



# Orofacial function of persons having Spinal muscular atrophy

## Report from questionnaires

The survey comprises 28 questionnaires.

**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.

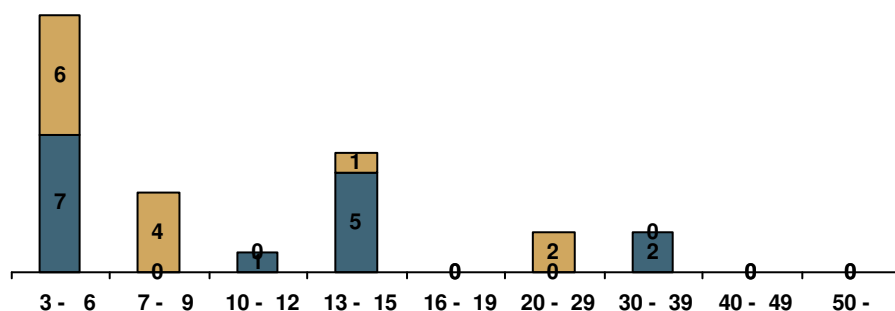
### Sources

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



Number: 28

Ages: 3 - 38

Sex: M (15) F (13)

## Additional diagnoses

Medical impairment	Yes	No	Missing data
Inborn heart defect	0	27	1
Other cardiovascular disease	0	28	0
Epilepsy	0	28	0
Asthma	3	25	0
Need of respiratory support	13	14	1
Allergy	5	23	0

Neuropsychiatric diagnosis	Yes	No	Missing data
ADHD/ADD	0	28	0
Autistic syndrome	0	28	0
Autistic traits	0	27	1
Asperger's syndrome	0	28	0
Tourette's syndrome	0	28	0

General disability	Yes	No	Missing data
Intellectual disability	0	28	0
Mobility dysfunction	1	6	21
Impaired vision	4	23	1
Impaired hearing	1	27	0
Difficulty in communicating	1	25	2

## About dental care and oral health

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

	Number
Yes, very much so	22
Yes, some what	5
No, not really	0
No, not at all	0
Missing data	1
<b>Sum:</b>	<b>28</b>

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

	Number
Three or more times per year	6
Twice per year	9
Once per year	8
Less than once per year	4
Missing data	1
<b>Sum:</b>	<b>28</b>

### When were your teeth last X-rayed?

	Number
During the past two years	12
More than two years ago	3
Never had my teeth X-rayed	9
Missing data	4
<b>Sum:</b>	<b>28</b>

## Do you look after your teeth in a good way?

	Number
Yes, very much so	15
Yes, some what	11
No, not really	1
No, not at all	0
Missing data	1
<b>Sum:</b>	<b>28</b>

## Who brushes your teeth?

	Number
I always brush myself	5
Sometimes I brush myself	11
Someone else always helps me	12
Missing data	0
<b>Sum:</b>	<b>28</b>

## How often are your teeth brushed?

	Number
Three or more times per day	3
Twice per day	20
Once per day	3
Not every day	1
Missing data	1
<b>Sum:</b>	<b>28</b>

## About dental care and oral health

	Yes	No	Missing data
Does your mouth hurt?	0	27	1
Does your mouth feel dry?	0	26	2
Have you ever taken a serious hit to your permanent front teeth?	3	24	1
Do you feel that you have a divergent bite?	6	21	1
Have you had a brace?	4	24	0
Do you feel that you need orthodontics/a brace?	4	23	1

## Do you grind or press your teeth at night?

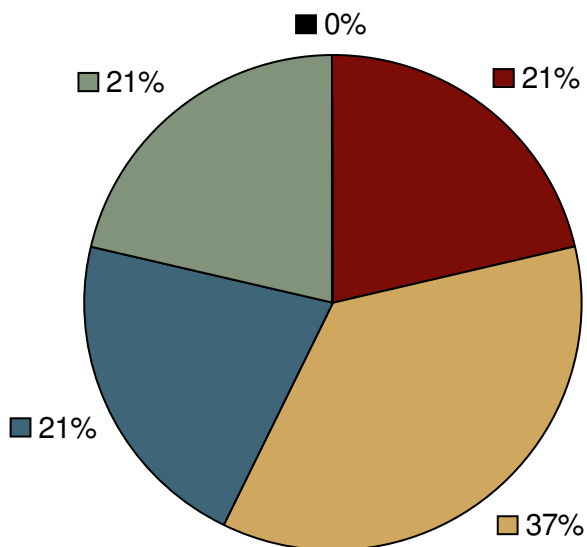
	Number
Never	25
Once or twice per week	3
Every night	0
Missing data	0
<b>Sum:</b>	<b>28</b>

## Do you grind or press your teeth during the day?

	Number
Never	25
Once or twice per week	3
Every day	0
Missing data	0
<b>Sum:</b>	<b>28</b>

## About eating

### Do you have any problems with eating?



	Number
Yes, very much so	6
Yes, some what	10
No, not really	6
No, not at all	6
Missing data	0
<b>Sum:</b>	<b>28</b>

	Yes	No	Missing data
Do you cough daily in connection with meals?	5	23	0
Do you gag daily in connection with meals?	2	26	0
Do you get acid reflux daily?	1	27	0
Do you throw up often (at least twice per week)?	1	27	0
Do you have a poor appetite?	4	23	1
Does it take a long time before you can swallow a mouthful?	11	16	1
Do you press your tongue forward when you swallow so that food ends up outside the mouth?	1	27	0
Do you find it difficult to chew, i.e. grind food using your molars?	10	18	0
Do you find it difficult to take food from the spoon using your lips?	5	23	0
Have you had problems with food and drink leaking out through the corners of your mouth?	2	26	0
Does food tend to remain in your mouth after meals?	2	25	1
Do you get nutrition in any other way than through your mouth?	10	18	0

## About drooling

### Do you drool?

	Number
Never drool	21
Drool sometimes – not every day	5
Drool often – every day	1
Constant drooling	0
Missing data	1
<b>Sum:</b>	<b>28</b>

### How much do you drool?

	Number
Slight drooling, only on the lips	3
Moderate drooling, on lip and chin	2
Profuse drooling, on clothes	0
Very profuse drooling, on hands/objects	0
Missing data	1
<b>Sum:</b>	<b>6</b>

### Is your drooling a problem for you?

	Number
Yes, very much so	0
Yes, some what	0
No, not really	4
No, not at all	1
Missing data	1
<b>Sum:</b>	<b>6</b>

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

	Number
Yes, very much so	0
Yes, some what	0
No, not really	1
No, not at all	4
Missing data	1
<b>Sum:</b>	<b>6</b>