



# Orofacial function of persons having Williams syndrome

## Report from questionnaires

The survey comprises 28 questionnaires.

**Synonyms:** William-Beuren syndrome.

**Estimated occurrence:** 5-10:100 000 live births.

**Etiology:** Deletion on chromosome 7, most often caused by a new mutation.

**General symptoms:** A congenital cardiac defect in the form of supravalvular aortic stenosis (SVAS) is common, and many children have raised levels of blood calcium (hypercalcemia). Individuals with Williams syndrome have varying degrees of intellectual disability, and delayed motor and language development. Most have a particular behaviour profile including anxiety in certain situations and autism and autistic traits are common. Squinting and other vision problems are frequent.

**Orofacial/odontological symptoms:** Characteristic facial features are associated with the diagnosis. Dental problems in the form of the absence of some tooth buds, irregular and/or small and/or widely-spaced teeth, enamel aberrations. Many of these children have some kind of malocclusion and muscle laxity in and around the mouth. Orofacial problems including sucking and chewing, drooling and over sensitivity of the oral cavity are frequent.

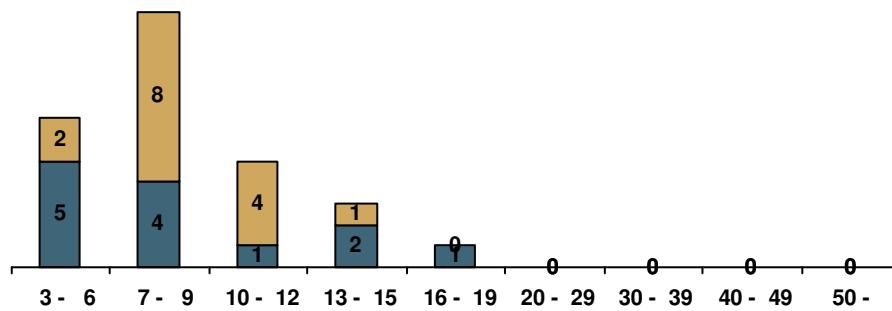
### **Orofacial/ odontological treatment:**

- 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.
- Feeding and swallowing difficulties are investigated and treated by a specialist team at the hospital or multidisciplinary treatment centre.
- Orofacial therapy and oral motor skills training and stimulation in cases of difficulties with eating, speech or drooling may be relevant.
- Speech, language and communication training are often required.
- When treating medically compromised patients always contact their doctors for medical advice.

### **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: 28

Ages: 4 - 19

Sex: M (13) F (15)

## Additional diagnoses

Medical impairment	Yes	No	Missing data
Inborn heart defect	21	5	2
Other cardiovascular disease	2	22	4
Epilepsy	1	27	0
Asthma	3	24	1
Need of respiratory support	0	28	0
Allergy	6	21	1

Neuropsychiatric diagnosis	Yes	No	Missing data
ADHD/ADD	1	25	2
Autistic syndrome	2	23	3
Autistic traits	9	17	2
Asperger's syndrome	0	26	2
Tourette's syndrome	0	26	2

General disability	Yes	No	Missing data
Intellectual disability	15	4	9
Mobility dysfunction	16	6	6
Impaired vision	10	14	4
Impaired hearing	4	23	1
Difficulty in communicating	13	12	3

## About dental care and oral health

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

	Number
Yes, very much so	18
Yes, some what	6
No, not really	1
No, not at all	0
Missing data	3
<b>Sum:</b>	<b>28</b>

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

	Number
Three or more times per year	14
Twice per year	6
Once per year	5
Less than once per year	2
Missing data	1
<b>Sum:</b>	<b>28</b>

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

	Number
During the past two years	14
More than two years ago	2
Never had my teeth X-rayed	10
Missing data	2
<b>Sum:</b>	<b>28</b>

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

	Number
Yes, very much so	14
Yes, some what	12
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	1
Sometimes I brush myself	11
Someone else always helps me	15
Missing data	1
<b>Sum:</b>	<b>28</b>

## How often are your teeth brushed?

	Number
Three or more times per day	0
Twice per day	25
Once per day	2
Not every day	0
Missing data	1
<b>Sum:</b>	<b>28</b>

## About dental care and oral health

	Yes	No	Missing data
Does your mouth hurt?	2	23	3
Does your mouth feel dry?	4	22	2
Have you ever taken a serious hit to your permanent front teeth?	2	25	1
Do you feel that you have a divergent bite?	11	15	2
Have you had a brace?	1	26	1
Do you feel that you need orthodontics/a brace?	9	17	2

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

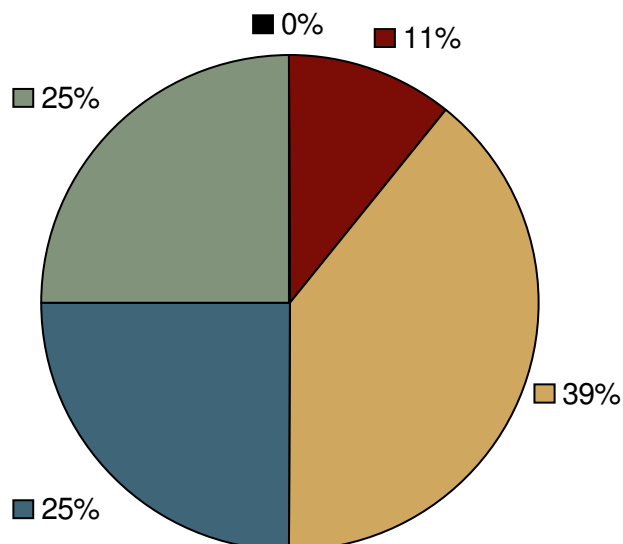
	Number
Never	26
Once or twice per week	1
Every night	0
Missing data	1
<b>Sum:</b>	<b>28</b>

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

	Number
Never	27
Once or twice per week	0
Every day	0
Missing data	1
<b>Sum:</b>	<b>28</b>

## About eating

### Do you have any problems with eating?



	Number
Yes, very much so	3
Yes, some what	11
No, not really	7
No, not at all	7
Missing data	0
<b>Sum:</b>	<b>28</b>

	Yes	No	Missing data
Do you cough daily in connection with meals?	1	25	2
Do you gag daily in connection with meals?	3	25	0
Do you get acid reflux daily?	1	26	1
Do you throw up often (at least twice per week)?	1	26	1
Do you have a poor appetite?	5	23	0
Does it take a long time before you can swallow a mouthful?	4	23	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?	8	15	5
Do you find it difficult to take food from the spoon using your lips?	3	24	1
Have you had problems with food and drink leaking out through the corners of your mouth?	4	22	2
Does food tend to remain in your mouth after meals?	0	27	1
Do you get nutrition in any other way than through your mouth?	0	28	0

## About drooling

### Do you drool?

	Number
Never drool	16
Drool sometimes – not every day	8
Drool often – every day	3
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	7
Profuse drooling, on clothes	0
Very profuse drooling, on hands/objects	0
Missing data	1
<b>Sum:</b>	<b>11</b>

### Is your drooling a problem for you?

	Number
Yes, very much so	0
Yes, some what	3
No, not really	3
No, not at all	4
Missing data	1
<b>Sum:</b>	<b>11</b>

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

	Number
Yes, very much so	1
Yes, some what	5
No, not really	2
No, not at all	2
Missing data	1
<b>Sum:</b>	<b>11</b>