



Orofacial function of persons having Aicardi syndrome

Report from observation charts

The survey comprises 13 observation charts.

Estimated occurrence: Aicardi syndrome is estimated to occur in 1/500,000 births. In Sweden, 13 individuals with the syndrome have been identified.

Aetiology: A change (mutation) on the short arm of the X chromosome. The exact location has not been fully specified.

General symptoms: Aicardi affects girls and women. Various brain defects may occur, and give rise to different symptoms. Between the ages of 3 and 6 months these girls develop epileptic seizures of the infantile spasm type. Later, different kinds of seizures commonly occur, and the epilepsy may be life-long. Most individuals with Aicardi syndrome have severe mental retardation. Most never learn to speak or walk. Eye defects are common, with consequent visual impairment. There may be other deformities as well.

Orofacial/odontological symptoms: Eating difficulties, drooling and tooth grinding with wear and tear on the teeth are common. Poor muscle tonus of the orofacial musculature is also common, and tends to impact on the development of face and jaws. Thus occlusal (bite) anomalies are frequent, for example, post-normal occlusion (Class II malocclusion) and a large horizontal overjet. There is an increased risk of tooth cavities (caries) and gingivitis.

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.
- Feeding and swallowing difficulties should be investigated and treated by a specialist team at the hospital (by a nutrition or dysphagia team) or at a multidisciplinary treatment center.
- Tooth grinding should be followed up, and possibly treated with a splint.
- 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 occlusal anomalies and whether corrective treatment is necessary.
- Orofacial therapy, stimulation of the musculature around the mouth, is often needed.
- Learning alternative and augmentative ways to communicate may be important.

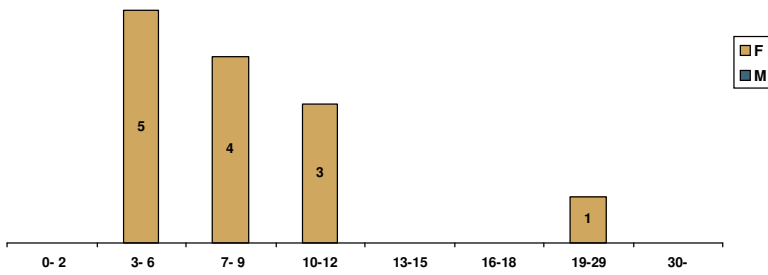
Sources:

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

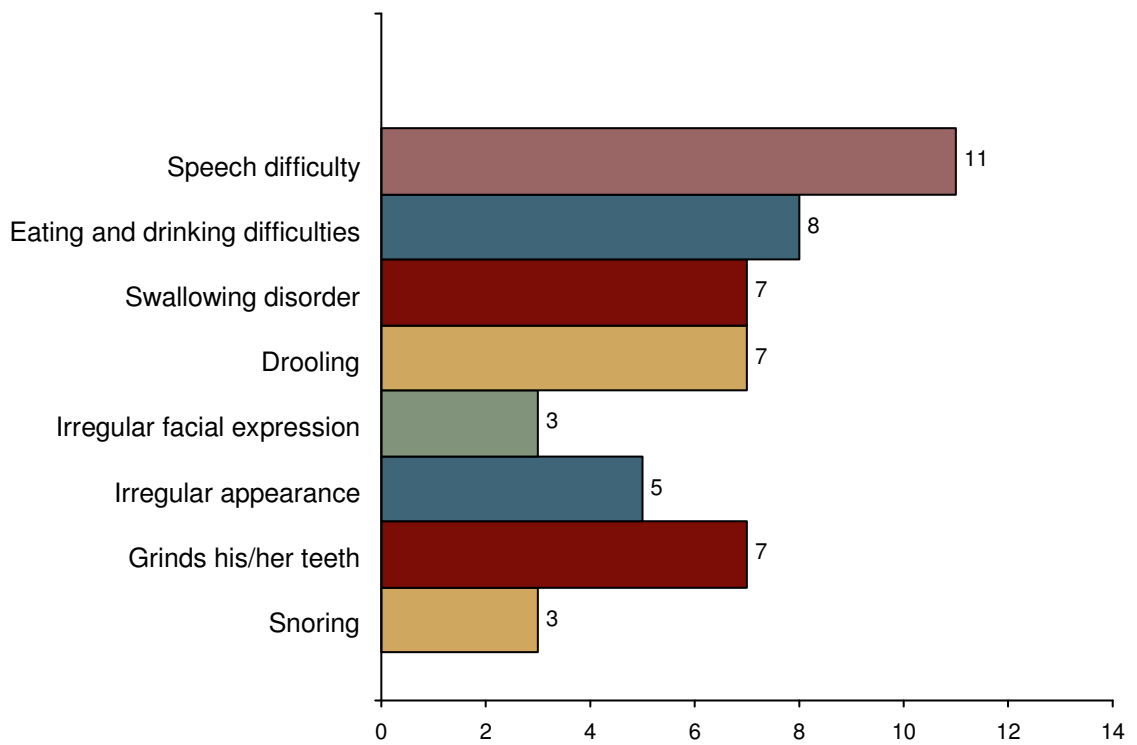


Number: 12

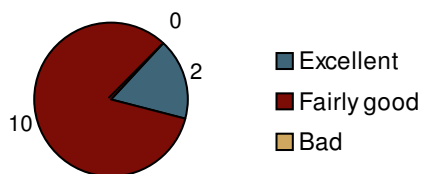
Ages: 4 -- 27 years

Sex: M (0) + F (12)

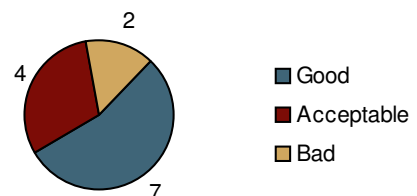
Orofacial problems



Oral health

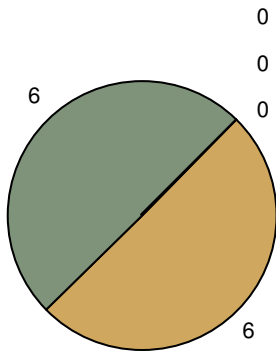


Oral hygiene



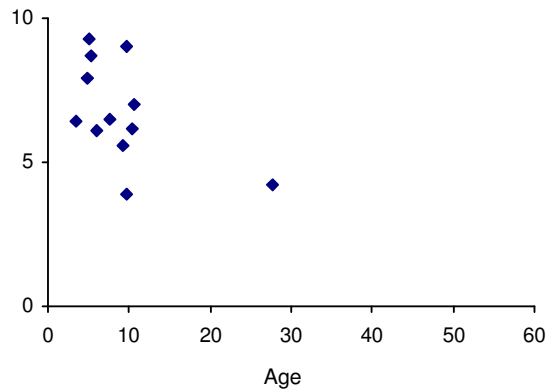
Behaviour in the treatment situation

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



- (0) ■ Examination is possible without problem
- (0) ■ Examination is possible without problems, some reaction is observed
- (6) ■ Examination can continue if adjusted to patient's reactions
- (6) ■ 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

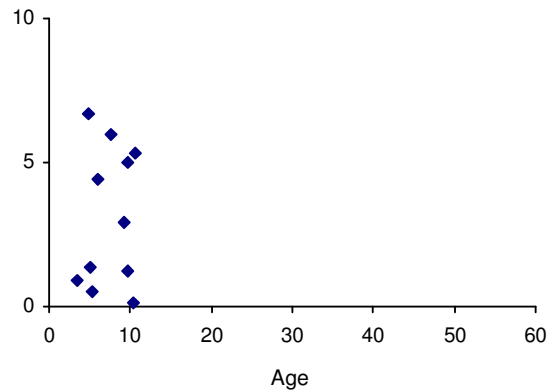
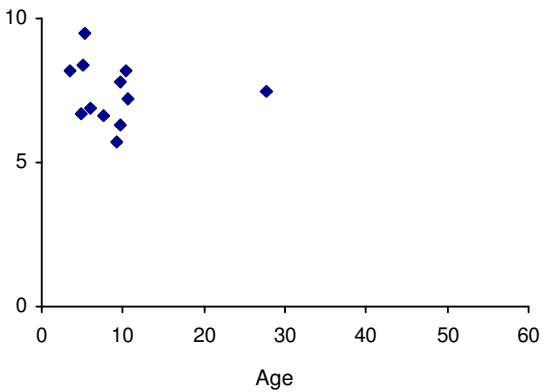


If there are treatment problems:

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=12	Boys/Men N=0	Girls/Women N=12	Not evaluated
Mask-like expression	11	0	11	0
Reduced mobility in neck	11	0	11	0
Reduced stability in neck	11	0	11	0
Speech difficulty	11	0	11	0
Low muscle tone in lower lip	10	0	10	0
Low muscle tone in upper lip	10	0	10	0
Reduced mobility in tongue	10	0	10	1
Low muscle tone in tongue	9	0	9	1
Drooling	7	0	7	0
Grinds his/her teeth	7	0	7	1
Open mouth at rest	7	0	7	0
Facial asymmetry	6	0	6	0
Gingivitis	6	0	6	0
Horizontal over-bite 6 mm or more	6	0	6	0
Other oral habits	6	0	6	0
Proclined upper incisors	6	0	6	1
Abrasion - significant	5	0	5	1
M mentalis is overactive	5	0	5	0
Post normal bite	5	0	5	2
Upper lip is inactive and raised	5	0	5	0
Deep bite with gingival contact	4	0	4	2
Hypomineralisation	4	0	4	1
Lower lip is flaccid and inactive	4	0	4	0
Spacing	4	0	4	1
Supra-gingival calculus	4	0	4	0
Proclined lower incisors	3	0	3	1
Tongue between front teeth when swallowing	3	0	3	1
Abrasion - insignificant	2	0	2	1
Difficulty in breathing	2	0	2	0
Facial palsy	2	0	2	0
Hyper-plastic gingivitis	2	0	2	1
Hyper-sensitive in oral cavity	2	0	2	1
Over crowding	2	0	2	0
Posturing forward	2	0	2	1
Tongue presses against lower front teeth	2	0	2	1
Tongue presses against upper front teeth	2	0	2	1
Tooth anomaly	2	0	2	1