



Orofacial function of persons having Aicardi syndrome

Report from questionnaires

The survey comprises 13 Questionnaires.

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.

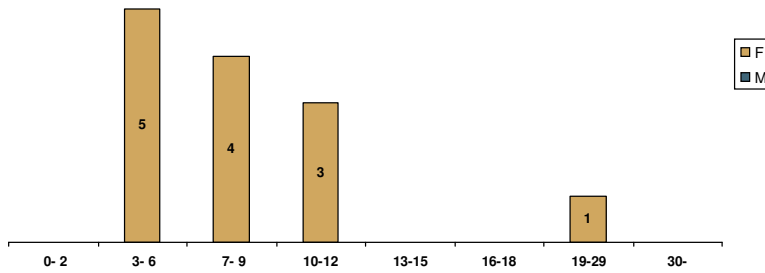
Sources:

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

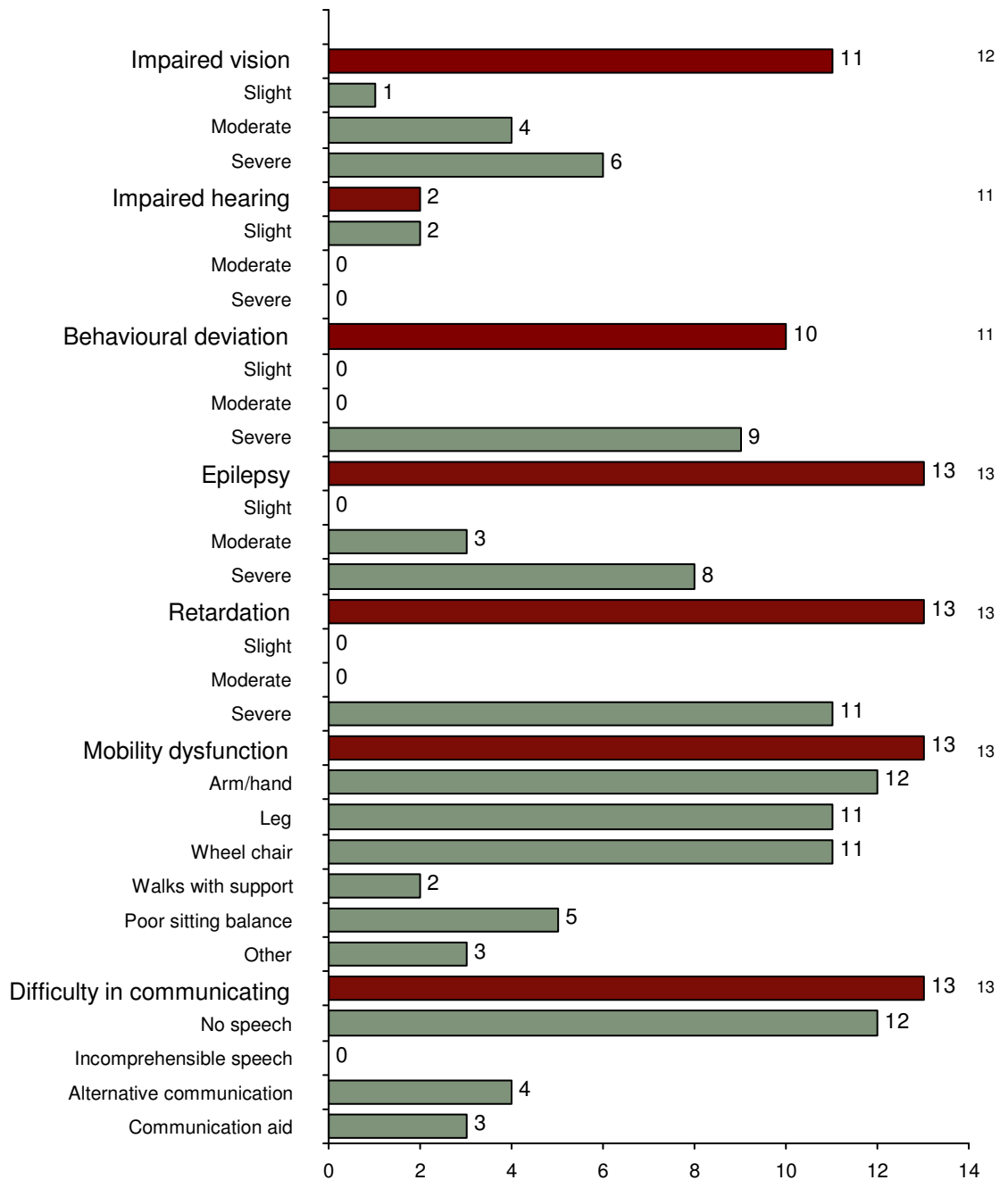


Number: 13

Ages: 4 -- 27 years

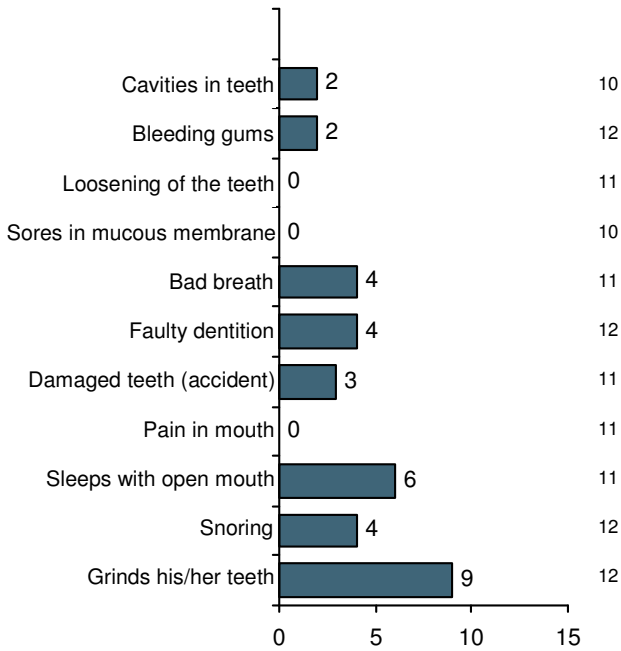
Sex: M (0) + F (13)

General disabilities

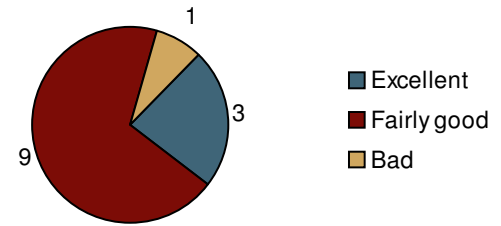


About dental health

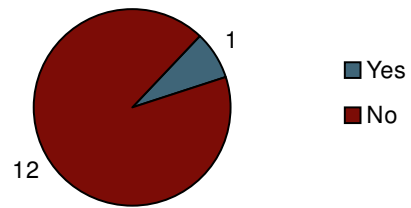
About dental health - problems



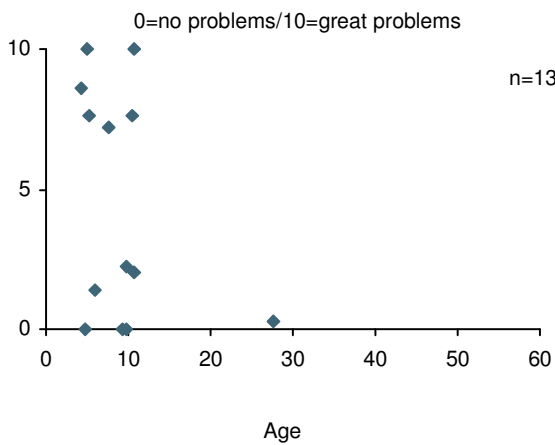
Oral health



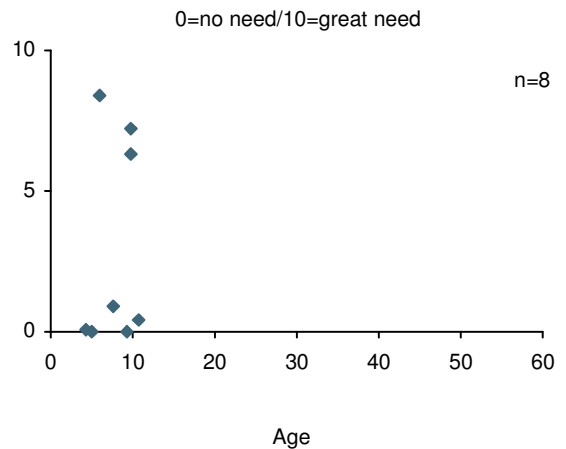
Has NN received orthodontic treatment?



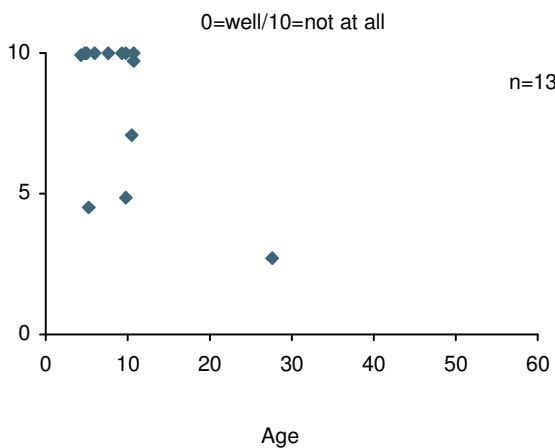
How does NN experience the dental care received?



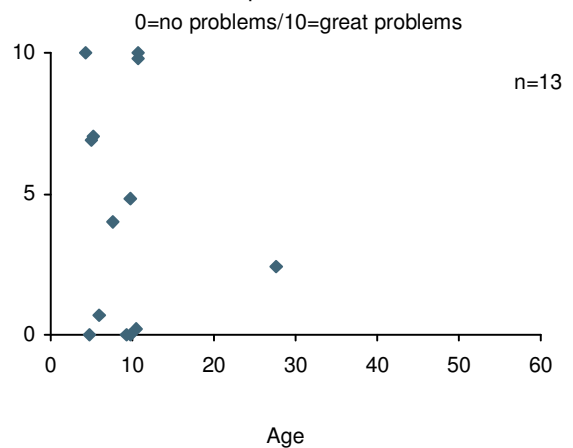
NN is considered to be in need of orthodontic treatment



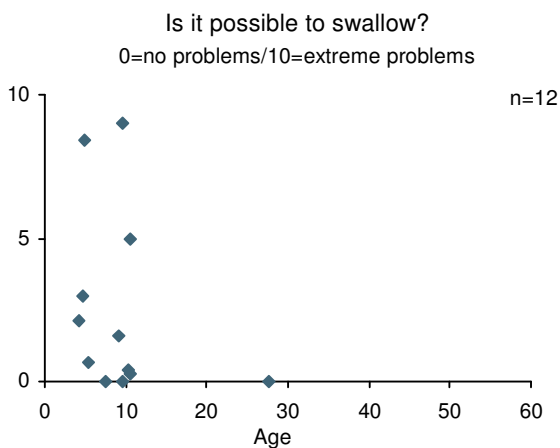
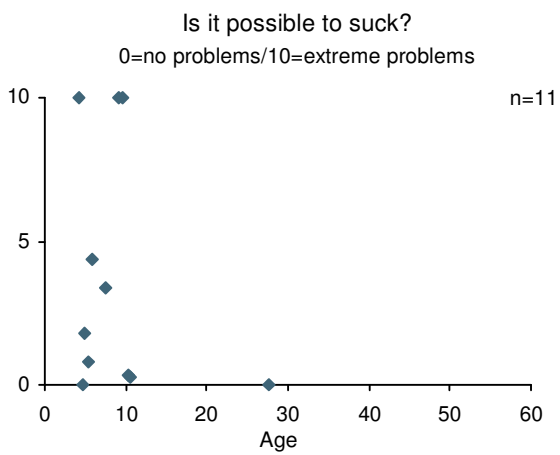
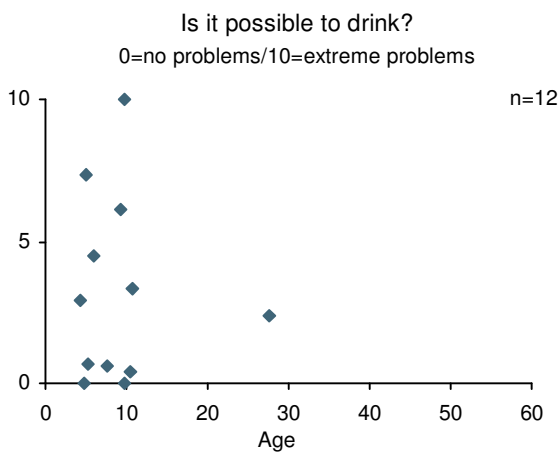
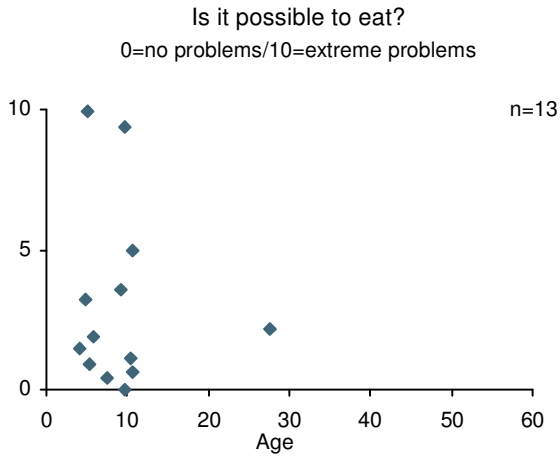
How does NN manage to brush his/her teeth?



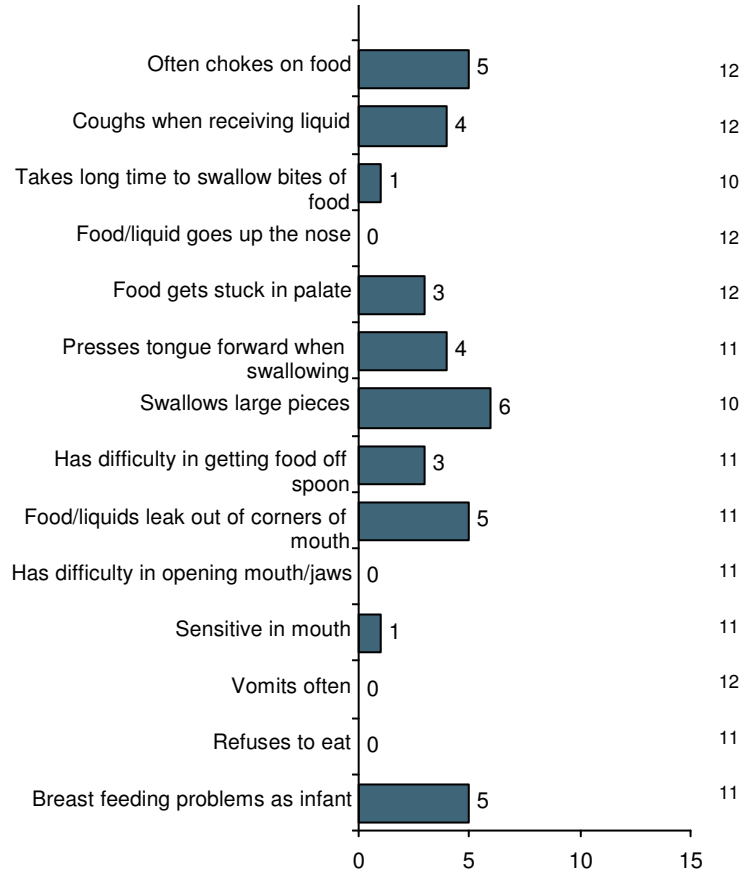
If help is needed with brushing teeth, how does it proceed?



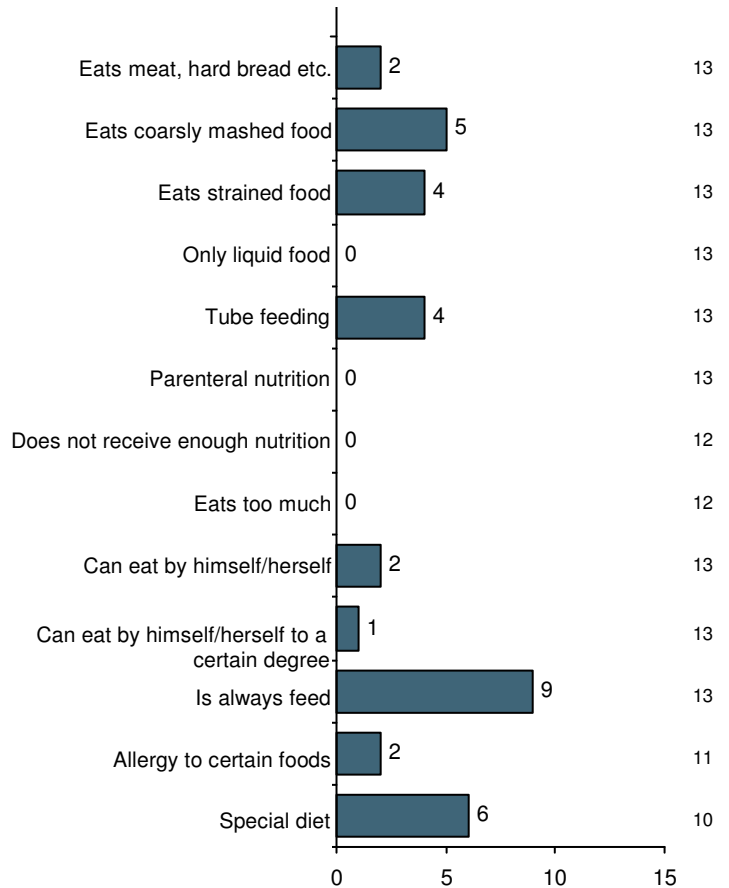
About eating habits



About eating habits - problems



Food habits



About drooling

