



# Orofacial function of persons having Kabuki syndrome

## Report from observation charts

The survey comprises 15 observation charts.

**Synonyms:** Kabuki Makeup Syndrome, Niikawa-Kuroki Syndrome

**Estimated occurrence:** 3:100 000 live births.

**Etiology:** Genetic, most often new mutation

**General symptoms:** Mild to moderate developmental delay, growth retardation, skeletal anomalies, chubby finger pads and various malformations of bodily organs, such as the heart and urinary tract.

**Orofacial / odontological symptoms:** There are facial features that are characteristic for the diagnosis. Facial expression is often flattened and speech is unclear. Difficulties in the ability to suck, chew and swallow, as well as gastroesophageal reflux are common. Many have a thin and narrow upper lip and some have pits or raised blisters on the lower lip. High palate is common, as is cleft palate; cleft lip and jaw also occur. Deformities in tooth shape and number are noted in most cases; usually it is the front teeth in the upper or lower jaw that are missing. The teeth may be conical or tapered (screwdriver form). Tooth eruption may be delayed.

### Orofacial/odontological treatment:

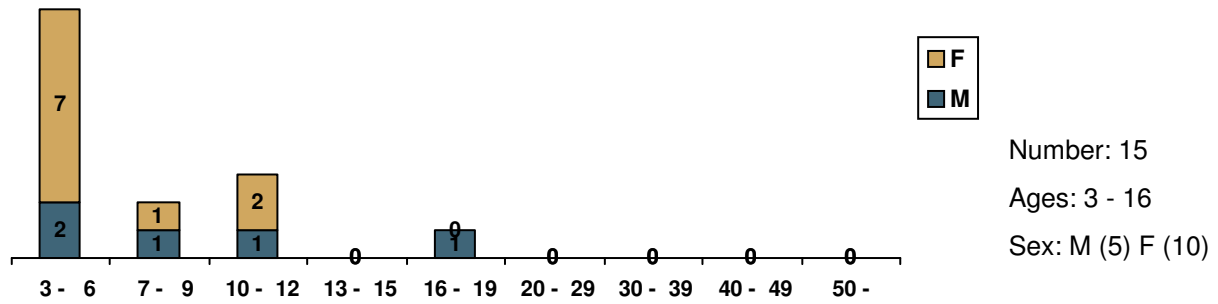
- Early contact with dental services for intensified prophylactic care and oral hygiene information is essential.
- Regular check-ups of dental and jaw development. Orthodontist should be consulted when needed.
- In cases of defective palate, a specialist team will be needed for follow up and treatment.
- When treating medically compromised patients always contact their doctor for medical advice (heart diseases etc).
- Training in oral motor skills and extra stimulation in cases of eating disorders are often required.

### 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.

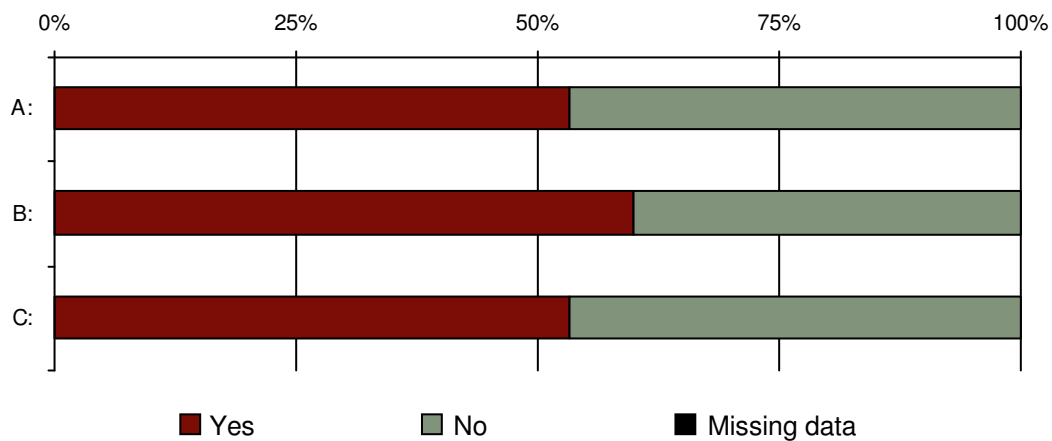
The Documentation from the Ågrenska Center.

### Age distribution



### Overview

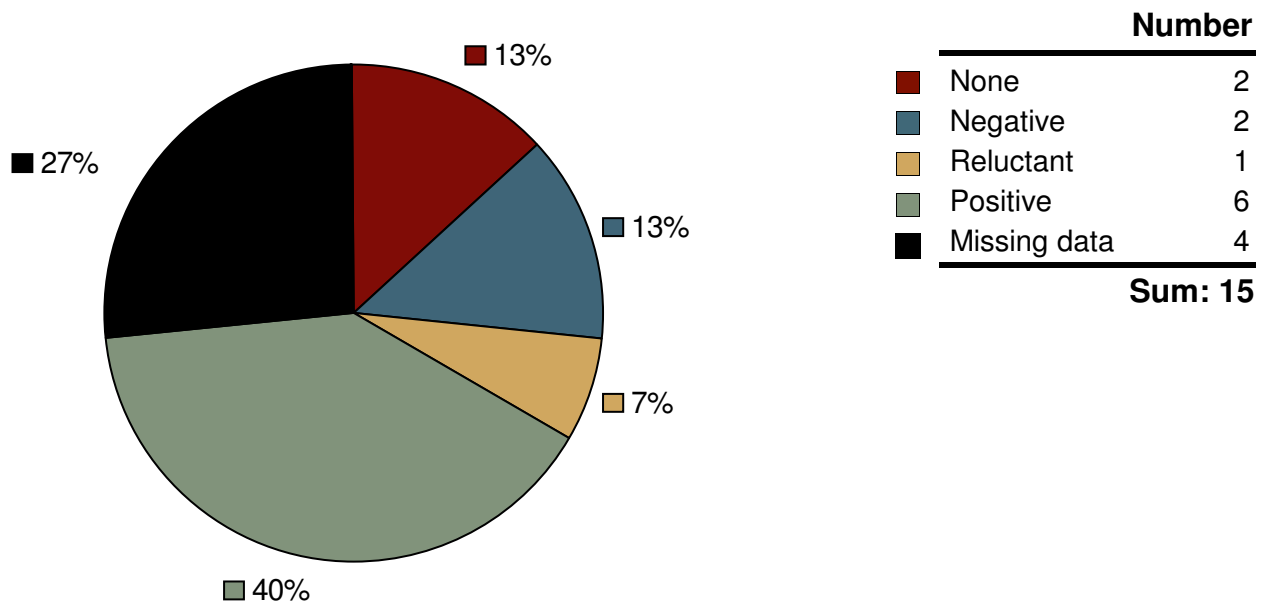
	Yes	No	Missing data	N
A: Incomprehensible speech/No speech	8	7	0	15
B: Eating and drinking difficulties <sup>1</sup>	9	6	0	15
C: Profuse drooling, on clothes <sup>1</sup>	8	7	0	15



Note that the diagram is based upon less than 100 individuals.

<sup>1</sup>: Compiled using questionnaire

## Acceptance of dental examination



## Caries

	3-6 years	7-12 years	13-19 years	Adults
<b>deft<sup>1</sup></b>				
Examined	1	2		
Number of individuals with deft=0	1	2		
Mean	0,0	0,0		
Standard deviation	0,0	0,0		
Missing data	8	3		
<b>DMFT<sup>2</sup></b>				
Examined		3	1	0
Number of individuals with DMFT=0		3	0	
Standard deviation		0,0	0,0	
Mean		0,0	1,0	
Missing data		2	0	

1: Number of carious or filled deciduous teeth

2: Number of carious or filled permanent teeth

## Occlusal relationship

	<b>Number</b>
Neutral bite	8
Post normal	2
Pre normal	1
Missing data	4
<b>Sum: 15</b>	

## Maximum jaw opening

Children younger  
than 10 years

	<b>Number</b>
- 20	0
21 - 30	0
31 - 40	1
41 - 50	2
51 -	0
Missing data	8
<b>Sum: 11</b>	

Children, 10 years or  
older, and adults

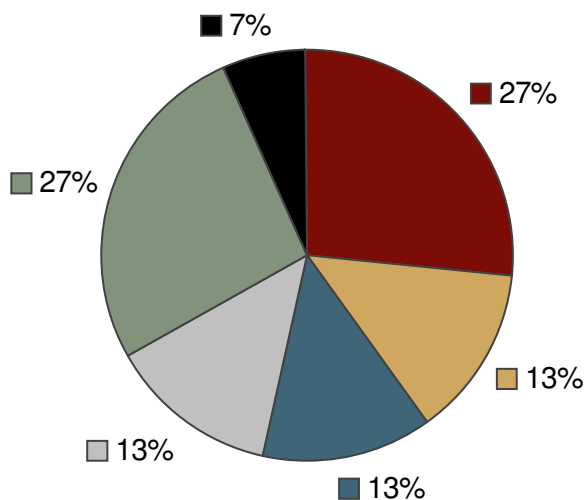
	<b>Number</b>
- 20	0
21 - 30	0
31 - 40	2
41 - 50	0
51 -	0
Missing data	2
<b>Sum: 4</b>	

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



### Speech difficulty



	<b>Number</b>
No speech	4
Very incomprehensible	2
Incomprehensible speech	2
Slightly indistinct speech	2
No problems	4
Missing data	1
<b>Sum: 15</b>	

Clinical findings	Yes-answers			
	Total N=15 (%)	Boys/Men N=5 (%)	Girls/Women N=10 (%)	Missing data
Low muscle tone in lips	12 (86)	4 (80)	8 (89)	1
Open mouth at rest	9 (60)	3 (60)	6 (60)	0
Impaired tongue motility	8 (53)	1 (20)	7 (70)	0
Reduced stability in neck	4 (27)	0 ( )	4 (40)	0
Cranio-facial abnormality	4 (27)	2 (40)	2 (20)	0
Cleft lip and palate	4 (27)	2 (40)	2 (20)	0
Narrow palate	4 (29)	1 (25)	3 (30)	1
Frontal open bite	4 (36)	0 ( )	4 (57)	4
Spacing	3 (30)	1 (25)	2 (33)	5