



Orofacial function of persons having Achondroplasia

Report from questionnaires

The survey comprises 19 questionnaires.

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.

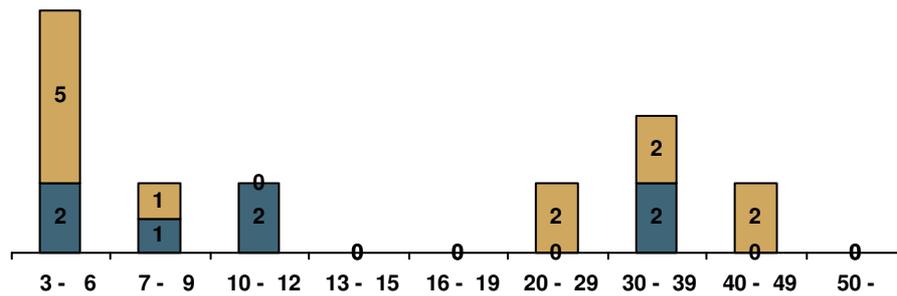
Orofacial/odontological treatment:

- 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: 19

Ages: 3 - 48

Sex: M (7) F (12)

Additional diagnoses

Medical impairment	Yes	No	Missing data
Inborn heart defect	0	19	0
Other cardiovascular disease	0	19	0
Epilepsy	1	18	0
Asthma	1	18	0
Need of respiratory support	2	17	0
Allergy	3	16	0

Neuropsychiatric diagnosis	Yes	No	Missing data
ADHD/ADD	0	19	0
Autistic syndrome	0	19	0
Autistic traits	0	19	0
Asperger's syndrome	0	19	0
Tourette's syndrome	0	19	0

General disability	Yes	No	Missing data
Intellectual disability	1	17	1
Mobility dysfunction	7	10	2
Impaired vision	4	12	3
Impaired hearing	8	8	3
Difficulty in communicating	2	15	2

About dental care and oral health

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

	Number
Yes, very much so	14
Yes, some what	2
No, not really	2
No, not at all	1
Missing data	0
Sum:	19

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

	Number
Three or more times per year	4
Twice per year	1
Once per year	10
Less than once per year	4
Missing data	0
Sum:	19

When were your teeth last X-rayed?

	Number
During the past two years	12
More than two years ago	0
Never had my teeth X-rayed	6
Missing data	1
Sum:	19

Do you look after your teeth in a good way?

	Number
Yes, very much so	5
Yes, some what	14
No, not really	0
No, not at all	0
Missing data	0
Sum:	19

Who brushes your teeth?

	Number
I always brush myself	10
Sometimes I brush myself	5
Someone else always helps me	4
Missing data	0
Sum:	19

How often are your teeth brushed?

	Number
Three or more times per day	0
Twice per day	19
Once per day	0
Not every day	0
Missing data	0
Sum:	19



About dental care and oral health

	Yes	No	Missing data
Does your mouth hurt?	1	16	2
Does your mouth feel dry?	2	14	3
Have you ever taken a serious hit to your permanent front teeth?	1	17	1
Do you feel that you have a divergent bite?	4	13	2
Have you had a brace?	5	14	0
Do you feel that you need orthodontics/a brace?	4	12	3

Do you grind or press your teeth at night?

	Number
Never	12
Once or twice per week	5
Every night	1
Missing data	1
Sum:	19

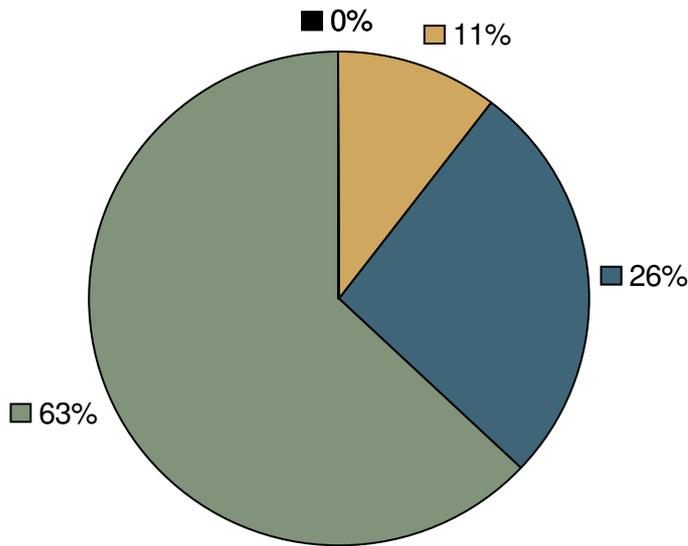
Do you grind or press your teeth during the day?

	Number
Never	16
Once or twice per week	2
Every day	0
Missing data	1
Sum:	19



About eating

Do you have any problems with eating?



	Number
Yes, very much so	0
Yes, some what	2
No, not really	5
No, not at all	12
Missing data	0
Sum:	19

	Yes	No	Missing data
Do you cough daily in connection with meals?	1	18	0
Do you gag daily in connection with meals?	0	19	0
Do you get acid reflux daily?	0	19	0
Do you throw up often (at least twice per week)?	0	19	0
Do you have a poor appetite?	4	15	0
Does it take a long time before you can swallow a mouthful?	3	16	0
Do you press your tongue forward when you swallow so that food ends up outside the mouth?	0	18	1
Do you find it difficult to chew, i.e. grind food using your molars?	1	9	9
Do you find it difficult to take food from the spoon using your lips?	0	12	7
Have you had problems with food and drink leaking out through the corners of your mouth?	0	12	7
Does food tend to remain in your mouth after meals?	0	12	7
Do you get nutrition in any other way than through your mouth?	0	19	0



About drooling

Do you drool?

	Number
Never drool	17
Drool sometimes – not every day	2
Drool often – every day	0
Constant drooling	0
Missing data	0
Sum:	19

How much do you drool?

	Number
Slight drooling, only on the lips	1
Moderate drooling, on lip and chin	1
Profuse drooling, on clothes	0
Very profuse drooling, on hands/objects	0
Missing data	0
Sum:	2

Is your drooling a problem for you?

	Number
Yes, very much so	0
Yes, some what	0
No, not really	0
No, not at all	2
Missing data	0
Sum:	2

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

	Number
Yes, very much so	0
Yes, some what	0
No, not really	0
No, not at all	2
Missing data	0
Sum:	2