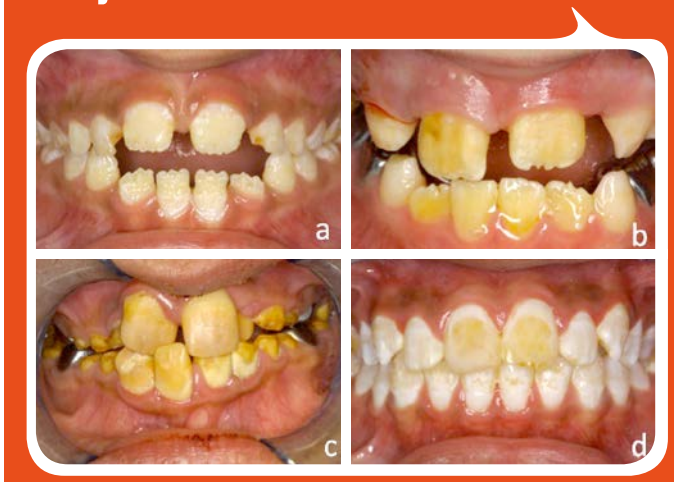
Amelogenesis Imperfecta



The diagnosis or the clinical phenotype in images

Different forms of amelogenesis imperfecta exist, depending on the type of enamel defect. When the defect is quantitative, the AI is called hypoplastic (a, b). This can go as far as a total absence of enamel (b); if the defect is qualitative, the amelogenesis imperfecta is referred to as hypomineralized (c) (soft and crumbly, rough enamel) or hypomature (d) (chalky white appearance but hard). The different types of defects can coexist on the same tooth or in the same mouth, or they can vary from one individual to another in the same family.

The disease

What is amelogenesis imperfecta?

Amelogenesis imperfecta (AI) represents a group of developmental conditions affecting the structure and clinical appearance of the enamel of all or nearly all the primary or permanent teeth. Diagnosis is based on the family history, pedigree plotting and meticulous clinical observation. At present, genetic diagnosis is only available as a research tool.

How many people are affected by amelogenesis imperfecta?

The prevalence of the disease is approximately 1 in 14,000. The data vary from one population to the next.

What causes the disease?

Amelogenesis imperfecta is a hereditary disease that exists in isolation or in association with other symptoms in syndromes. All modes of transmission are possible (autosomal dominant, recessive or X-linked).
The known genes that are implicated are: **AMELX (Xp22.3-p22.1); ENAM (4q21);AMBN(4q21);MMP20(11q22.3-q23);KLK4(19q13.3-q13.4);WDR72 (15q21.3); FAM83H (8q24.3); FAM20A (17q24.2); DLX3 (17q21.3-q22); TP63 (3q27-q29); CNNM4 (2q11.2); ROGDI (16p13.3); C4orf26 (4q21.1); SLC24A4 (14q32.12); LAMB3 (1q32); ITGB6 (2q24.2); STIM1 (11p15.5).** Many other responsible genes remain to be discovered.

AI is sometimes accompanied by other dental abnormalities such as taurodontism, dentin abnormalities, tooth eruption disturbances, tooth resorption, a dysmorphic maxillary basal bone in the form of a skeletal anterior open-bite, and gingival hyperplasia.

What are the clinical manifestations of the disease?

Amelogenesis imperfecta can be subdivided into different forms, which are hypoplastic, hypomineralized and hypomature.

The hypoplastic form – quantitative defects
This form is manifested by a thin or no enamel coating and the presence of pits or striae. Areas of normal enamel are found. However, the enamel is hard and translucent, sometimes with a rough or stippled texture. The enamel is not prone to wear.

The hypomineralized form – qualitative defect
The enamel is of a normal thickness. It is soft, intrinsically yellowish-brown and chips quickly.

The hypomature form – qualitative defect
The thickness of the enamel is normal, relatively hard, with little or no contrast with the dentin and its colour ranges from chalky white to yellowish-brown.

Amelogenesis imperfecta may also be clinically associated with rare syndromes and diseases:
For example, it may be associated with
- Nephrocalcinosis (kidney disorder), which may be asymptomatic
- Platyspondyly (vertebral anomalies), short stature
- Cone-rod dystrophy (photoreceptive cells of the eye)
- Epilepsy and mental deterioration
- Onycholysis (nail abnormalities)
- Curly hair and bone abnormalities.

Treatment, care and prevention

Amelogenesis imperfecta may be a sign of a more systemic disease, and patients should be examined in particular for the associated signs. This anomaly must be reported to the doctor in charge of the child or adult patient, i.e. the paediatrician, family doctor, geneticist, etc. This medical diagnosis, reinforced by the observations of the dentist, is important for the affected individual and his or her family.

From the point of view of dental care, crucial elements of successful treatment include prevention through the implementation of an oral health program, protection of the existing teeth and multidisciplinary treatment of these dental abnormalities.

In **Alsace** there is a national **Reference Centre for OroDental Manifestations of Rare Diseases** that can be contacted for diagnosis, advises on the treatment and care of patients and/or referrals. In Alsace and France the prevalence of these disorders is unknown. **You can help increase knowledge of this dental disorder and of the associated rare diseases by participating in the registration of patients** in the D[4]/Phenodent patient registry. The creation of this registry was approved by the Consultative Committee for Data Processing in Health Research (CCTIRS) on September 11, 2008 and was authorized by the French Data Protection Authority (CNIL) on May 18, 2009 (registration no. 908416).

Patients and their families may participate in the **InterregV project, "RARENET"**. The objectives of this project are to characterize the clinical manifestations and problems linked to the disease, evaluate their incidence and repercussions in terms of quality of life, attempt to establish a link between certain symptoms and the implicated genes and discover new genes responsible for this disease.

Patient contact

If you encounter any patients affected by the diseases described above
contact@rarenet.eu
The anonymity of the physician, patient and family will be protected in the patient registration process, which will furthermore be carried out with the patient's consent.

Find out more

www.rarenet.eu, INTERREG research project supported by the Faculty of Oral Medicine of the University of Strasbourg.

Orphanet, the portal for rare diseases and orphan drugs, under amelogenesis imperfecta

Centre de Référence des Manifestations Odontologiques de Maladies Rares

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