

## FAM83H gene

family with sequence similarity 83 member H

### Normal Function

The *FAM83H* gene provides instructions for making a protein whose function is not well understood. The protein is found in several types of cells, including specialized cells called ameloblasts. Ameloblasts produce tooth enamel, which is the hard, calcium-rich material that forms the protective outer layer of each tooth. The FAM83H protein is thought to be involved in the formation of enamel, although its role in this process is unknown.

### Health Conditions Related to Genetic Changes

#### Amelogenesis imperfecta

At least 20 mutations in the *FAM83H* gene have been found in an autosomal dominant form of a disorder of tooth development called amelogenesis imperfecta. Autosomal dominant inheritance means that one copy of the *FAM83H* gene in each cell is altered. All of the known *FAM83H* gene mutations lead to production of an abnormally short protein. While the normal protein is found in the fluid-filled space inside the cell (the cytoplasm), the altered protein is found in the cell's nucleus. Researchers speculate that the altered protein produced from the mutated copy of the gene interferes with the function of the normal protein produced from the non-mutated copy of the gene (such mutations are described as "dominant-negative"). However, researchers do not know how the altered protein leads to teeth with unusually thin, rough, yellowish-brown enamel.

### Other Names for This Gene

- AI3
- FA83H\_HUMAN
- FAM83H variant 1
- family with sequence similarity 83, member H
- FLJ46072
- protein FAM83H

## Additional Information & Resources

### Tests Listed in the Genetic Testing Registry

- Tests of FAM83H ([https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=286077\[geneid\]](https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=286077[geneid]))

### Scientific Articles on PubMed

- PubMed (<https://pubmed.ncbi.nlm.nih.gov/?term=%28FAM83H%5BTIAB%5D%29+OR+%28AI3%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%29>)

### Catalog of Genes and Diseases from OMIM

- FAMILY WITH SEQUENCE SIMILARITY 83, MEMBER H; FAM83H (<https://omim.org/entry/611927>)

### Gene and Variant Databases

- NCBI Gene (<https://www.ncbi.nlm.nih.gov/gene/286077>)
- ClinVar ([https://www.ncbi.nlm.nih.gov/clinvar?term=FAM83H\[gene\]](https://www.ncbi.nlm.nih.gov/clinvar?term=FAM83H[gene]))

## References

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### **Genomic Location**

The *FAM83H* gene is found on chromosome 8 (<https://medlineplus.gov/genetics/chromosome/8/>).

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