MMP20 Gene

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matrix metallopeptidase 20

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1. Introduction

The *MMP20* gene provides instructions for making a protein called enamelysin, which is essential for normal tooth development. Enamelysin 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.

Certain proteins are needed to shape and organize the crystals as they form, but these proteins must be removed for enamel to harden normally. Enamelysin cuts (cleaves) other proteins involved in enamel formation, such as amelogenin and ameloblastin, into smaller pieces. Cleavage of these proteins makes them easier to remove when they are no longer needed.

2. Health Conditions Related to Genetic Changes

2.1. Amelogenesis imperfecta

At least seven mutations in the *MMP20* gene have been identified in people with an autosomal recessive form of a disorder of tooth development called amelogenesis imperfecta. Autosomal recessive inheritance means that two copies of the *MMP20* gene in each cell are altered. The *MMP20* gene mutations involved in this condition prevent cells from producing functional enamelysin. Without this protein's function, amelogenin and other proteins are not cleaved during enamel formation. Because these proteins remain in the enamel, it does not harden during its formation. The resulting enamel is soft and has an abnormal crystal structure. Teeth with this defective enamel are abnormally rough, discolored, and prone to breakage.

3. Other Names for This Gene

- enamel metalloproteinase
- matrix metallopeptidase 20 (enamelysin)
- matrix metalloproteinase 20
- MMP-20
- MMP20_HUMAN

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