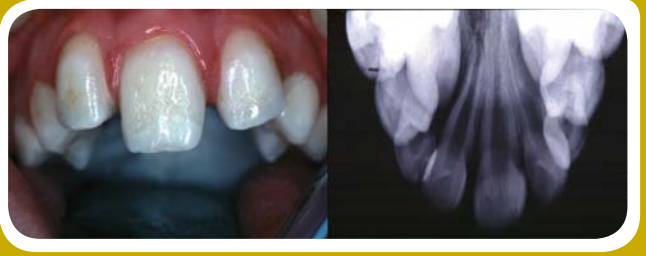
Solitary Median Maxillary Central Incisor Syndrome



The diagnosis or the clinical phenotype in images

The disease

What is solitary median maxillary central incisor syndrome?

Solitary median maxillary central incisor (SMMCI) syndrome is a developmental defect of the median line of the craniofacial bones and a minor clinical manifestation of holoprosencephaly (cerebral disorder).

How many people are affected by this dental disorder?

Currently, the incidence of the disease is estimated at 1 in 50,000 births.

What causes the disease?

It is a genetic disorder. Various genes, implicated in holoprosencephaly or in other median defects, are responsible for this dental disorder: SHH (7q36), SIX3 (2p21), ZIC2 (13q32), TGIF (18p11.3), PTCH (9q22.3), GLI2 (2q14) and SALL4 (20q13.13-q13.2).

What are the clinical manifestations of this disorder?

The solitary median maxillary central incisor, either primary or permanent, has a symmetrically-shaped crown. It is neither left nor right; it develops and erupts precisely in the midline of the maxillary dental arch.
The median maxillary frenum may also be missing, as the palate is V-shaped.
Other abnormalities are generally associated, which are clinical manifestations of rare syndromes or diseases:
- Hypotelorism (eyes very close together), convergent strabismus
- A malformation of the nasal cavities (either choanal atresia, midnasal stenosis or congenital pyriform aperture stenosis)
- Short stature is observed in half of the children
- Growth hormone deficiency
- Hypopituitarism
- Mild to severe learning disabilities

Associated clinically observable abnormalities may include heart disease, cleft lip and/or palate and less frequently, microcephaly, deafness, oesophageal atresia, cervical hemivertebrae, cervical dermoid cysts, hypothyroidism, scoliosis and absent kidney.

Treatment, care and prevention

The presence of a solitary median maxillary central incisor is a sign of a systemic disease and should result in a search in particular for signs of holoprosencephaly. 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 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 **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

contact@genosmile.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.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 solitary median maxillary central incisor syndrome

Centre de Référence des Manifestations Odontologiques de Maladies Rares

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D[4] Diagnosing Dental Defects Database

phenodent.org

ORO-DENTAL RARE DISEASES

MALADIES RARES Manifestations bucco-dentaires
SELTENE KRANKHEITEN Manifestationen im Mund- und Zahnbereich

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