



## Orofacial function of persons having Williams syndrome Report from observation charts

The survey comprises 88 observation charts.

**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.

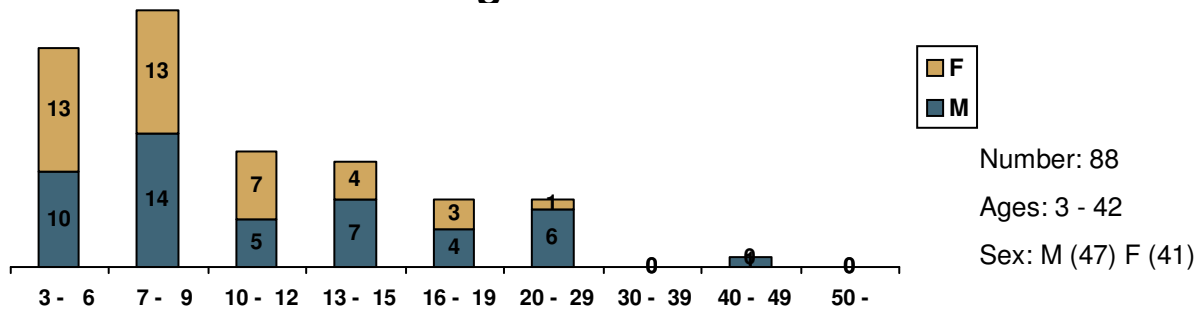
### **Orofacial/ odontological treatment:**

- 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:**

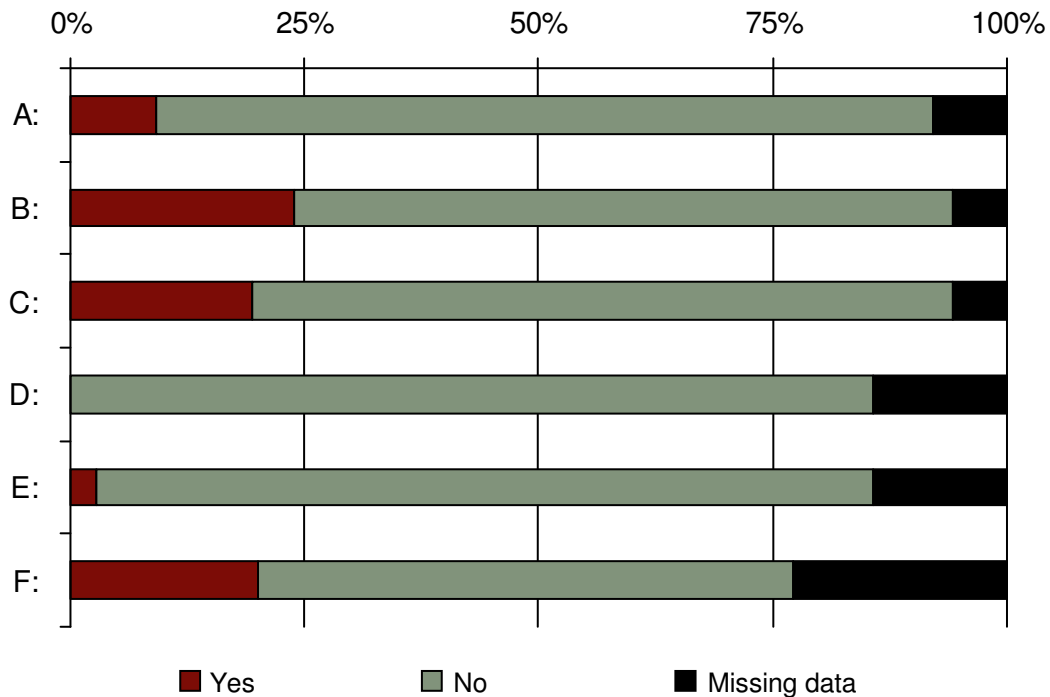
The rare disease database of the Swedish National Board of Health and Welfare.  
The MHC database - The Mun-H-Center database on oral health and orofacial function in rare diseases.  
The Documentation from the Ågrenska Center.

### Age distribution



### Overview

	Yes	No	Missing data	N
A: Incomprehensible speech/No speech	8	73	7	88
B: Eating and drinking difficulties <sup>1</sup>	21	62	5	88
C: Profuse drooling, on clothes <sup>1</sup>	17	66	5	88
D: Breathing difficulties <sup>1 2</sup>	0	30	5	35
E: Grinding every day <sup>1 2</sup>	1	29	5	35
F: Severe malocclusions <sup>2</sup>	7	20	8	35



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

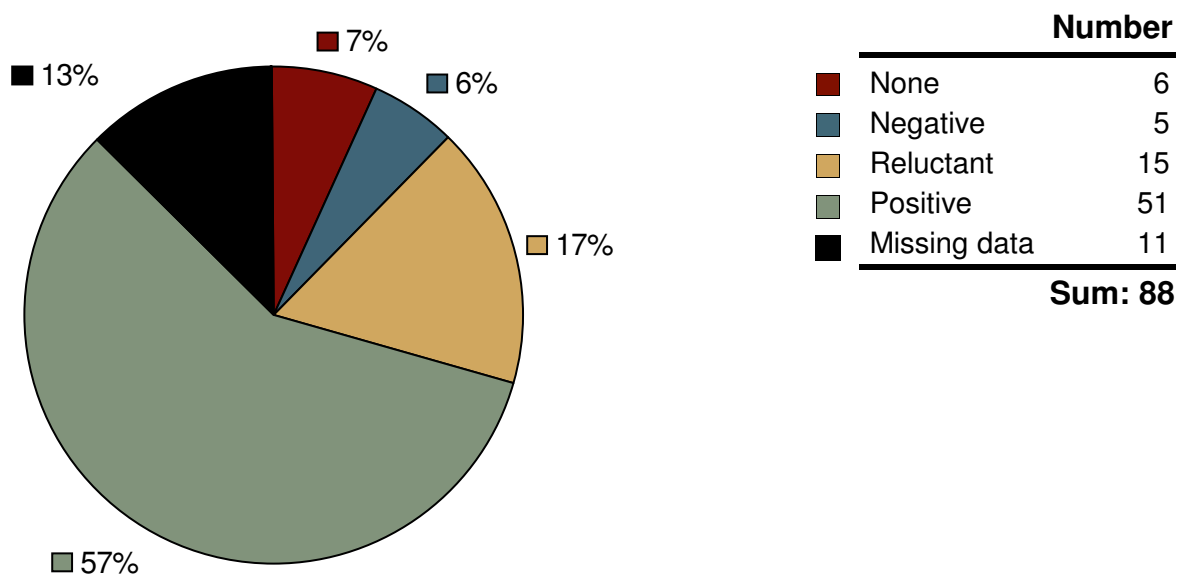
### Oral health index (indices)<sup>1</sup>

		0	1	2	3	4	5	6	Missing data	N
Calc	Calculus	22	0	0	0	0	0	0	13	35
GI	Gingivitis	15	3	0	2	1	0	0	14	35
Plaq	Coating	14	4	1	1	1	0	0	14	35
Toot	Tooth wear	20	1	0	0				14	35

- 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		19	27	
Number of individuals with deft=0		11	18	
Mean		1,2	1,2	
Standard deviation		1,7	2,2	
Missing data		4	12	
<b>DMFT<sup>2</sup></b>				
Examined		32	17	7
Number of individuals with DMFT=0		25	11	2
Standard deviation		1,3	1,4	4,3
Mean		0,6	0,8	3,7
Missing data		7	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	45
Post normal	17
Pre normal	20
Missing data	6
<b>Sum: 88</b>	

## Maximum jaw opening

Children younger than 10 years

	<b>Number</b>
- 20	0
21 - 30	2
31 - 40	18
41 - 50	9
51 -	1
Missing data	20
<b>Sum: 50</b>	

Children, 10 years or older, and adults

	<b>Number</b>
- 20	0
21 - 30	0
31 - 40	11
41 - 50	15
51 -	8
Missing data	4
<b>Sum: 38</b>	

## Profile<sup>1</sup>

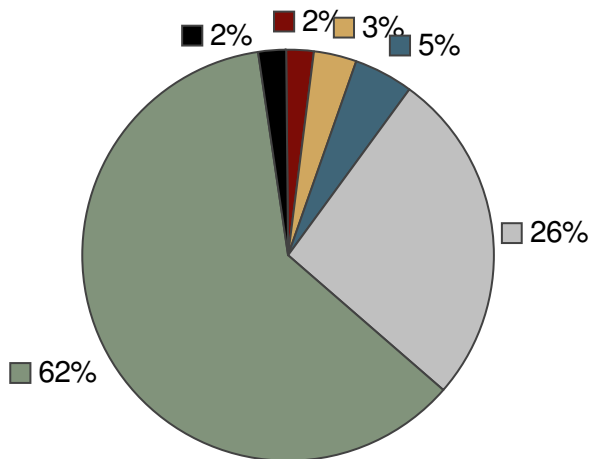
	<b>Number</b>
Normal	24
Convex	8
Concave	0
Missing data	3
<b>Sum: 35</b>	

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

	<b>Number</b>
Normal	10
Increased	12
Reduced	0
Missing data	13
<b>Sum: 35</b>	

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

## Speech difficulty



	<b>Number</b>
No speech	2
Very incomprehensible	3
Incomprehensible speech	4
Slightly indistinct speech	23
No problems	54
Missing data	2
<b>Sum: 88</b>	

Clinical findings	Yes-answers			
	Total N=88 (%)	Boys/Men N=47 (%)	Girls/Women N=41 (%)	Missing data
Open mouth at rest	65 (76)	36 (80)	29 (71)	2
Low muscle tone in lips	58 (67)	31 (67)	27 (68)	2
Spacing	33 (41)	20 (47)	13 (34)	7
Frontal open bite	23 (27)	15 (34)	8 (20)	4
High palate	20 (24)	11 (26)	9 (22)	4
M mentalis overactive	17 (20)	11 (26)	6 (15)	5
Narrow palate	14 (17)	9 (20)	5 (13)	4
Impaired tongue motility	12 (14)	11 (24)	1 (3)	2
Over crowding	12 (15)	10 (23)	2 (5)	7
Low muscle tone in masticatory muscles	11 (14)	8 (19)	3 (9)	11
Intra oral hypo-sensitivity	7 (8)	5 (11)	2 (5)	4
Facial asymmetry	7 (8)	5 (11)	2 (5)	4
Deep bite with gingival contact	5 (6)	3 (7)	2 (5)	5
Mucous membrane changes	5 (6)	5 (11)	0 (0)	8
High muscle tone in lips	4 (5)	2 (5)	2 (5)	8
High muscle tone in tongue	4 (5)	2 (4)	2 (5)	1
Short tongue frenulum	4 (5)	3 (7)	1 (2)	2
Macroglossia	3 (3)	2 (4)	1 (2)	1