DSPP Gene

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Dentin Sialophosphoprotein: The DSPP gene provides instructions for making a protein called dentin sialophosphoprotein.

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1. Normal Function

Soon after it is produced, this protein is cut into two smaller proteins: dentin sialoprotein and dentin phosphoprotein. These proteins are components of dentin, which is a bone-like substance that makes up the protective middle layer of each tooth. A third smaller protein produced from dentin sialophosphoprotein, called dentin glycoprotein, was identified in pigs but has not been found in humans.

Although the exact functions of the DSPP-derived proteins are unknown, these proteins appear to be essential for normal tooth development. Dentin phosphoprotein is thought to be involved in the normal hardening of collagen, the most abundant protein in dentin. Specifically, dentin phosphoprotein may play a role in the deposition of mineral crystals among collagen fibers (mineralization).

The *DSPP* gene is also active in the inner ear, although it is unclear whether it plays a role in normal hearing.

2. Health Conditions Related to Genetic Changes

2.1 Dentinogenesis Imperfecta

More than 20 mutations in the *DSPP* gene have been identified in people with dentinogenesis imperfecta. These genetic changes are responsible for two forms of this disorder, type II and type III. Mutations in this gene also cause dentin dysplasia type II, a disorder with signs and symptoms very similar to those of dentinogenesis imperfecta. However, dentin dysplasia type II affects the primary (baby) teeth much more than the permanent teeth. Some researchers believe that this type of dentin dysplasia and dentinogenesis imperfecta types II and III are actually forms of a single disorder.

About half of *DSPP* gene mutations affect dentin sialoprotein, altering its transport in cells. The remaining mutations affect dentin phosphoprotein, interfering with its normal production and/or secretion. As a result of these abnormalities of *DSPP*-related proteins, teeth have abnormally soft dentin. Teeth with defective dentin are discolored, weak, and prone to breakage and decay.

Although the *DSPP* gene is active in the inner ear, it is unclear whether *DSPP* gene mutations are related to the hearing loss found in a few older individuals with dentinogenesis imperfecta type II.

2.2 Nonsyndromic Hearing Loss

3. Other Names for This Gene

- dentin glycoprotein
- dentin phosphophoryn
- dentin phosphoprotein
- dentin phosphoryn
- dentin sialoprotein

- DFNA39
- DGI1
- DGP
- DPP
- DSP
- DSPP_HUMAN
- DTDP2

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