



Orofacial function of persons having Williams syndrome Report from observation charts

The survey comprises 69 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.

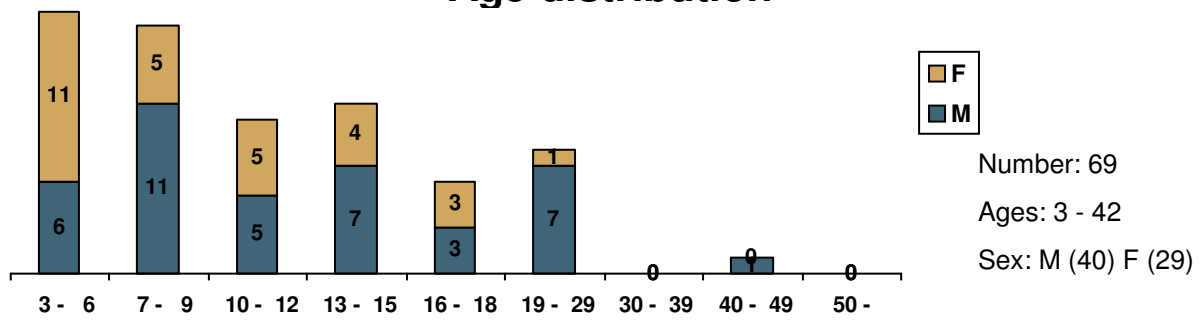
Source:

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.

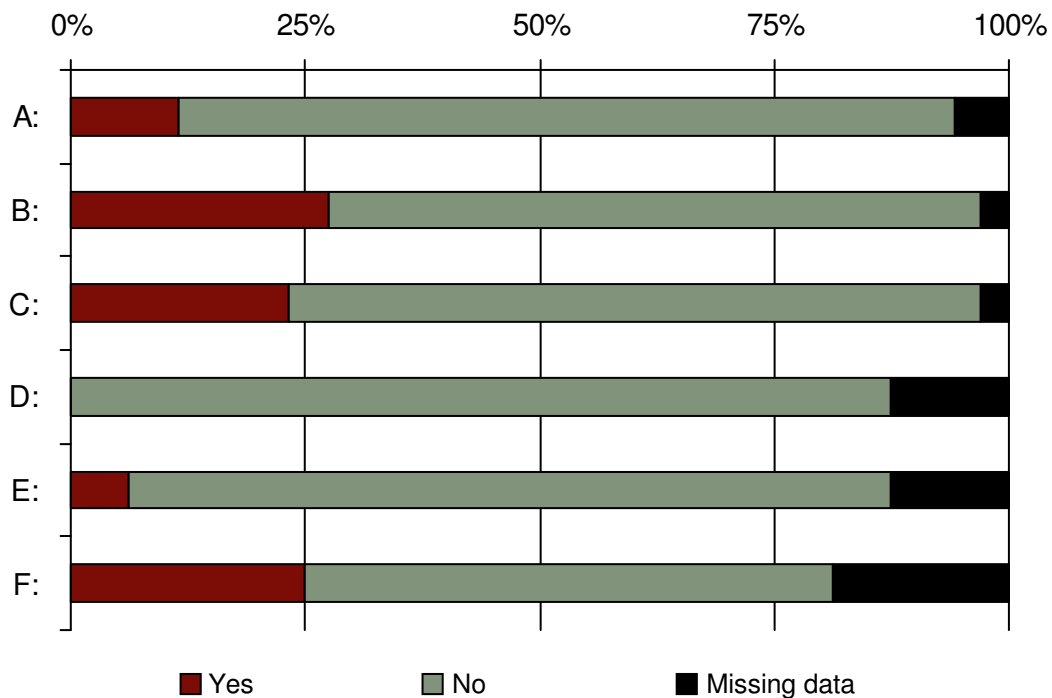
The Newsletter from the Ågrenska center.

Age distribution



Overview

	Yes	No	Missing data	N
A: Incomprehensible speech/No speech	8	57	4	69
B: Eating and drinking difficulties ¹	19	48	2	69
C: Abundant drooling ¹	16	51	2	69
D: Breathing difficulties ^{1 2}	0	14	2	16
E: Grinding every day ^{1 2}	1	13	2	16
F: Severe malocclusions ²	4	9	3	16



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)¹

		0	1	2	3	4	5	6	Missing data	N
Calc	Calculus	13	0	0	0	0	0	0	3	16
GI	Gingivitis	6	3	0	2	1	0	0	4	16
Plaq	Coating	7	3	0	1	1	0	0	4	16
Toot	Tooth wear	11	1	0	0				4	16

Ca 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

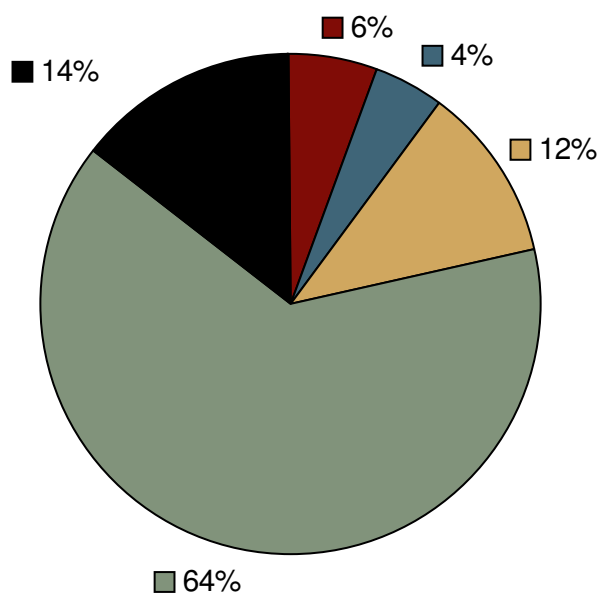
1: Marked tooth wear of the enamel, possibly exceeding into dentin

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.

¹: Oral health index (indices) was (were) introduced in the observations in 2008

Acceptance of dental examination



	Number
None	4
Negative	3
Reluctant	8
Positive	44
Missing data	10
Sum:	69

Caries

	3-6 years	7-12 years	13-19 years	Adults
deft¹				
Examined	17	20		
Number of individuals with deft=0	9	11		
Mean	1,4	1,6		
Standard deviation	1,7	2,4		
Missing data	0	6		
DMFT²				
Examined		24	17	7
Number of individuals with DMFT=0		18	11	2
Standard deviation		1,3	1,4	4,3
Mean		0,6	0,8	3,7
Missing data		2	1	1

1: Number of carious or filled deciduous teeth

2: Number of carious or filled permanent teeth

Occlusal relationshi

	Number
Neutral bite	33
Post normal	16
Pre normal	18
Missing data	2
Sum: 69	

Maximum jaw opening

Children younger
than 10 years

	Number
- 20	0
21 - 30	2
31 - 40	12
41 - 50	4
51 -	1
Missing data	14
Sum: 33	

Children, 10 years or older,
and adults

	Number
- 20	0
21 - 30	0
31 - 40	10
41 - 50	14
51 -	8
Missing data	4
Sum: 36	

Profile¹

	Number
Normal	12
Convex	4
Concave	0
Missing data	0
Sum: 16	

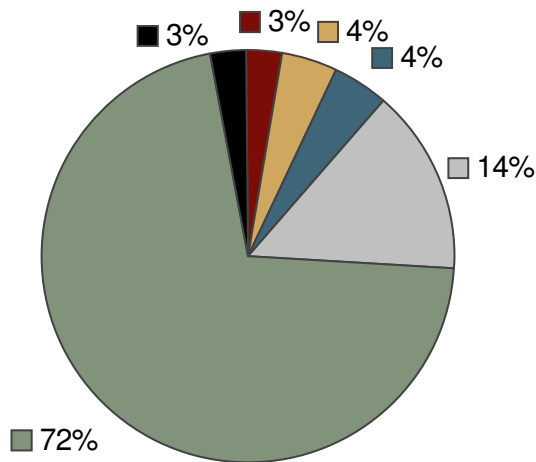
Mandibular plane¹

	Number
Normal	4
Increased	3
Reduced	0
Missing data	9
Sum: 16	

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

Orofacial function

Speech difficulty



	Number
No speech	2
Very incomprehensible speech	3
Incomprehensible speech	3
Slightly indistinct speech	10
No problems	49
Missing data	2
Sum: 69	

Clinical findings	Yes-answers			
	Total N=69 (%)	Boys/Men N=40 (%)	Girls/Women N=29 (%)	Missing data
Open mouth at rest	50 (74)	30 (77)	20 (69)	1
Low muscle tone in lips	43 (62)	25 (63)	18 (62)	0
M mentalis overactive	17 (25)	11 (29)	6 (21)	2
Impaired tongue motility	8 (12)	8 (20)	0 ()	1
Low muscle tone in masticatory muscles	6 (9)	5 (13)	1 (4)	3
Intra oral hypo-sensitivity	6 (9)	4 (10)	2 (7)	1
Mucous membrane changes	5 (7)	5 (13)	0 ()	2
Deep bite with gingival contact	4 (6)	3 (8)	1 (3)	2
High muscle tone in tongue	4 (6)	2 (5)	2 (7)	0
High muscle tone in lips	4 (6)	2 (5)	2 (7)	2
Short tongue frenulum	3 (4)	3 (8)	0 ()	1
Cleft lip and palate	2 (3)	2 (5)	0 ()	0
Low muscle tone in tongue	2 (3)	0 ()	2 (7)	1
Cranio-facial abnormality	1 (2)	1 (3)	0 ()	3
Reduced stability in neck	1 (1)	1 (3)	0 ()	0
Asymmetric tongue	1 (1)	1 (3)	0 ()	0
Extreme mobility in TMJ	1 (1)	1 (3)	0 ()	2