



# Orofacial function of persons having Angelman syndrome

## Report from observation charts

The survey comprises 94 observation charts.

**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.

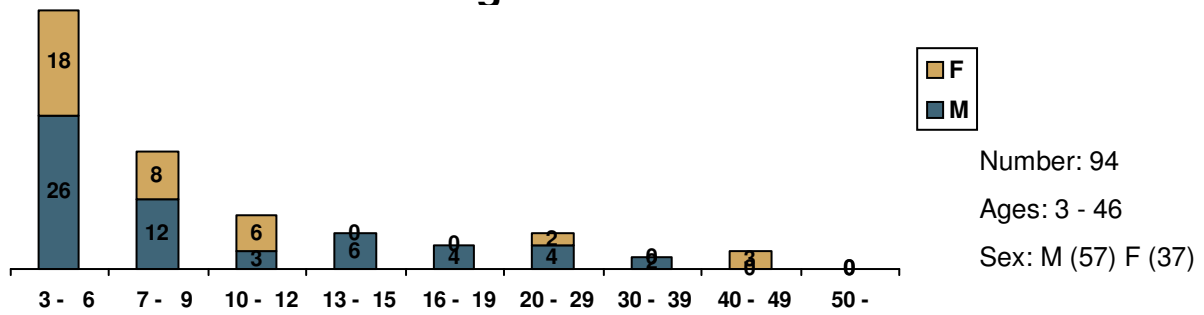
### Source:

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.

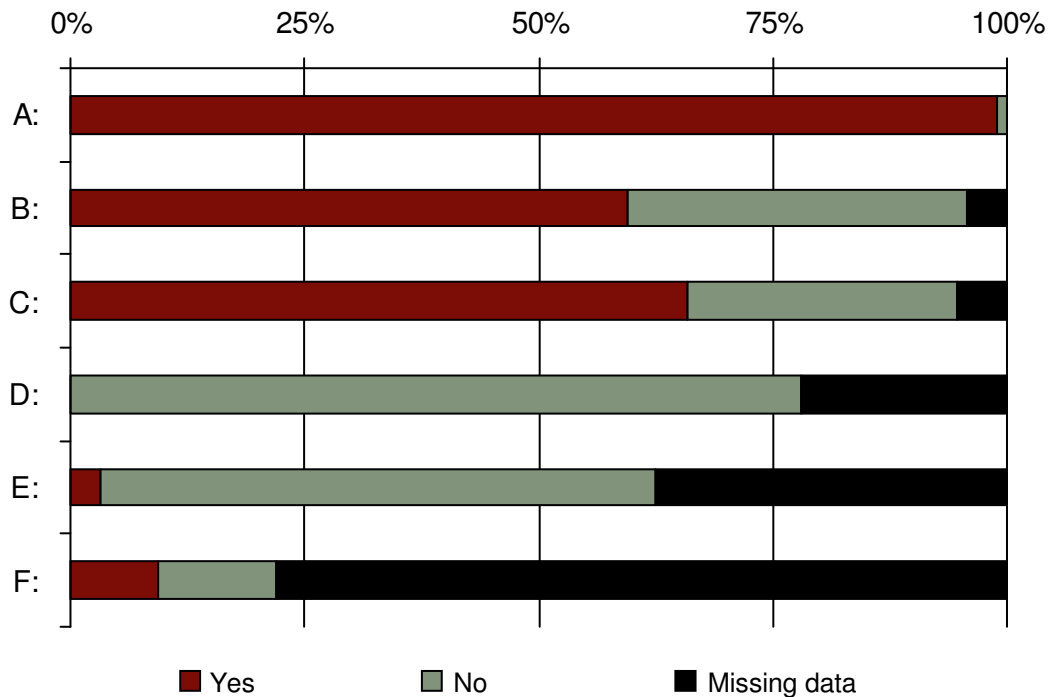
The Documentation from the Ågrenska Center.

### Age distribution



### Overview

	Yes	No	Missing data	N
A: Incomprehensible speech/No speech	93	1	0	94
B: Eating and drinking difficulties <sup>1</sup>	56	34	4	94
C: Profuse drooling, on clothes <sup>1</sup>	62	27	5	94
D: Breathing difficulties <sup>1 2</sup>	0	25	7	32
E: Grinding every day <sup>1 2</sup>	1	19	12	32
F: Severe malocclusions <sup>2</sup>	3	4	25	32



Note that the diagram is based upon less than 100 individuals.

1: Compiled using questionnaire

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



## Oral health

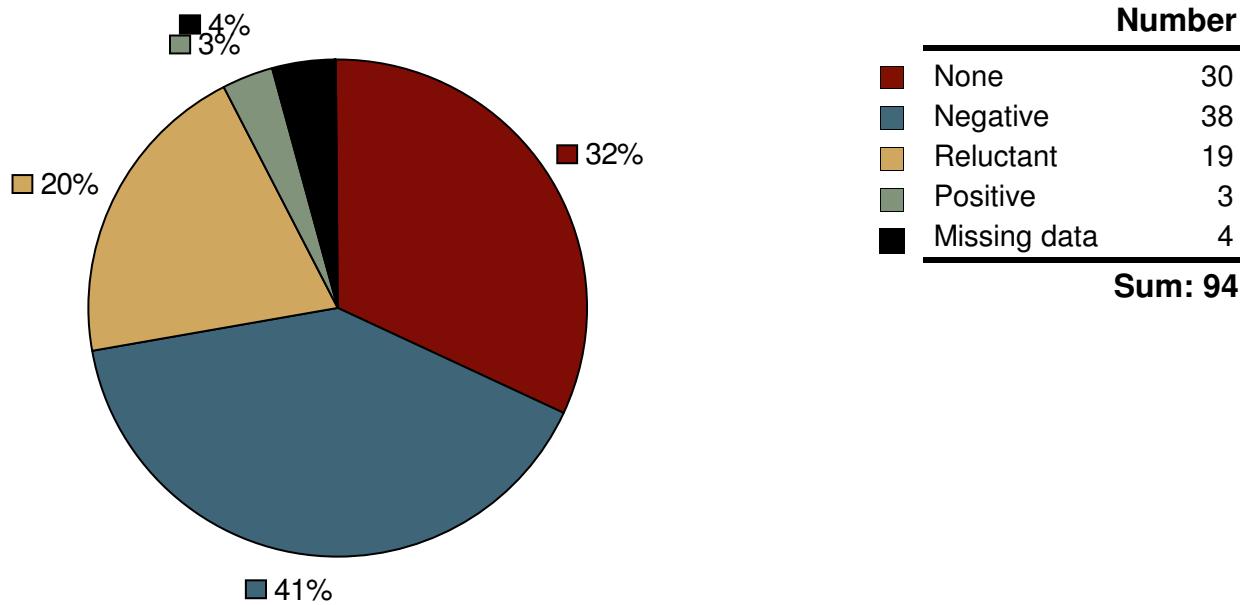
### Oral health index (indices)<sup>1</sup>

		0	1	2	3	4	5	6	Missing data	N
Calc	Calculus	5	0	0	0	0	0	0	27	32
GI	Gingivitis	4	0	0	0	0	2	0	26	32
Plaq	Coating	3	1	0	0	0	1	1	26	32
Toot	Tooth wear	3	1	0	0				28	32

- 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.

<sup>1</sup>: 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
<b>deft<sup>1</sup></b>				
Examined	26	13		
Number of individuals with deft=0	25	10		
Mean	0,1	0,5		
Standard deviation	0,6	1,1		
Missing data	18	16		
<b>DMFT<sup>2</sup></b>				
Examined		15	9	10
Number of individuals with DMFT=0		14	7	1
Standard deviation		1,5	0,7	3,2
Mean		0,4	0,3	3,2
Missing data		14	1	1

1: Number of carious or filled deciduous teeth

2: Number of carious or filled permanent teeth

## Occlusal relationship

	<b>Number</b>
Neutral bite	30
Post normal	16
Pre normal	25
Missing data	23
<b>Sum: 94</b>	

## Maximum jaw opening

Children younger than 10 years

	<b>Number</b>
- 20	0
21 - 30	0
31 - 40	6
41 - 50	5
51 -	0
Missing data	53
<b>Sum: 64</b>	

Children, 10 years or older, and adults

	<b>Number</b>
- 20	0
21 - 30	0
31 - 40	1
41 - 50	6
51 -	0
Missing data	23
<b>Sum: 30</b>	

## Profile<sup>1</sup>

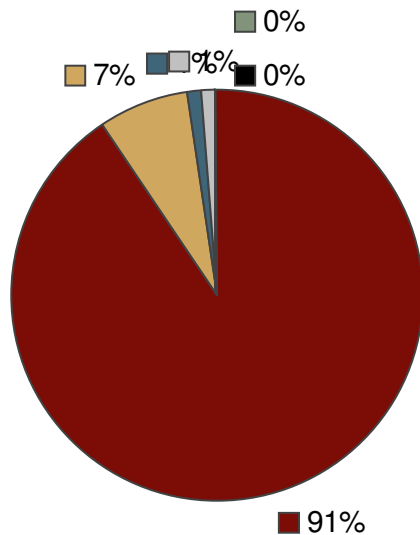
	<b>Number</b>
Normal	28
Convex	2
Concave	1
Missing data	1
<b>Sum: 32</b>	

## Mandibular plane<sup>1</sup>

	<b>Number</b>
Normal	9
Increased	3
Reduced	0
Missing data	20
<b>Sum: 32</b>	

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

### Speech difficulty



	<b>Number</b>
<span style="color: #800000;">■</span> No speech	85
<span style="color: #C08040;">■</span> Very incomprehensible	7
<span style="color: #408080;">■</span> Incomprehensible speech	1
<span style="color: #A0A0A0;">■</span> Slightly indistinct speech	1
<span style="color: #808080;">■</span> No problems	0
<span style="color: #000000;">■</span> Missing data	0
<b>Sum: 94</b>	

Clinical findings	Yes-answers			
	Total N=94 (%)	Boys/Men N=57 (%)	Girls/Women N=37 (%)	Missing data
Open mouth at rest	78 (83)	45 (79)	33 (89)	0
Low muscle tone in lips	70 (75)	37 (66)	33 (89)	1
Impaired tongue motility	61 (69)	35 (63)	26 (79)	5
Frontal open bite	48 (56)	24 (45)	24 (75)	9
Spacing	40 (48)	26 (51)	14 (44)	11
Low muscle tone in tongue	39 (45)	24 (44)	15 (47)	7
Narrow palate	10 (14)	4 (9)	6 (22)	22
High palate	8 (11)	2 (4)	6 (20)	19
Macroglossia	7 (8)	4 (7)	3 (9)	7
Low muscle tone in masticatory muscles	7 (11)	5 (12)	2 (9)	30
Reduced stability in neck	7 (8)	5 (9)	2 (6)	8
M mentalis overactive	6 (7)	4 (8)	2 (6)	6
Intra oral hypo-sensitivity	5 (7)	1 (2)	4 (13)	20
Facial asymmetry	4 (4)	2 (4)	2 (6)	4
High muscle tone in tongue	3 (4)	1 (2)	2 (6)	9
Mucous membrane changes	3 (4)	2 (5)	1 (4)	27