



Orofacial function of persons having Beckwith-Wiedemann syndrome

Report from observation charts

The survey comprises 22 observation charts.

Synonym: EMG syndrome (Exomphalus = umbilical hernia, Macroglossia = enlarged tongue, Gigantism = overgrowth)

Estimated prevalence: 5-10:100 000 live births.

Etiology: Research indicates an imbalanced regulation of the genes on the short arm of chromosome 11. Two probable defective genes for BWS are the gene for insulin-like growth factor 2 (IGF2) and gene H19. Most cases of BWS are sporadic but approximately 15% are familial, with autosomal dominant inheritance.

General symptoms: Increased growth during foetal life and early childhood cause overgrowth and considerable enlargement of most abdominal organs and the musculature. Enlarged tongue musculature is striking. About 25 percent of the children have asymmetric growth. Congenital umbilical hernia is common. Other malformations that may occur are cardiac malformation, inguinal hernia and defects of the urinary and sexual organs. 5-10% of the children develop tumours. The most common tumour is Wilms' tumour, which originates in the kidney. The main medical problems in infancy are umbilical hernia, the risk of severe low blood sugar and respiratory insufficiency due to enlarged tongue.

Orofacial/odontological symptoms: In extreme cases, enlargement of the tongue may lead to impaired breathing and/or feeding difficulties. If the tongue is too large for the oral cavity this may influence speech and cause frontal open bite. The midface may be underdeveloped, and a cleft palate may occur.

Orofacial/odontological treatment:

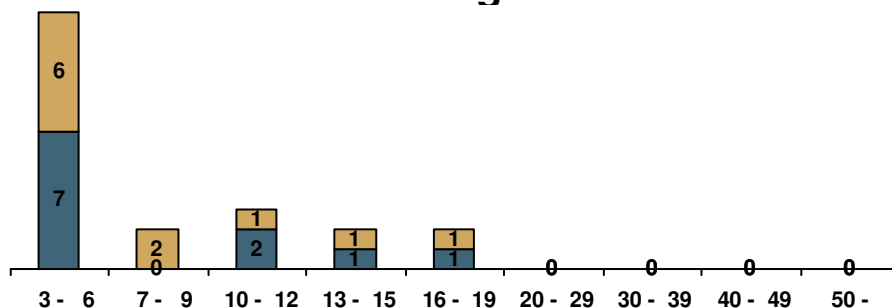
- Plastic surgery may be required when the tongue is considerably enlarged.
- Regular check-ups of dental and jaw development. Orthodontist should be consulted when needed.
- In cases of cleft palate, a specialist team will be needed for follow up and treatment.
- When treating medically compromised patients always contact their doctors for medical advice (bleeding problems, heart diseases etc).

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.



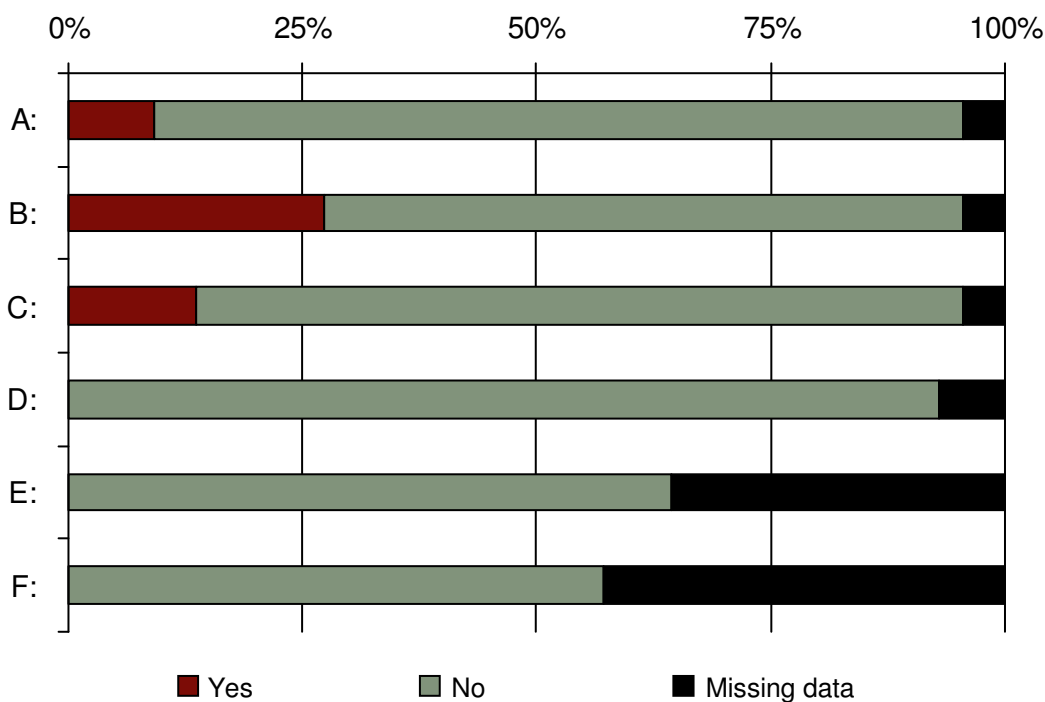
Age distribution



Number: 22
 Ages: 3 - 17
 Sex: M (11) F (11)

Overview

	Yes	No	Missing data	N
A: Incomprehensible speech/No speech	2	19	1	22
B: Eating and drinking difficulties ¹	6	15	1	22
C: Profuse drooling, on clothes ¹	3	18	1	22
D: Breathing difficulties ^{1 2}	0	13	1	14
E: Grinding every day ^{1 2}	0	9	5	14
F: Severe malocclusions ²	0	8	6	14



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

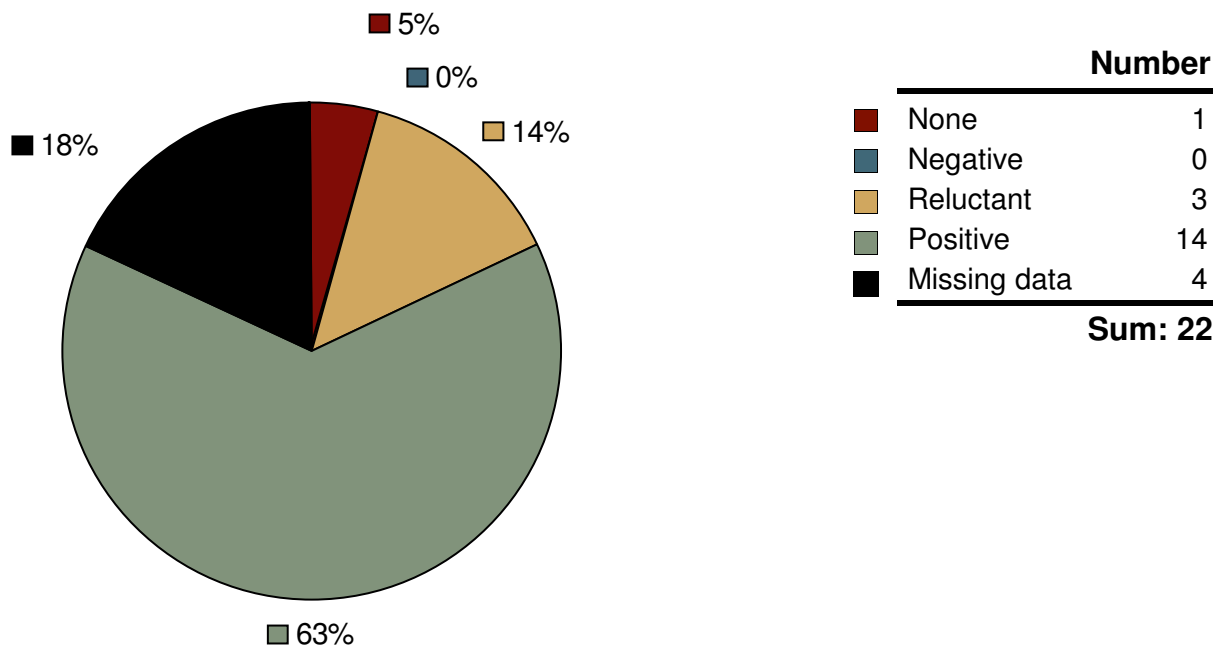
Oral health index (indices)¹

		0	1	2	3	4	5	6	Missing data	N
Calc	Calculus	10	0	0	0	0	0	0	4	14
GI	Gingivitis	10	0	0	0	0	0	0	4	14
Plaq	Coating	7	2	1	0	0	0	0	4	14
Toot	Tooth wear	10	0	0	0				4	14

- 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



Caries

	3-6 years	7-12 years	13-19 years	Adults
deft¹				
Examined	11	5		
Number of individuals with deft=0	9	4		
Mean	0,8	0,2		
Standard deviation	1,8	0,4		
Missing data	2	0		
DMFT²				
Examined		5	3	0
Number of individuals with DMFT=0		4	0	
Standard deviation		0,4	1,9	
Mean		0,2	4,3	
Missing data		0	1	

1: Number of carious or filled deciduous teeth

2: Number of carious or filled permanent teeth

Occlusal relationship

	Number
Neutral bite	12
Post normal	0
Pre normal	8
Missing data	2
<hr/>	
	Sum: 22

Maximum jaw opening

Children younger than 10 years

	Number
- 20	0
21 - 30	0
31 - 40	0
41 - 50	6
51 -	1
Missing data	8
<hr/>	
	Sum: 15

Children, 10 years or older, and adults

	Number
- 20	0
21 - 30	0
31 - 40	1
41 - 50	2
51 -	4
Missing data	0
<hr/>	
	Sum: 7

Profile¹

	Number
Normal	12
Convex	0
Concave	0
Missing data	2
<hr/>	
	Sum: 14

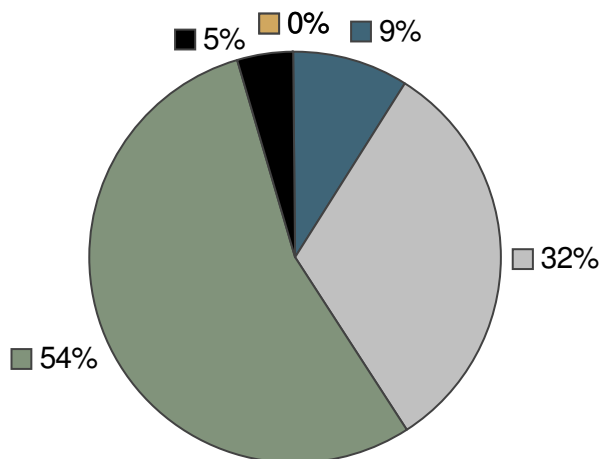
Mandibular plane¹

	Number
Normal	9
Increased	3
Reduced	0
Missing data	2
<hr/>	
	Sum: 14

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



Speech difficulty



	Number
No speech	0
Very incomprehensible	0
Incomprehensible speech	2
Slightly indistinct speech	7
No problems	12
Missing data	1
Sum: 22	

Clinical findings	Yes-answers			
	Total N=22 (%)	Boys/Men N=11 (%)	Girls/Women N=11 (%)	Missing data
Macroglossia	17 (81)	8 (80)	9 (82)	1
Frontal open bite	11 (55)	6 (60)	5 (50)	2
Open mouth at rest	8 (38)	4 (40)	4 (36)	1
Spacing	8 (40)	5 (50)	3 (30)	2
Facial asymmetry	7 (35)	4 (40)	3 (30)	2
Asymmetric tongue	5 (28)	4 (44)	1 (11)	4
Impaired tongue motility	4 (19)	1 (10)	3 (27)	1