CONDITION

Amelogenesis imperfecta

Learn how amelogenesis imperfecta affects tooth enamel, including its inherited causes, symptoms, diagnosis, dental treatment, complications, and follow-up care.

WHAT IT IS
Group of inherited tooth enamel conditions

BODY SYSTEM
Teeth and mouth

MAIN SYMPTOMS
Abnormal enamel, discolored teeth, enamel chipping or wear, tooth sensitivity

WHAT CAUSES IT
Inherited gene changes

HOW IT IS DIAGNOSED
Dental examination and X-rays

MAIN TREATMENT
Preventive and restorative dental care

CAN IT BE CURED?
No, enamel cannot grow back

IS IT AN EMERGENCY?
Sometimes, facial or neck swelling affecting breathing or swallowing

LONG-TERM OUTLOOK
Lifelong dental follow-up and repairs often needed

Overview

Amelogenesis imperfecta is a group of inherited conditions that affect the formation of tooth enamel. Enamel is the hard outer covering that protects the visible part of each tooth. In amelogenesis imperfecta, enamel may be too thin, too soft, poorly hardened, or a combination of these.

The condition is often shortened to AI. It can affect both primary teeth, sometimes called baby teeth, and permanent teeth. Most or all teeth are usually involved in a similar pattern, although the severity may vary from one tooth to another.

Enamel cannot grow back after a tooth has erupted. For this reason, early dental care is important. Treatment can protect teeth, reduce sensitivity, improve chewing, and support a person's confidence in their appearance. Management often continues from childhood into

adulthood as the mouth grows and dental needs change.

Dentists classify amelogenesis imperfecta according to how the enamel is affected. The main patterns include:

These categories help describe the teeth, but genetic testing has shown that the condition is more complex than appearance alone may suggest.

- Hypoplastic: Enamel is harder than in some other forms but is unusually thin, pitted, or grooved.
- Hypomaturation: Enamel reaches a near-normal thickness but does not mature fully, making it weaker or more likely to chip.
- Hypocalcified: Enamel forms at close to normal thickness but remains soft and may wear away soon after teeth erupt.
- Mixed patterns: Features can overlap. Some people also have enlarged pulp chambers within the teeth, a feature called taurodontism.

Symptoms

The appearance and strength of teeth can differ greatly among people with amelogenesis imperfecta. Even relatives with the same genetic change may not have identical dental problems.

Possible signs and symptoms include:

Some people develop an anterior open bite, meaning the front teeth do not touch when the back teeth are together. Tooth eruption may also be delayed, and some teeth may remain trapped beneath the gum or bone.

Sensitivity can make brushing uncomfortable. However, reducing brushing may allow more plaque to collect and can increase the chance of gum inflammation or tooth decay. A dentist can suggest ways to make daily cleaning more comfortable.

Dental appearance may affect speaking, eating in public, smiling, or social confidence. These effects are part of the condition's health impact and can be discussed with the dental team.

- Enamel that is thin, rough, pitted, grooved, or uneven
- Teeth that look yellow, brown, gray, cream, or unusually white
- Rapid chipping, crumbling, or wearing of enamel
- Sensitivity to hot, cold, sweet foods, air, or toothbrushing

- Pain when chewing if the protective enamel has been lost
- Teeth that appear small because the enamel layer is thin
- Difficulty keeping rough tooth surfaces clean
- Buildup of plaque, tartar, or stains
- Changes in the way the upper and lower teeth meet

Causes and risk factors

Amelogenesis imperfecta results from changes in genes involved in making, organizing, hardening, or maturing enamel. Different genes act at different stages of enamel development, which helps explain the range of appearances.

Inheritance patterns vary by gene. The condition may be inherited in an autosomal dominant, autosomal recessive, or X-linked pattern.

In plain language:

A genetic change can also arise for the first time in one person, so the absence of a known family history does not rule out amelogenesis imperfecta.

The condition is not caused by poor brushing, food choices, or anything a parent did during pregnancy. These factors can affect dental health, but they do not cause inherited enamel formation defects.

In some people, enamel changes occur as part of a broader genetic syndrome that may affect other parts of the body. One example is enamel-renal syndrome, which can involve calcium deposits in the kidneys. Most people with enamel abnormalities do not have this syndrome. Medical evaluation may be considered when dental findings, family history, genetic results, or other symptoms suggest a wider condition.

- Autosomal dominant means a person may develop the condition after inheriting one altered copy of a gene from either parent.
- Autosomal recessive means altered copies are usually inherited from both parents. The parents often do not have dental symptoms themselves.
- X-linked means the altered gene is on the X chromosome. The pattern and severity can differ between males and females.

Diagnosis

A dentist usually begins with a detailed examination. The pattern across the mouth is important because amelogenesis imperfecta generally affects many teeth and may involve both primary and permanent teeth.

The evaluation may include:

Dentin is the less hard tissue beneath enamel. On X-rays, enamel normally appears more dense than dentin. In some forms of amelogenesis imperfecta, that contrast is reduced. X-rays can also show unerupted teeth, enlarged pulp chambers, impacted teeth, or other developmental findings.

Genetic testing may identify the responsible gene change. It can help clarify the type of inheritance, provide information for relatives, or show whether the enamel problem is part of a syndrome. Testing does not find a cause in every person, and it is not always required to plan dental care. A genetic counselor can explain the possible results and what they may mean for family members.

Other conditions can resemble amelogenesis imperfecta. Dentists may consider excess fluoride exposure during tooth development, illness or nutritional problems affecting enamel, trauma or infection involving a primary tooth, and molar-incisor hypomineralization. Unlike inherited amelogenesis imperfecta, many of these conditions affect only certain teeth or appear in a less even pattern.

- The age at which dental changes were first noticed
- Whether primary teeth were affected
- A family history of similar tooth appearance or early dental treatment
- The thickness, hardness, color, and surface texture of the enamel
- Tooth sensitivity, wear, breakage, and bite problems
- Dental X-rays to compare enamel with the underlying dentin

Treatment and management

Treatment is based on age, the type and severity of enamel damage, sensitivity, bite, and which teeth have erupted. Care may involve a general dentist, pediatric dentist, prosthodontist, orthodontist, periodontist, oral surgeon, or clinical geneticist. A prosthodontist specializes in rebuilding and replacing teeth.

The first goals are usually to reduce discomfort, protect exposed tooth structure, and support effective cleaning. Preventive care may include professional cleaning, fluoride products, desensitizing products, sealants, and advice about brushing without causing unnecessary

abrasion. The dental team can recommend products suited to the enamel and the person's age.

Frequent acidic drinks or snacks can add chemical wear to already vulnerable teeth. Regular exposure to sugary foods and drinks also supports decay-causing bacteria. Practical dietary changes may help protect restorations and remaining tooth structure without requiring an overly restrictive diet.

Restorative treatment depends on how much sound enamel remains. Options may include tooth-colored bonding materials, fillings, sealants, veneers, or crowns. Bonding can be more difficult when enamel is soft or poorly mineralized, so the dentist may choose materials and techniques based on the specific enamel pattern.

For children, treatment is often staged. Temporary or repairable restorations can protect teeth while the jaws and permanent teeth continue to develop. Preformed metal crowns may be used on some back teeth when broader protection is needed. Tooth-colored restorations may be used where appearance is especially important.

In adolescents and adults, more durable crowns or other restorations may be considered after growth, tooth eruption, gum health, and bite have been assessed. Extensive treatment may require careful planning to avoid removing more tooth structure than necessary. Dental implants or removable replacement teeth may be options for selected adults when teeth cannot be maintained, but they are not required for everyone.

Orthodontic treatment may help with an open bite, crowding, or impacted teeth. Brackets may be harder to attach to abnormal enamel, and treatment must be coordinated with plans for crowns or bonding. Some bite problems require combined orthodontic and surgical assessment.

Pain control and anxiety management also matter. People who have had repeated dental procedures may feel worried about future care. Dentists can explain each stage, arrange shorter visits, use local anesthesia when appropriate, and discuss other support based on the planned procedure.

Complications

Without enough protection, weak enamel may wear down and expose dentin. This can increase sensitivity and may shorten the teeth or alter the bite. Severe wear can eventually approach the pulp, the soft inner tissue containing nerves and blood vessels.

Rough surfaces and discomfort during brushing can contribute to plaque buildup, gum inflammation, and tooth decay. Amelogenesis imperfecta does not mean that every person will

develop cavities, but damaged surfaces may be harder to protect and restore.

Other possible complications include broken restorations, repeated need for dental repair, difficulty chewing, altered speech, and problems with tooth eruption. Jaw-joint or muscle discomfort may develop when substantial tooth wear changes the bite.

Dental infection can occur if decay, a crack, or severe wear reaches the pulp. Increasing tooth pain, pain on biting, pus, gum swelling, fever, or facial swelling should be assessed promptly.

Call emergency services or seek emergency care for facial or neck swelling that affects breathing or swallowing, rapidly spreading swelling, severe weakness or confusion with an infection, or major facial trauma. These are different from routine sensitivity or a chipped restoration, which still deserve dental follow-up but are not usually emergencies.

Follow up

Amelogenesis imperfecta usually requires lifelong dental follow-up because enamel does not repair itself and restorations can wear or break. The timing of visits should be individualized. People with active sensitivity, rapid wear, erupting permanent teeth, orthodontic treatment, or extensive restorations may need closer monitoring.

Routine visits may include checking for new wear, decay, gum inflammation, changes in the bite, and damage to crowns or fillings. In children, the plan is revised as permanent teeth erupt and the jaws grow. In adults, follow-up focuses on preserving tooth structure and maintaining restorations, gum health, and comfortable function.

Contact a dentist sooner than a planned visit for a newly broken tooth or restoration, worsening sensitivity, persistent toothache, pain when biting, gum swelling, or difficulty eating. Early repair may prevent a smaller problem from becoming more complex.

If genetic testing suggests a syndromic form, follow-up may also involve a medical clinician or genetic counselor. Relatives may choose to seek dental or genetic assessment, but testing decisions are personal.

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