

AMELX gene

amelogenin X-linked

Normal Function

The *AMELX* gene provides instructions for making a protein called amelogenin, which is essential for normal tooth development. Amelogenin 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 amelogenin is not well understood, it appears to separate and support the crystals as they grow. Amelogenin is removed from the developing crystals when it is no longer needed, leaving mature enamel that contains very little protein.

One copy of the amelogenin gene is located on each of the sex chromosomes (the X and Y chromosomes). The *AMELX* gene, which is located on the X chromosome, makes almost all of the body's amelogenin. The copy of the amelogenin gene on the Y chromosome, *AMELY*, makes very little amelogenin and is not needed for enamel formation.

Health Conditions Related to Genetic Changes

Amelogenesis imperfecta

At least 23 mutations in the *AMELX* gene have been identified in people with an X-linked form of a disorder of tooth development called amelogenesis imperfecta. (X-linked disorders are caused by mutations in genes on the X chromosome.) Some *AMELX* gene mutations lead to the production of an abnormal version of the amelogenin protein that can interfere with the formation and organization of enamel crystals. Other *AMELX* gene mutations prevent one copy of the gene from producing any amelogenin protein at all. Enamel cannot form properly without an adequate amount of amelogenin.

Males have a single copy of the X chromosome in each cell. Males who inherit an altered copy of the *AMELX* gene have very little amelogenin and develop almost no enamel to cover and protect their teeth. (The normal *AMELY* gene on the Y chromosome does not provide enough amelogenin to compensate.) Females have two copies of the X chromosome in each cell. Females who inherit one altered copy of the *AMELX* gene are less severely affected than males because they have a normal copy

of the gene on the other X chromosome to produce amelogenin. Their tooth enamel may have structural defects such as a distinctive pattern of vertical grooves.

Other Names for This Gene

- AIH1
- ALGN
- amelogenin (amelogenesis imperfecta 1, X-linked)
- amelogenin, X-linked
- AMELX_HUMAN
- AMG
- AMGL
- AMGX

Additional Information & Resources

Tests Listed in the Genetic Testing Registry

- Tests of AMELX ([https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=265\[geneid\]](https://www.ncbi.nlm.nih.gov/gtr/all/tests/?term=265[geneid]))

Scientific Articles on PubMed

- PubMed (<https://pubmed.ncbi.nlm.nih.gov/?term=%28%28AMELX%5BTIAB%5D%29+OR+%28amelogenin%5BTI%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

- AMELOGENIN; AMELX (<https://omim.org/entry/300391>)

Gene and Variant Databases

- NCBI Gene (<https://www.ncbi.nlm.nih.gov/gene/265>)
- ClinVar ([https://www.ncbi.nlm.nih.gov/clinvar?term=AMELX\[gene\]](https://www.ncbi.nlm.nih.gov/clinvar?term=AMELX[gene]))

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Genomic Location

The *AMELX* gene is found on the X chromosome (<https://medlineplus.gov/genetics/chromosome/x/>).

Last updated May 1, 2015