Orofacial function of persons having Achondroplasia

Report from observation charts

The survey comprises 56 observation charts.

Estimated occurrence: 5:100 000 live births.

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. Achondroplasia is the most common type of diagnosis with a short stature and the mean length for men is 132 cm and for women 125 cm. 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. Due to a narrow spinal cord there is an increased risk for neurological symptoms such as pain, sensory loss or pricking sensation.

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). Frequent ear infections may cause hearing impairment. 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.

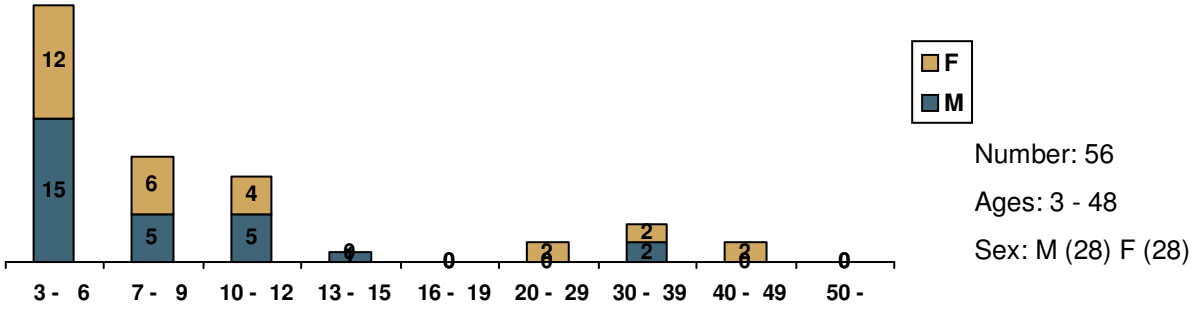
Orofacial/odontological treatment:

- It is important that these children receive dental preventive treatment, with extra prophylactic care and information on oral hygiene from an early age.
- Regular check-ups of dental and jaw development. Orthodontist should be consulted when needed.
- 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 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



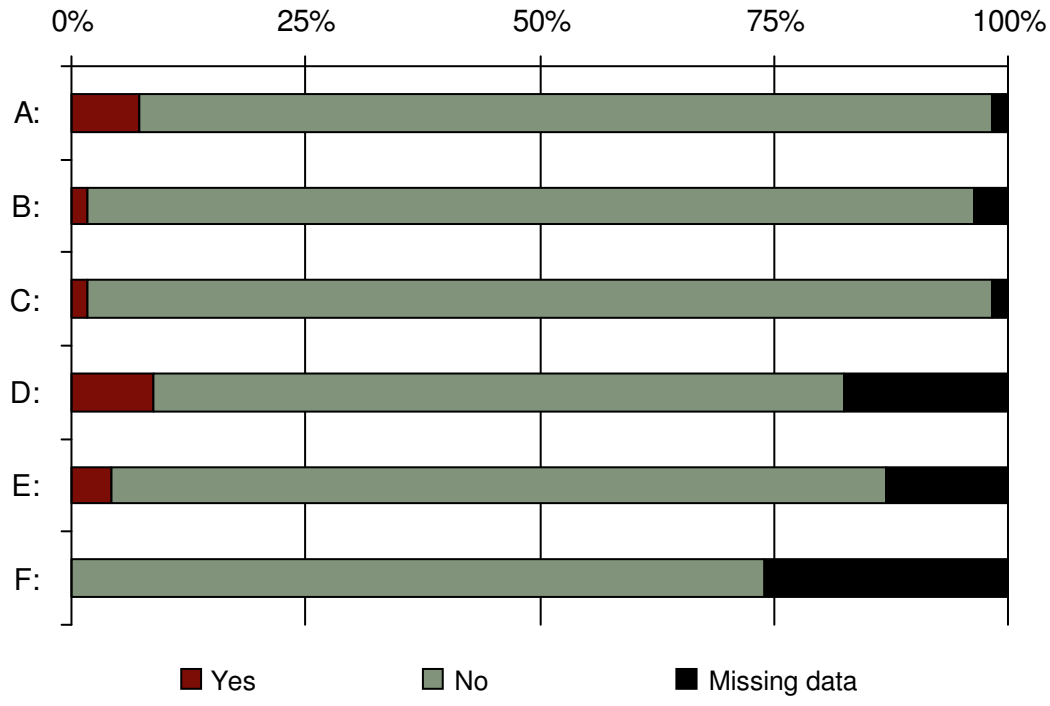
Number: 56

Ages: 3 - 48

Sex: M (28) F (28)

Overview

		Yes	No	Missing data	N
A:	Incomprehensible speech/No speech	4	51	1	56
B:	Eating and drinking difficulties[1]	1	53	2	56
C:	Profuse drooling, on clothes[1]	1	54	1	56
D:	Breathing difficulties[1] [2]	2	17	4	23
E:	Grinding every day[1] [2]	1	19	3	23
F:	Severe malocclusions[2]	0	17	6	23



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)[1]

		0	1	2	3	4	5	6	Missing data	N
Calc	Calculus	20	3	0	0	0	0	0	0	23
GI	Gingivitis	18	5	0	0	0	0	0	0	23
Plaq	Coating	19	4	0	0	0	0	0	0	23
Toot	Tooth wear	19	4	0	0				0	23

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.

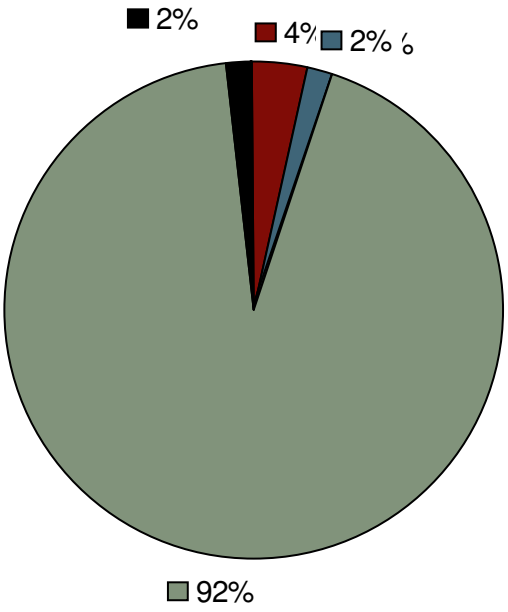
PI Plaque index is based on the presence of visible plaque on the buccal surface of 6 index teeth. 0 indicates that there is no plaque, 6 indicates plaque on all index teeth.

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.

1: Oral health index (indices) was (were) introduced in the observations in 2008

Acceptance of dental examination



	Number
None	2
Negative	1
Reluctant	0
Positive	52
Missing data	1
Sum:	**56**

Caries

	3-6 years	7-12 years	13-19 years	Adults
deft[1]				
Examined	25	18		
Number of individuals with deft=0	21	13		
Mean	0,8	0,5		
Standard deviation	2,3	1,0		
Missing data	2	2		
DMFT[2]				
Examined		17	1	8
Number of individuals with DMFT=0		14	0	0
Standard deviation		1,0	0,0	5,7
Mean		0,4	1,0	7,8
Missing data		3	0	0

1: Number of carious or filled deciduous teeth

2: Number of carious or filled permanent teeth

Occlusal relationship

	Number
Neutral bite	40
Post normal	1
Pre normal	13
Missing data	2
	Sum: 56

Maximum jaw opening

Children younger than 10 years

	Number
- 20	0
21 - 30	4
31 - 40	15
41 - 50	7
51 -	1
Missing data	11
	Sum: 38

Children, 10 years or older, and adults

	Number
- 20	0
21 - 30	0
31 - 40	0
41 - 50	14
51 -	4
Missing data	0
	Sum: 18

Profile[1]

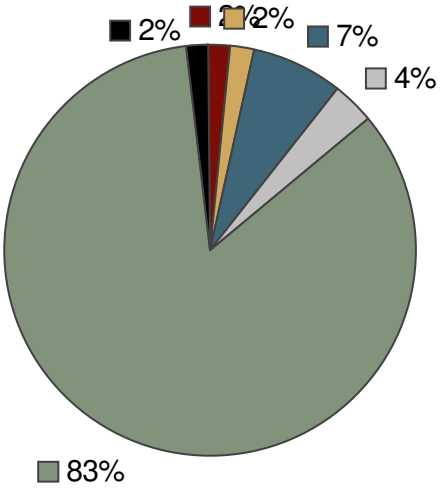
	Number
Normal	9
Convex	0
Concave	11
Missing data	3
	Sum: 23

Mandibular plane[1]

	Number
Normal	10
Increased	3
Reduced	0
Missing data	10
	Sum: 23

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

Speech difficulty



	Number
No speech	1
Very incomprehensible	1
Incomprehensible speech	4
Slightly indistinct speech	2
No problems	47
Missing data	1

Sum: 56

Clinical findings	Yes-answers Total N=56 (%)	Boys/Men N=28 (%)	Girls/Women N=28 (%)	Missing data
Open mouth at rest	**25** (45)	14 (50)	11 (41)	1
Frontal open bite	**24** (43)	11 (39)	13 (46)	0
Narrow palate	**11** (21)	3 (12)	8 (31)	4
Cranio-facial abnormality	**9** (23)	3 (15)	6 (30)	16
High palate	**9** (18)	3 (12)	6 (23)	5
Over crowding	**6** (11)	2 (7)	4 (14)	1
M mentalis overactive	**4** (7)	2 (7)	2 (7)	0
Spacing	**4** (7)	2 (7)	2 (7)	1
Facial asymmetry	**3** (5)	1 (4)	2 (7)	0
Low muscle tone in lips	**3** (5)	1 (4)	2 (7)	1