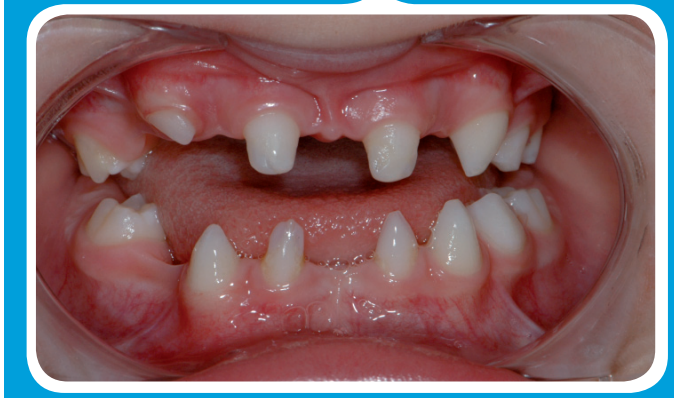
Oligodontia



The clinical phenotype in images

The disease - The clinical sign

What is oligodontia?

Oligodontia is a dental abnormality in which the patient is missing teeth. It is a hereditary disorder characterized by agenesis of more than 6 primary/permanent teeth, excluding the wisdom teeth.
Not only the number of missing teeth, but also the type of missing teeth, must be considered. The teeth most often absent are the terminal teeth of a series (premolars (32% to 15%), maxillary lateral incisors (27%) and third molars (25%). The canines, first and second molars (1%) and maxillary central incisors (0.05%) are rarely absent; particular attention must be paid to this sign. The type of teeth that are missing is not coincidental; rather, it is directly related to the dentition pattern.

How many people are affected by the disease?

The prevalence of oligodontia (number of teeth missing ≥6) is estimated at 0.09%. By way of comparison, the prevalence of dental agenesis varies from 4.5% to 10%. Agenesis of at least one incisor identifies 65% of affected patients, as early as the age at which these teeth erupt, i.e. around 8 years old. Many patients affected by oligodontia do not present any other associated ectodermal signs.

What causes the disease?

This genetic disorder is usually transmitted through autosomal dominant inheritance with complete penetrance and variable expression. It is caused by a disruption of the mechanisms that control the dentition pattern or the progression of dental development. Many genes are concerned.

Oligodontia is caused by mutations of divergent homeobox genes coding for transcription factors, such as MSX1 and PAX9 responsible for the maxillary and mandible territorial organisation. WNT10A gene mutations are responsible of autosomal recessive forms of hypohidrotic ectodermal dysplasia, odonto-onycho-dermal and Schöpf-Schulz-Passarge syndromes as well as of a substantial number (30 to 50% cases according to studies) of non-syndromic oligodontia.
Mutations in the gene coding for the AXIN2 protein, a regulator of the Wnt signalling pathway, are implicated in the association between oligodontia and a predisposition to colorectal cancer.
Oligodontia may also exist in association with cleft lip and/or palate in other syndromes, such as the Van der Woude syndrome (IRF6 gene). The MSX1 gene is also implicated in this association. Individuals from the same family may present either with varying degrees of oligodontia, or with a cleft lip and/or palate, or with both (variable expression).

Oligodontia may be associated with other symptoms affecting ectodermal structures such as skin, nails, hair, sweat glands, saliva glands, mammary glands and the nasolacrimal duct, and it may be clinically associated with ectodermal dysplasia, a huge group of rare diseases. In that case, many genes of the Nf-kappab and Wnt pathways are implicated. Some of them, such as EDA and EDARADD are also responsible for isolated oligodontia, without associated manifestations.
It can also be associated with other syndromes, such as Down syndrome.

What are the clinical manifestations of the disease?

Oligodontia is associated with teeth of an abnormal size (smaller) and shape Oligodontia(conical, taurodontism). Enamel abnormalities and delayed eruption are frequent.
Oligodontia is also associated with deficient secretion of saliva (30% of cases).
Other ectodermal signs are present in 50% of patients. 10% of patients also have reduced functioning of their sweat glands and hair or nail abnormalities.

Treatment, care and prevention

The presence of oliogdontia may be a sign of a systemic disease and must result in a search in particular for 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 patient 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 and multidisciplinary treatment of these dental abnormalities. One of the key objectives is to favour the academic and social integration of patients. By way of example, the prosthetic rehabilitation of a child with oligodontia begins before the age of 2 years, with the prosthesis being replaced progressively as the child grows.
Prosthetic implantation has recently begun to be covered under the French healthcare system following an assessment made by an expert.
There are possibilities for treatment reimbursement:

- before growth has stopped (2 to 4 implants in the symphyseal region)
- after growth has stopped (12 maxillary / mandibular implants).

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 amelogenesis imperfecta

Centre de Référence des Manifestations Odontologiques de Maladies Rares

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