

## ENAM gene

enamelin

### Normal Function

The *ENAM* gene provides instructions for making a protein called enamelin, which is essential for normal tooth development. Enamelin is involved in the formation of enamel, which is the hard, white material that forms the protective outer layer of each tooth. Enamel is composed mainly of mineral crystals. These microscopic crystals are arranged in organized bundles that give enamel its strength and durability. Although the exact function of enamelin is not well understood, this protein plays a key role in the formation and growth of crystals in developing enamel.

### Health Conditions Related to Genetic Changes

#### Amelogenesis imperfecta

At least 14 mutations in the *ENAM* gene have been identified in people with a disorder of tooth development called amelogenesis imperfecta. Mutations in this gene cause autosomal dominant and autosomal recessive forms of this condition.

In the autosomal dominant form, one copy of the *ENAM* gene in each cell is altered. These mutations have a variety of effects on enamel formation. Some of these mutations reduce the amount of enamel produced from one copy of the gene. Other mutations lead to the production of an abnormally short version of enamelin that is missing critical regions. A reduced amount of enamelin or an altered version of the protein can lead to severe problems with developing enamel or cause milder defects such as shallow pits or horizontal grooves in the teeth.

In the autosomal recessive form of amelogenesis imperfecta, two copies of the *ENAM* gene in each cell are altered. These mutations result in the production of an abnormal version of enamelin that prevents enamel from developing properly. People who inherit two mutated copies of the *ENAM* gene have severe defects in their enamel; as a result, this protective covering may be very thin or completely absent.

### Other Names for This Gene

- ADAI
- AIH2

- ENAM\_HUMAN

## **Additional Information & Resources**

### Tests Listed in the Genetic Testing Registry

- Tests of ENAM ([https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=10117\[geneid\]](https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=10117[geneid]))

### Scientific Articles on PubMed

- PubMed (<https://pubmed.ncbi.nlm.nih.gov/?term=%28%28ENAM%5BTIAB%5D%29+OR+%28enamelin%5BTIAB%5D%29%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+1800+days%22%5Bdp%5D%29%29%29>)

### Catalog of Genes and Diseases from OMIM

- ENAMELIN; ENAM (<https://omim.org/entry/606585>)

### Gene and Variant Databases

- NCBI Gene (<https://www.ncbi.nlm.nih.gov/gene/10117>)
- ClinVar ([https://www.ncbi.nlm.nih.gov/clinvar?term=ENAM\[gene\]](https://www.ncbi.nlm.nih.gov/clinvar?term=ENAM[gene]))

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## Genomic Location

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

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