Orofacial function of persons having
Apert syndrome
Report from observation charts

The survey comprises 21 observation charts.

Synonym: Acrocephalosyndactyly type 1

Estimated occurrence: 1:100 000 live births.

Etiology: Autosomal dominant genetic trait with aberration on chromosome 10. Most often spontaneous mutation.

General symptoms: Cranial malformations and malformations of the hands and feet. Fusion of the cervical vertebrae is frequent. Children with Apert Syndrome often have delayed speech and language development, as well as learning disabilities. They also generally have vision and hearing problems. Severe acne is common while growing up.

Orofacial/odontological symptoms: Premature fusion of the bones at the base of the skull (craniosynostosis) gives rise to craniofacial malformations. The palate is often high and narrow, and some children are born with a cleft soft palate. Crowded teeth and an underbite are common. In a recently published study irregularities were noted in the region of the dentino-enamel junction, which could affect caries progression and also make dental management more difficult. Constriction in the upper respiratory system may lead to breathing difficulties and sleep apnea (frequently stopping breathing when asleep). Craniofacial malformations lead to articulation difficulties and impair chewing ability.

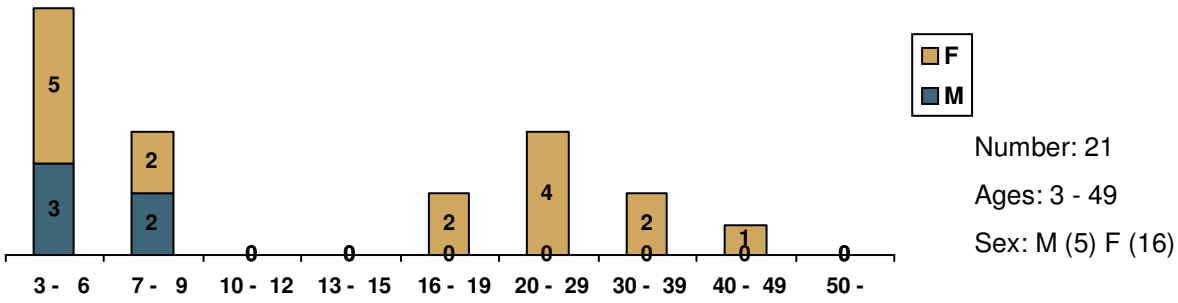
Orofacial/odontological treatment:

- In cases of craniofacial malformations, a specialist team will be needed for follow up and treatment.
- Most individuals with Apert syndrome require both orofacial surgery and extensive orthodontia..
- Many of these individuals require supplementary prophylactic dental care.
- Speech and language disabilities are dealt with by a speech therapist
- Snoring problems should be followed up by a physician.

Source

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.
Surman TL, Logan RM, Townsend GC, Anderson PJ. Oral features in Apert syndrome: a histological investigation

Age distribution



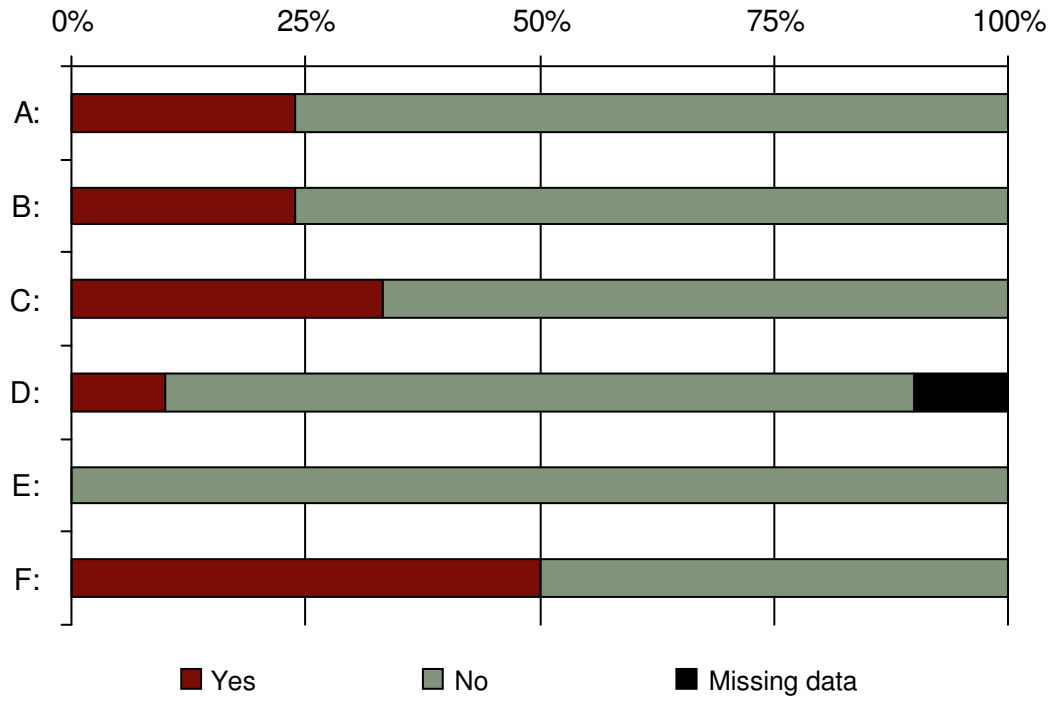
Number: 21

Ages: 3 - 49

Sex: M (5) F (16)

Overview

		Yes	No	Missing data	N
A:	Incomprehensible speech/No speech	5	16	0	21
B:	Eating and drinking difficulties[1]	5	16	0	21
C:	Profuse drooling, on clothes[1]	7	14	0	21
D:	Breathing difficulties[1] [2]	1	8	1	10
E:	Grinding every day[1] [2]	0	10	0	10
F:	Severe malocclusions[2]	5	5	0	10



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	7	2	0	0	0	0	0	1	10
GI	Gingivitis	7	0	0	0	1	0	1	1	10
Plaq	Coating	4	2	1	0	0	1	1	1	10
Toot	Tooth wear	7	2	0	0				1	10

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.

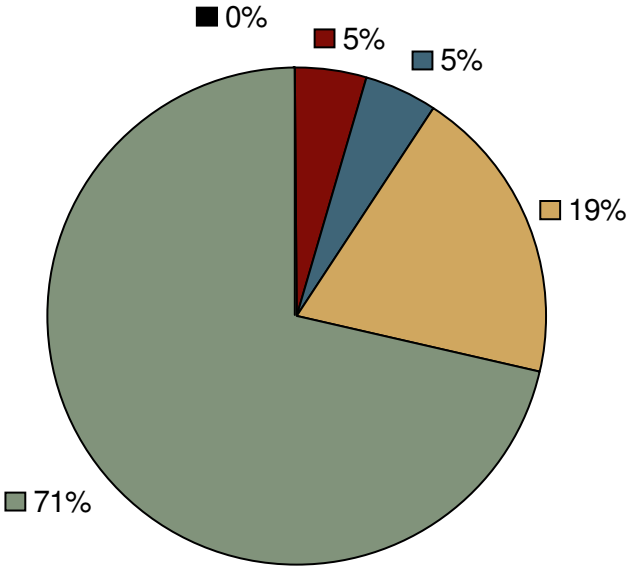
PI Plaque index is based on the presence of visible plaque on the buccal surface of 6 index teeth. 0 indicates that there is no plaque, 6 indicates plaque on all index teeth.

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	1
Negative	1
Reluctant	4
Positive	15
Missing data	0
Sum:	**21**

Caries

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

1: Number of carious or filled deciduous teeth

2: Number of carious or filled permanent teeth

Occlusal relationship

	Number
Neutral bite	6
Post normal	0
Pre normal	14
Missing data	1
	Sum: 21

Maximum jaw opening

Children younger than 10 years

	Number
- 20	0
21 - 30	2
31 - 40	5
41 - 50	2
51 -	0
Missing data	3
	Sum: 12

Children, 10 years or older, and adults

	Number
- 20	0
21 - 30	0
31 - 40	3
41 - 50	5
51 -	1
Missing data	0
	Sum: 9

Profile[1]

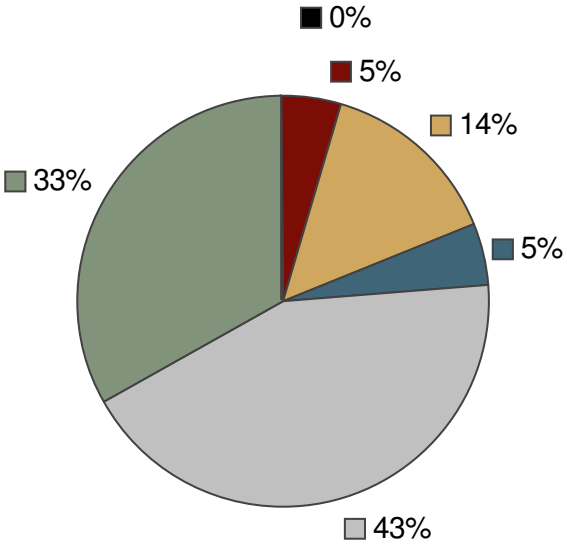
	Number
Normal	4
Convex	0
Concave	6
Missing data	0
	Sum: 10

Mandibular plane[1]

	Number
Normal	5
Increased	5
Reduced	0
Missing data	0
	Sum: 10

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

Speech difficulty



	Number
No speech	1
Very incomprehensible	3
Incomprehensible speech	1
Slightly indistinct speech	9
No problems	7
Missing data	0
Sum:	**21**

Clinical findings	Yes-answers Total N=21 (%)	Boys/Men N=5 (%)	Girls/Women N=16 (%)	Missing data
Cranio-facial abnormality	**21** (100)	5 (100)	16 (100)	0
Narrow palate	**19** (100)	4 (100)	15 (100)	2
Open mouth at rest	**16** (76)	3 (60)	13 (81)	0
Frontal open bite	**13** (65)	5 (100)	8 (53)	1
Over crowding	**11** (55)	3 (60)	8 (53)	1
High palate	**9** (45)	1 (20)	8 (53)	1
Cleft lip and palate	**8** (38)	2 (40)	6 (38)	0
Hyperplastic alveolar ridge	**7** (78)	2 (100)	5 (71)	12
Facial asymmetry	**6** (30)	1 (20)	5 (33)	1
M mentalis overactive	**5** (24)	1 (20)	4 (25)	0
Mucous membrane changes	**4** (20)	1 (20)	3 (20)	1