



Orofacial function of persons having Apert syndrome

Report from observation charts

The survey comprises 15 observation charts.

Synonym: Acrocephalosyndactyly type 1

Estimated occurrence: 1:100 000 live births.

Etiology: Autosomal dominant genetic trait. Most often spontaneous mutation. Diagnosis based on symptoms.

General symptoms: Cranial and malformations, 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 gives rise to craniofacial malformations. The head is broad and elongated, the midface underdeveloped, and the eyes bulge. The palate is often high and narrow, and some children are born with a cleft soft palate. Crowded teeth and an underbite are common. 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 Sweden, treatment of craniofacial malformations is centralized, and carried out at Sahlgrenska University Hospital, Göteborg.

- Most individuals with Apert syndrome require both orofacial surgery and extensive orthodontia.
- Speech and language disabilities are dealt with by a speech therapist.
- Many of these individuals require supplementary prophylactic dental care.
- Snoring problems should be followed up by a physician.

Source

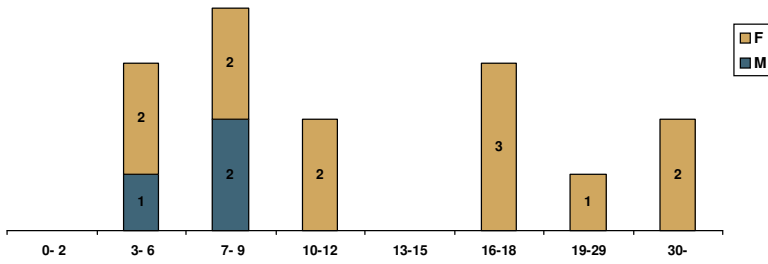
The rare disease database of the Swedish National Board of Health and Welfare.

The MHC database - The Mun-H-Center database of orofacial manifestations in rare diseases.

The Newsletter of the Ågrenska Center.

Apert syndrom – folder from Smågruppscentrum

Age distribution

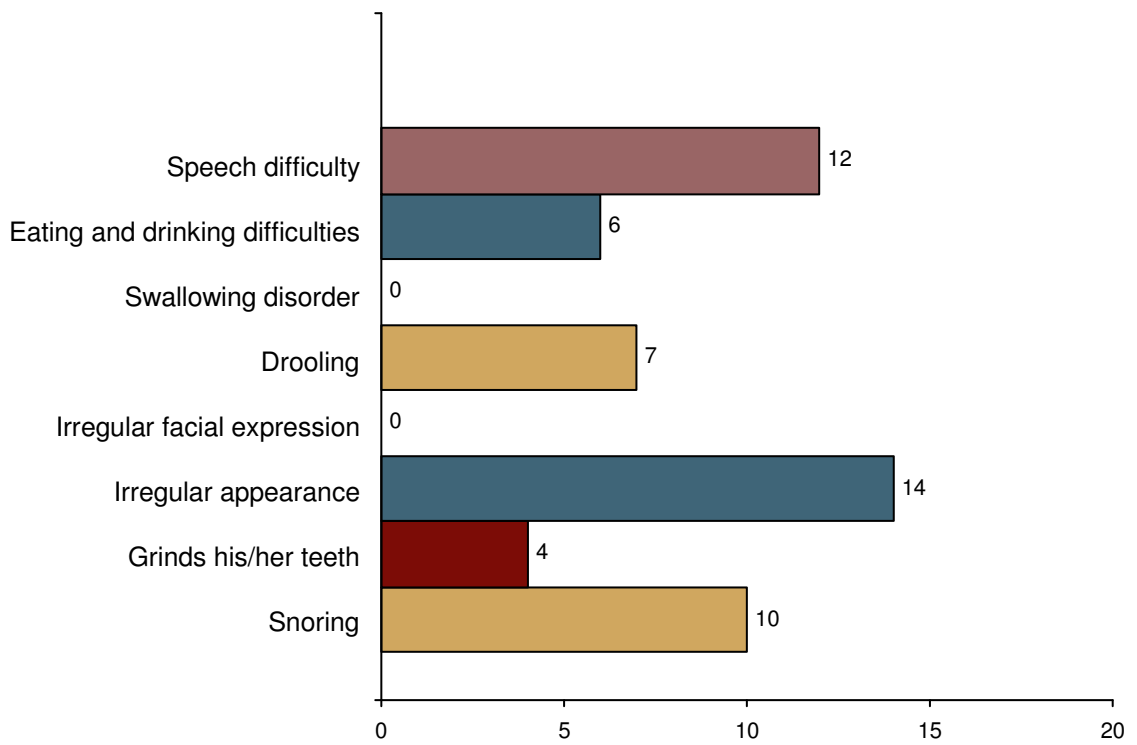


Number: 15

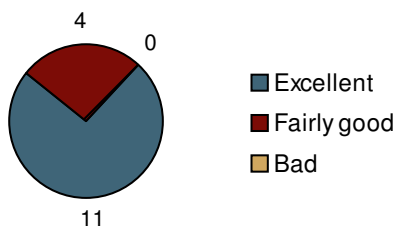
Ages: 4 -- 34 years

Sex: M (3) + F (12)

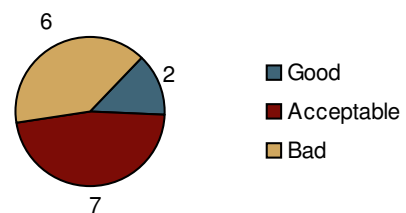
Orofacial problems



Oral health

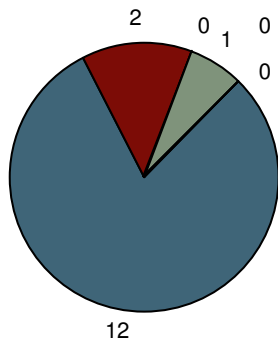


Oral hygiene



Behaviour in the treatment situation

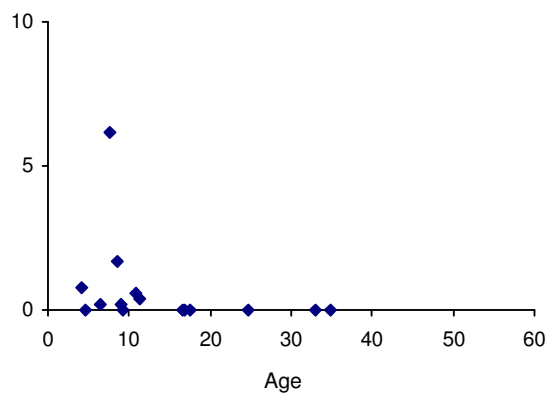
How calm and co-operative is the patient at time of examination?



- (12) ■ 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
- (1) ■ Reactions are considerable and examination is obviously affected
- (0) ■ Examination is practically impossible to complete
- (0) ■ Patient refuses examination

How does the patient cope with treatment in general?

0=no problems/10=great problems

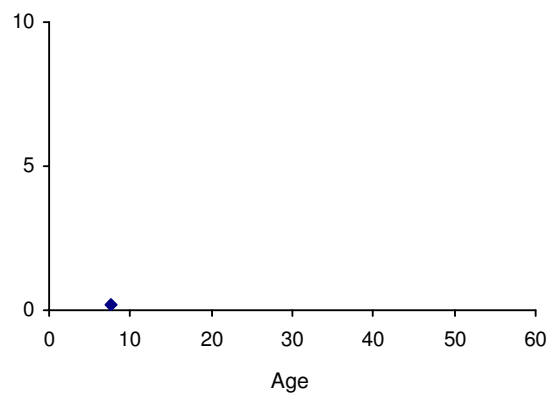
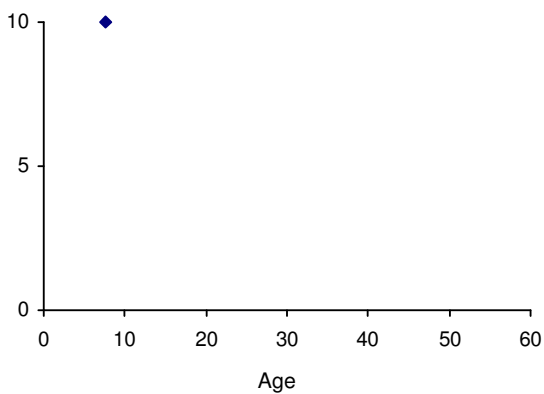


If there are treatment problems:

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=15	Boys/Men N=3	Girls/Women N=12	Not evaluated
Cranio-facial abnormality	15	3	12	0
Narrow palate	14	2	12	1
Gingivitis	13	2	11	0
Concave facial profile	12	3	9	0
Open mouth at rest	12	2	10	0
Pre normal bite	12	2	10	1
Speech difficulty	12	3	9	0
Upper jaw seems small	12	3	9	0
Frontal open bite	11	3	8	0
Over crowding	11	2	9	0
Cross bite	8	1	7	1
High palate	8	1	7	0
Divided uvula	7	2	5	4
Drooling	7	1	6	0
M mentalis is overactive	7	1	6	0
Molar contact only	7	2	5	0
Retroclined lower incisors	7	1	6	1
Mucous membrane change	6	1	5	0
Horizontal over-bite 6 mm or more	5	1	4	1
Lower jaw seems small	5	2	3	1
Retroclined upper incisors	5	1	4	1
Supra-gingival calculus	5	1	4	0
Cleft palate	4	1	3	1
Frontal inversion	4	1	3	1
Retained teeth	4	1	3	0
Gingival hyperplasia	3	0	3	0
Grinds his/her teeth	3	2	1	0
Hypomineralisation	3	0	3	0
Philtrum seems short	3	0	3	0
Tongue between front teeth	3	1	2	0
Tongue between front teeth when swallowing	3	1	2	0
Tooth anomaly	3	1	2	0
Deviation on opening	2	0	2	0
Facial asymmetry	2	1	1	0
Hyper-plastic gingivitis	2	0	2	0
Mouth breathing	2	1	1	1
Abrasion - insignificant	1	1	0	0
Corner of mouth lowered	1	0	1	0
Edge to edge bite	1	0	1	1
Gingival retraction	1	0	1	0
Long face	1	1	0	2
Periodontitis	1	0	1	0
Philtrum seems long	1	0	1	0
Proclined upper incisors	1	0	1	1
Reduced mobility in soft palate	1	1	0	6
Reduced mobility in tongue	1	1	0	1
Scissor bite	1	0	1	1
Sub-gingival calculus	1	0	1	0
Supernumeraries	1	1	0	0