

Orofacial function of persons having Kabuki syndrome

Report from observation charts



15 observation charts

Synonym Kabuki Makeup Syndrome, Niikawa-Kuroki Syndrome

ICD-10 Q87.8

Estimated occurrence 3:100,000 live births.

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.

Oral 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), meaning that they are widely spaced. Tooth eruption may be delayed.

- Oral 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.

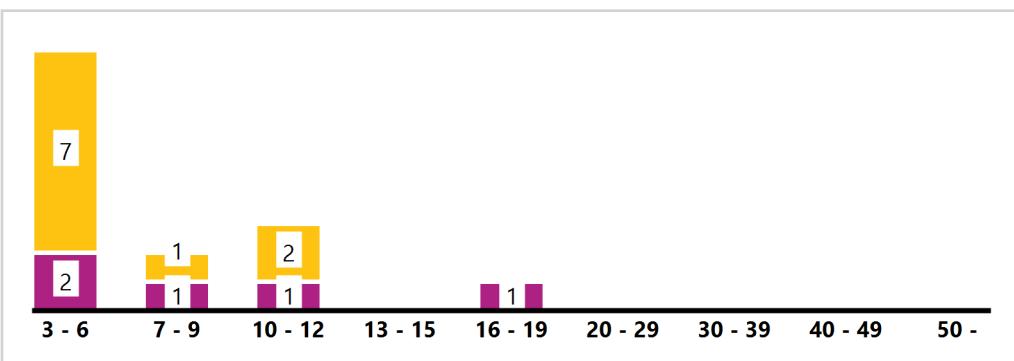
Sources The MHC database Rare diseases Dokumentation-Ågrenska

Age distribution

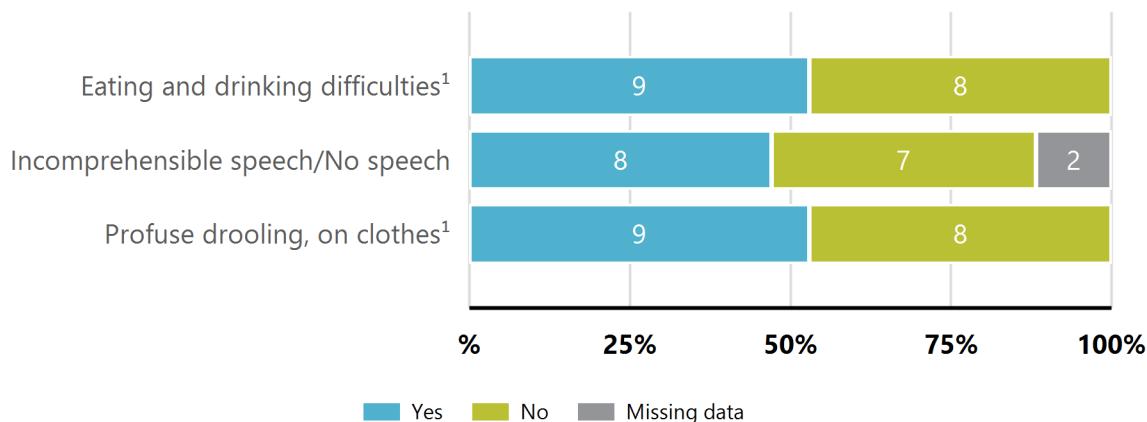
Count: 15

Ages: 3 - 16

♂ (5)
♀ (10)



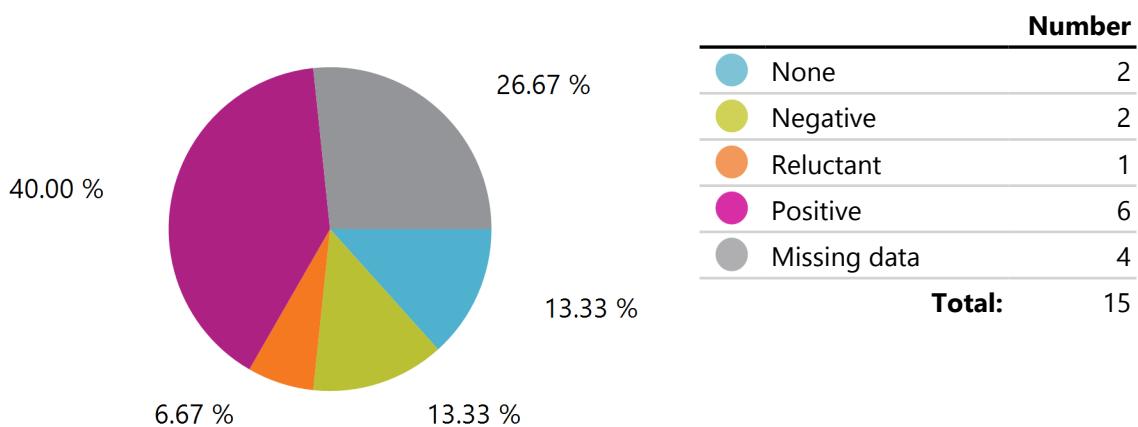
Summary



Certain caution should be observed when interpreting the chart because the number of individuals is less than 100.

¹ Reported via questionnaire

Acceptance of dental examination



Oral health

Caries

		3-6 years	7-12 years	13-19 years
deft Number of carious or filled deciduous teeth	Examined	9	5	
	Number of individuals with deft=0	9	5	
	Mean	0.0	0.0	
	Standard deviation		0.0	
DMFT Number of carious or filled permanent teeth	No data	8	3	
	Examined		5	1
	Number of individuals with DMFT=0		5	
	Mean		0.0	1.0
	Standard deviation		0.0	
	No data		2	0

Oral health index (indices)¹

	0	1	2	3	4	5	6	Missing data	Number
Calculus	2	0	0	0	0	0	0	13	5
Gingivitis	2	0	0	0	0	0	0	13	5
Plaque	2	0	0	0	0	0	0	13	5
Tooth wear	3	0	0	0	0	0	0	12	5

CALCULUS

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.

GINGIVITIS

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.

PLAQUE

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.

TOOTH WEAR

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

Occlusal relationship

	Number
Neutral bite	8
Overbite	2
Underbite	1
Missing data	4
Total:	15

Maximum jaw opening

Children younger than 10 years

	Number
-20	0
21-30	0
31-40	1
41-50	2
51-	0
Missing data	8
Total:	11

Children, 10 years or older, and adults

	Number
-20	0
21-30	0
31-40	2
41-50	0
51-	0
Missing data	2
Total:	4

Profile¹

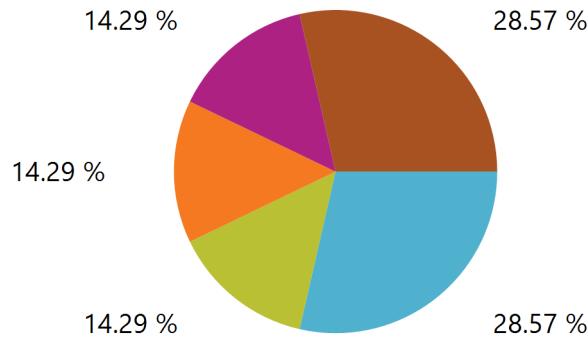
	Number
Normal	4
Convex	0
Concave	0
Missing data	1
Total:	5

Mandibular plane¹

	Number
Normal	2
Increased	0
Reduced	0
Missing data	3
Total:	5

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

Speech problems



	Number
No speech	4
Very difficult to understand	2
Difficult to understand	2
Slightly indistinct speech	2
No problems	4
Total:	14

Clinical findings

Number of yes-answers

	Total = 15 (%)	$\text{♂} = 5 (%)$	$\text{♀} = 10 (%)$	Missing Data
	12 (80)	4 (80)	8 (80)	1
Open mouth at rest	9 (60)	3 (60)	6 (60)	0
Impaired tongue motility	8 (53)	1 (20)	7 (70)	0
Frontal open bite	4 (27)	0 (0)	4 (40)	4
Narrow palate	4 (27)	1 (20)	3 (30)	1
Reduced stability in neck	4 (27)	0 (0)	4 (40)	0
Spacing	3 (20)	1 (20)	2 (20)	5
High palate	2 (13)	0 (0)	2 (20)	1
Low muscle tone in tongue	2 (13)	1 (20)	1 (10)	0
Low muscle tone in masticatory muscles	1 (7)	0 (0)	1 (10)	7
Deep bite with gingival contact	1 (7)	0 (0)	1 (10)	3