

VELOCARDIOFACIAL SYNDROME

The name Velocardiofacial Syndrome comes from the Latin words "velum" meaning palate, "cardio" meaning heart, and "facies" having to do with the face thus incorporating the most common features of cleft palate, heart defects, and a characteristic facial appearance.

Unfortunately the testing for this syndrome has only been available since 1994 and the variability in the presentation of children with this syndrome means that they are often overlooked by professionals. A diagnosis is usually made by a clinical geneticist based on the clinical presentation of the person and the genetic testing for the deletion. A "syndrome" means a pattern of features occurring together. People with VCFS usually present with a few or many of the following features:

Congenital heart defects

Mental illness

Palate and throat problems-

Feeding and speech problems,

Nasal speech

Facial features including almond-shaped eyes, slender nose with bulbous nasal tip, small mouth

Hyperparathyroidism-low calcium & high phosphate levels, Seizures

Absent or small thymus gland

Dental issues

Immune deficiency

Hearing problems

Learning difficulties -mild to severe

Developmental delay

Low muscle tone

Gastric reflux and

regurgitation when young

Research shows that there are 188 possible anomalies associated with VCFS.

ORAL ISSUES:

Children affected with a 22q11.2 deletion may display certain dental characteristics such as higher number of cavities or poor quality teeth, which may decay quickly. Gastric reflux can contribute to the deterioration of the teeth.

There are 2 main diseases, which are preventable - Gum Disease and Tooth Decay. By preventing these diseases needless suffering and threat to general health is avoided.

Children with a 22q11.2 deletion can be more prone to disease because of:

- Poor quality teeth that may decay more easily;
- Difficulty with feeding - i.e. eating smaller amounts but more frequently; Gastric reflux.

Many children with the deletion also require medication and/or calorie supplements and some of these have a high sugar content. This magnifies the need for dental care to protect the teeth and encourage good long-term oral hygiene habits.

For children with heart defects, decayed teeth or inflamed gums also increases the risk of getting a disease of the heart called "infective endocarditis" which is an infection of the inner lining of the heart.

You can visit www.vcfsfa.org.au for more information regarding VCFS.