



Orofacial function of persons having LCHAD

Report from observation charts

The survey comprises 5 observation charts.

Synonym: Long Chain 3-Hydroxy-Akyl-CoA Dehydrogenase Deficiency

Estimated occurrence: Uncommon diagnosis. Thirteen known cases in Sweden (2004).

Etiology: Autosomal recessive inheritance with a defect in chromosome 2. Deficiency in an enzyme (LCHAD-enzyme) that oxidizes long chain fatty acids.

General symptoms: The disorder often causes hypoglycemia (low blood sugar), lethargy, hypotonia (low muscle tone) and affects the heart and liver. Many are susceptible to infection. Abnormalities in the retina may cause visual impairment and sensitivity to light. Brain damage and development delay may occur as a result of the child becoming seriously ill. Frequent high-carbohydrate meals are necessary and a fat-free diet is recommended. Nearly all those with LCHAD have a gastrostomy.

Orofacial/odontological symptoms: There are no known specific symptoms related to the teeth or bite in LCHAD. Eating difficulties and frequent vomiting are common. Many children who are enterally fed eat only small portions by mouth and lack appetite. When general development is delayed, even speech and oral motor development may be affected.

Orofacial/odontological treatment:

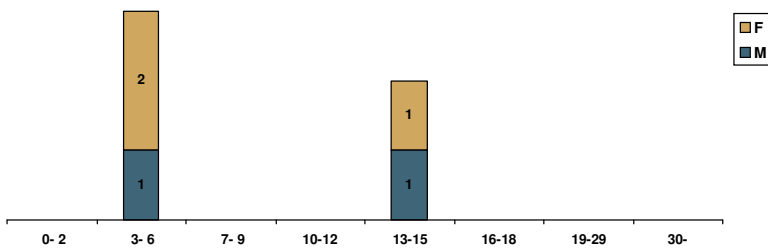
- Children with eating difficulties often need extra dental care, e.g. help with oral hygiene and fluoride treatment.
- Persons with heart defects may require prophylactic antibiotics prior to oral interventions where bleeding is to be expected.
- Eating and swallowing difficulties should be investigated and treated by a specialist team at the hospital (nutrition team or dysphagia team) or multidisciplinary treatment center.
- Orofacial therapy and oral motor skill training and stimulation is recommended for eating difficulties.
- Speech and language difficulties are treated by a speech-language pathologist.

Sources:

The MHC database - The Mun-H-Center database of orofacial manifestations associated with rare disorders.

The Newsletter of the Ågrenska Center.

Age distribution

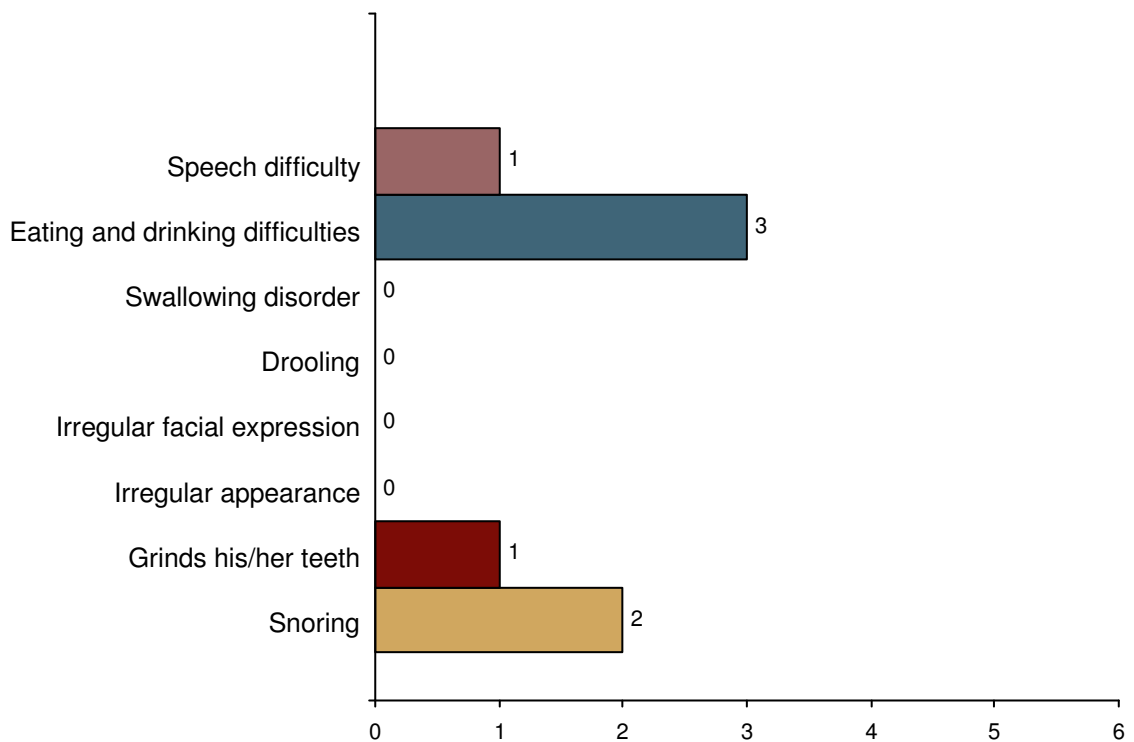


Number: 5

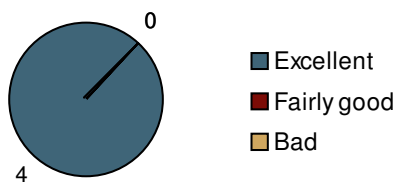
Ages: 3 -- 14 years

Sex: M (2) + F (3)

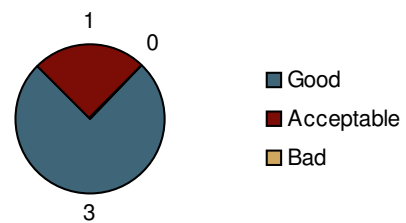
Orofacial problems



Oral health

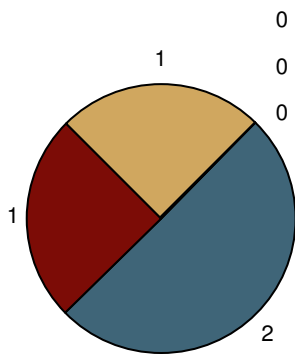


Oral hygiene



Behaviour in the treatment situation

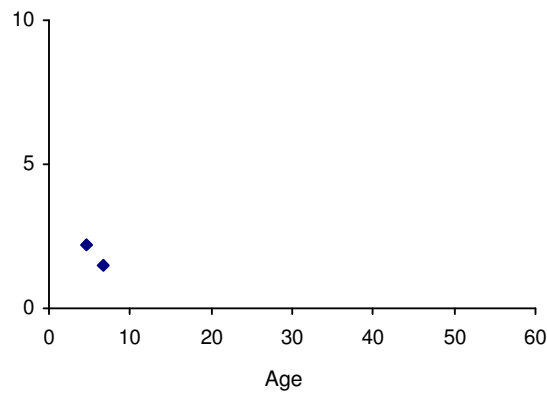
How calm and co-operative is the patient at time of examination?



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

0=/10=

0=/10=

Age

Age



Clinical findings	Total N=5	Boys/Men N=2	Girls/Women N=3	Not evaluated
Cross bite	1	0	1	1
Dental trauma	1	1	0	1
Hypomineralisation	1	1	0	1
Mucous membrane change	1	0	1	1
Speech difficulty	1	1	0	0