



Orofacial function of persons having CDG syndrome

Report from questionnaires

The survey comprises 16 Questionnaires.

Synonyms: Congenital Disorder of Glycosylation, previously termed Carbohydrate Deficient Glycoprotein.

Estimated occurrence: In Sweden, 1:50–70 000 live births, i.e. on average 1–2 children/year. It is estimated that there are about 100 children and adults with this syndrome. There are a variety of forms of CDG. The most common form, CDG1A, accounts for more than 70% of known cases.

Etiology: CDG1A is caused by mutations (changes) in the PMM2-gene that result in an impaired ability of the cells to synthesize or process glycoproteins. The gene is located on the short arm (p) of chromosome 16 (chromosome 16p13). The disease is inherited autosomal recessively.

General symptoms: CDG is a metabolic disorder that affects all parts of the body, but particularly leads to functional disturbances in the central and peripheral nervous systems. Common symptoms include varying degrees of intellectual disability; delayed gross motor development; low muscle tone (hypotonia); impaired balance and physical coordination; muscle weakness, primarily in the legs); and impaired vision. During infancy, common problems include feeding difficulties, diarrhoea and poor weight gain. Epilepsy is somewhat more common in children with CDG than in healthy ones. Speech is in most cases extremely impaired, whereas comprehension is quite good. Most will require a wheelchair or rollator. Spinal and thoracic deformities eventually arise. The medical complications stabilize in adolescence.

Orofacial/odontological symptoms: Eating difficulties and frequent vomiting are common, especially during infancy. Impaired oral motor function and low muscle tone can cause speech difficulties, eating difficulties, drooling and occlusal (bite) anomalies.

Orofacial/odontological treatment:

- Early contact with dental services for intensified prophylactic care and oral hygiene information is essential.
- An orthodontist should be consulted when the child is 7-9 years of age to plan for possible occlusal corrective treatment.
- Speech, language and communication training is often necessary

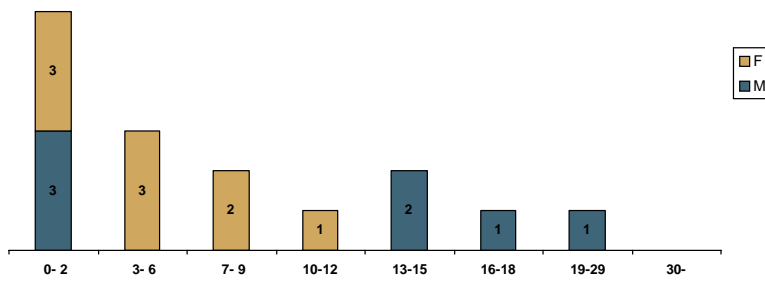
Source:

MHC-basen - Mun-H-Center's database of orofacial manifestations in rare disorders.

Ågrenska's Newsletter (in Swedish)

Läkartidningen (Journal of the Swedish Medical Association.), 1998 Dec 9; 95(50) 5742-5748. (in Swedish)

Age distribution

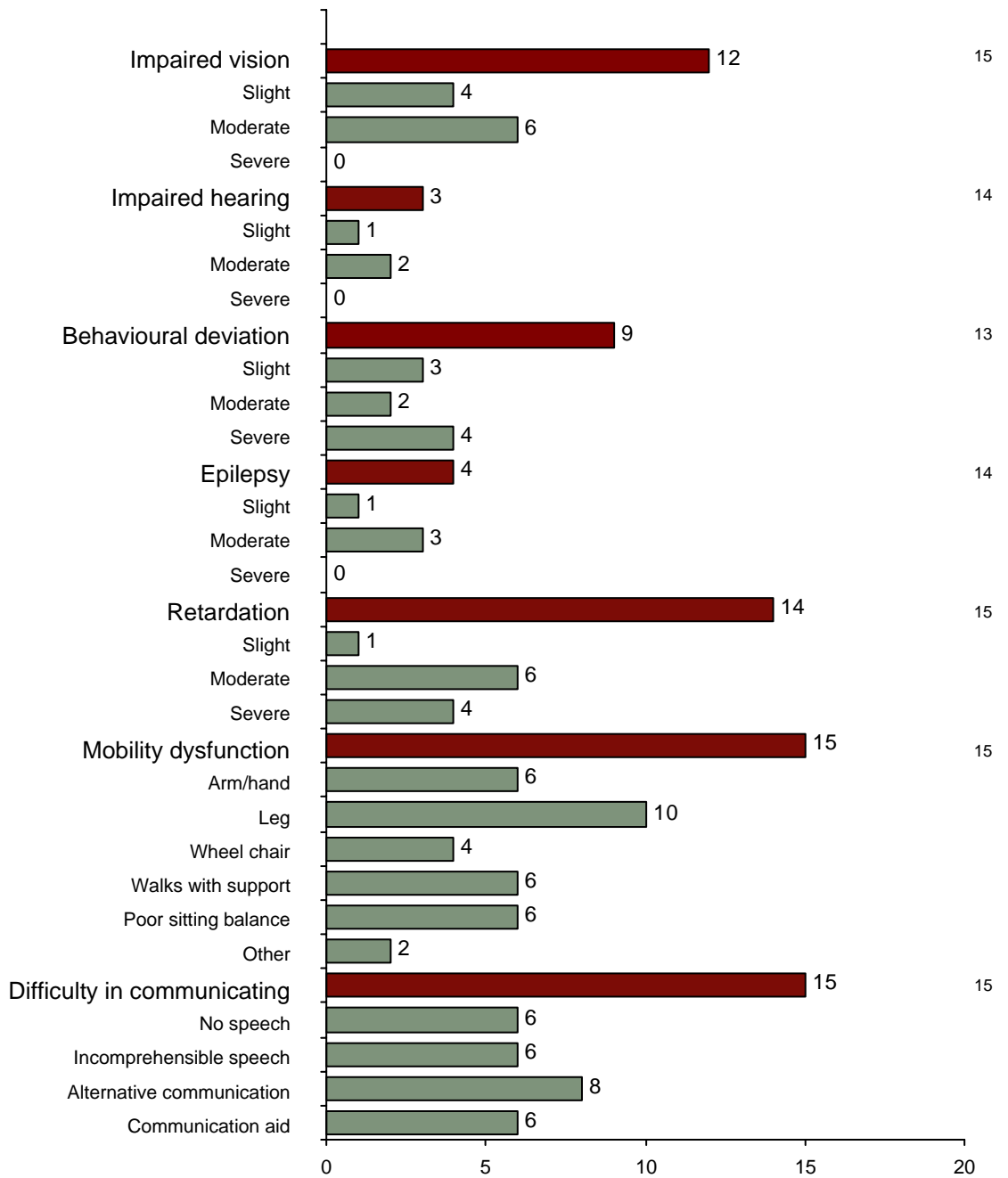


Number: 16

Ages: 1 -- 19 years

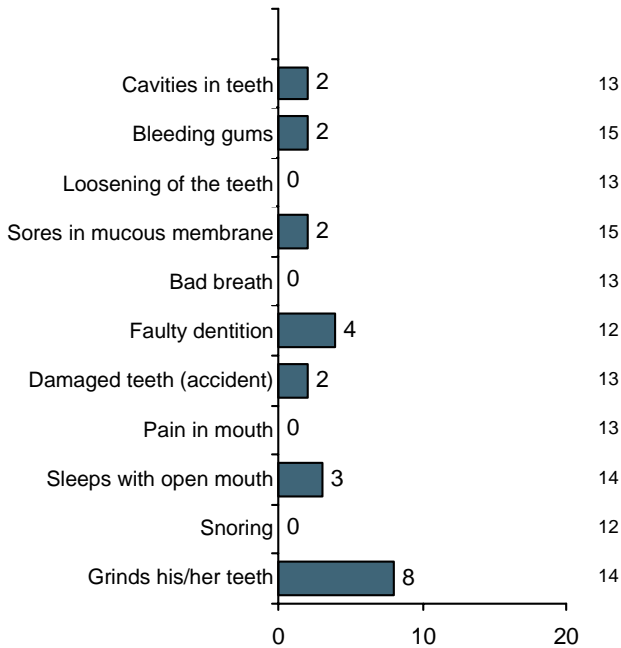
Sex: M (7) + F (9)

General disabilities

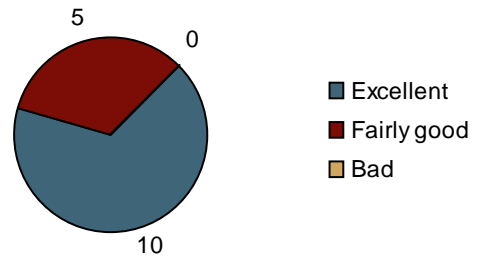


About dental health

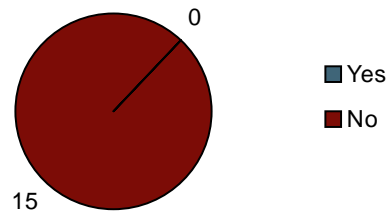
About dental health - problems



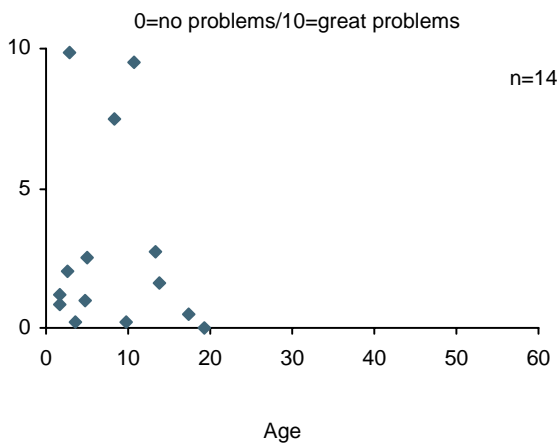
Oral health



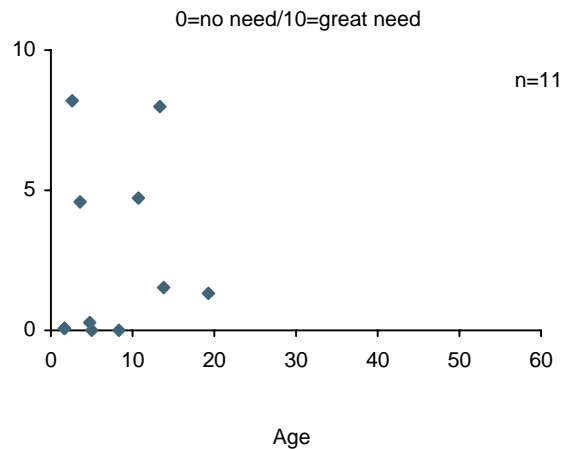
Has NN received orthodontic treatment?



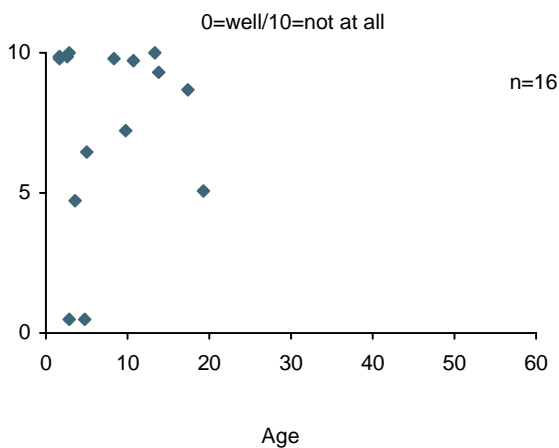
How does NN experience the dental care received?



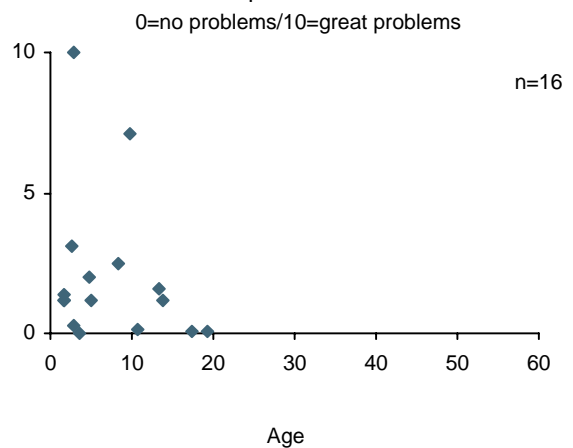
NN is considered to be in need of orthodontic treatment



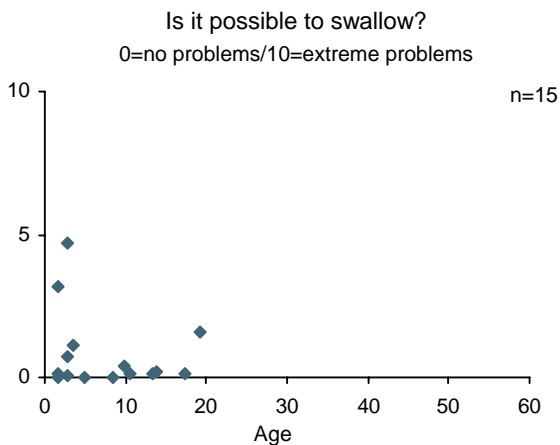
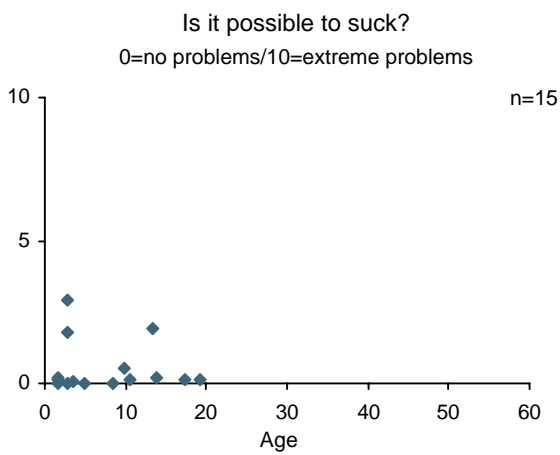
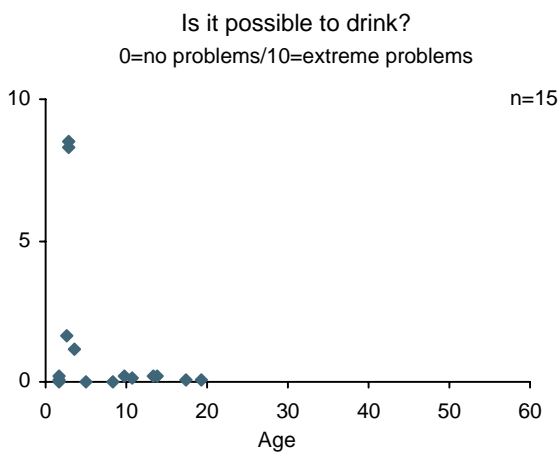
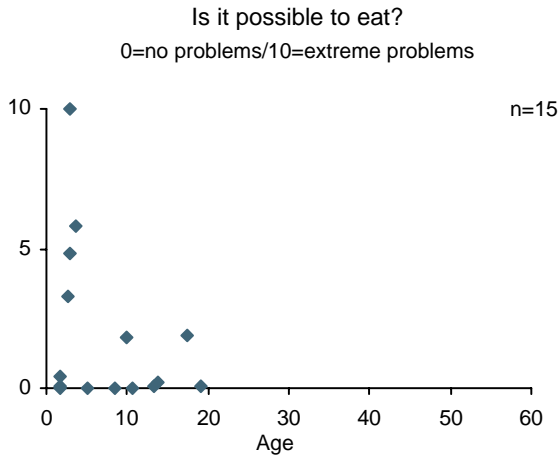
How does NN manage to brush his/her teeth?



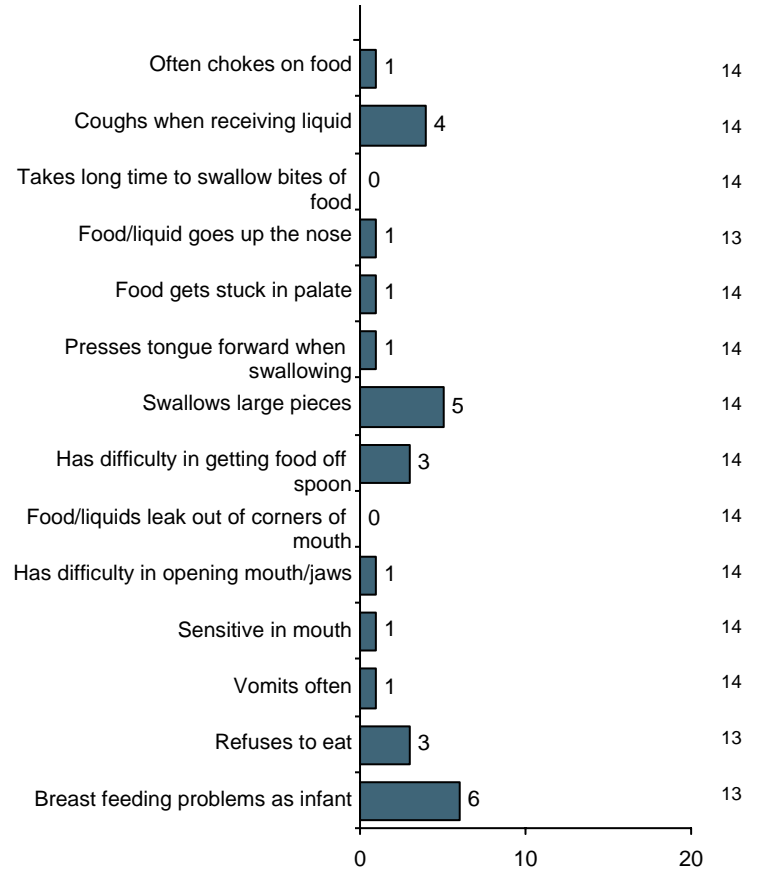
If help is needed with brushing teeth, how does it proceed?



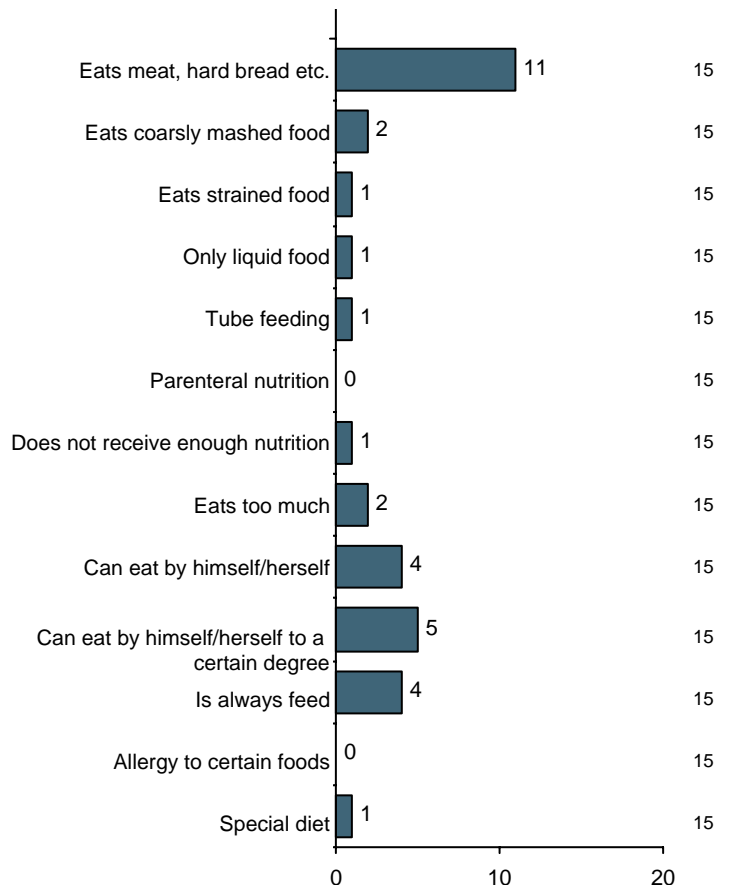
About eating habits



About eating habits - problems



Food habits



About drooling

