



# Orofacial function of persons having 22q11 deletion syndrome

## Report from observation charts

The survey comprises 107 observation charts.

**Synonyms:** CATCH 22, 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:

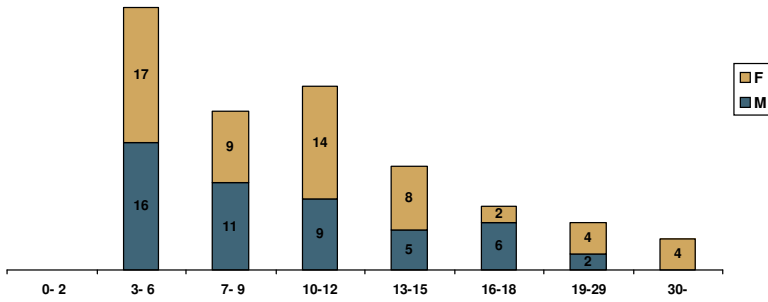
- It is important that these children come in early contact with the dental services for extra preventive dental care and information about oral hygiene. Frequent infections, poor nutrition, and poorly mineralized enamel all increase the risk of caries.
- In cases of defective palate, a specialist team 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

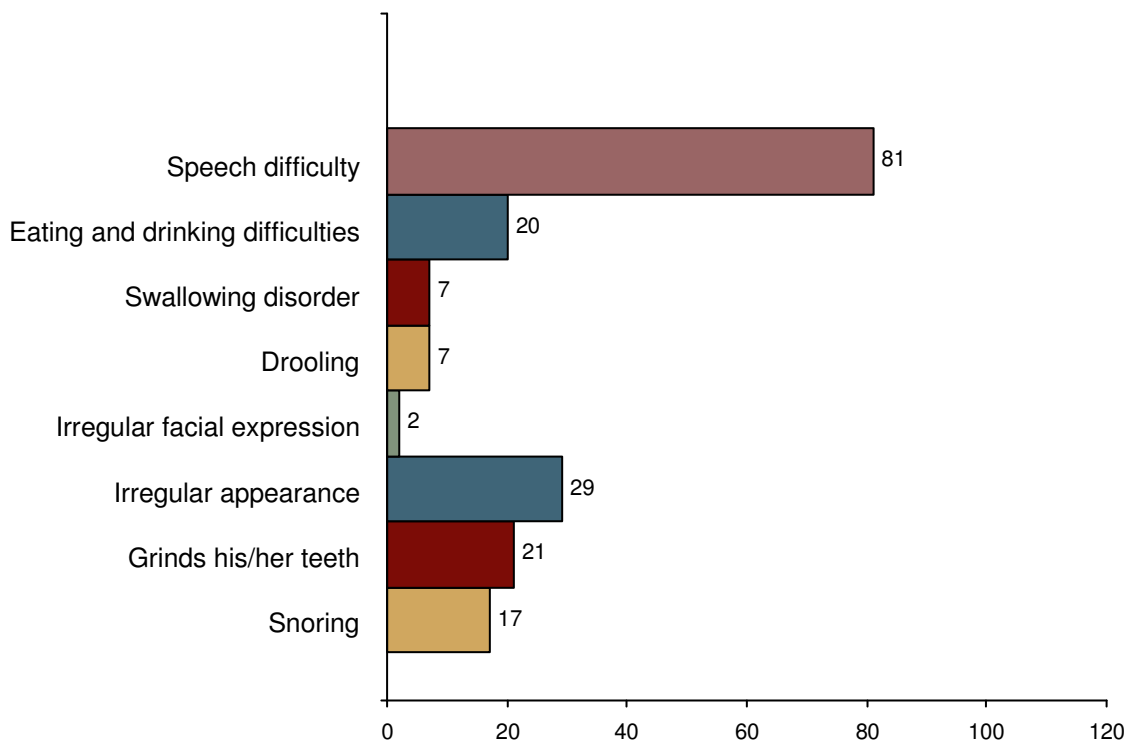


Number: 106

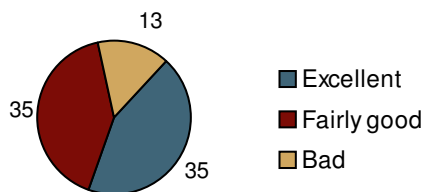
Ages: 3 -- 33 years

Sex: M (49) + F (57)

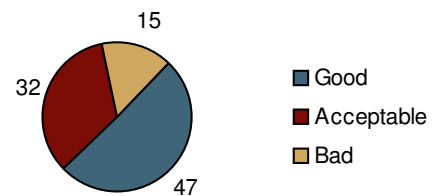
Orofacial problems



Oral health

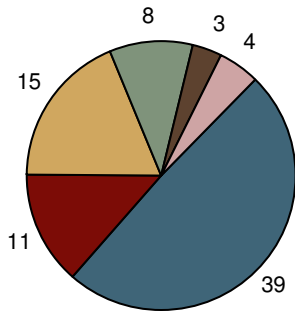


Oral hygiene



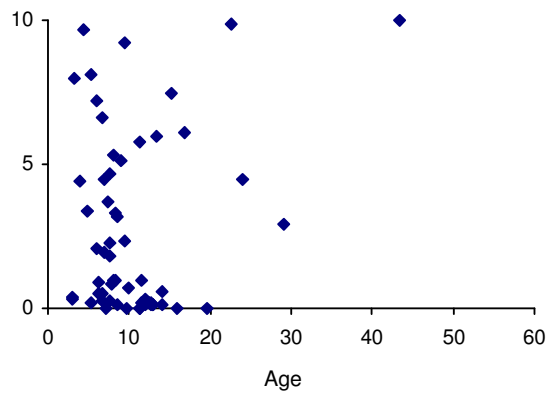
## Behaviour in the treatment situation

How calm and co-operative is the patient at time of examination?



- (39) ■ Examination is possible without problem
- (11) ■ Examination is possible without problems, some reaction is observed
- (15) ■ Examination can continue if adjusted to patient's reactions
- (8) ■ Reactions are considerable and examination is obviously affected
- (3) ■ Examination is practically impossible to complete
- (4) ■ Patient refuses examination

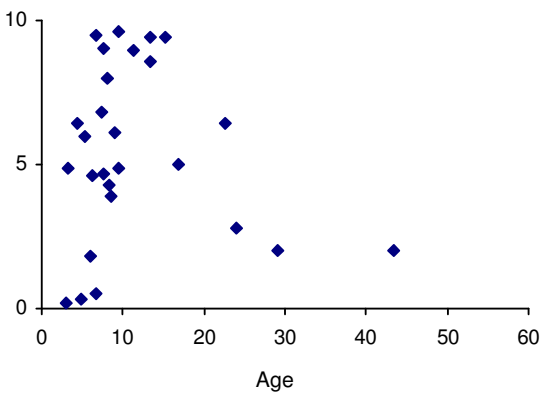
How does the patient cope with treatment in general?  
0=no problems/10=great problems



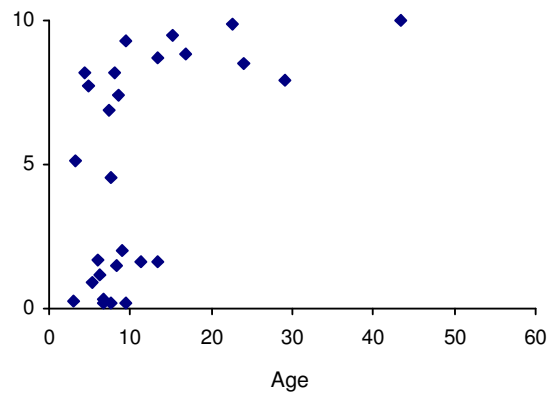
If there are treatment problems:

To what degree are the problems, if any, due to NN's handicap? To what degree are the problems, if any, due to NN's fear?

0=not at all/10=to a very high degree



0=not at all/10=to a very high degree



<b>Clinical findings</b>	<b>Total N=106</b>	<b>Boys/Men N=49</b>	<b>Girls/Women N=57</b>	<b>Not evaluated</b>
Speech difficulty	85	39	46	2
Hypomineralisation	46	16	30	11
Mask-like expression	33	18	15	9
Open mouth at rest	30	13	17	8
Gingivitis	27	12	15	9
Corner of mouth lowered	25	6	19	11
Post normal bite	24	12	12	13
Enamel hypoplasia	20	9	11	14
Frontal open bite	20	9	11	13
Other oral habits	20	8	12	11
Cross bite	19	10	9	11
Reduced mobility in soft palate	18	9	9	37
Dental trauma	15	7	8	11
High palate	14	7	7	13
Mouth breathing	14	7	7	15
Spacing	14	6	8	14
Over crowding	13	4	9	15
Grinds his/her teeth	12	1	11	14
Tooth anomaly	11	4	7	16
Cleft palate	10	3	7	13
Abrasion - insignificant	9	4	5	11
Deviation on opening	9	5	4	16
Drooling	9	3	6	15
Hypodontia	9	4	5	22
Dry mouth	8	2	6	21
Horizontal over-bite 6 mm or more	8	3	5	15
Proclined upper incisors	8	2	6	14
Deep bite with gingival contact	7	3	4	13
Retroclined upper incisors	7	3	4	15
Deep bite without gingival contact	6	4	2	13
Facial asymmetry	6	3	3	13
Geographic tongue	6	2	4	11
Narrow palate	6	3	3	13
Reduced mobility in TMJ	6	3	3	15
Edge to edge bite	5	2	3	15
M mentalis is overactive	5	2	3	13
Mucous membrane change	5	1	4	10
Philtrum seems long	5	1	4	11
Philtrum seems short	5	1	4	10
Posturing forward	5	4	1	15
Reduced mobility in neck	5	1	4	9
Tongue plaque	5	1	4	10
Hyper-sensitive in oral cavity	4	3	1	10
Jaw cleft	4	1	3	16
Long face	4	2	2	12
Retroclined lower incisors	4	2	2	15
Upper jaw seems small	4	1	3	13
Wide palate	4	1	3	13