



Orofacial function of persons having Angelman syndrome

Report from observation charts

The survey comprises 64 observation charts.

Estimated occurrence: 80: 1 000 000 inhabitants.

Etiology: Genetic origin, often spontaneous mutation. Most of these children have some deletion of chromosomal material in the chromosome 15 inherited from the mother.

General symptoms: Delayed psychomotor development. Muscle laxity and balance problems. Severe mental retardation. Epilepsy. Abnormal language development, where understanding is often superior to the ability to express things in speech or using alternative means of communication.

Orofacial/odontological symptoms: A particular appearance, with a wide mouth and jaws, and a flat back of the head. Widely-spaced teeth are common, as are malocclusions in the form of a postnormal or prenatal bite and an open, frontal bite. Oral motor skills poorly developed, and there is muscular laxity (hypotonia). The tip of the tongue is often inserted between the front teeth. Sucking, eating and drinking difficulties and drooling are all frequent problems. Most individuals with Angelman syndrome never acquire speech. Many like to suck and bite their hands and objects. Daytime tooth grinding is common.

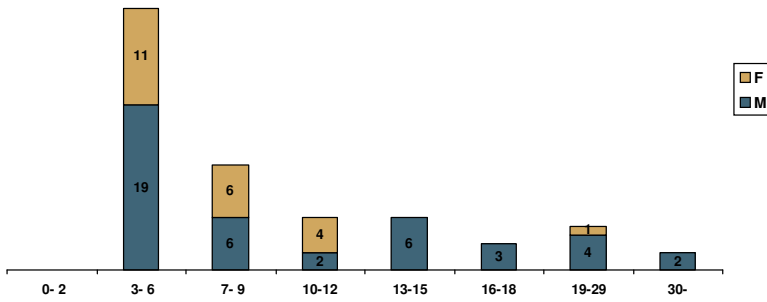
- Orofacial/odontological treatment:
- Problems in managing oral hygiene and tooth brushing justify extra preventive dental care.
- Oral motor skill training and extra stimulation are justified.
- Communication skills training is frequently essential.
- Tooth grinding should be followed up, and be managed with a splint when necessary.
- 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 to correct bite problems.

Source:

The rare disease database of the Swedish National Board of Health and Welfare.
The MHC database - The Mun-H-Center database of orofacial manifestations in rare diseases.

The Newsletter of the Ågrenska Center.

Age distribution

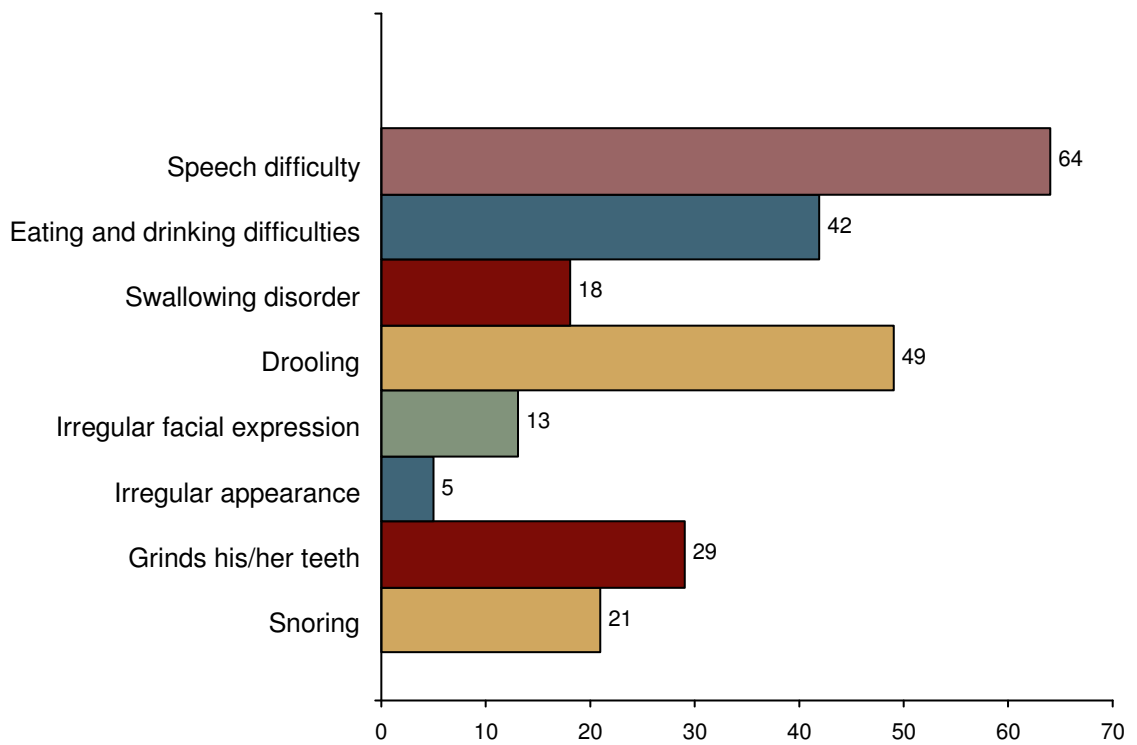


Number: 64

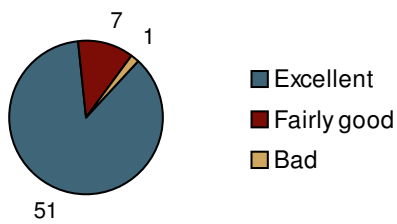
Ages: 3 -- 31 years

Sex: M (42) + F (22)

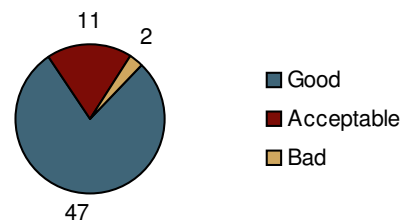
Orofacial problems



Oral health

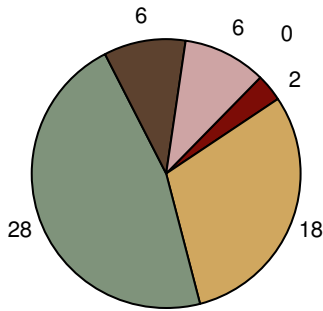


Oral hygiene



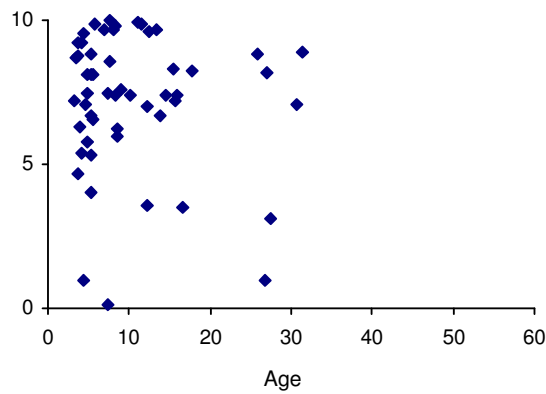
Behaviour in the treatment situation

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



- (0) ■ Examination is possible without problem
- (2) ■ Examination is possible without problems, some reaction is observed
- (18) ■ Examination can continue if adjusted to patient's reactions
- (28) ■ Reactions are considerable and examination is obviously affected
- (6) ■ Examination is practically impossible to complete
- (6) ■ Patient refuses examination

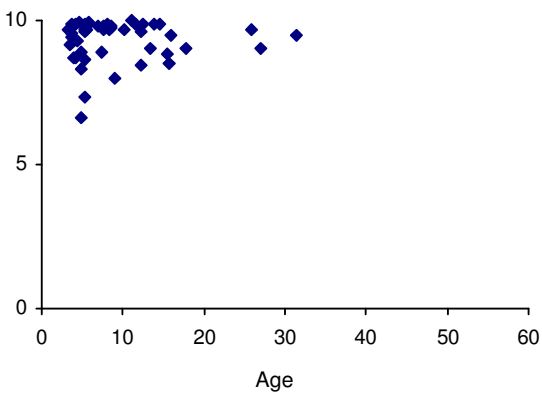
How does the patient cope with treatment in general?
0=no problems/10=great problems



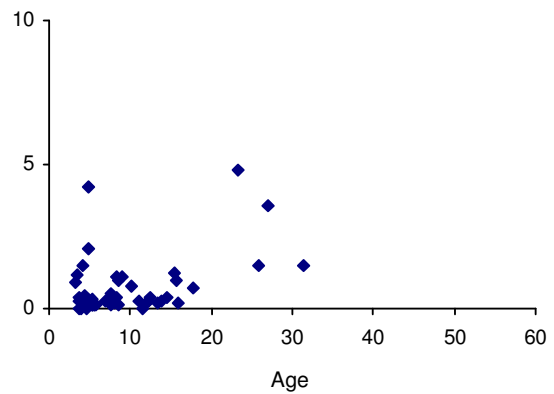
If there are treatment problems:

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

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



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





Clinical findings	Total N=64	Boys/Men N=42	Girls/Women N=22	Not evaluated
Speech difficulty	64	42	22	0
Open mouth at rest	51	32	19	0
Drooling	47	33	14	1
Low muscle tone in upper lip	43	23	20	1
Other oral habits	43	24	19	2
Low muscle tone in lower lip	39	22	17	1
Reduced mobility in tongue	36	22	14	2
Frontal open bite	33	17	16	1
Mask-like expression	31	20	11	1
Spacing	30	21	9	1
Lower jaw seems large	29	19	10	1
Low muscle tone in tongue	28	17	11	2
Tongue between front teeth	28	16	12	2
Tongue between front teeth when swallowing	25	13	12	3
Upper jaw seems large	24	17	7	1
Wide palate	24	17	7	3
Pre normal bite	22	12	10	0
Gingivitis	20	13	7	4
Grinds his/her teeth	20	12	8	12
Lower lip is flaccid and inactive	20	8	12	1
Abrasion - insignificant	15	8	7	8
Post normal bite	15	10	5	1
Mouth breathing	14	9	5	20
Hypomineralisation	12	10	2	10
Molar contact only	12	5	7	2
Upper lip is inactive and raised	12	6	6	1
Cross bite	10	6	4	5
Dental trauma	10	5	5	6
Horizontal over-bite 6 mm or more	10	7	3	3
Philtrum seems short	10	7	3	0
Proclined upper incisors	10	7	3	2
Supra-gingival calculus	9	6	3	8
Abrasion - significant	7	5	2	7
Concave facial profile	7	3	4	0
Occlusal reflex	7	3	4	10
Tongue seems large	7	4	3	1
Frontal inversion	6	4	2	3
M mentalis is overactive	6	4	2	0
Narrow palate	6	3	3	4
Edge to edge bite	5	3	2	2
Low muscle tone in masticatory muscles	5	4	1	6
Upper jaw seems small	5	2	3	2
Deep bite without gingival contact	4	4	0	2
Facial asymmetry	4	2	2	0
Long face	4	1	3	1
Reduced stability in neck	4	4	0	3
Sucks/bites on lower lip	4	3	1	0