



Orofacial function of persons having Prader-Willi syndrome

Report from observation charts

The survey comprises 72 observation charts.

Estimated occurrence: 6-8:100 000 live births.

Etiology: Chromosomal aberration on chromosome 15. The gene or genes that cause the syndrome have not yet been identified.

General symptoms: There are great variations between different individuals and different ages. Pronouncedly poor muscle tone/hypotonicity from birth. After the age of one or two, distinct eating disorder occur, with an insatiable appetite despite a low need of energy (approximately 50-60% of what is considered normal). Without a special diet, this results in extreme obesity and an increased risk of type II diabetes. Most of these individuals are mildly to moderately intellectually impaired and delayed motor development is common. There is an increased risk of behavior problems. Individuals with PWS often have short stature and underdeveloped sexual organs (hypogonadism).

Orofacial/odontological symptoms: Characteristic facial features are associated with the diagnosis. Muscle laxity in newborns leads to most PWS infants being unable to be breast-fed. Many require a special bottle or tube feeding. Delayed speech and language development and problems with oral motor skills are common. Many suffer from dry mouth. Narrow nasal and throat airways may result in snoring and an increased risk of sleep apnea (frequent suspension of breathing while asleep). Delayed teething, defective tooth enamel, an excessive number of teeth and small teeth have all been reported. A high, arched palate and malocclusion are common. Some individuals have tooth grinding. There is an increased risk of caries owing to the eating disorder and dry mouth.

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.
- Tooth grinding should be followed up, and be managed with a splint when necessary.
- Training in oral motor skills may be relevant.
- Speech, language and communication training are often justified.
- Snoring problems should be followed up by a physician.

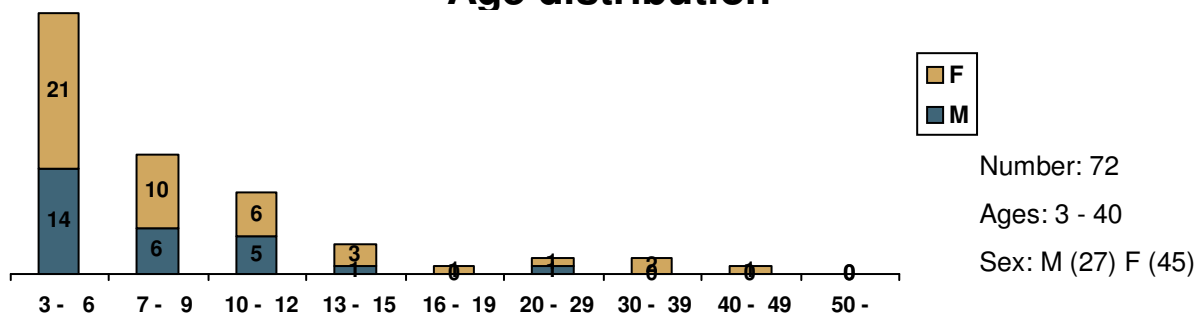
Sources:

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.

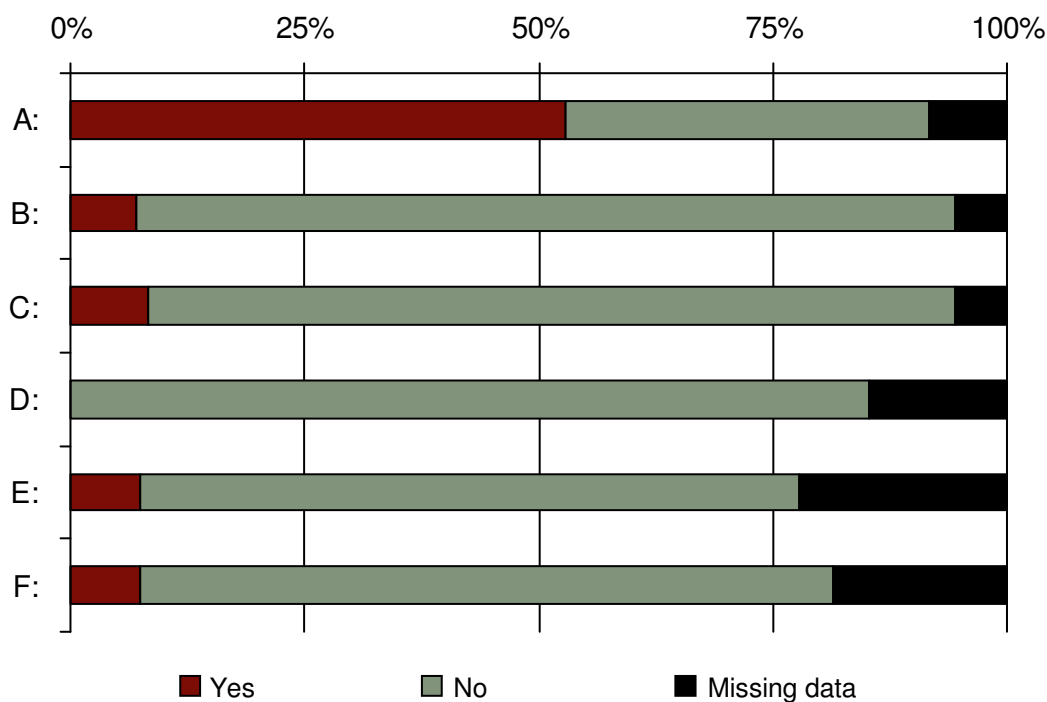


Age distribution



Overview

	Yes	No	Missing data	N
A: Incomprehensible speech/No speech	38	28	6	72
B: Eating and drinking difficulties ¹	5	63	4	72
C: Profuse drooling, on clothes ¹	6	62	4	72
D: Breathing difficulties ^{1 2}	0	23	4	27
E: Grinding every day ^{1 2}	2	19	6	27
F: Severe malocclusions ²	2	20	5	27



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

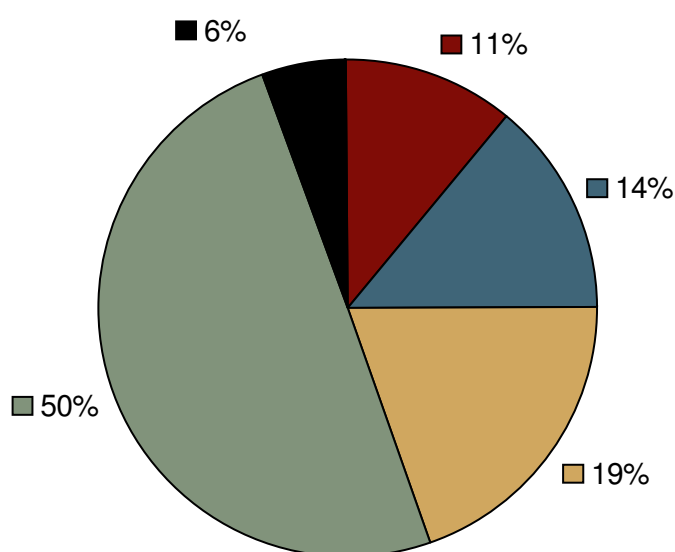
Oral health index (indices)¹

		0	1	2	3	4	5	6	Missing data	N
Calc	Calculus	11	0	0	0	0	0	0	16	27
GI	Gingivitis	11	1	1	0	0	0	0	14	27
Plaq	Coating	8	1	1	0	3	0	1	13	27
Toot	Tooth wear	10	6	2	0				9	27

- 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.

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

Acceptance of dental examination



	Number
None	8
Negative	10
Reluctant	14
Positive	36
Missing data	4
Sum:	72

Caries

	3-6 years	7-12 years	13-19 years	Adults
deft¹				
Examined	21	18		
Number of individuals with deft=0	20	18		
Mean	0,0	0,0		
Standard deviation	0,2	0,0		
Missing data	14	9		
DMFT²				
Examined		19	3	5
Number of individuals with DMFT=0		18	2	0
Standard deviation		0,4	0,5	4,5
Mean		0,1	0,3	6,8
Missing data		8	2	0

1: Number of carious or filled deciduous teeth

2: Number of carious or filled permanent teeth

Occlusal relationship

	Number
Neutral bite	49
Post normal	9
Pre normal	7
Missing data	7
<hr/>	
	Sum: 72

Maximum jaw opening

Children younger than 10 years

	Number
- 20	0
21 - 30	4
31 - 40	18
41 - 50	8
51 -	0
Missing data	21
<hr/>	
	Sum: 51

Children, 10 years or older, and adults

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

Profile¹

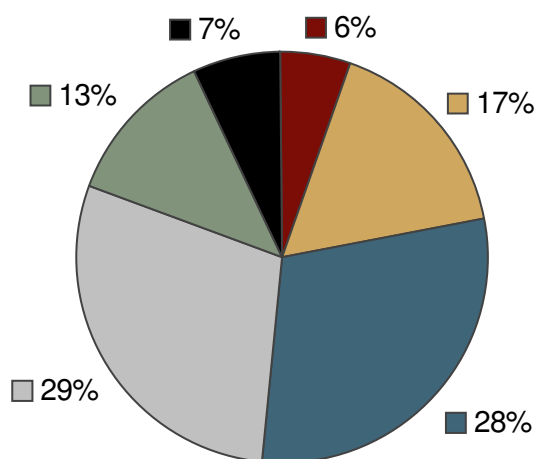
	Number
Normal	20
Convex	4
Concave	1
Missing data	2
<hr/>	
	Sum: 27

Mandibular plane¹

	Number
Normal	21
Increased	4
Reduced	0
Missing data	2
<hr/>	
	Sum: 27

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

Speech difficulty



	Number
No speech	4
Very incomprehensible	12
Incomprehensible speech	21
Slightly indistinct speech	21
No problems	9
Missing data	5
Sum: 72	

Clinical findings	Yes-answers			
	Total N=72 (%)	Boys/Men N=27 (%)	Girls/Women N=45 (%)	Missing data
Open mouth at rest	38 (55)	15 (56)	23 (55)	3
Low muscle tone in lips	37 (54)	14 (52)	23 (55)	3
Impaired tongue motility	26 (40)	12 (55)	14 (33)	7
Over crowding	13 (20)	4 (17)	9 (21)	7
Deep bite with gingival contact	12 (19)	4 (18)	8 (20)	10
M mentalis overactive	9 (13)	8 (30)	1 (2)	3
Low muscle tone in tongue	8 (12)	3 (13)	5 (12)	6
Low muscle tone in masticatory muscles	8 (14)	3 (15)	5 (14)	16
Short tongue frenulum	6 (9)	1 (5)	5 (12)	8
High palate	6 (9)	1 (4)	5 (13)	8
Spacing	6 (9)	3 (13)	3 (7)	6
Narrow palate	5 (8)	1 (4)	4 (11)	12
Mucous membrane changes	4 (7)	0 ()	4 (11)	15
Macroglossia	3 (5)	1 (5)	2 (5)	7