



Orofacial function of persons having Cystinosis

Report from questionnaires

The survey comprises 9 Questionnaires.

Synonyms: Nephropathic cystinosis

Estimated occurrence: The incidence in Sweden is unknown; however, an incidence of 1 child per 160,000 to 200,000 live births has been reported in other countries.

Etiology: Cystinosis is caused by a mutation in the so-called CTNS gene on chromosome 17. This mutation causes an amino acid, cystine, to accumulate in the lysosomes of the cells. This results in cystine crystals being stored in the kidneys, brain, eyes, bone marrow, lymph nodes and other organs. The disorder is inherited autosomal recessively.

General symptoms: The children are born healthy and appear generally normal during the first months. Poor appetite is occasionally observed in the child. Impaired kidney function eventually leads to severe kidney failure. Excessive urination and thirst commonly lead to evaluation and diagnosis. Kidney damage may result in the need for dialysis or kidney transplantation. The eyes and vision may be affected by storage of cystine crystals in the cornea and retina. The electrolyte balance and the body's supply of calcium and vitamin D are affected. Growth disorders are common. Today, cystinosis may be treated with medications (Cysteamine®, Cystagon®) that cause cystine to be released from the cells so that the body can rid itself of it. The medicine has some side-effects including nausea, vomiting, halitosis and abnormal skin odor. Growth hormone treatment is common. Treatment also involves ensuring that the child receives adequate amounts of fluids, electrolytes and nutrition. Many have very high energy requirements, which means that they need to eat a lot and often.

Orofacial/odontological symptom: Sucking, chewing and swallowing difficulties occur. Enteral feeding is common and sometimes necessary to ensure that the child's nutritional requirements are met.

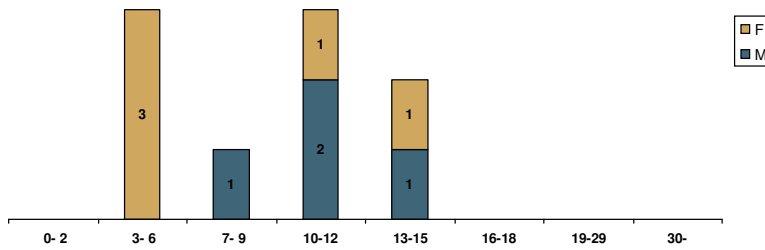
Orofacial/odontological treatment:

- Early contact with dental services for evaluation and treatment planning is important.
- Prevention is an important part of the child's dental care. Intensive prophylactic care is required and preferably performed by the same staff members at each visit. An increased risk for caries (cavities) probably exists in view of the child's dietary intake.
- Eating and swallowing difficulties should be investigated and treated by a specialist team (nutrition team or dysphagia team) at the hospital or at a habilitation unit (multidisciplinary treatment center).

Sources

The MHC database - The Mun-H-Center database of orofacial manifestations in rare disease: Ågrenska Newsletter on Cystinosis.

Age distribution

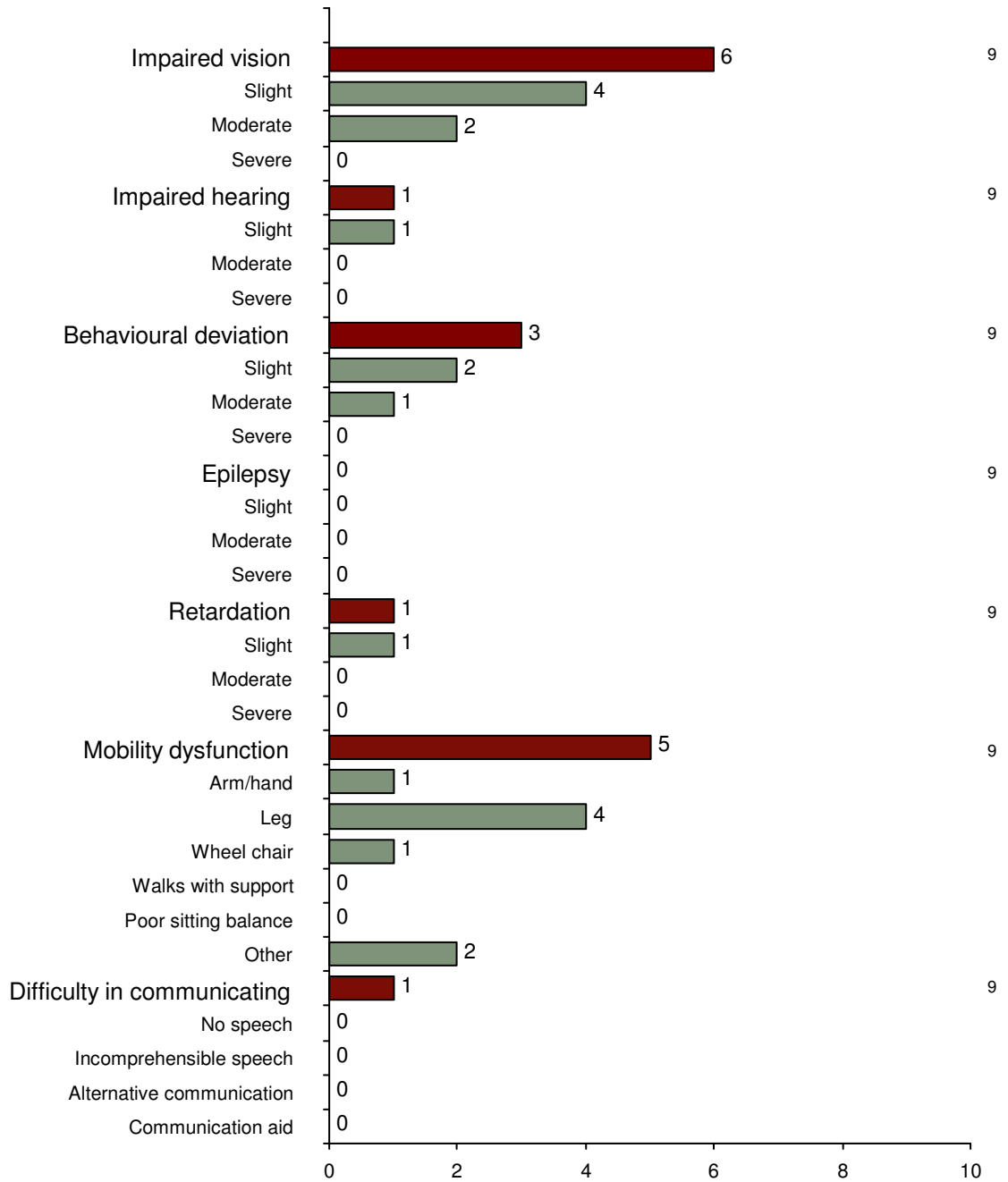


Number: 9

Ages: 3 -- 13 years

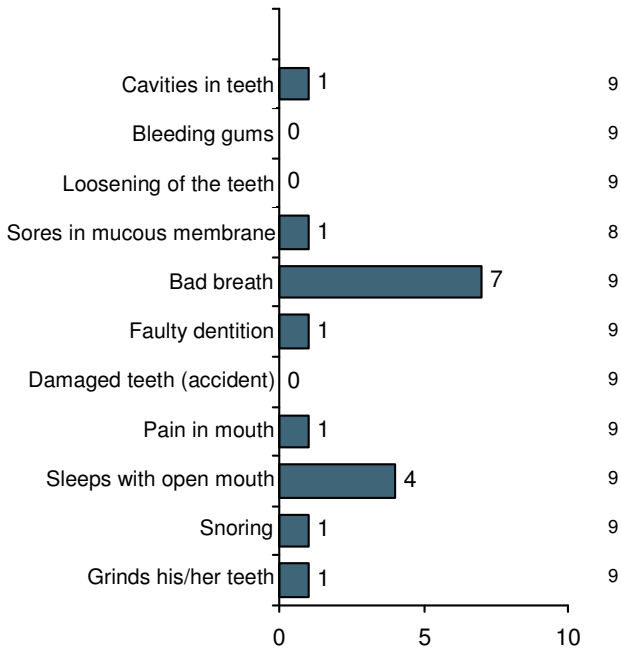
Sex: M (4) + F (5)

General disabilities

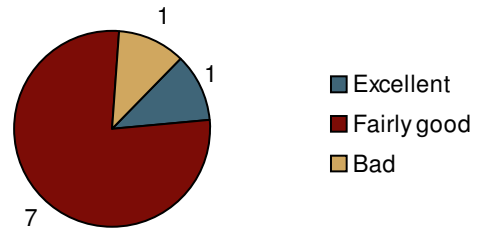


About dental health

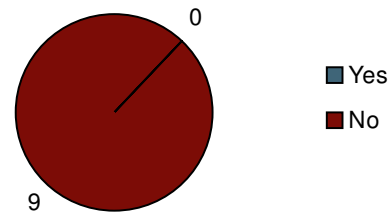
About dental health - problems



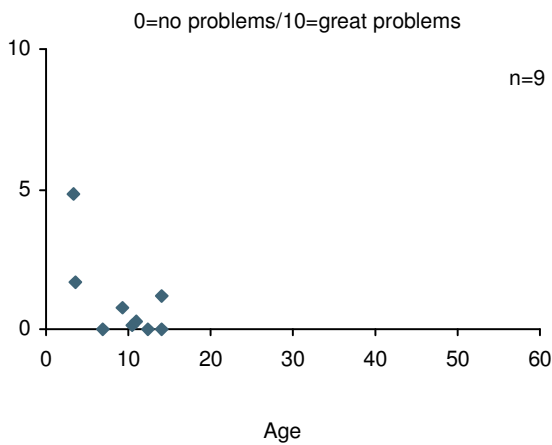
Oral health



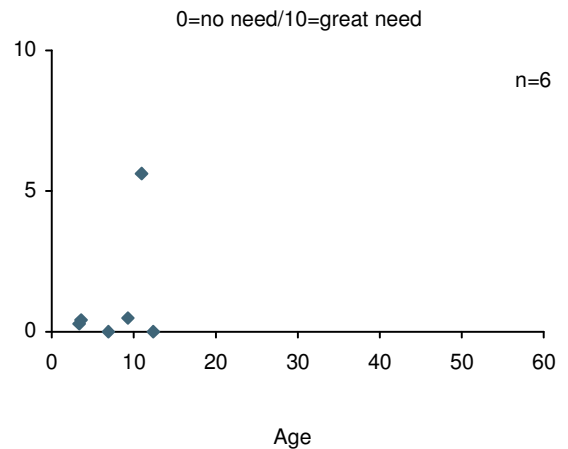
Has NN received orthodontic treatment?



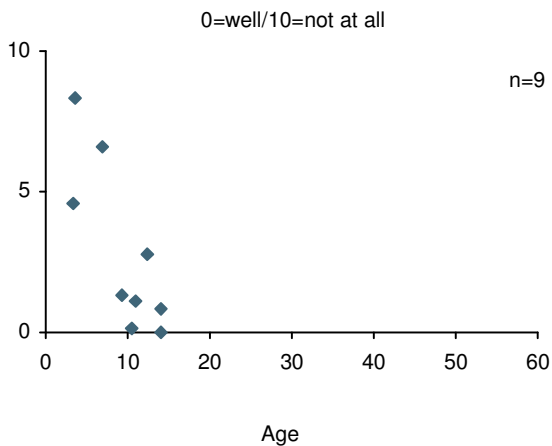
How does NN experience the dental care received?



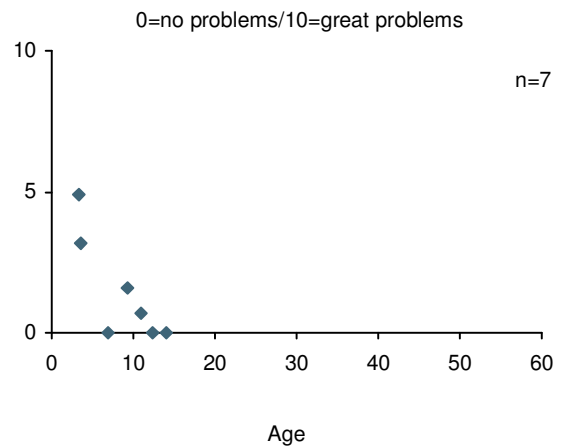
NN is considered to be in need of orthodontic treatment



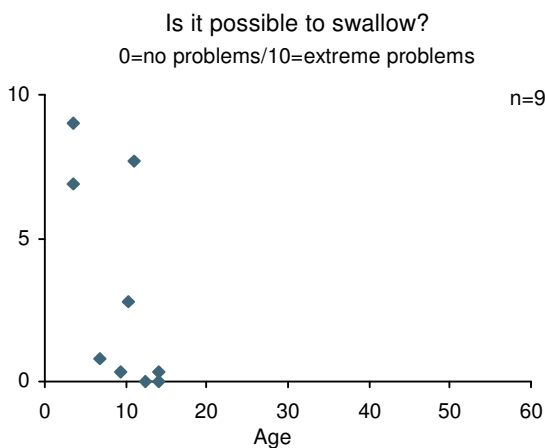
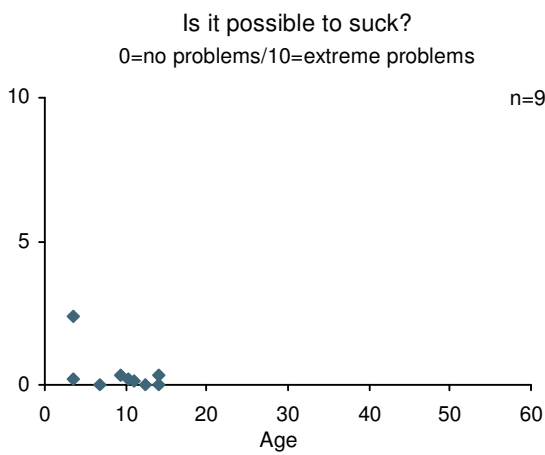
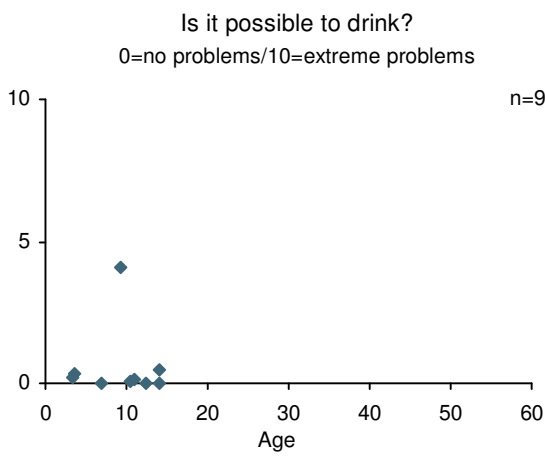
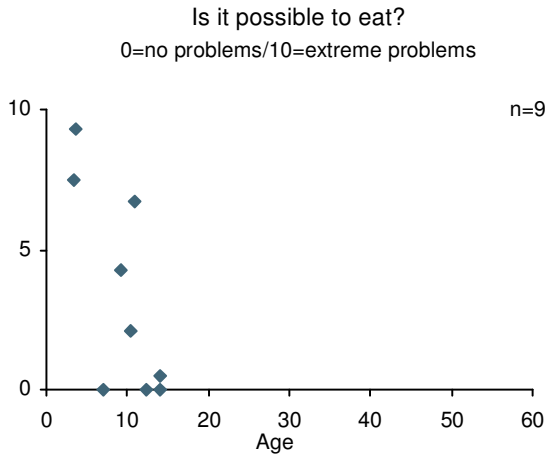
How does NN manage to brush his/her teeth?



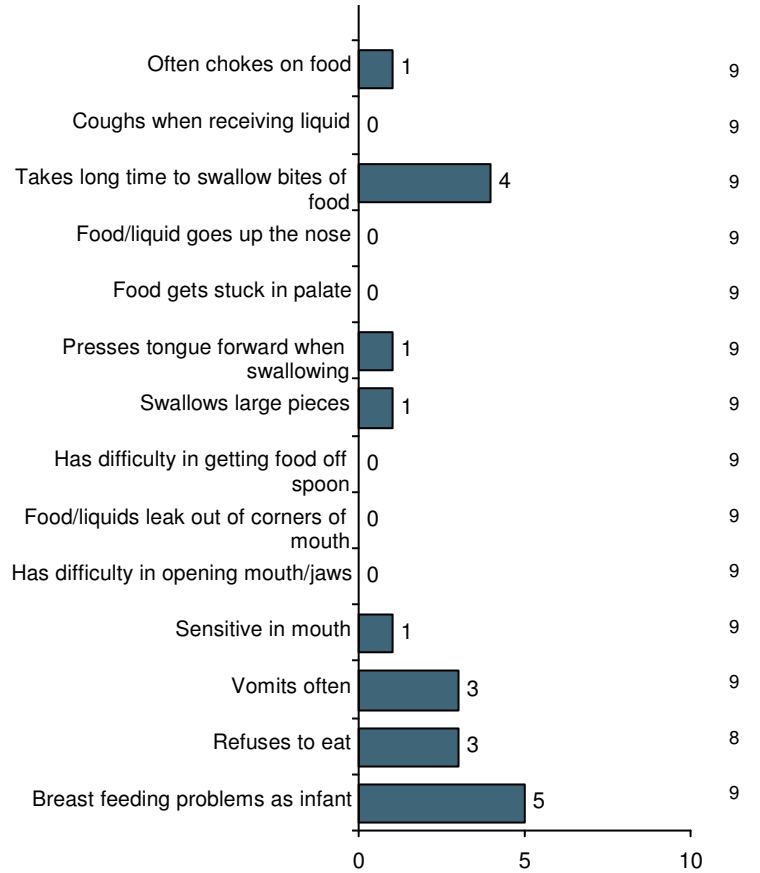
If help is needed with brushing teeth, how does it proceed?



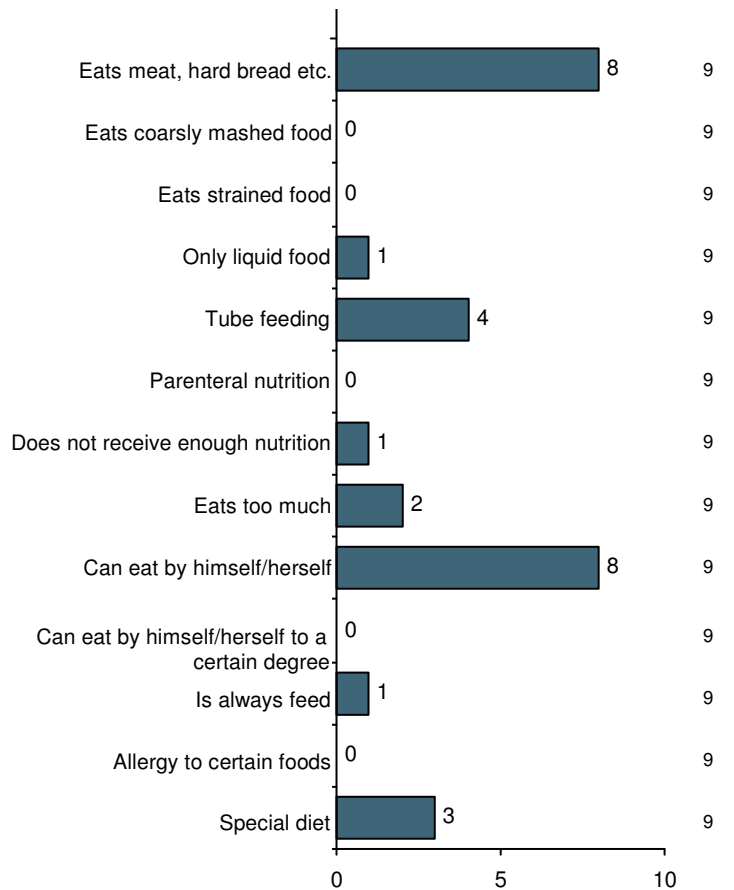
About eating habits



About eating habits - problems



Food habits



About drooling

