



# Orofacial function of persons having Neurofibromatosis 1

## Report from questionnaires

The survey comprises 48 questionnaires.

**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).

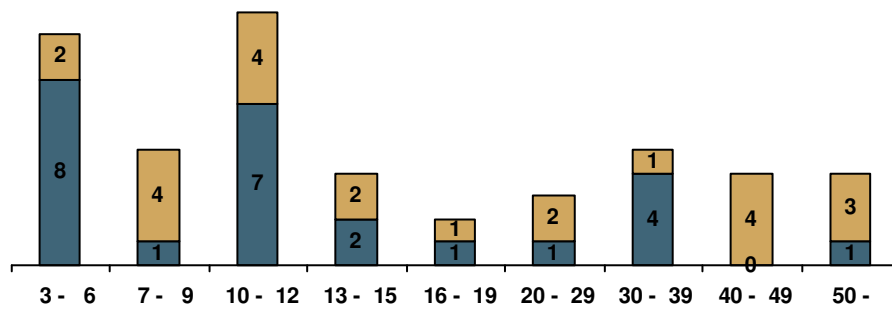
### **Orofacial/ odontological treatment:**

- 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



Number: 48

Ages: 4 - 75

Sex: M (25) F (23)

## Additional diagnoses

Medical impairment	Yes	No	Missing data
Inborn heart defect	0	46	2
Other cardiovascular disease	0	46	2
Epilepsy	2	44	2
Asthma	3	42	3
Need of respiratory support	2	44	2
Allergy	10	36	2

Neuropsychiatric diagnosis	Yes	No	Missing data
ADHD/ADD	6	36	6
Autistic syndrome	1	44	3
Autistic traits	2	41	5
Asperger's syndrome	0	45	3
Tourette's syndrome	0	45	3

General disability	Yes	No	Missing data
Intellectual disability	3	38	7
Mobility dysfunction	18	17	13
Impaired vision	10	20	18
Impaired hearing	5	36	7
Difficulty in communicating	7	31	10

## About dental care and oral health

### Do you feel that you receive the dental care you need?

	Number
Yes, very much so	29
Yes, some what	14
No, not really	1
No, not at all	0
Missing data	4
<b>Sum:</b>	<b>48</b>

### How many times per year do you normally seek dental care?

	Number
Three or more times per year	9
Twice per year	5
Once per year	24
Less than once per year	7
Missing data	3
<b>Sum:</b>	<b>48</b>

### When were your teeth last X-rayed?

	Number
During the past two years	29
More than two years ago	6
Never had my teeth X-rayed	5
Missing data	8
<b>Sum:</b>	<b>48</b>

## Do you look after your teeth in a good way?

	Number
Yes, very much so	24
Yes, some what	21
No, not really	2
No, not at all	0
Missing data	1
<b>Sum:</b>	<b>48</b>

## Who brushes your teeth?

	Number
I always brush myself	26
Sometimes I brush myself	15
Someone else always helps me	7
Missing data	0
<b>Sum:</b>	<b>48</b>

## How often are your teeth brushed?

	Number
Three or more times per day	4
Twice per day	42
Once per day	2
Not every day	0
Missing data	0
<b>Sum:</b>	<b>48</b>

## About dental care and oral health

	Yes	No	Missing data
Does your mouth hurt?	8	35	5
Does your mouth feel dry?	7	38	3
Have you ever taken a serious hit to your permanent front teeth?	5	41	2
Do you feel that you have a divergent bite?	6	39	3
Have you had a brace?	7	41	0
Do you feel that you need orthodontics/a brace?	11	31	6

## Do you grind or press your teeth at night?

	Number
Never	37
Once or twice per week	6
Every night	2
Missing data	3
<b>Sum:</b>	<b>48</b>

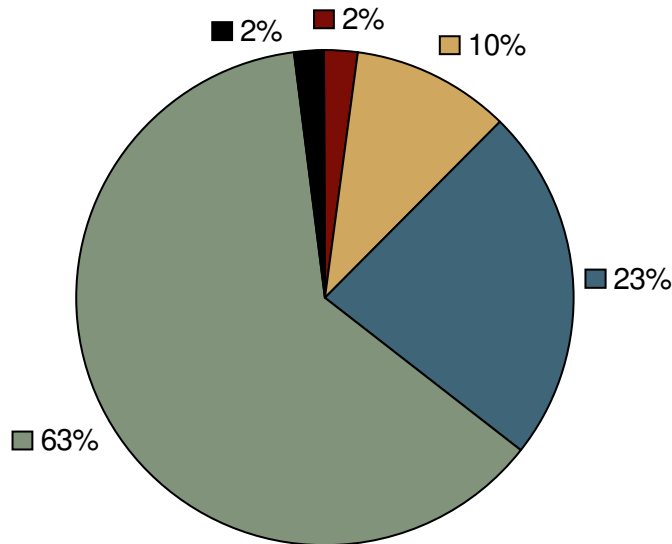
## Do you grind or press your teeth during the day?

	Number
Never	41
Once or twice per week	3
Every day	2
Missing data	2
<b>Sum:</b>	<b>48</b>



## About eating

### Do you have any problems with eating?



	Number
Yes, very much so	1
Yes, some what	5
No, not really	11
No, not at all	30
Missing data	1
<b>Sum:</b>	<b>48</b>

	Yes	No	Missing data
Do you cough daily in connection with meals?	1	45	2
Do you gag daily in connection with meals?	0	44	4
Do you get acid reflux daily?	2	42	4
Do you throw up often (at least twice per week)?	1	46	1
Do you have a poor appetite?	3	43	2
Does it take a long time before you can swallow a mouthful?	4	43	1
Do you press your tongue forward when you swallow so that food ends up outside the mouth?	2	43	3
Do you find it difficult to chew, i.e. grind food using your molars?	5	40	3
Do you find it difficult to take food from the spoon using your lips?	3	43	2
Have you had problems with food and drink leaking out through the corners of your mouth?	2	42	4
Does food tend to remain in your mouth after meals?	1	43	4
Do you get nutrition in any other way than through your mouth?	0	47	1



## About drooling

### Do you drool?

	Number
Never drool	39
Drool sometimes – not every day	3
Drool often – every day	1
Constant drooling	0
Missing data	5
<b>Sum:</b>	<b>48</b>

### How much do you drool?

	Number
Slight drooling, only on the lips	1
Moderate drooling, on lip and chin	3
Profuse drooling, on clothes	0
Very profuse drooling, on hands/objects	0
Missing data	0
<b>Sum:</b>	<b>4</b>

### Is your drooling a problem for you?

	Number
Yes, very much so	0
Yes, some what	1
No, not really	3
No, not at all	0
Missing data	0
<b>Sum:</b>	<b>4</b>

### Is your drooling a problem for your family or people around you?

	Number
Yes, very much so	0
Yes, some what	1
No, not really	1
No, not at all	2
Missing data	0
<b>Sum:</b>	<b>4</b>