



Orofacial function of persons having 22q11 deletion syndrome

Report from observation charts

The survey comprises 147 observation charts.

Synonyms: CATCH 22, Di George syndrome, Velocardiofacial syndrome

Estimated incidence: 25:100 000 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, clubfoot, 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: Certain facial features are characteristic for the diagnosis. 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:

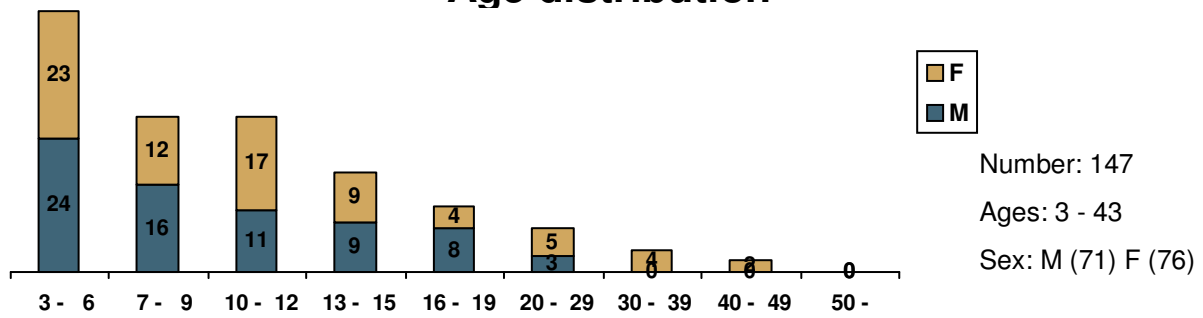
- It is important that these children come in early contact with the dental services for extra preventive dental care and information about oral hygiene. Frequent infections, poor nutrition, and poorly mineralized enamel all increase the risk of caries.
- X-ray to determine the presence of tooth buds 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.
- In cases of defective palate, a specialist team will be needed for follow up and treatment.
- When treating medically compromised patients always contact their doctors for medical advice (bleeding problems, heart diseases etc).
- Feeding and swallowing difficulties are investigated and treated by a specialist team at the hospital or multidisciplinary treatment center.

Source:

The rare disease database of the Swedish National Board of Health and Welfare.
The MHC database - The Mun-H-Center database on oral health and orofacial function in rare diseases.

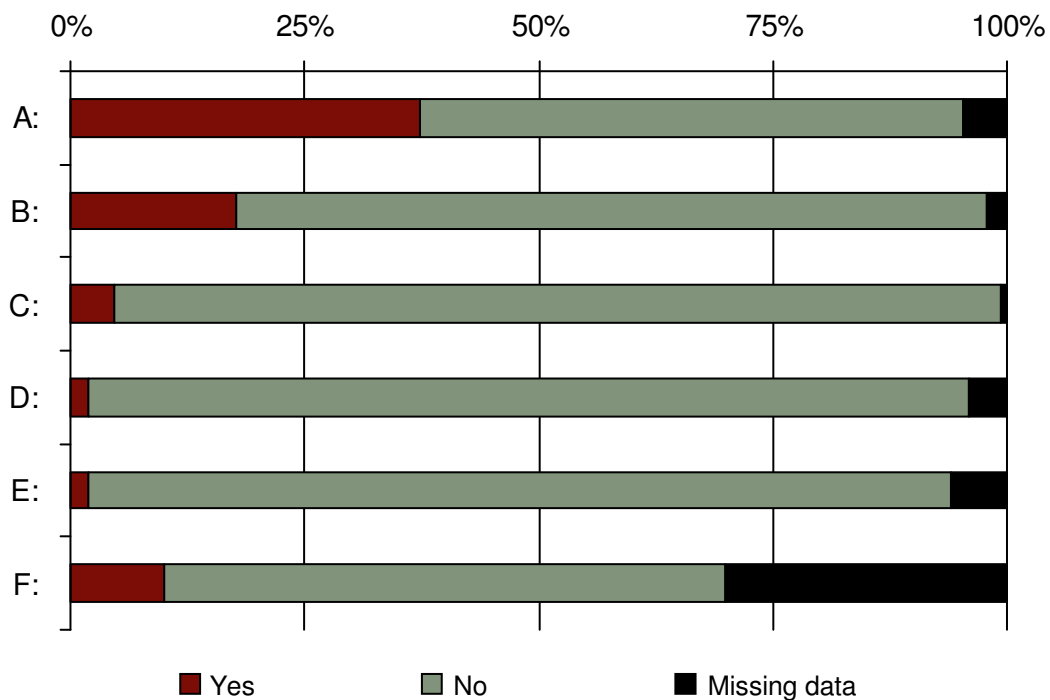
The Documentation from the Ågrenska Center.

Age distribution



Overview

	Yes	No	Missing data	N
A: Incomprehensible speech/No speech	55	85	7	147
B: Eating and drinking difficulties ¹	26	118	3	147
C: Profuse drooling, on clothes ¹	7	139	1	147
D: Breathing difficulties ^{1 2}	1	47	2	50
E: Grinding every day ^{1 2}	1	46	3	50
F: Severe malocclusions ²	5	30	15	50



1: Compiled using questionnaire

2: This variable was introduced in version 2 (2008) of the Observation chart.

Oral health

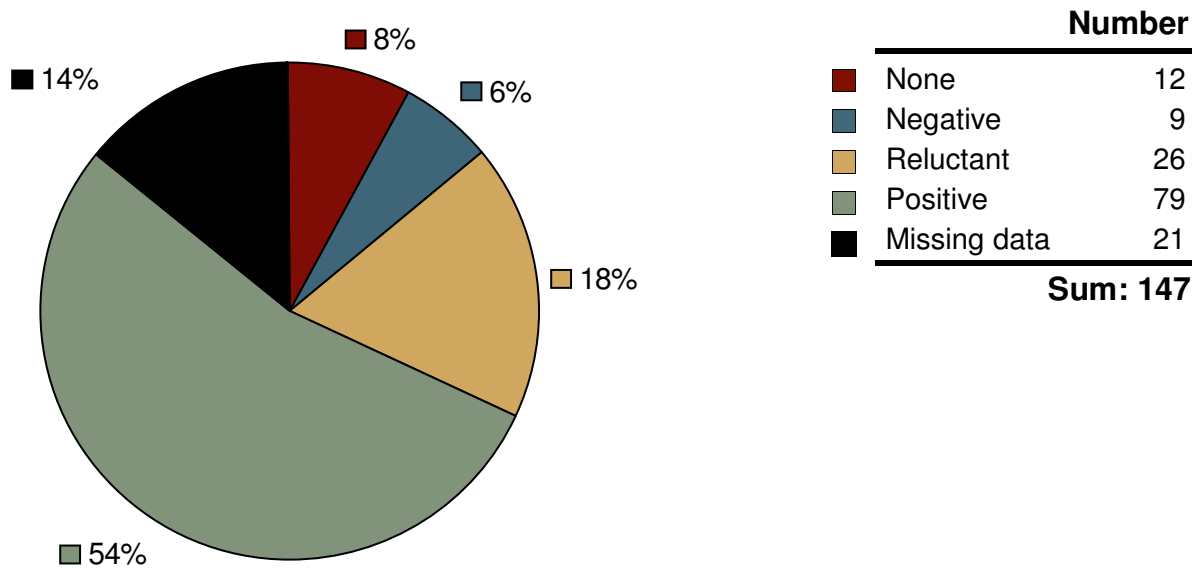
Oral health index (indices)¹

		0	1	2	3	4	5	6	Missing data	N
Calc	Calculus	34	0	0	1	1	0	1	13	50
GI	Gingivitis	23	2	3	0	2	0	5	15	50
Plaq	Coating	15	3	3	1	3	6	6	13	50
Toot	Tooth wear	23	7	6	0				14	50

- C Calculus index is based on the presence of visible calculus on the buccal surface of 6 index teeth. 0 indicates that there is no calculus at all, 6 indicates calculus on all index teeth.
- GI Gingivitis index is based on the presence of visible gingivitis on the buccal surface of 6 index teeth. 0 indicates that there is no bleeding, 6 indicates bleeding on all index teeth.
- PI Plaque index is based on the presence of visible plaque on the buccal surface of 6 index teeth. 0 indicates that there is no plaque, 6 indicates plaque on all index teeth.
- To Tooth wear index is a weighted summary of the degree of tooth wear on 6 different segments. Tooth wear is only evaluated in the permanent dentition, not in the primary teeth. The final index score is based on the degree of tooth wear found in most segments.
- 0: No tooth wear or minor wear of enamel in either of the segments
- 1: Marked tooth wear of the enamel, possibly exceeding into dentin
- 2: tooth wear in the dentine reaching up to 1/3 of the tooth crown
- 3: Tooth wear in the dentine reaching up to more than 1/3 of the tooth crown. If 3 is given in any segment then SI is 3.

¹: Oral health index (indices) was (were) introduced in the observations in 2008

Acceptance of dental examination



Caries

	3-6 years	7-12 years	13-19 years	Adults
deft¹				
Examined	38	40		
Number of individuals with deft=0	22	18		
Mean	1,7	2,2		
Standard deviation	3,2	2,8		
Missing data	9	16		
DMFT²				
Examined		42	25	14
Number of individuals with DMFT=0		27	9	1
Standard deviation		1,7	3,1	8,2
Mean		1,0	2,5	12,5
Missing data		14	5	0

1: Number of carious or filled deciduous teeth

2: Number of carious or filled permanent teeth

Occlusal relationship

	Number
Neutral bite	93
Post normal	32
Pre normal	4
Missing data	18
Sum: 147	

Maximum jaw opening

Children younger than 10 years

	Number
- 20	0
21 - 30	4
31 - 40	25
41 - 50	11
51 -	0
Missing data	35
Sum: 75	

Children, 10 years or older, and adults

	Number
- 20	0
21 - 30	2
31 - 40	17
41 - 50	22
51 -	7
Missing data	24
Sum: 72	

Profile¹

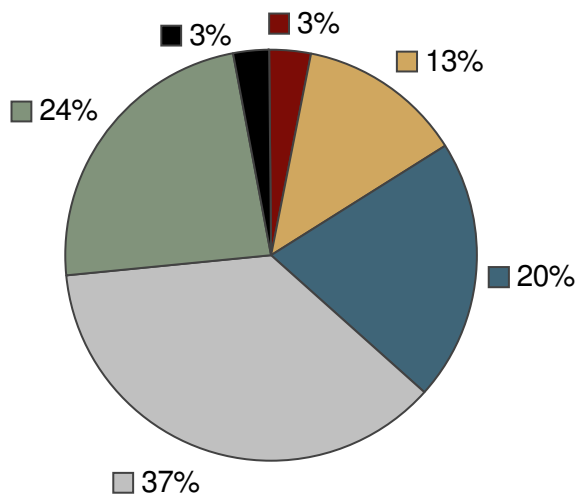
	Number
Normal	31
Convex	4
Concave	2
Missing data	13
Sum: 50	

Mandibular plane¹

	Number
Normal	25
Increased	3
Reduced	1
Missing data	21
Sum: 50	

1: This variable was introduced in version 2 (2008) of the Observation chart.

Speech difficulty



	Number
No speech	5
Very incomprehensible	19
Incomprehensible speech	30
Slightly indistinct speech	54
No problems	35
Missing data	4
Sum: 147	

Clinical findings	Yes-answers			
	Total N=147 (%)	Boys/Men N=71 (%)	Girls/Women N=76 (%)	Missing data
Open mouth at rest	45 (33)	22 (34)	23 (32)	9
Low muscle tone in lips	27 (20)	17 (27)	10 (14)	12
High palate	24 (18)	13 (22)	11 (16)	17
Frontal open bite	24 (19)	12 (21)	12 (17)	19
Cleft lip and palate	19 (15)	10 (17)	9 (13)	22
Over crowding	15 (12)	4 (7)	11 (16)	24
Spacing	14 (11)	5 (9)	9 (13)	22
Narrow palate	14 (11)	8 (14)	6 (9)	18
Deep bite with gingival contact	12 (9)	6 (10)	6 (9)	20
M mentalis overactive	11 (8)	4 (7)	7 (10)	17
Mucous membrane changes	9 (7)	4 (7)	5 (7)	17
Facial asymmetry	8 (6)	4 (7)	4 (6)	19
Reduced opening capacity	7 (6)	3 (6)	4 (6)	31
Impaired tongue motility	7 (6)	4 (7)	3 (4)	21
Low muscle tone in masticatory muscles	7 (6)	4 (7)	3 (5)	30
Intra oral hypo-sensitivity	4 (3)	3 (5)	1 (1)	15
Cranio-facial abnormality	4 (3)	2 (3)	2 (3)	19

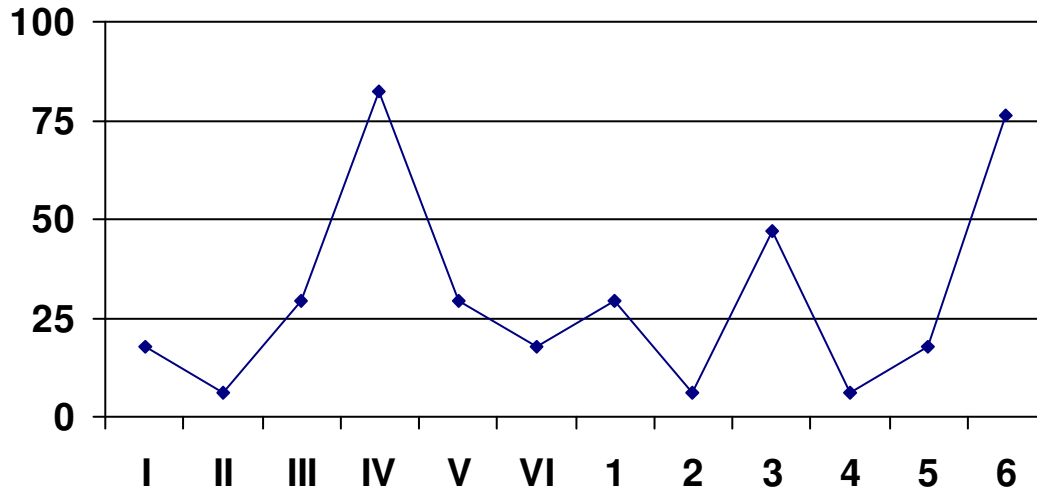
NOT-S

Total mean score: 3,41

Number: 17

Ages: 4 - 28

Sex: M (11) F (6)



NOT-S interview	Number	%
I : Sensory function	3	17,6
II : Breathing	1	5,9
III : Habits	5	29,4
IV : Chewing and swallowing	14	82,4
V : Drooling	5	29,4
VI : Dry mouth	3	17,6

NOT-S examination	Number	%
1 : Face at rest	5	29,4
2 : Nose breathing	1	5,9
3 : Facial expression	8	47,1
4 : Masticatory muscle and jaw function	1	5,9
5 : Oral motor function	3	17,6
6 : Speech	13	76,5