



Orofacial function of persons having Angelman syndrome

Report from questionnaires

The survey comprises 30 questionnaires.

Estimated occurrence: 8:100 000 live births.

Etiology: Genetic origin, often spontaneous mutation. Most of these children have some deletion of chromosomal material in the chromosome 15 inherited from the mother. The diagnosis is often decided according to well established international clinical criteria.

General symptoms: Delayed psychomotor development. Muscle laxity and balance problems. Severe mental retardation. Epilepsy. Abnormal language development, where understanding is often superior to the ability to express things in speech or using alternative means of communication. Other characteristics are hyperactivity and stereotypic behavior.

Orofacial/odontological symptoms: There is a particular appearance characteristic for the diagnosis. Widely-spaced teeth are common, as are malocclusions in the form of a post-normal or prenatal bite and an open, frontal bite. Daytime tooth grinding is common. Many like to suck and bite their hands and objects. Oral motor skills are poorly developed, and there is muscular laxity (hypotonia). Sucking, eating and drinking difficulties and drooling are all frequent problems. Most individuals with Angelman syndrome never acquire speech.

Orofacial/odontological treatment:

- It is important that individuals with this diagnosis receive dental preventive treatment, with extra prophylactic care and information on oral hygiene from an early age.
- Regular check-ups of dental and jaw development. Orthodontist should be consulted when needed.
- Tooth grinding should be followed up, and be managed with a splint when necessary.
- Oral motor skill training and extra stimulation are justified.
- Communication skills training is frequently essential.

Source:

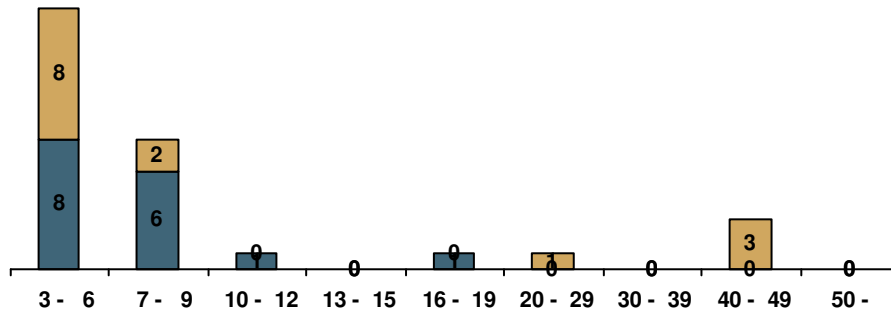
The rare disease database of the Swedish National Board of Health and Welfare.

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: 30
 Ages: 3 - 46
 Sex: M (16) F (14)

Additional diagnoses

Medical impairment	Yes	No	Missing data
Inborn heart defect	0	29	1
Other cardiovascular disease	0	29	1
Epilepsy	21	8	1
Asthma	4	25	1
Need of respiratory support	0	29	1
Allergy	6	23	1

Neuropsychiatric diagnosis	Yes	No	Missing data
ADHD/ADD	0	28	2
Autistic syndrome	3	26	1
Autistic traits	9	18	3
Asperger's syndrome	0	28	2
Tourette's syndrome	0	29	1

General disability	Yes	No	Missing data
Intellectual disability	3	0	27
Mobility dysfunction	3	1	26
Impaired vision	3	18	9
Impaired hearing	1	29	0
Difficulty in communicating	1	0	29

About dental care and oral health

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

	Number
Yes, very much so	12
Yes, some what	16
No, not really	2
No, not at all	0
Missing data	0
Sum:	30

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

	Number
Three or more times per year	8
Twice per year	9
Once per year	8
Less than once per year	4
Missing data	1
Sum:	30

When were your teeth last X-rayed?

	Number
During the past two years	5
More than two years ago	4
Never had my teeth X-rayed	20
Missing data	1
Sum:	30



Do you look after your teeth in a good way?

	Number
Yes, very much so	9
Yes, some what	17
No, not really	4
No, not at all	0
Missing data	0
Sum:	30

Who brushes your teeth?

	Number
I always brush myself	0
Sometimes I brush myself	1
Someone else always helps me	29
Missing data	0
Sum:	30

How often are your teeth brushed?

	Number
Three or more times per day	0
Twice per day	21
Once per day	6
Not every day	3
Missing data	0
Sum:	30



About dental care and oral health

	Yes	No	Missing data
Does your mouth hurt?	0	20	10
Does your mouth feel dry?	1	20	9
Have you ever taken a serious hit to your permanent front teeth?	5	25	0
Do you feel that you have a divergent bite?	9	14	7
Have you had a brace?	0	28	2
Do you feel that you need orthodontics/a brace?	0	24	6

Do you grind or press your teeth at night?

	Number
Never	15
Once or twice per week	8
Every night	1
Missing data	6
Sum:	30

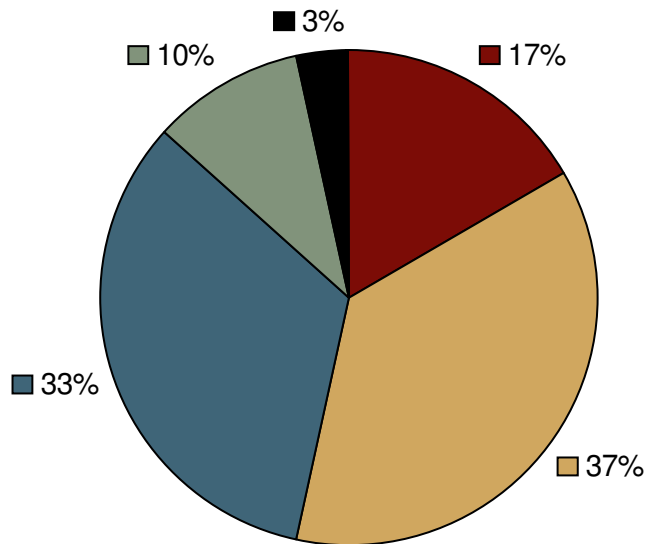
Do you grind or press your teeth during the day?

	Number
Never	16
Once or twice per week	10
Every day	1
Missing data	3
Sum:	30



About eating

Do you have any problems with eating?



	Number
Yes, very much so	5
Yes, some what	11
No, not really	10
No, not at all	3
Missing data	1
Sum:	30

	Yes	No	Missing data
Do you cough daily in connection with meals?	6	24	0
Do you gag daily in connection with meals?	4	25	1
Do you get acid reflux daily?	4	21	5
Do you throw up often (at least twice per week)?	4	26	0
Do you have a poor appetite?	5	24	1
Does it take a long time before you can swallow a mouthful?	2	25	3
Do you press your tongue forward when you swallow so that food ends up outside the mouth?	11	15	4
Do you find it difficult to chew, i.e. grind food using your molars?	15	13	2
Do you find it difficult to take food from the spoon using your lips?	10	20	0
Have you had problems with food and drink leaking out through the corners of your mouth?	16	13	1
Does food tend to remain in your mouth after meals?	10	17	3
Do you get nutrition in any other way than through your mouth?	2	28	0



About drooling

Do you drool?

	Number
Never drool	1
Drool sometimes – not every day	7
Drool often – every day	11
Constant drooling	8
Missing data	3
Sum:	30

How much do you drool?

	Number
Slight drooling, only on the lips	1
Moderate drooling, on lip and chin	12
Profuse drooling, on clothes	8
Very profuse drooling, on hands/objects	5
Missing data	0
Sum:	26

Is your drooling a problem for you?

	Number
Yes, very much so	3
Yes, some what	9
No, not really	9
No, not at all	2
Missing data	3
Sum:	26

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

	Number
Yes, very much so	3
Yes, some what	11
No, not really	11
No, not at all	1
Missing data	0
Sum:	26