



CENTER

# Orofacial function of persons having 22q11 deletion syndrome

## Report from questionnaires

The survey comprises 86 Questionnaires.

**Synonyms:** 22q11-deletion syndrome, Di George syndrome, Velocardiofacial syndrome, Conotruncal anomaly face syndrome, Shprintzen syndrome.

**Estimated incidence:** 1:4 000 live births.

**Etiology:** Chromosomal deletion of a small amount of material on the long arm (q) of chromosome 22. Autosomal dominant heredity. In most cases the 22q11-deletion syndrome is a spontaneous mutation.

### General symptoms:

- C** Cardiac defect, various congenital cardiac malformations
- A** Abnormal faces, deviant facial features
- T** Thymic hypoplasia/aplasia, very small thymus gland or none at all, increased risk of infection
- C** Cleft palate, most commonly covert (submucous) clefting
- H** Hypocalcemia, calcium deficiency attributable to poorly functioning or absent parathyroid glands
- 22** deletion of chromosomal material on chromosome 22

Other malformations including deformity of the kidneys, club foot, hearing and vision problems as well as behavioral aberrations and learning difficulties may occur. There are varying combinations of symptoms, as well as a substantial variation in degree of severity.

**Orofacial/odontological symptoms:** Typical facial features may include narrow, downslanting palpebral fissures, low nasal bridge, arched upper lip and flat cheekbones (malar hypoplasia). Hypernasal speech is a very common problem. Eating disorder may occur. Aberrations in tooth mineralisation taking the form of spotting or pitting of the tooth enamel are common. Deformed teeth, known as peg shaped teeth, and occasional missing tooth buds are more frequent than in healthy individuals. Delayed dental development is also often found. An increased incidence of lingua geografica and a tendency to bleed easily from the oral mucosa have been reported, as well as poor oral hygiene, frequent caries and gingivitis (inflamed gums).

### Orofacial/odontological treatment:

- It is important that these children come in early contact with the dental services for extra preventive dental care and information about oral hygiene. Frequent infections, poor nutrition, and poorly mineralized enamel all increase the risk of caries.
- In cases of defective palate, a specialist team will be needed for follow up and treatment.
- X-ray to determine the presence of tooth buds may be needed around the age of 7 to 9.
- An orthodontist should be consulted between the ages of 7 and 9 in order to identify dental aberrations or malocclusions and to plan any necessary orthodontic treatment.
- Individuals with cardiac defects may require prophylactic antibiotics when oral interventions associated with bleeding are undertaken.

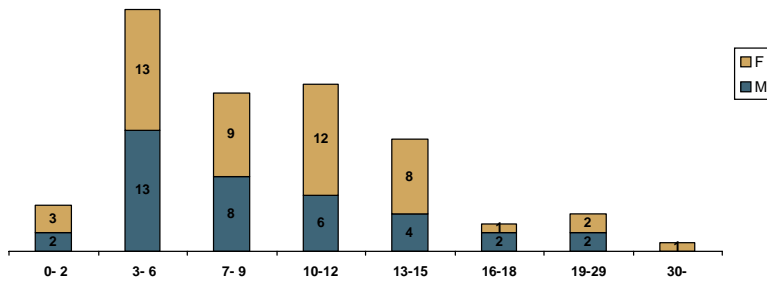
### Source:

The rare disease database of the Swedish National Board of Health and Welfare.

The MHC database - The Mun-H-Center database of orofacial manifestations in rare diseases.



Age distribution

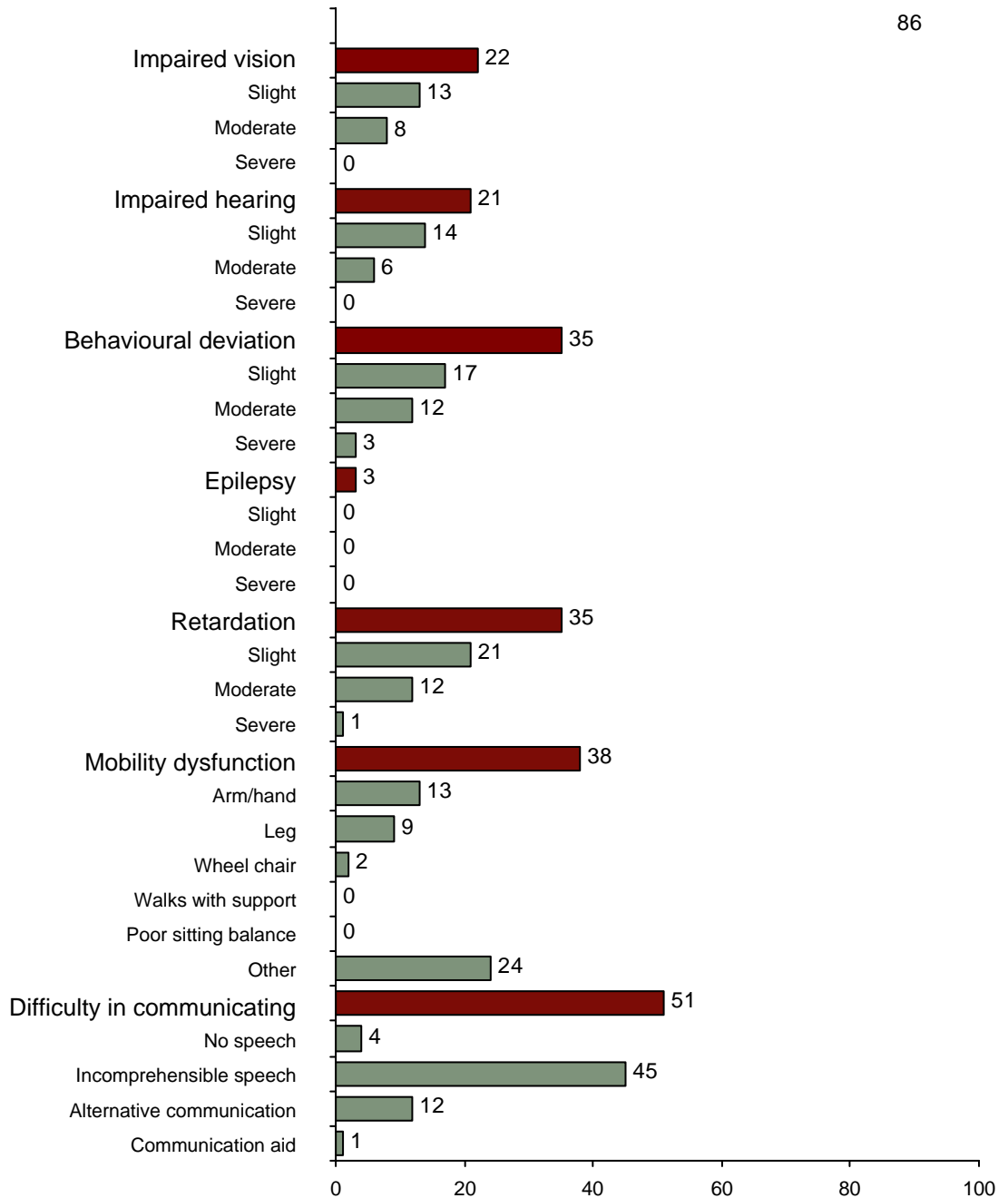


Number: 86

Ages: 0 -- 33 years

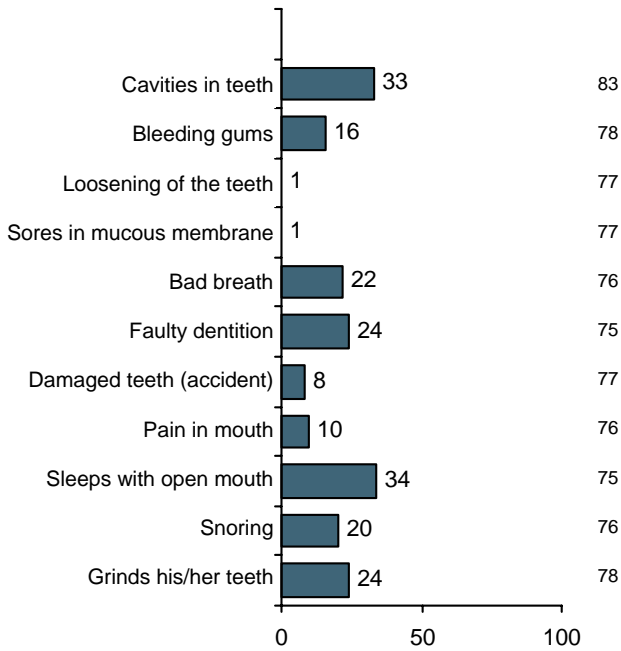
Sex: M (37) + F (49)

General disabilities

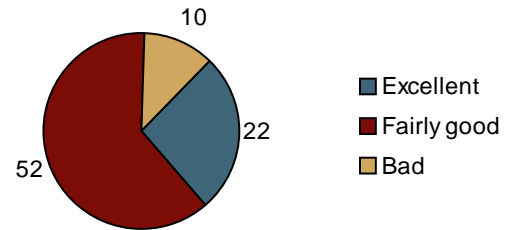


# About dental health

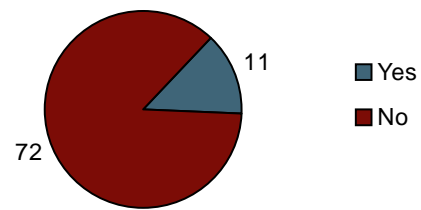
About dental health - problems



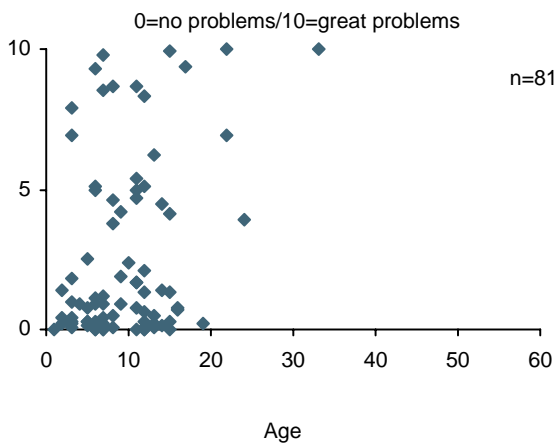
Oral health



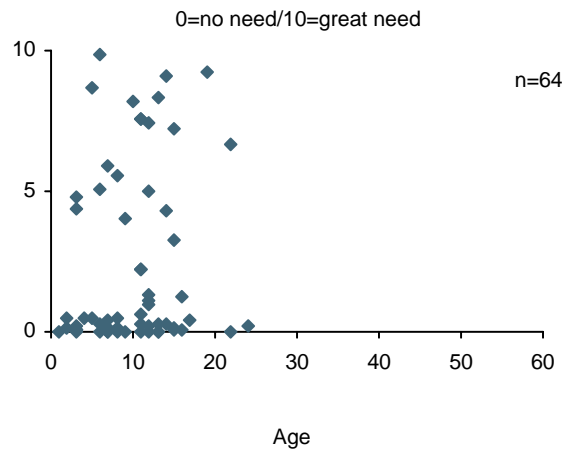
Has NN received orthodontic treatment?



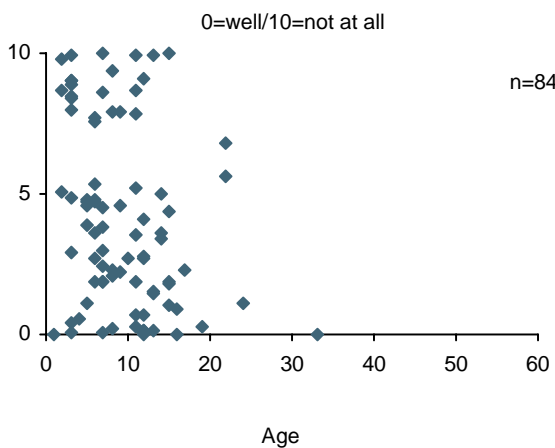
How does NN experience the dental care received?



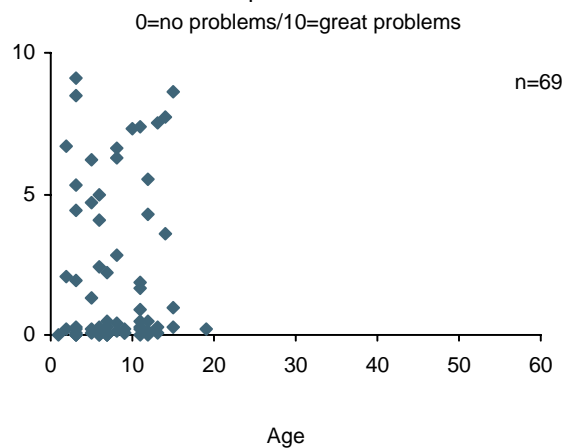
NN is considered to be in need of orthodontic treatment



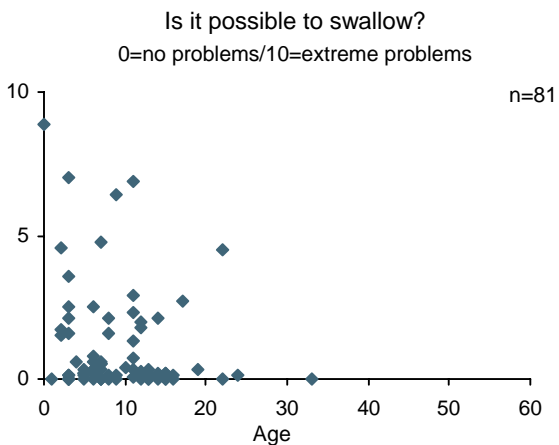
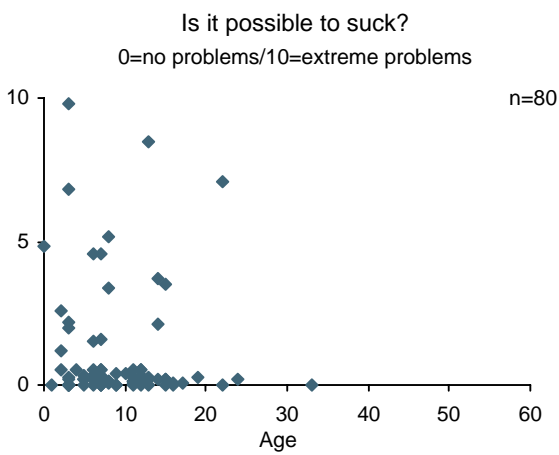
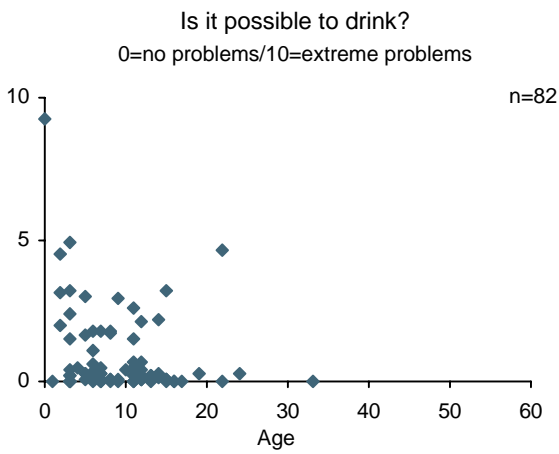
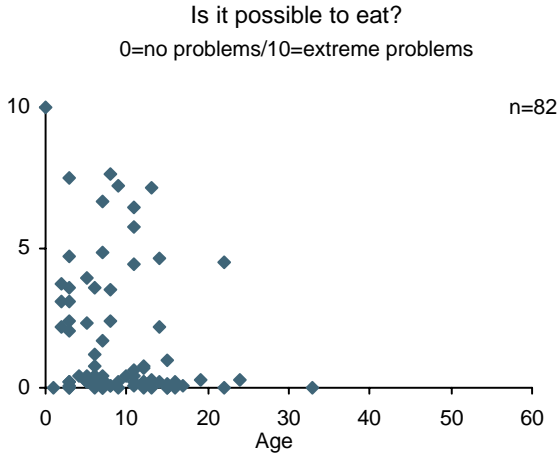
How does NN manage to brush his/her teeth?



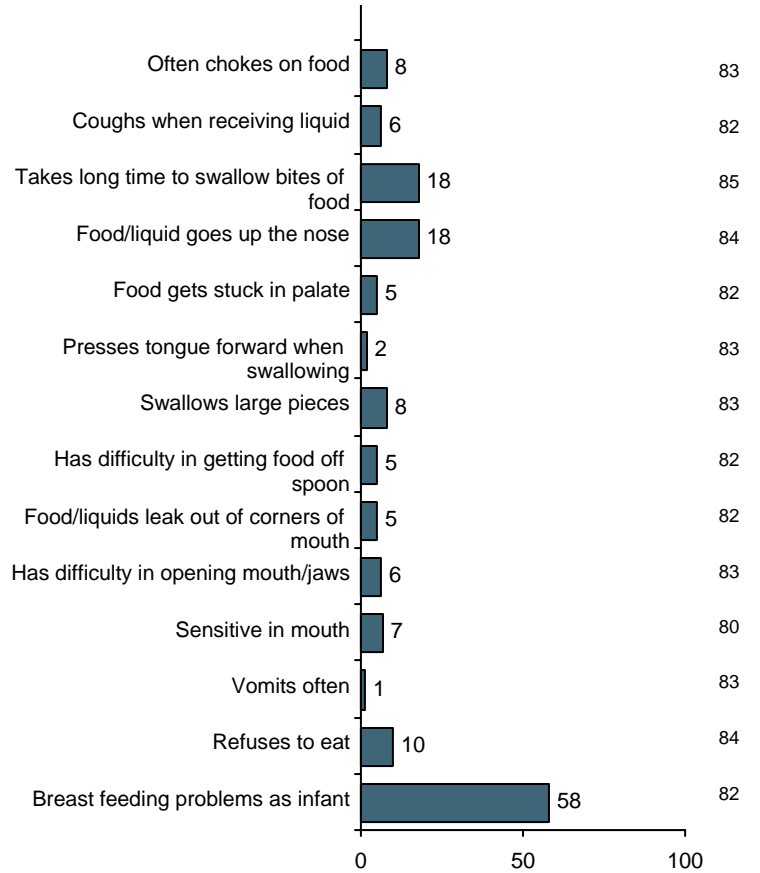
If help is needed with brushing teeth, how does it proceed?



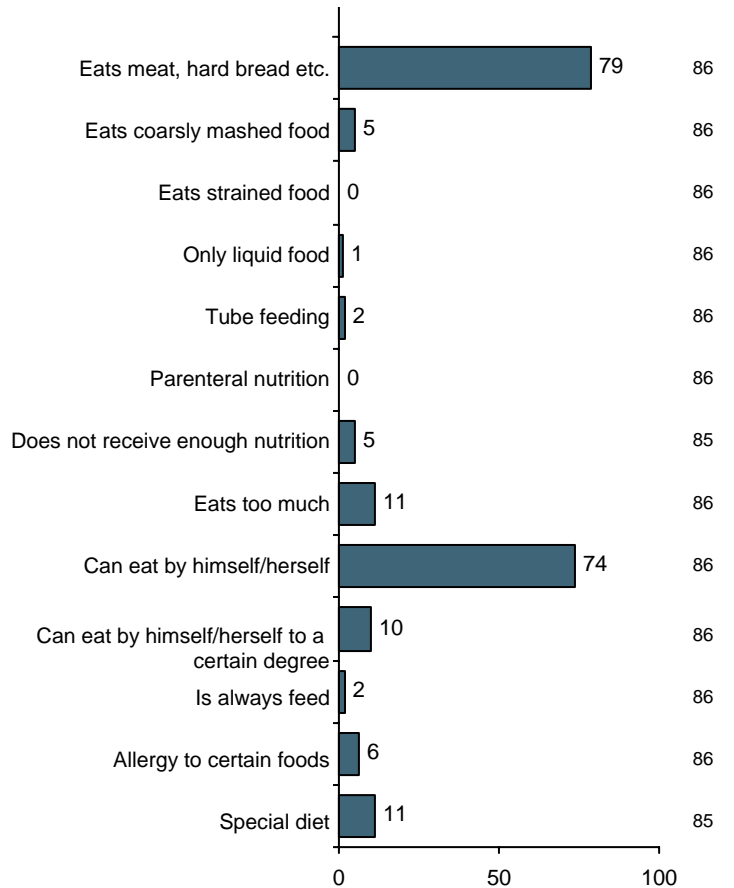
# About eating habits



## About eating habits - problems



## Food habits





# About drooling

