



## Orofacial function of persons having Spinal muscular atrophy Report from observation charts

The survey comprises 49 observation charts.

**Synonyms:** SMA I (Werdnig-Hoffmann disease, SMA II, SMA III (Kugelberg-Welander disease).

**Estimated occurrence:** 4-6 children diagnosed per year in Sweden (9000 000 inhabitants).

**Aeteiology:** Defect on the SMN1-gene on chromosome 5. SMA types I, II and III are inherited via autosomal recessive inheritance. Spinal muscular atrophy (SMA) is caused by destruction of the motor neurons in the diencephalon section of the brain, the medulla and the anterior horn cells of the spinal cord, resulting in muscular weakness and atrophy.

**General symptoms:** Muscular weakness and atrophy are most pronounced in the proximal musculature, including the chest, back and neck muscles. Intellectual development is not affected.

SMA I (Werdnig-Hoffmann disease): Symptoms from birth, or prior to the age of 6 months. Respiratory function is severely affected, and these children are at high risk from infections.

SMA II (known as the intermediate form): Symptoms present around the age of 6-18 months. Muscular weakness is often more pronounced in the legs than in the arms. Scoliosis (curvature of the spine) is common. Respiratory function may be affected.

SMA III (Kugelberg-Welander disease): Presents around the age of 2 years. Weak musculature in the trunk (proximal), back problems, and ambulatory difficulties are common and increase with age.

Sometimes, SMA 0 is used for the severe congenital type and SMA IV for the milder type with adult onset.

**Orofacial/odontological symptoms:** Infants with SMA I are unable, owing to their weak neck musculature, to lift their heads. Musculature weakness in the throat affects the ability to suck and to swallow. The musculature of the tongue is also weak, and fasciculations (slight trembling) in the tongue may occur. Adolescents and adults with SMA sometimes develop an impaired jaw opening capacity. Some have malocclusion

### **Orofacial/odontological treatment:**

- Early contact with dental services for intensified prophylactic care and oral hygiene information is essential.
- Regular check-ups of dental and jaw development. Orthodontist should be consulted when needed.
- When the jaws do not open properly, the function of the jaw joint should be investigated, and appropriate treatment thereafter prescribed.
- Feeding and swallowing difficulties are investigated and treated by a specialist team at the hospital or multidisciplinary treatment center.

### **Sources**

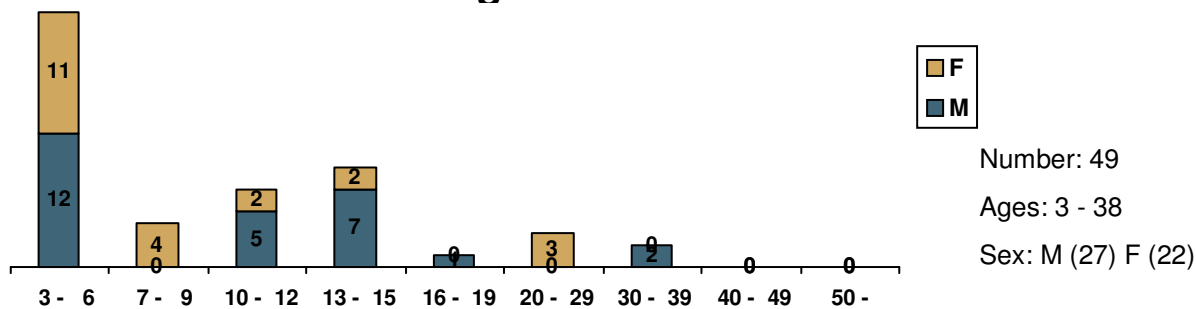
The rare disease database of the Swedish National Board of Health and Welfare.

The MHC database - The Mun-H-Center database on oral health and orofacial function in rare diseases.

The Documentation from the Ågrenska Center.

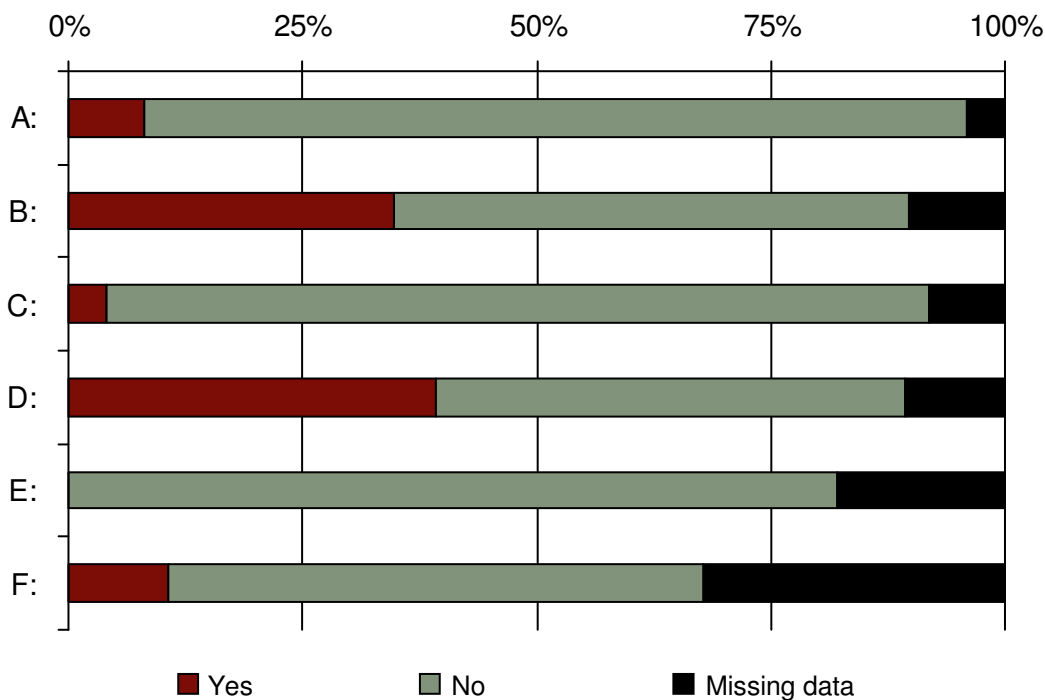


### Age distribution



### Overview

	Yes	No	Missing data	N
A: Incomprehensible speech/No speech	4	43	2	49
B: Eating and drinking difficulties <sup>1</sup>	17	27	5	49
C: Profuse drooling, on clothes <sup>1</sup>	2	43	4	49
D: Breathing difficulties <sup>1 2</sup>	11	14	3	28
E: Grinding every day <sup>1 2</sup>	0	23	5	28
F: Severe malocclusions <sup>2</sup>	3	16	9	28



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

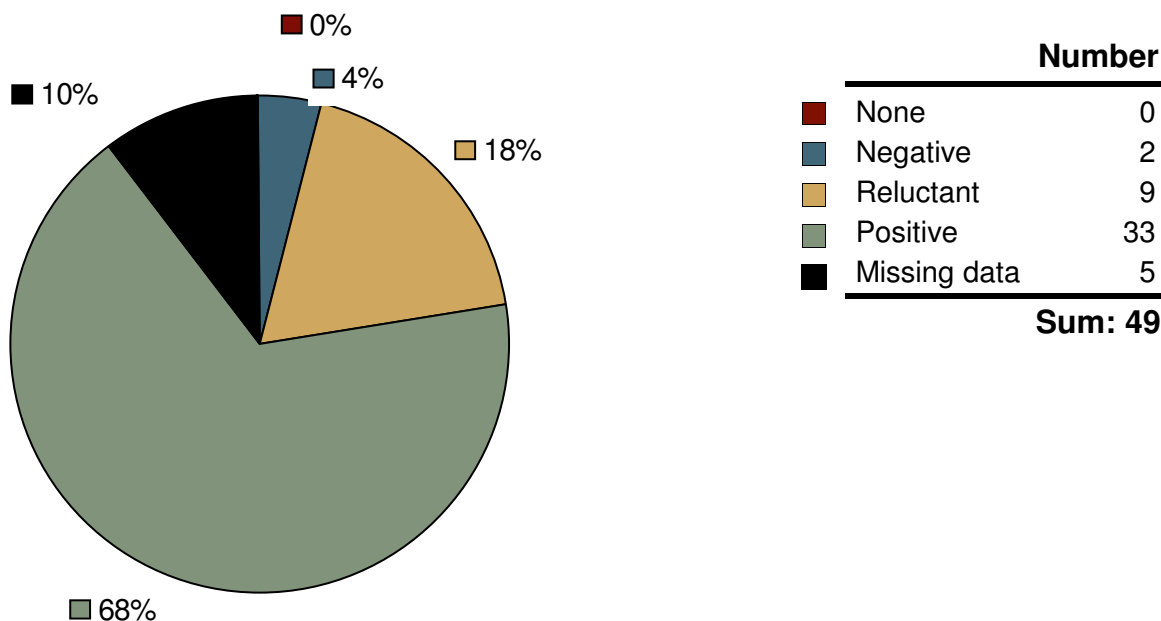
### Oral health index (indices)<sup>1</sup>

		0	1	2	3	4	5	6	Missing data	N
Calc	Calculus	17	1	3	0	0	0	0	7	28
GI	Gingivitis	14	1	5	1	0	0	0	7	28
Plaq	Coating	11	3	1	4	2	0	0	7	28
Toot	Tooth wear	20	0	0	0				8	28

- 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

	<u>3-6 years</u>	<u>7-12 years</u>	<u>13-19 years</u>	<u>Adults</u>
<b>deft<sup>1</sup></b>				
Examined	12	7		
Number of individuals with deft=0	12	4		
Mean	0,0	1,3		
Standard deviation	0,0	1,8		
Missing data	11	4		
<b>DMFT<sup>2</sup></b>				
Examined		9	8	2
Number of individuals with DMFT=0		6	7	0
Standard deviation		0,7	0,7	0,5
Mean		0,4	0,3	3,5
Missing data		2	2	3

1: Number of carious or filled deciduous teeth  
 2: Number of carious or filled permanent teeth

## Occlusal relationship

	<b>Number</b>
Neutral bite	28
Post normal	12
Pre normal	4
Missing data	5
<hr/>	
	<b>Sum: 49</b>

## Maximum jaw opening

Children younger than 10 years

	<b>Number</b>
- 20	0
21 - 30	4
31 - 40	6
41 - 50	7
51 -	2
Missing data	8
<hr/>	
	<b>Sum: 27</b>

Children, 10 years or older, and adults

	<b>Number</b>
- 20	6
21 - 30	4
31 - 40	5
41 - 50	5
51 -	0
Missing data	2
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	<b>Sum: 22</b>

## Profile<sup>1</sup>

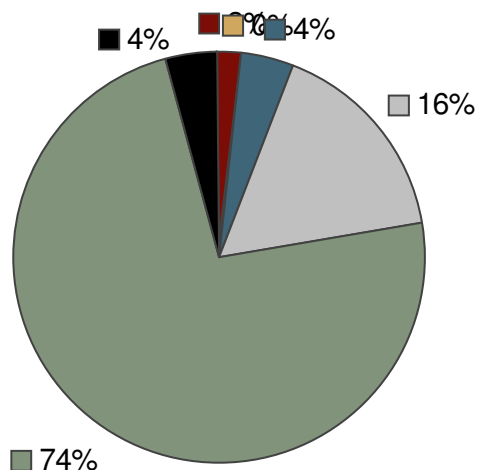
	<b>Number</b>
Normal	19
Convex	4
Concave	2
Missing data	3
<hr/>	
	<b>Sum: 28</b>

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

	<b>Number</b>
Normal	17
Increased	7
Reduced	0
Missing data	4
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	<b>Sum: 28</b>

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

### Speech difficulty



	<b>Number</b>
No speech	1
Very incomprehensible	0
Incomprehensible speech	2
Slightly indistinct speech	8
No problems	36
Missing data	2
<b>Sum: 49</b>	

Clinical findings	Yes-answers			
	Total N=49 (%)	Boys/Men N=27 (%)	Girls/Women N=22 (%)	Missing data
Reduced stability in neck	23 (48)	14 (52)	9 (43)	1
Reduced opening capacity	17 (37)	10 (40)	7 (33)	3
Impaired tongue motility	15 (31)	9 (35)	6 (27)	1
Frontal open bite	15 (33)	8 (33)	7 (33)	4
Open mouth at rest	13 (27)	6 (22)	7 (32)	0
Low muscle tone in tongue	13 (28)	7 (28)	6 (27)	2
Low muscle tone in lips	7 (15)	5 (19)	2 (10)	2
Low muscle tone in masticatory muscles	6 (14)	3 (13)	3 (15)	6
M mentalis overactive	5 (11)	3 (12)	2 (10)	2
Spacing	5 (11)	3 (13)	2 (10)	5
Narrow palate	4 (9)	1 (4)	3 (14)	5
Over crowding	4 (9)	3 (13)	1 (5)	5
Mucous membrane changes	4 (9)	3 (13)	1 (5)	4
High palate	3 (7)	0 (0)	3 (14)	5

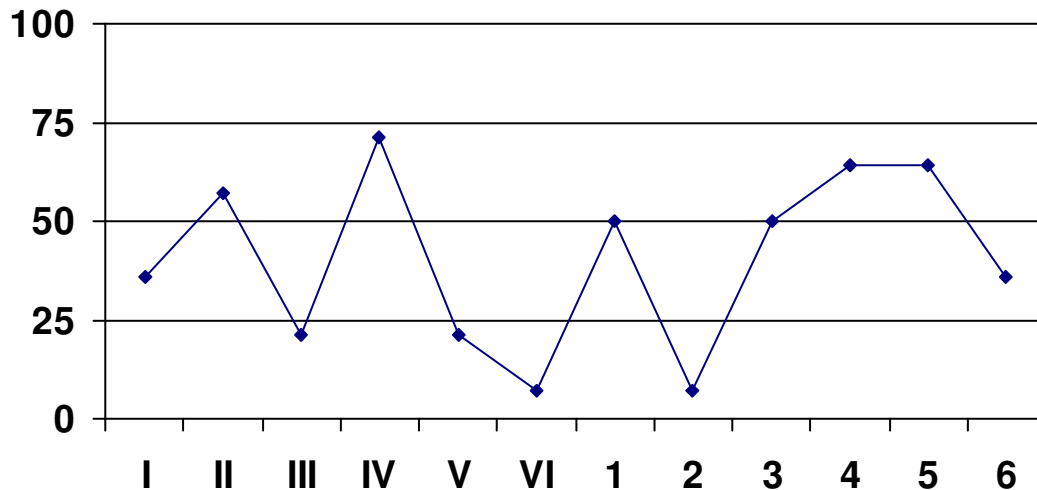
## NOT-S

Total mean score: 4,86

Number: 14

Ages: 3 - 38

Sex: M (7) F (7)



NOT-S interview	Number	%
I : Sensory function	5	35,7
II : Breathing	8	57,1
III : Habits	3	21,4
IV : Chewing and swallowing	10	71,4
V : Drooling	3	21,4
VI : Dry mouth	1	7,1

NOT-S examination	Number	%
1 : Face at rest	7	50,0
2 : Nose breathing	1	7,1
3 : Facial expression	7	50,0
4 : Masticatory muscle and jaw function	9	64,3
5 : Oral motor function	9	64,3
6 : Speech	5	35,7