



## Orofacial function of persons having Goldenhar syndrome Report from observation charts

The survey comprises 18 observation charts.

**Synonyms:** Oculo-auriculo-vertebral spectrum (OAVS), hemifacial microsomia.

**Estimated occurrence:** 1:50 000 live births.

**Etiology:** Unknown

**General symptoms:** The following deformities may occur:

*Craniofacial:* facial asymmetry and underdeveloped jaws (hemifacial microsomia).

*Auricular:* underdeveloped ears and ear canals, "tags" of skin or cartilage in front of the ear.

*Eyes:* white lumps on the edge of the cornea (epibulbar dermoids) and eyelid deformities (eyelid colobomas).

*Skeletal:* vertebral anomalies.

*Cardiac:* The most common defect is ventricular septum defect (VSD) and Fallot's anomaly. Hearing impairment and squinting are common.

**Orofacial/odontological symptoms:** Skeletal asymmetry, and a small lower jaw. Facial palsy may occur. Sucking difficulties are common in newborns, own to the craniofacial deformities, narrow airway and/or cardiac defect. Many of these children have to be tube fed. Even older children and adults may have eating difficulties. Narrow airways may result in snoring problems and sleep apnea (frequent suspension of breathing while asleep).

### Orofacial/ odontological treatment:

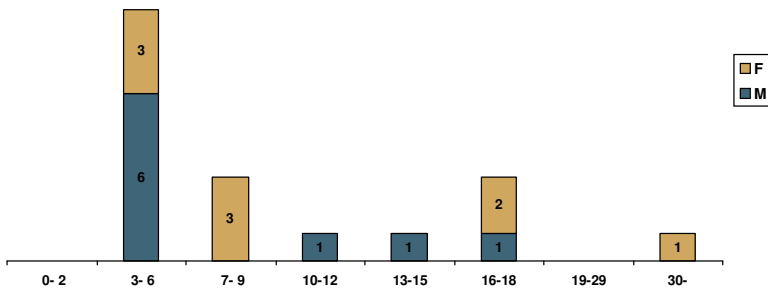
- Children with eating difficulties often require extra dental care, including help with oral hygiene and fluoride treatments.
- Eating and swallowing problems will need to be investigated and treated by hospital specialist teams (either a nutrition team or a dysphagia team, or by other multidisciplinary treatment specialists.)
- Snoring problems should be followed up by a physician.
- Individuals with cardiac defects may require prophylactic antibiotics when oral interventions associated with bleeding are undertaken.
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### Sources:

The rare disease database of the Swedish National Board of Health and Welfare.

The MHC database - The Mun-H-Center database of orofacial manifestations in rare diseases.

Age distribution

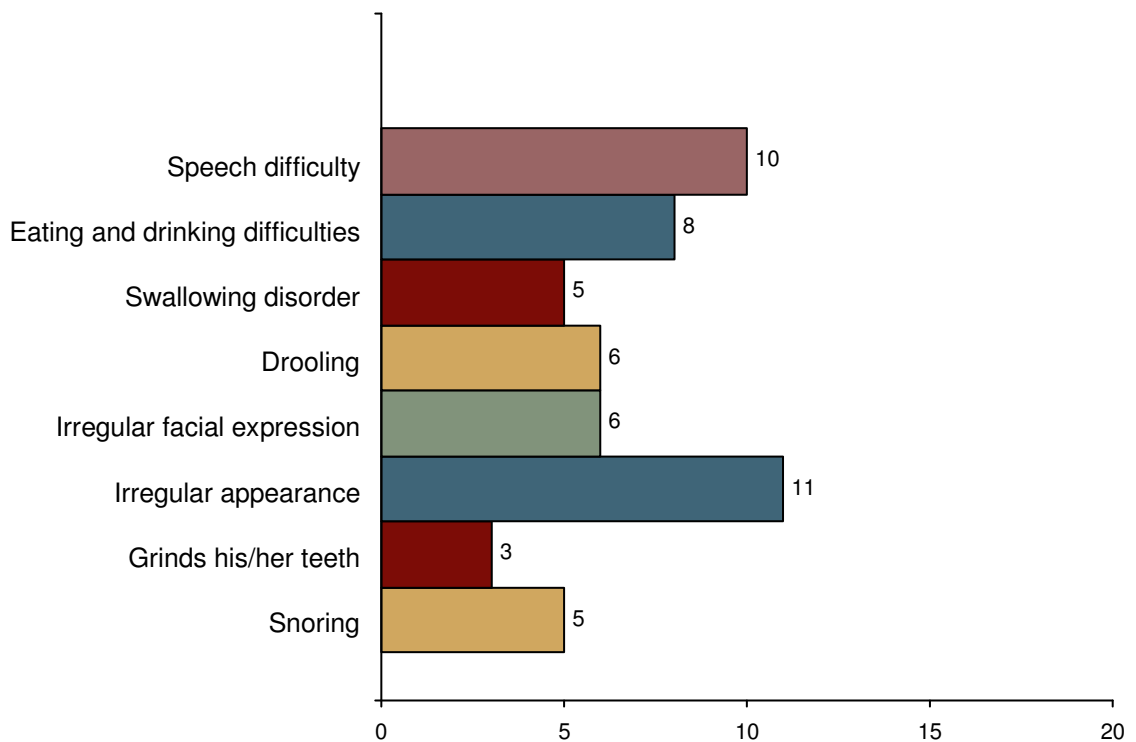


Number: 17

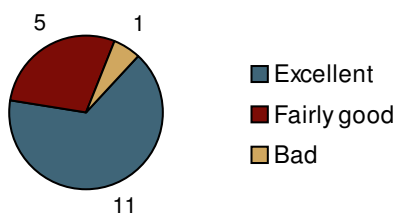
Ages: 3 -- 17 years

Sex: M (9) + F (8)

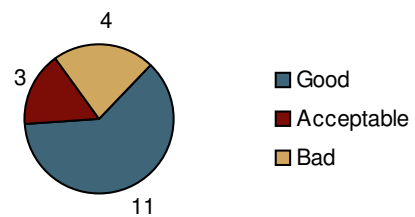
Orofacial problems



Oral health

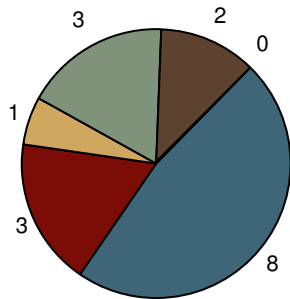


Oral hygiene



## Behaviour in the treatment situation

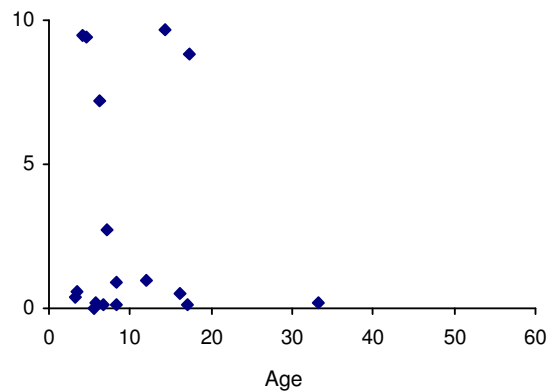
How calm and co-operative is the patient at time of examination?



- (8) ■ Examination is possible without problem
- (3) ■ Examination is possible without problems, some reaction is observed
- (1) ■ Examination can continue if adjusted to patient's reactions
- (3) ■ Reactions are considerable and examination is obviously affected
- (2) ■ Examination is practically impossible to complete
- (0) ■ Patient refuses examination

How does the patient cope with treatment in general?

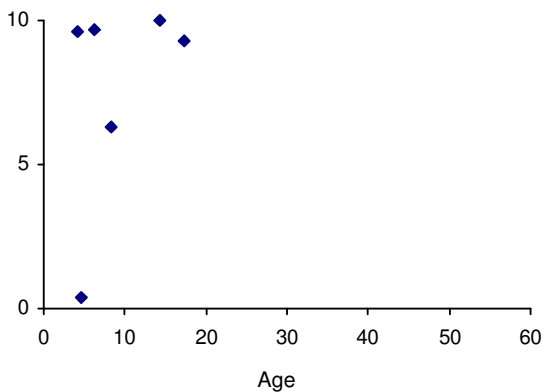
0=no problems/10=great problems



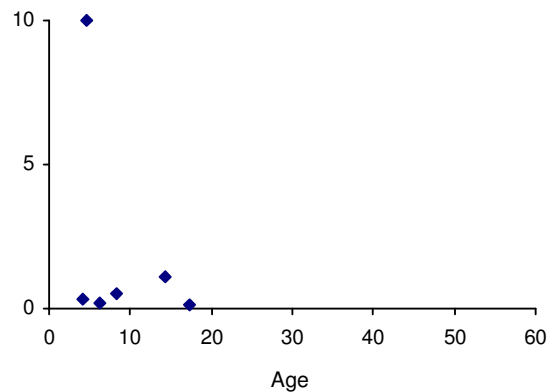
If there are treatment problems:

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



<b>Clinical findings</b>	<b>Total N=17</b>	<b>Boys/Men N=9</b>	<b>Girls/Women N=8</b>	<b>Not evaluated</b>
Facial asymmetry	14	8	6	0
Cranio-facial abnormality	13	8	5	1
Deviation on opening	10	6	4	0
Speech difficulty	10	8	2	0
Gingivitis	9	5	4	0
Drooling	7	6	1	0
Over crowding	7	2	5	0
Facial palsy	6	5	1	0
Frontal open bite	6	4	2	0
Hypomineralisation	6	3	3	0
Open mouth at rest	6	4	2	0
Post normal bite	6	5	1	0
Reduced mobility in tongue	6	4	2	0
Cross bite	5	3	2	0
Misshaped occlusal plane	5	4	1	1
Difficulty in breathing	4	2	2	0
Grinds his/her teeth	4	3	1	0
High palate	4	2	2	0
Lower jaw seems small	4	2	2	0
Narrow palate	4	1	3	0
Other oral habits	4	4	0	0
Supra-gingival calculus	4	3	1	0
Frontal inversion	3	2	1	0
Horizontal over-bite 6 mm or more	3	1	2	0
Hyper-plastic gingivitis	3	2	1	0
M mentalis is overactive	3	2	1	0
Mouth breathing	3	1	2	2
Pre normal bite	3	1	2	0
Reduced mobility in TMJ	3	2	1	0
Upper jaw seems small	3	2	1	0
Deep bite without gingival contact	2	1	1	0
Dental trauma	2	1	1	0
Divided uvula	2	2	0	2
Jaw cleft	2	2	0	0
Lip cleft	2	2	0	0
Low muscle tone in upper lip	2	2	0	0
Philtrum seems long	2	2	0	0
Proclined upper incisors	2	0	2	0
Scissor bite	2	2	0	0
Soft palate is asymmetric	2	0	2	2
Tooth anomaly	2	1	1	1