

Dentinogenesis Imperfecta and Dentin Dysplasia



The diagnosis or the clinical phenotype in images

Clinical examination

The following aspects should be analysed during the clinical examination: the colour of the teeth (amber, grey or even purple) wear with loss of vertical dimension and the possible presence of abscesses, due to the penetration of bacteria in the exposed dentinal tubules.

The disease

What is dentinogenesis imperfecta?

Dentinogenesis imperfecta and dentin dysplasia are diseases characterized by an abnormal formation and thus abnormal structure of the dentin, generally affecting both primary and permanent teeth. There are three types of dentinogenesis imperfecta (DGI-I, DGI-II, DGI-III) and two types of dentin dysplasia (DD-I, DD-II).

How many people are affected by amelogenesis imperfecta?

According to certain studies, the prevalence of the disease is 1 in 6,000 to 8,000.

What causes the disease?

Dentinogenesis imperfecta - Shield's type II (DGHII) is an autosomal dominant hereditary disease caused by mutations in the DSPP gene (dentin sialophosphoprotein) coding for dentin sialoprotein and dentin phosphoprotein. The same gene is implicated in type III dentinogenesis imperfecta and in type II

dentin dysplasia (DD-II). These different forms are allelic.

Dentinogenesis imperfecta (type I DGI-I) may be associated with certain forms of osteogenesis imperfecta. Mutations in the genes coding for type I collagen, COL1A1 and COL1A2, or coding for collagen-modifying enzymes and chaperone proteins (CRTAP, LEPRE1, PPIB, FKBP10, SERPINH1) cause this disease.

What are the clinical manifestations of the disease?

Dentinogenesis imperfecta

The teeth, in both primary and permanent dentition, are typically amber-coloured and translucent, and are very prone to wear.

The crowns are globular. The enamel chips quickly, leaving the dentin bare. Infectious episodes are frequent.

On radiographs, the crowns are bell-shaped with a very pronounced cervical constriction; the roots are short and thin, and the irregular and defective production of dentin results in rapid pulp obliteration. Teeth without cavities sometimes present periapical radiolucency.

DGI-I: This form is associated with osteogenesis imperfecta, a disease characterized by brittle bones (fractures, osteoporosis, reduced bone mass and bone deformations) and sometimes blue sclera.

DGI-II: The permanent teeth are less affected than the primary teeth. The bones are not affected. However, this disease can be associated with hearing loss.

DGI-III: This form is found in an isolated population in Brandywine in the United States (Maryland).

Dentinogenesis imperfecta is also found in certain syndromes, such as Ehlers-Danlos syndrome, Goldblatt syndrome, Schimke immuno-osseous dysplasia, brachio-skeletal-genital syndrome, the syndrome associating short stature with severe microdontia, opalescent teeth and rootless molars.

Dentin dysplasia

DD-I: The teeth have a normal clinical appearance.

On radiographs, the roots are pointy and short or inexistent, with apical cone shape constriction.

DD-II: In this pathology, only the mineralization of the dentin of the primary teeth is abnormal. The primary teeth are opalescent with obliterated pulp chambers, whereas the permanent teeth have a normal colour, but present pulp calcifications.

Treatment, care and prevention

Dentinogenesis imperfecta may be a sign of a more systemic disease, and patients should be examined in particular for associated signs such as hearing loss, bone fractures, etc. 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, with the implementation of an oral health program, protection of the existing teeth (protection; the realisation of prefabricated paediatric crowns on the primary molars, and then on the permanent molars; maintenance of the arch length and of the vertical dimension of occlusion; endodontic treatments are very difficult or even impossible) and the 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 **Interreg IV/OS A27 project, "Oro-Dental Manifestations of Rare Diseases"**. 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

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.genosmile.eu, INTERREG/OS research project supported by the Faculty of Oral Medicine of the University of Strasbourg.

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

Centre de Référence des Manifestations Odontologiques de Maladies Rares

Pôle de Médecine et Chirurgie Bucco-Dentaires
Hôpitaux Universitaires de Strasbourg,
1 place de l'Hôpital, F-67000 Strasbourg Cedex France.
cref-odonto@chru-strasbourg.fr
Tel +33 (0)3.88.11.69.10
(Fax -18)
www.chru-strasbourg.fr

