Dentinogenesis imperfecta

Description

Dentinogenesis imperfecta is a disorder of tooth development. This condition causes the teeth to be discolored (most often a blue-gray or yellow-brown color) and translucent. Teeth are also weaker than normal, making them prone to rapid wear, breakage, and loss. These problems can affect both primary (baby) teeth and permanent teeth.

Researchers have described three types of dentinogenesis imperfecta with similar dental abnormalities. Type I occurs in people who have osteogenesis imperfecta, a genetic condition in which bones are brittle and easily broken. Dentinogenesis imperfecta type II and type III usually occur in people without other inherited disorders. A few older individuals with type II have had progressive high-frequency hearing loss in addition to dental abnormalities, but it is not known whether this hearing loss is related to dentinogenesis imperfecta.

Some researchers believe that dentinogenesis imperfecta type II and type III, along with a condition called dentin dysplasia type II, are actually forms of a single disorder. The signs and symptoms of dentin dysplasia type II are very similar to those of dentinogenesis imperfecta. However, dentin dysplasia type II affects the primary teeth much more than the permanent teeth.

Frequency

Dentinogenesis imperfecta affects an estimated 1 in 6,000 to 8,000 people.

Causes

Mutations in the *DSPP* gene have been identified in people with dentinogenesis imperfecta type II and type III. Mutations in this gene are also responsible for dentin dysplasia type II. Dentinogenesis imperfecta type I occurs as part of osteogenesis imperfecta, which is caused by mutations in one of several other genes (most often the *COL1A1* or *COL1A2* genes).

The *DSPP* gene provides instructions for making two proteins that are essential for normal tooth development. These proteins are involved in the formation of dentin, which is a bone-like substance that makes up the protective middle layer of each tooth. *DSPP* gene mutations alter the proteins made from the gene, leading to the production of abnormally soft dentin. Teeth with defective dentin are discolored, weak, and more likely

to decay and break. It is unclear whether *DSPP* gene mutations are related to the hearing loss found in a few older individuals with dentinogenesis imperfecta type II.

Learn more about the gene associated with Dentinogenesis imperfecta

- DSPP

Inheritance

This condition is inherited in an autosomal dominant pattern, which means one copy of the altered gene in each cell is sufficient to cause the disorder.

Other Names for This Condition

- DGI
- Hereditary opalescent dentin

Additional Information & Resources

Genetic Testing Information

- Genetic Testing Registry: Dentinogenesis imperfecta type 2 (https://www.ncbi.nlm.nih.gov/gtr/conditions/C2973527/)
- Genetic Testing Registry: Dentinogenesis imperfecta type 3 (https://www.ncbi.nlm.nih.gov/gtr/conditions/C0399378/)

Genetic and Rare Diseases Information Center

- Dentinogenesis imperfecta (https://rarediseases.info.nih.gov/diseases/6258/index)
- Dentinogenesis imperfecta type 2 (https://rarediseases.info.nih.gov/diseases/12796/index)
- Dentinogenesis imperfecta type 3 (https://rarediseases.info.nih.gov/diseases/10144/index)

Patient Support and Advocacy Resources

- National Organization for Rare Disorders (NORD) (https://rarediseases.org/)

Clinical Trials

- ClinicalTrials.gov (https://clinicaltrials.gov/search?cond=%22Dentinogenesis imperfecta%22)

Catalog of Genes and Diseases from OMIM

- DENTIN DYSPLASIA, TYPE II; DTDP2 (https://omim.org/entry/125420)
- DENTINOGENESIS IMPERFECTA 1; DGI1 (https://omim.org/entry/125490)
- DENTINOGENESIS IMPERFECTA, SHIELDS TYPE III (https://omim.org/entry/125500)

Scientific Articles on PubMed

- PubMed (https://pubmed.ncbi.nlm.nih.gov/?term=%28Dentinogenesis+Imperfecta%5BMAJR%5D%29+AND+%28dentinogenesis+imperfecta%5BTIAB%5D%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+1800+days%22%5Bdp%5D)

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