumulations can cause cell damage leading to cell death. Individuals with CLN7 disease have gradual nerve cell loss in certain parts of the brain, which likely leads to the signs and symptoms of this condition.

Other Names for This Gene

- ceroid-lipofuscinosis neuronal protein 7
- CLN7
- major facilitator superfamily domain-containing protein 8
- MFSD8_HUMAN
- MGC33302

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of MFSD8 (https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=256471[geneid])

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?term=%28MFSD8%5BTIAB%5D%29+OR+%28CLN7%5BTIAB%5D%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+3600+days%22%5Bdp%5D)

Catalog of Genes and Diseases from OMIM

- MAJOR FACILITATOR SUPERFAMILY DOMAIN-CONTAINING PROTEIN 8; MFSD8 (https://omim.org/entry/611124)

Gene and Variant Databases

- NCBI Gene (https://www.ncbi.nlm.nih.gov/gene/256471)
- ClinVar (https://www.ncbi.nlm.nih.gov/clinvar?term=MFSD8[gene])

References

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Genomic Location

The *MFSD8* gene is found on chromosome 4 (https://medlineplus.gov/genetics/chromosome/4/).

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