



Orofacial function of persons having Cornelia de Lange syndrome

Report from observation charts

The survey comprises 26 observation charts.

Synonyms: Brachmann-de Lange syndrom, de Lange syndrom, Typus degenerativus Amstelodamensis.

Estimated occurrence: 10-30:100 000 live births.

Etiology: Not completely known. The majority have a defect (mutation) on the NIPBL-gene on chromosome 5.

General symptoms: Children with the syndrome are low birth weight children. Characteristic symptoms of the syndrome include excessive body hair, delayed growth, particular facial features, short arms and legs with typical deformities of the lower arms and hands. Recently, a division between two types of CdL has begun to be made. Type I is the classic type, often with severe mental retardation, and is diagnosed immediately at birth. Type II is a considerably milder form, often not diagnosed until the age of two to three. Some children with CdL have autistic characteristics and are self-destructive. Vision and hearing problems are common. Congenital cardiac defects, deformities of the gastrointestinal system and epilepsy may also be present.

Orofacial/odontological symptoms: Characteristic facial features are associated with the diagnosis. Small jaws, widely-spaced teeth, small teeth, open bite and cleft lip and palate may occur. Tooth grinding and missing teeth are common. Infants often have difficulties with respiration and feeding. Gastroesophageal reflux and frequent vomiting is common and increase the risk for dental erosion and caries. Oral motor dysfunction may cause difficulties with eating and drooling. Most individuals with CdL syndrome never acquire speech. The voice is sometimes hoarse.

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.
- Feeding and swallowing difficulties are investigated and treated by a specialist team at the hospital or multidisciplinary treatment center.
- Oral motor skill training and communication practice are often required.
- When treating medically compromised patients always contact their doctors for medical advice (bleeding problems, heart diseases etc.).

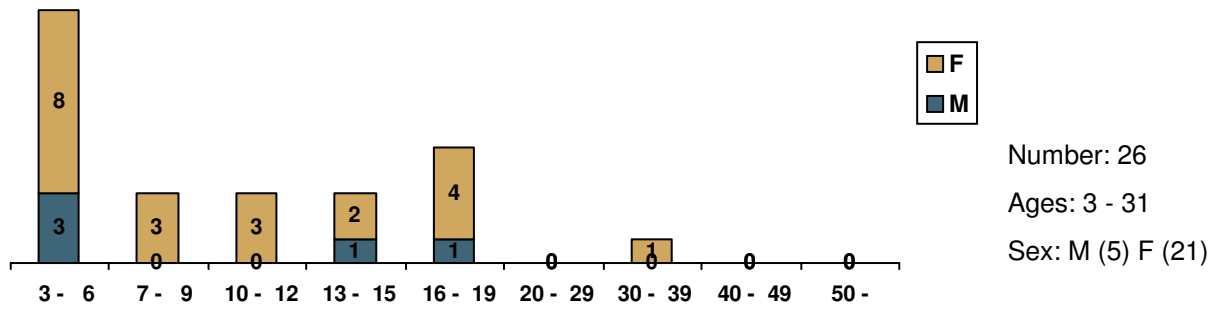
Sources

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.

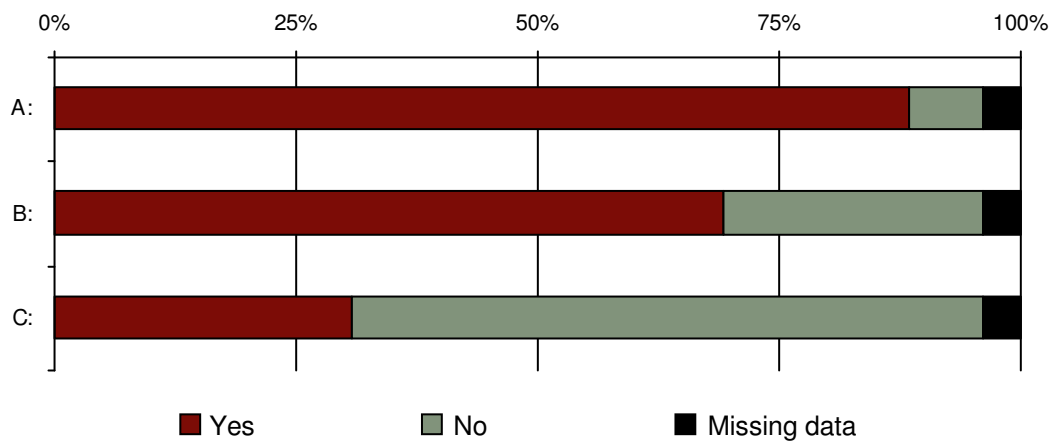
The Documentation from the Ågrenska Center.

Age distribution



Overview

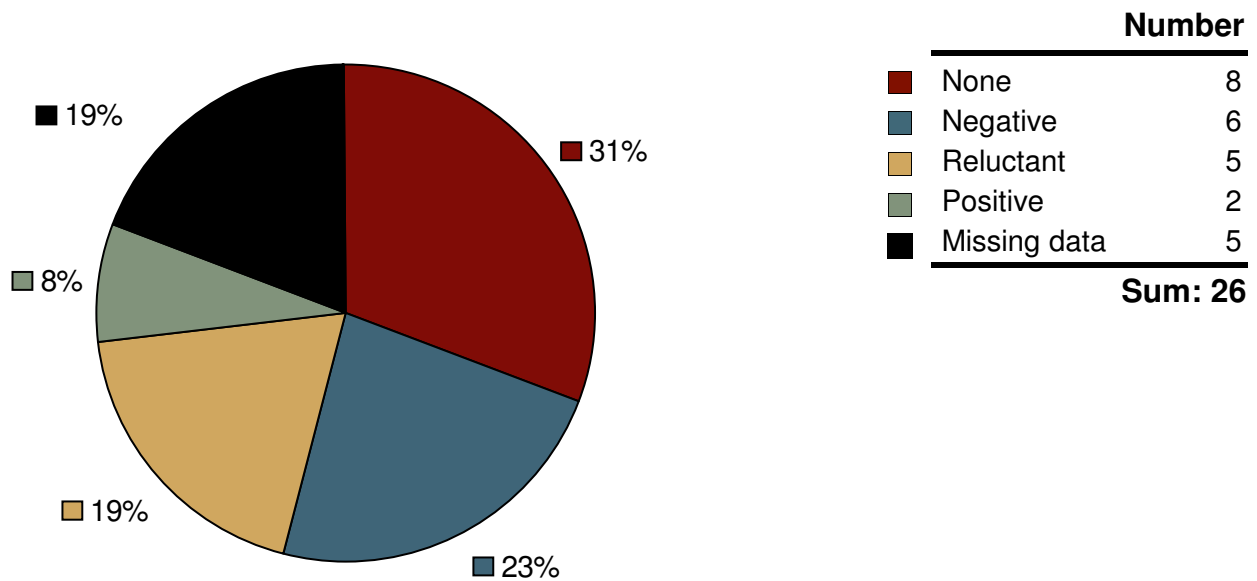
	Yes	No	Missing data	N
A: Incomprehensible speech/No speech	23	2	1	26
B: Eating and drinking difficulties ¹	18	7	1	26
C: Profuse drooling, on clothes ¹	8	17	1	26



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

¹: Compiled using questionnaire

Acceptance of dental examination



Caries

	3-6 years	7-12 years	13-19 years	Adults
deft¹				
Examined	6	5		
Number of individuals with deft=0	6	4		
Mean	0,0	0,2		
Standard deviation	0,0	0,4		
Missing data	5	1		
DMFT²				
Examined		5	6	1
Number of individuals with DMFT=0		4	5	0
Standard deviation		1,6	2,6	0,0
Mean		0,8	1,2	10,0
Missing data		1	2	0

1: Number of carious or filled deciduous teeth

2: Number of carious or filled permanent teeth

Occlusal relationship

	Number
Neutral bite	11
Post normal	5
Pre normal	2
Missing data	8
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	Sum: 26

Maximum jaw opening

Children younger
than 10 years

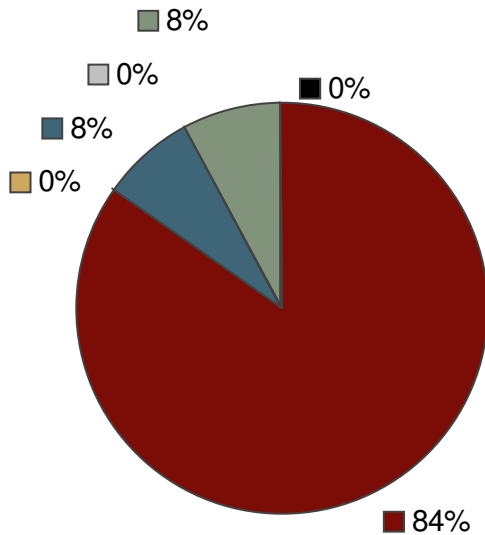
	Number
- 20	1
21 - 30	2
31 - 40	1
41 - 50	0
51 -	0
Missing data	10
<hr/>	
	Sum: 14

Children, 10 years or
older, and adults

	Number
- 20	0
21 - 30	0
31 - 40	2
41 - 50	2
51 -	0
Missing data	8
<hr/>	
	Sum: 12

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

Speech difficulty



	Number
■ No speech	22
■ Very incomprehensible	0
■ Incomprehensible speech	2
■ Slightly indistinct speech	0
■ No problems	2
■ Missing data	0
Sum: 26	

Clinical findings	Yes-answers			
	Total N=26 (%)	Boys/Men N=5 (%)	Girls/Women N=21 (%)	Missing data
Impaired tongue motility	15 (63)	3 (75)	12 (60)	2
Open mouth at rest	11 (42)	1 (20)	10 (48)	0
Low muscle tone in lips	11 (42)	1 (20)	10 (48)	0
Narrow palate	7 (32)	1 (25)	6 (33)	4
M mentalis overactive	6 (25)	0 ()	6 (32)	2
Reduced stability in neck	5 (21)	1 (25)	4 (20)	2
Over crowding	5 (23)	1 (25)	4 (22)	4
Spacing	5 (24)	1 (33)	4 (22)	5
Low muscle tone in tongue	4 (17)	0 ()	4 (21)	3
Frontal open bite	4 (17)	0 ()	4 (21)	3
Cleft lip and palate	3 (12)	1 (20)	2 (10)	0
High muscle tone in lips	3 (12)	0 ()	3 (14)	0
High palate	3 (14)	1 (25)	2 (11)	4