Orofacial function of persons having
Neurofibromatosis 1
Report from observation charts

The survey comprises 51 observation charts.

Synonym: Recklinghausen's disease

Estimated occurrence: 1:3,000 inhabitants (Neurofibromatosis type 1)

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.

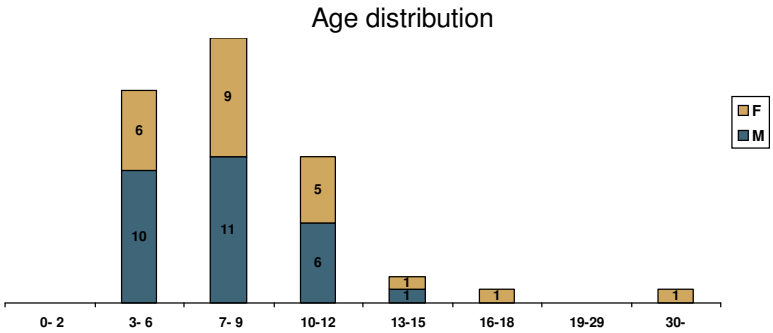
General symptoms: Café au lait spots and neurofibromas of the skin are characteristic. The latter are benign tumors that develop in the supportive tissue around the nerves. There may be up to several hundred of them. One-third of affected individuals develop plexiform neurofibromas, which are not nearly 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:

- Many of these children require extra preventive dental care.
- Training in oral motor skills and extra stimulation in cases of eating disorders, speech difficulties and drooling may be necessary.
- Speech, language and communication training are often justified.
- Snoring problems should be followed up by a physician.
- An orthodontist should be consulted between the ages of 7 and 9 in order to identify dental aberrations or malocclusions and to plan any necessary orthodontic treatment.

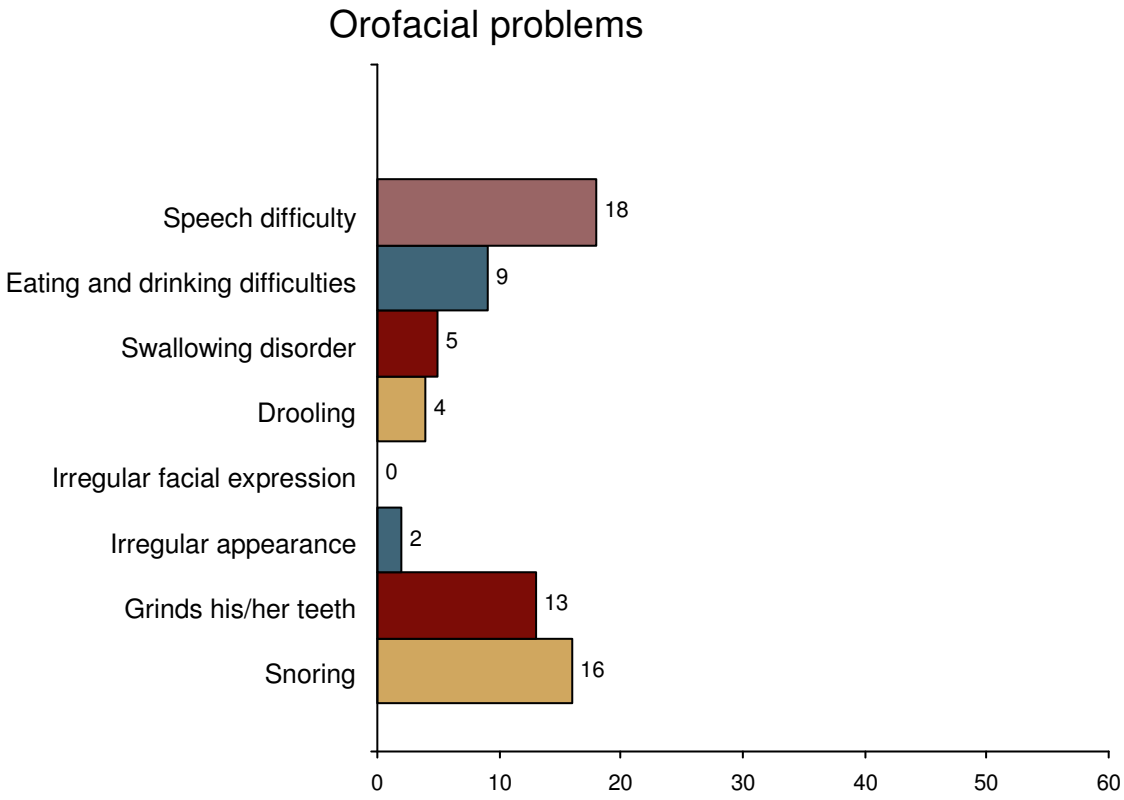
Sources:
The MHC database - The Mun-H-Center database of orofacial manifestations in rare diseases.
The Newsletter of the Ågrenska Center.

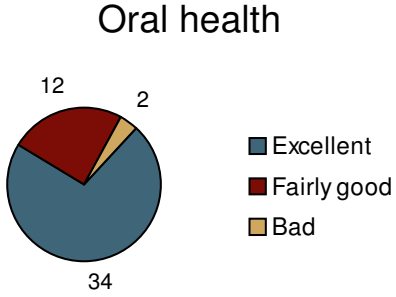


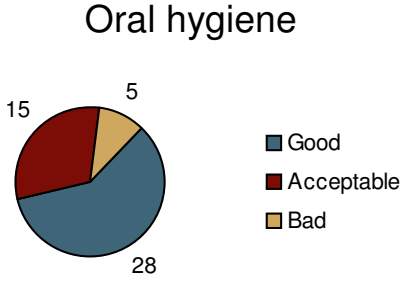
Number: 51

Ages: 3 -- 51 years

Sex: M (28) + F (23)

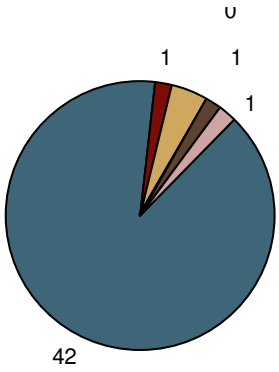






Behaviour in the treatment situation

How calm and co-operative is the patient at time of examination?



(42) Examination is possible w ithout problem

(1) Examination is possible w ithout problems, some reaction is observed

(2) Examination can continue if adjusted to patient's reactions

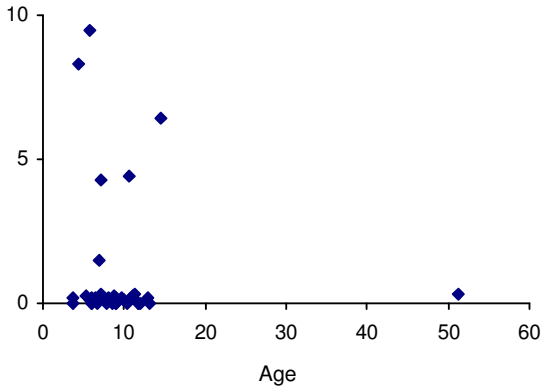
(0) Reactions are considerable and examination is obviously affected

(1) Examination is practically impossible to complete

(1) Patient refuses examination

How does the patient cope with treatment in general?

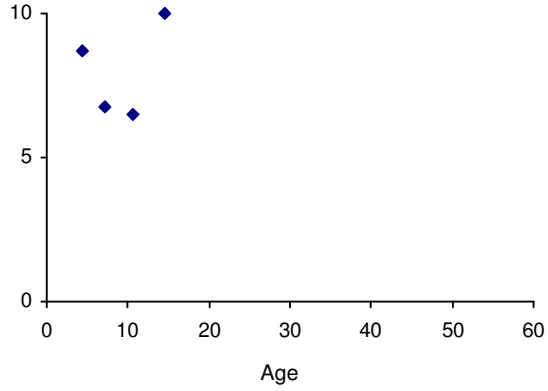
0=no problems/10=great problems



If there are treatment problems:

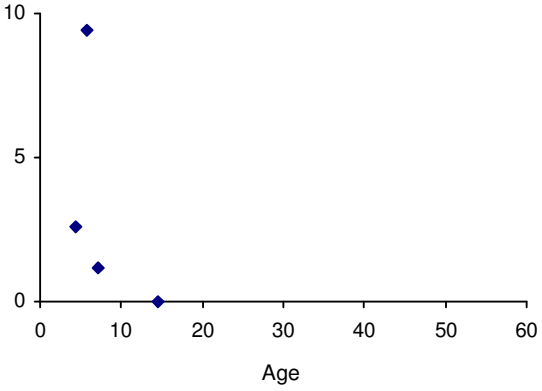
To what degree are the problems, if any, due to NN's handicap?

0=not at all/10=to a very high degree



To what degree are the problems, if any, due to NN's fear?

0=not at all/10=to a very high degree



Clinical findings	Total N=51	Boys/Men N=28	Girls/Women N=23	Not evaluated
Gingivitis	23	12	11	0
Speech difficulty	18	9	9	0
Mucous membrane change	17	10	7	1
Facial asymmetry	14	9	5	0
Pre normal bite	14	9	5	1
Hypomineralisation	11	6	5	0
Grinds his/her teeth	10	8	2	0
Spacing	10	6	4	0
Cross bite	8	6	2	0
Edge to edge bite	8	7	1	0
Over crowding	8	5	3	0
Dental trauma	7	5	2	0
Other oral habits	7	4	3	0
Abrasion - insignificant	6	3	3	0
Open mouth at rest	6	2	4	1
Post normal bite	6	2	4	1
Reduced mobility in tongue	6	4	2	0
Deviation on opening	5	3	2	0
Drooling	5	2	3	1
Frontal open bite	5	2	3	0
M mentalis is overactive	5	3	2	1
Posturing forward	5	3	2	1
Deep bite without gingival contact	4	1	3	0
Lower jaw seems large	4	3	1	0
Abrasion - significant	3	2	1	1
Frontal inversion	3	3	0	0
High palate	3	2	1	0
Horizontal over-bite 6 mm or more	3	0	3	0
Low muscle tone in lower lip	3	2	1	0
Low muscle tone in upper lip	3	2	1	0
Molar contact only	3	1	2	0
Narrow palate	3	2	1	0
Retroclined lower incisors	3	1	2	0