



Orofacial function of persons having 22q11 deletion syndrome

Report from questionnaires

The survey comprises 53 questionnaires.

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:

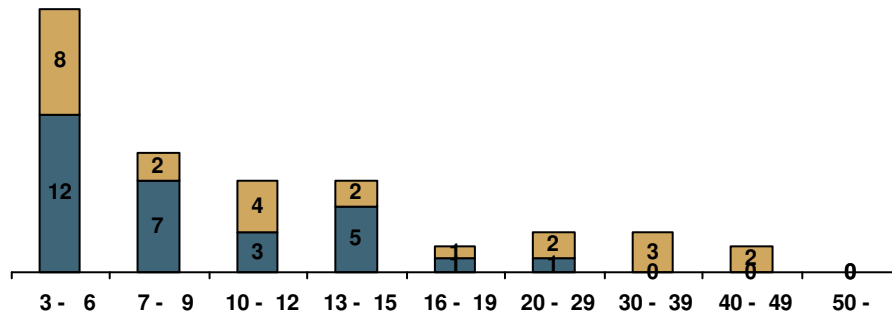
- It is important that these children come in early contact with the dental services for extra preventive dental care and information about oral hygiene. Frequent infections, poor nutrition, and poorly mineralized enamel all increase the risk of caries.
- X-ray to determine the presence of tooth buds 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



Number: 53

Ages: 3 - 44

Sex: M (29) F (24)

Additional diagnoses

Medical impairment	Yes	No	Missing data
Inborn heart defect	29	23	1
Other cardiovascular disease	2	47	4
Epilepsy	3	49	1
Asthma	15	37	1
Need of respiratory support	1	50	2
Allergy	16	36	1

Neuropsychiatric diagnosis	Yes	No	Missing data
ADHD/ADD	11	37	5
Autistic syndrome	7	41	5
Autistic traits	12	36	5
Asperger's syndrome	0	48	5
Tourette's syndrome	0	49	4

General disability	Yes	No	Missing data
Intellectual disability	16	27	10
Mobility dysfunction	19	21	13
Impaired vision	11	30	12
Impaired hearing	14	28	11
Difficulty in communicating	20	18	15

About dental care and oral health

Do you feel that you receive the dental care you need?

	Number
Yes, very much so	40
Yes, some what	9
No, not really	2
No, not at all	0
Missing data	2
Sum:	53

How many times per year do you normally seek dental care?

	Number
Three or more times per year	25
Twice per year	11
Once per year	13
Less than once per year	3
Missing data	1
Sum:	53

When were your teeth last X-rayed?

	Number
During the past two years	36
More than two years ago	3
Never had my teeth X-rayed	11
Missing data	3
Sum:	53

Do you look after your teeth in a good way?

	Number
Yes, very much so	26
Yes, some what	26
No, not really	1
No, not at all	0
Missing data	0
Sum:	53

Who brushes your teeth?

	Number
I always brush myself	17
Sometimes I brush myself	14
Someone else always helps me	21
Missing data	1
Sum:	53

How often are your teeth brushed?

	Number
Three or more times per day	3
Twice per day	45
Once per day	4
Not every day	0
Missing data	1
Sum:	53



About dental care and oral health

	Yes	No	Missing data
Does your mouth hurt?	4	44	5
Does your mouth feel dry?	10	34	9
Have you ever taken a serious hit to your permanent front teeth?	7	44	2
Do you feel that you have a divergent bite?	7	43	3
Have you had a brace?	5	47	1
Do you feel that you need orthodontics/a brace?	5	37	11

Do you grind or press your teeth at night?

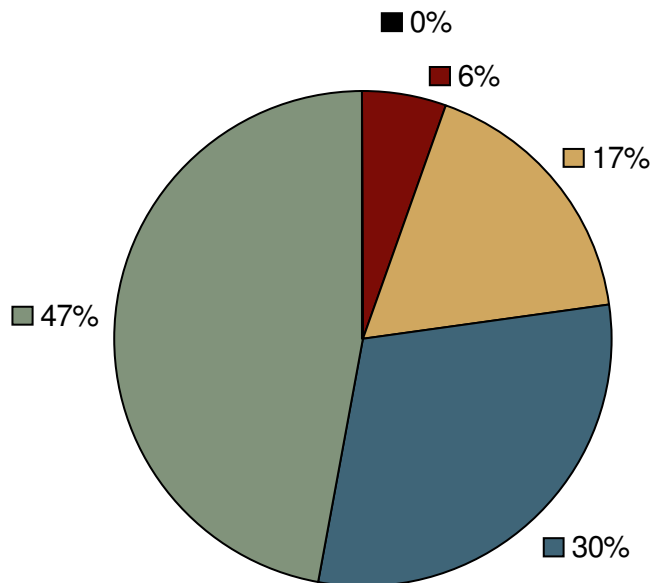
	Number
Never	27
Once or twice per week	15
Every night	8
Missing data	3
Sum:	53

Do you grind or press your teeth during the day?

	Number
Never	46
Once or twice per week	4
Every day	3
Missing data	0
Sum:	53

About eating

Do you have any problems with eating?



	Number
Yes, very much so	3
Yes, some what	9
No, not really	16
No, not at all	25
Missing data	0
Sum:	53

	Yes	No	Missing data
Do you cough daily in connection with meals?	3	49	1
Do you gag daily in connection with meals?	5	48	0
Do you get acid reflux daily?	3	43	7
Do you throw up often (at least twice per week)?	0	52	1
Do you have a poor appetite?	10	38	5
Does it take a long time before you can swallow a mouthful?	14	38	1
Do you press your tongue forward when you swallow so that food ends up outside the mouth?	2	47	4
Do you find it difficult to chew, i.e. grind food using your molars?	9	38	6
Do you find it difficult to take food from the spoon using your lips?	3	49	1
Have you had problems with food and drink leaking out through the corners of your mouth?	5	48	0
Does food tend to remain in your mouth after meals?	7	44	2
Do you get nutrition in any other way than through your mouth?	9	43	1



About drooling

Do you drool?

	Number
Never drool	40
Drool sometimes – not every day	9
Drool often – every day	1
Constant drooling	0
Missing data	3
Sum:	53

How much do you drool?

	Number
Slight drooling, only on the lips	5
Moderate drooling, on lip and chin	3
Profuse drooling, on clothes	1
Very profuse drooling, on hands/objects	1
Missing data	0
Sum:	10

Is your drooling a problem for you?

	Number
Yes, very much so	0
Yes, some what	2
No, not really	3
No, not at all	5
Missing data	0
Sum:	10

Is your drooling a problem for your family or people around you?

	Number
Yes, very much so	0
Yes, some what	1
No, not really	5
No, not at all	4
Missing data	0
Sum:	10