



# Orofacial function of persons having Neurofibromatosis 1

## Report from observation charts

The survey comprises 88 observation charts.

**Synonym:** Recklinghausen's disease

**Estimated occurrence:** 30:100 000 inhabitants.

**Etiology:** The locus of the gene that causes this syndrome is the long arm of chromosome 17. Autosomal dominant heredity. 50-60% of the occurrences are spontaneous mutations.

**General symptoms:** Café au lait spots and neurofibromas of the skin are characteristic. The latter are benign tumors that develop in the supportive tissue around the nerves. There may be up to several hundred of them. One-third of affected individuals develop plexiform neurofibromas, which are not nearly as restricted as neurofibromas of the skin. This disease, or more correctly disorder, may also impair the ocular and auditory nerves, the central nervous system, and skeletal development. Endocrine tumours occur. Children with this diagnosis should have annual physical examinations. Some developmental delay, learning difficulties and concentration problems may occur, as well as epilepsy. Scoliosis is occasionally found.

**Orofacial/odontological symptoms:** Neurofibromas may occur in the oral mucous membranes, but these are normally not treated unless they grow or become uncomfortable. Neurofibromas may also occur in the jawbone, where they may be found with the aid of general radiographs. If they are found, they should be checked at regular intervals. There may be enlarged papillae on the tongue. Early tooth eruption has been reported, as well as abnormal tooth positions. Eating and speech difficulties and drooling are found, as well as occasional sleep apnea (frequent suspension of breathing while asleep).

### **Orofacial/ odontological treatment:**

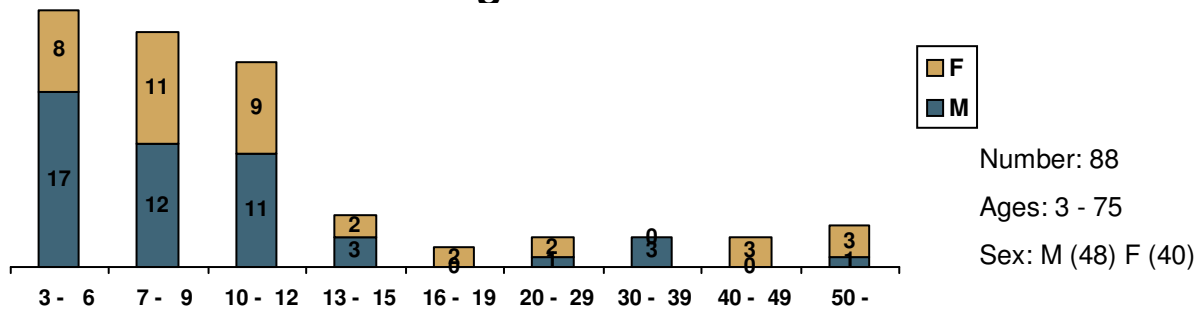
- Early contact with dental services for intensified prophylactic care and oral hygiene information is essential.
- Regular check-ups of dental and jaw development. Orthodontist should be consulted when needed.
- Oral motor training and stimulation may be relevant in cases of eating difficulties, speech impairment and drooling
- 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.

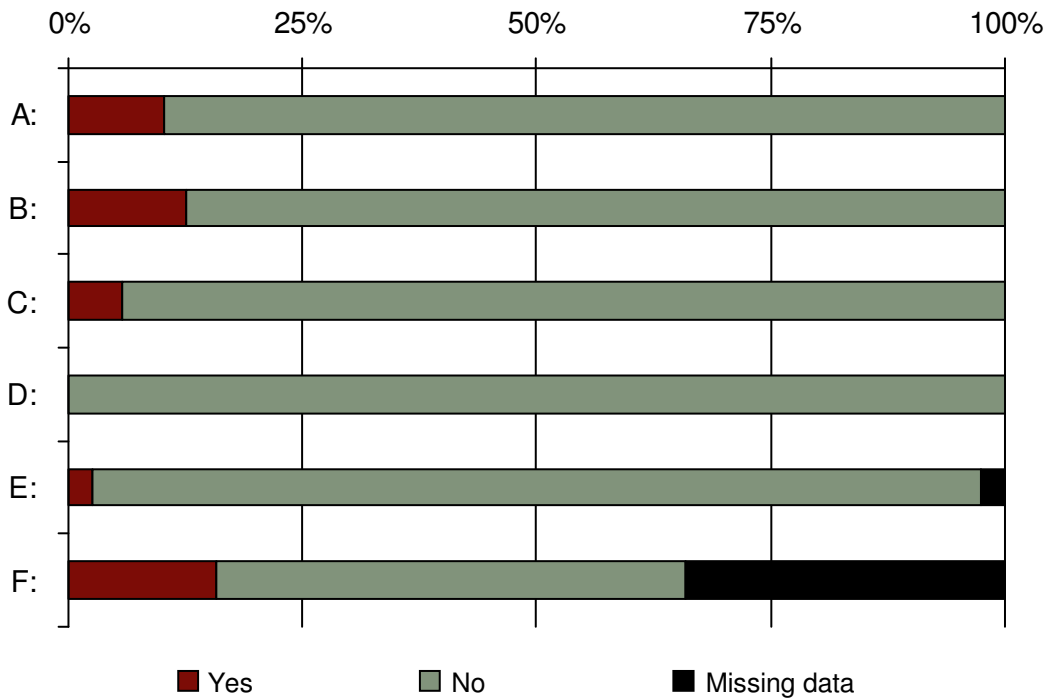


### Age distribution



### Overview

	Yes	No	Missing data	N
A: Incomprehensible speech/No speech	9	79	0	88
B: Eating and drinking difficulties <sup>1</sup>	11	77	0	88
C: Profuse drooling, on clothes <sup>1</sup>	5	83	0	88
D: Breathing difficulties <sup>1 2</sup>	0	38	0	38
E: Grinding every day <sup>1 2</sup>	1	36	1	38
F: Severe malocclusions <sup>2</sup>	6	19	13	38



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

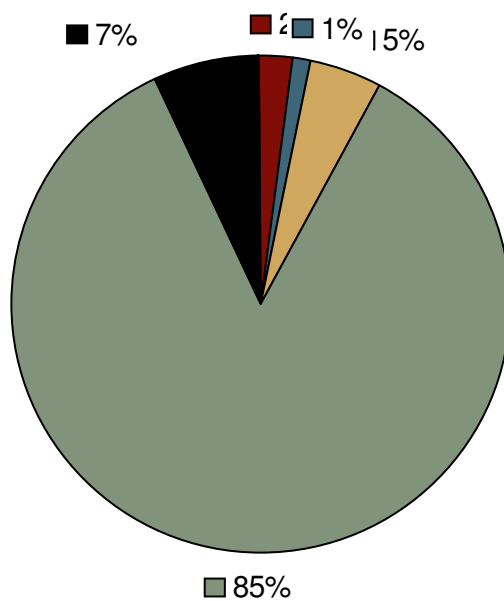
### Oral health index (indices)<sup>1</sup>

		0	1	2	3	4	5	6	Missing data	N
Calc	Calculus	23	7	0	0	0	0	0	8	38
GI	Gingivitis	14	9	1	3	2	1	0	8	38
Plaq	Coating	18	6	2	2	1	1	1	7	38
Toot	Tooth wear	25	4	1	0				8	38

- 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.

<sup>1</sup>: Oral health index (indices) was (were) introduced in the observations in 2008

## Acceptance of dental examination



	Number
None	2
Negative	1
Reluctant	4
Positive	75
Missing data	6
<b>Sum:</b>	<b>88</b>

## Caries

	3-6 years	7-12 years	13-19 years	Adults
<b>deft<sup>1</sup></b>				
Examined	20	30		
Number of individuals with deft=0	17	16		
Mean	0,3	1,3		
Standard deviation	1,1	1,8		
Missing data	5	13		
<b>DMFT<sup>2</sup></b>				
Examined		39	6	12
Number of individuals with DMFT=0		32	4	1
Standard deviation		0,8	0,8	6,7
Mean		0,3	0,5	9,5
Missing data		4	1	1

1: Number of carious or filled deciduous teeth

2: Number of carious or filled permanent teeth

## Occlusal relationship

	<b>Number</b>
Neutral bite	53
Post normal	10
Pre normal	21
Missing data	4
<b>Sum: 88</b>	

## Maximum jaw opening

Children younger than 10 years

	<b>Number</b>
- 20	0
21 - 30	1
31 - 40	10
41 - 50	26
51 -	6
Missing data	5
<b>Sum: 48</b>	

Children, 10 years or older, and adults

	<b>Number</b>
- 20	0
21 - 30	0
31 - 40	7
41 - 50	18
51 -	14
Missing data	1
<b>Sum: 40</b>	

## Profile<sup>1</sup>

	<b>Number</b>
Normal	26
Convex	2
Concave	3
Missing data	7
<b>Sum: 38</b>	

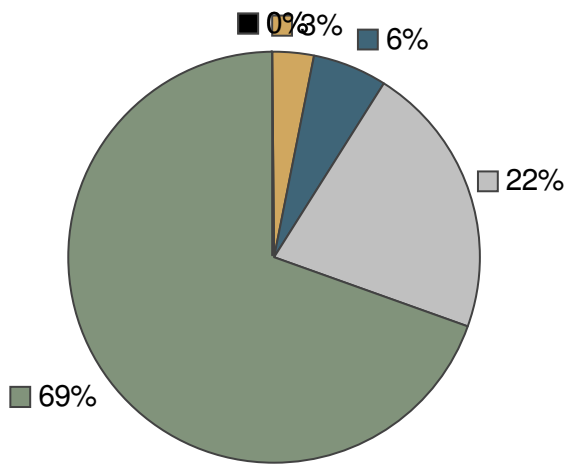
## Mandibular plane<sup>1</sup>

	<b>Number</b>
Normal	25
Increased	4
Reduced	0
Missing data	9
<b>Sum: 38</b>	

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



### Speech difficulty



	<b>Number</b>
No speech	0
Very incomprehensible	3
Incomprehensible speech	5
Slightly indistinct speech	19
No problems	61
Missing data	0
<b>Sum: 88</b>	

Clinical findings	Yes-answers			
	Total N=88 (%)	Boys/Men N=48 (%)	Girls/Women N=40 (%)	Missing data
Mucous membrane changes	25 (30)	14 (30)	11 (29)	4
Facial asymmetry	14 (16)	9 (19)	5 (13)	1
Open mouth at rest	14 (16)	7 (15)	7 (18)	2
Spacing	13 (16)	7 (16)	6 (16)	6
Over crowding	12 (14)	7 (16)	5 (13)	5
Frontal open bite	11 (13)	5 (11)	6 (15)	3
Low muscle tone in lips	10 (12)	7 (16)	3 (8)	3
Impaired tongue motility	10 (12)	7 (15)	3 (8)	3
High palate	7 (8)	4 (9)	3 (8)	5
M mentalis overactive	6 (7)	3 (6)	3 (8)	3
Narrow palate	4 (5)	3 (6)	1 (3)	4
Low muscle tone in masticatory muscles	3 (4)	3 (7)	0 (0)	6
Deep bite with gingival contact	3 (4)	3 (7)	0 (0)	5