# CLN7 Disease

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CLN7 disease is an inherited disorder that primarily affects the nervous system. The signs and symptoms of this condition typically begin between ages 2 and 7. The initial features usually include recurrent seizures (epilepsy) and the loss of previously acquired skills (developmental regression). Affected children also develop 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.

Keywords: genetic conditions

## 1. Introduction

CLN7 disease is one of a group of disorders known as neuronal ceroid lipofuscinoses (NCLs), which may also be collectively referred to as Batten disease. All these disorders affect the nervous system and typically cause worsening problems with vision, movement, and thinking ability. The different NCLs are distinguished by their genetic cause. Each disease type is given the designation "CLN," meaning ceroid lipofuscinosis, neuronal, and then a number to indicate its subtype.

## 2. Frequency

The incidence of CLN7 disease is unknown; more than 70 cases have been described in the scientific literature. CLN7 disease was first diagnosed in the Turkish population and was thought to be limited to individuals in that group. However, CLN7 disease has now been identified in people around the world. Collectively, all forms of NCL affect an estimated 1 in 100,000 individuals worldwide.

## 3. Causes

Mutations in the *MFSD8* gene cause CLN7 disease. 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. Based on the structure of the protein, MFSD8 probably transports molecules across the lysosomal membrane, but the specific molecules it moves have not been identified.

*MFSD8* gene mutations likely lead to the production of a protein with altered structure or function. It is unclear how an altered MFSD8 protein leads to the severe neurological features of CLN7 disease. CLN7 disease, like other NCLs, 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.

### 3.1. The Gene Associated with CLN7 Disease

- MFSD8

## 4. Inheritance

This condition is inherited in an autosomal recessive pattern, which means both copies of the gene in each cell have mutations. The parents of an individual with an autosomal recessive condition each carry one copy of the mutated gene, but they typically do not show signs and symptoms of the condition.

## 5. Other Names for This Condition

- CLN7
- CLN7 disease, late infantile
- MFSD8-related neuronal ceroid lipofuscinosis

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