# Dental aspects and VCFS (22q11.2 deletion syndrome)
## What we know

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There is only some knowledge about specific dental and oral health issues in 22q11.2 deletion syndrome. Apart from a few publications involving larger patient cohorts, the majority of publications are still limited to case reports.

Persons with VCFS have an increased risk for impaired oral health. In particular, children with 22q11.2 deletion are reported to more frequently have aberrations in enamel formation (enamel hypoplasia – surface defect, thin but normally mineralized enamel often seen as a pit; and enamel hypomineralization – normal thickness of enamel but lower mineralization often seen as opacities), delay in tooth eruption, and hypodontia (missing teeth). Much of the deviations in enamel are related to endocrine problems, mainly hypocalcemia.

Teeth start to develop a long time before birth (during the first stage of development in the second fetal month) and the development and mineralization go on until all teeth are fully erupted during the teenage period. In fact the development continues even longer as the wisdom teeth do not erupt until adulthood and further, the dentine inside all teeth continues to develop through out life. Thus, the effect of calcium metabolism on the teeth in people with 22q11.2 deletion syndrome remains a

concern for a long period of time. However, most defects in enamel are seen in the primary or deciduous teeth.

Hypodontia, in 22q11.2 deletion syndrome, is usually manifested as missing one tooth. There is insufficient data on which teeth are affected most frequently. In some cases orthodontic treatment is required, for example in cases where there is only one upper central incisor. However, for the majority of patients with hypodontia of a single tooth, orthodontic treatment is not necessary and the bite will develop satisfactory by itself.

For many people with 22q11.2 deletion syndrome there seems to be an increased risk of dental caries. One study has shown impaired salivary secretion rate and buffer capacity in patients with 22q11.2 deletion syndrome as well as increased saliva protein and IgA concentrations, reduced output of sodium, potassium, calcium, phosphate, and bicarbonate in saliva, and more cariogenic bacteria. The underlying mechanism of this is not fully understood. However, as both the quality and quantity of saliva are implicated it is plausible that salivary glands could be affected in terms of numbers, size, and function. More research is needed to learn more about this.

There are may be several issues involved when a person has dental caries problems. For example having problems with frequent infections as well as having feeding problems may affect the dietary intake in a way that increases the risk for dental caries; e.g. higher frequency of intake of food and beverages and increased consumptions of products rich in carbohydrates or frequent small meals or treats to raise appetite and interest in food are far from ideal from a dental health point of view.

There is very little knowledge about behavioral aspects in relation to dental care, but dental anxiety is frequently observed in the clinical situation. Studies are lacking concerning gingival and periodontal health. This is something for future studies to look at.

As the oral cavity is affected it is advisable that children diagnosed with 22q11.2 deletion have an odontological evaluation, if possible by a pediatric dentist, at the time of diagnosis. In the presence of palatal abnormalities, an evaluation should also be made by an orthodontist as an active member of a cleft palate craniofacial team. Adults who are diagnosed should also be examined by a dentist. The dentist should look for:

- Enamel aberrations
- Dental caries
- Gingivitis and oral hygiene
- Bite development and possible hypodontia (age appropriate assessment)
- Saliva properties, foremost saliva secretion rate.

Based on the age of the patient, evaluation and treatment plan have to be individualized. Regular dental visits are important, preferably with annual checkups. Focus should be on providing information to the patient/family and to prevent of oral health problems. A majority of the patients will need extended chair side dental prevention. Repeated evaluation of bite development and saliva properties during childhood and adolescence is advocated. In adult saliva should preferably be re-evaluated every fifth year.

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