



Orofacial function of persons having Achondroplasia Report from observation charts

The survey comprises 33 observation charts.

Estimated occurrence: Approximately 100 individuals have been diagnosed in Sweden. Two to three children per year are born in Sweden with a diagnosis of achondroplasia, which corresponds to about 11-12 per million inhabitants.

Aetiology: Achondroplasia is caused by a mutation on chromosome 4. It is inherited as an autosomal dominant trait. A new change in the genetic code, a spontaneous mutation, is the cause of the syndrome in 80-90% of cases.

General symptoms: Achondroplasia is a disease of the skeleton that affects growth. Children with achondroplasia are born with short arms and legs. Total body length is usually within the bounds of the normal, but on the short side. The skull is larger than average. After the first few months, the baby's growth slows down dramatically, and as early as by the age of 9 months, the child is seriously short for age. As he or she continues to grow, the arms and legs grow slowly, while trunk growth is closer to normal. Other characteristics include reduced muscle strength, joint laxity (particularly the knees), and difficulties in balancing the head.

Orofacial/odontological symptoms: While skull growth is greater than average, the face does not keep pace. This is especially true of the mid-face, and may result in narrowing of the respiratory canals, which, in turn, increases the risk of snoring, ear infections and sleep apnea (cessation of breathing during sleep). The small size of the mid-face affects the upper jaw with growth impairment, which often results in a protruding jaw. The teeth are of normal size, but both upper and lower teeth may be crowded. An open frontal bite may also occur – a distance between the upper and lower front teeth may make it difficult to take bites prior to chewing.

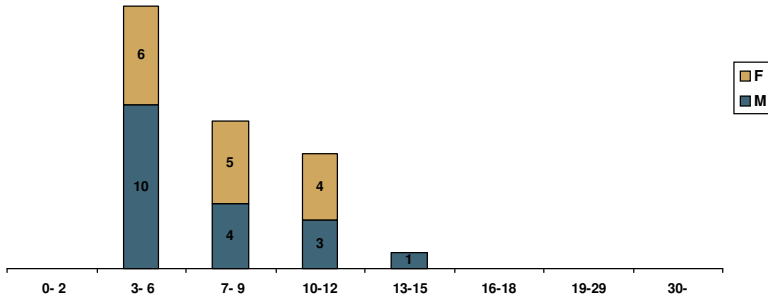
Orofacial/odontological treatment:

- It is important that these children come into early contact with the dental services for extra preventive dental care, information about oral hygiene, and monitoring of occlusal development.
- An orthodontist should be consulted when the child is between the ages of 7 and 9 in order to determine whether there are any dental or bite anomalies and whether corrective treatment is necessary.
- In cases of severe malocclusion, orthodontic treatment in combination with craniofacial surgery may be needed in late adolescence.
- Problems associated with snoring and sleep apnea should be followed up by a physician.

Sources

The rare diseases database of the Swedish National Board of Health and Welfare.
The MHC database – The database of Mun-H-Center on orofacial manifestations associated with rare diseases.
The newsletter of the Ågrenska Center.

Age distribution

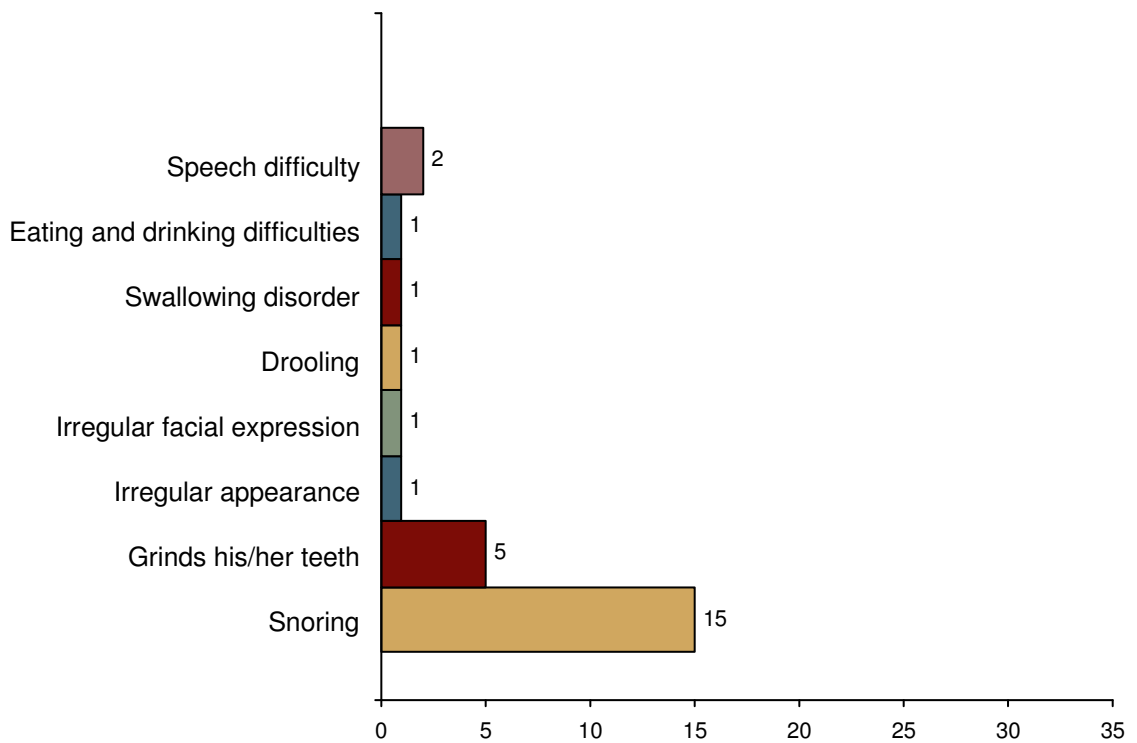


Number: 33

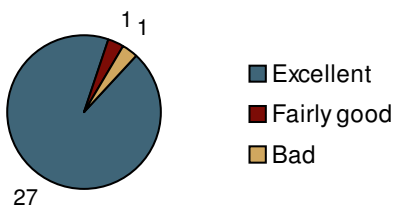
Ages: 3 -- 15 years

Sex: M (18) + F (15)

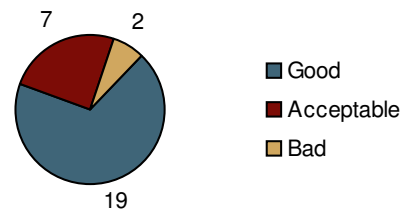
Orofacial problems



Oral health

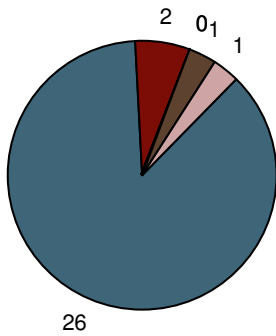


Oral hygiene



Behaviour in the treatment situation

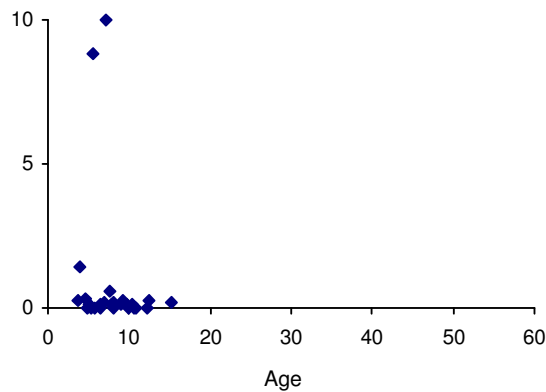
How calm and co-operative is the patient at time of examination?



- (26) ■ Examination is possible without problem
- (2) ■ Examination is possible without problems, some reaction is observed
- (0) ■ 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

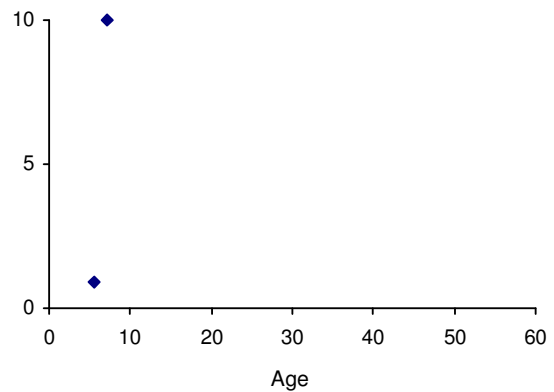
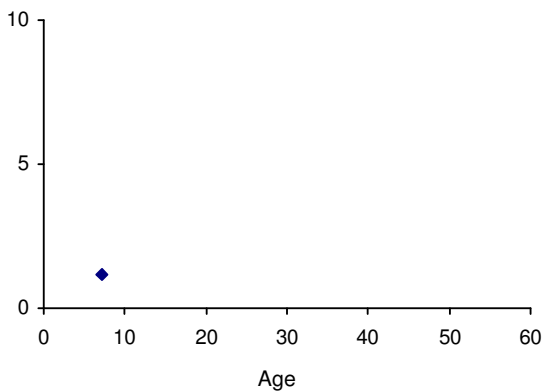


If there are treatment problems:

To what degree are the problems, if any, due to NN's handicap? 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=33	Boys/Men N=18	Girls/Women N=15	Not evaluated
Frontal open bite	16	8	8	3
Open mouth at rest	15	10	5	4
Concave facial profile	12	6	6	4
Mouth breathing	12	9	3	6
Pre normal bite	11	4	7	4
Hypomineralisation	10	5	5	5
Retroclined lower incisors	8	3	5	4
Narrow palate	7	2	5	6
Cross bite	5	2	3	4
Gingivitis	5	2	3	5
High palate	5	2	3	6
Philtrum seems short	5	1	4	4
Frontal inversion	4	3	1	4
Edge to edge bite	3	2	1	4
Molar contact only	3	0	3	4
Other oral habits	3	0	3	3
Over crowding	3	1	2	4
Tongue between front teeth	3	1	2	4
Abrasion - insignificant	2	2	0	5
Dental trauma	2	0	2	5
Difficulty in breathing	2	1	1	7
Facial asymmetry	2	0	2	3
Grinds his/her teeth	2	0	2	4
Horizontal over-bite 6 mm or more	2	2	0	4
Long face	2	1	1	3
M mentalis is overactive	2	1	1	3
Reduced mobility in neck	2	0	2	2
Spacing	2	1	1	4
Speech difficulty	2	1	1	3
Upper jaw seems small	2	1	1	4
Corner of mouth lowered	1	0	1	4
Cranio-facial abnormality	1	0	1	3
Deep bite with gingival contact	1	1	0	4
Enamel hypoplasia	1	1	0	5
Low muscle tone in lower lip	1	0	1	4
Lower jaw seems large	1	1	0	4
Lower jaw seems small	1	1	0	4
Philtrum seems long	1	1	0	4
Post normal bite	1	1	0	4
Posturing forward	1	0	1	4
Proclined upper incisors	1	0	1	4
Short tongue frenulum	1	0	1	5
Supra-gingival calculus	1	0	1	5
Tongue between front teeth when swallowing	1	1	0	4
Tongue presses against lower front teeth	1	0	1	4
Tongue rests over molars	1	0	1	4
Tongue seems large	1	0	1	4
Upper jaw seems large	1	1	0	4
Upper lip is inactive and raised	1	0	1	4
Wide palate	1	1	0	6