



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: 2:100 000 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:

- Many individuals with Goldenhar syndrome will require jaw surgery and orthodontic treatment.
- In cases of craniofacial deformities, a specialist team will be needed for follow up and treatment.
- Children with eating difficulties often require extra dental care, including help with oral hygiene and fluoride treatments.
- Feeding and swallowing difficulties are investigated and treated by a specialist team at the hospital or multidisciplinary treatment center.
- Snoring problems should be followed up by a physician.
- When treating medically compromised patients always contact their doctors for medical advice.

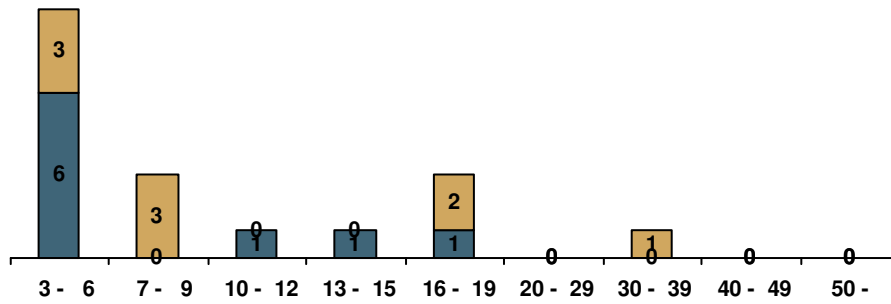
Sources:

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

The MHC database - The Mun-H-Center database on oral health and orofacial function in rare diseases.



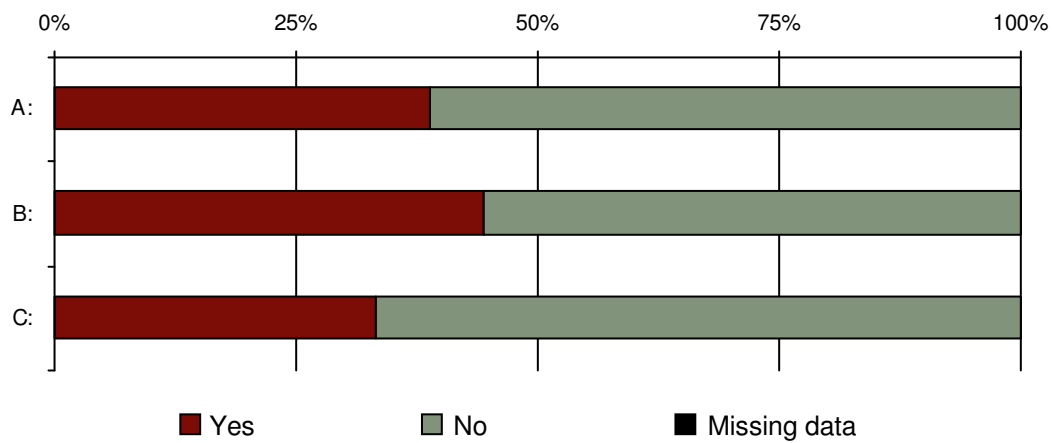
Age distribution



Number: 18
 Ages: 3 - 33
 Sex: M (9) F (9)

Overview

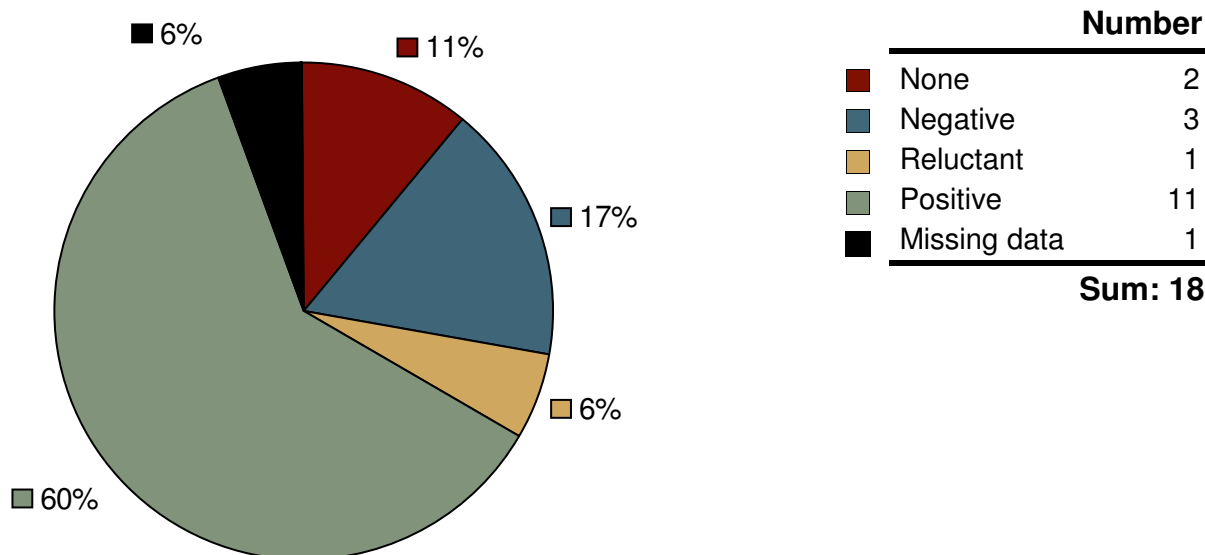
	Yes	No	Missing data	N
A: Incomprehensible speech/No speech	7	11	0	18
B: Eating and drinking difficulties ¹	8	10	0	18
C: Profuse drooling, on clothes ¹	6	12	0	18



Note that the diagram is based upon less than 100 individuals.

¹: Compiled using questionnaire

Acceptance of dental examination



Caries

	3-6 years	7-12 years	13-19 years	Adults
deft¹				
Examined	8	4		
Number of individuals with deft=0	7	1		
Mean	0,3	0,8		
Standard deviation	0,7	0,4		
Missing data	1	0		
DMFT²				
Examined		4	4	1
Number of individuals with DMFT=0		2	2	0
Standard deviation		1,0	1,7	0,0
Mean		1,0	1,5	2,0
Missing data		0	0	0

1: Number of carious or filled deciduous teeth

2: Number of carious or filled permanent teeth

Occlusal relationship

	Number
Neutral bite	9
Post normal	6
Pre normal	3
Missing data	0
Sum: 18	

Maximum jaw opening

Children younger
than 10 years

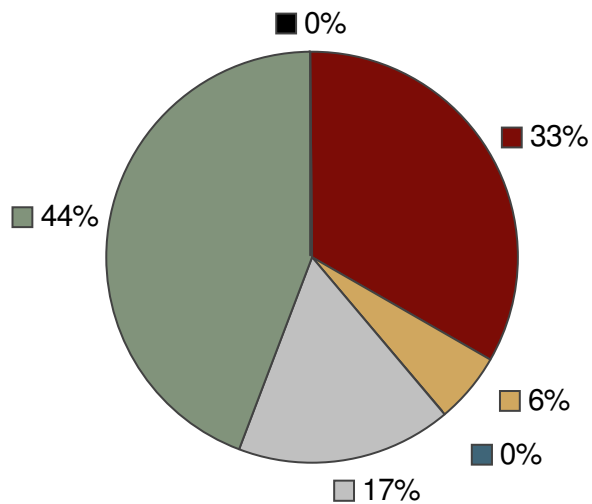
	Number
- 20	0
21 - 30	3
31 - 40	3
41 - 50	3
51 -	0
Missing data	3
Sum: 12	

Children, 10 years or
older, and adults

	Number
- 20	0
21 - 30	0
31 - 40	3
41 - 50	1
51 -	0
Missing data	2
Sum: 6	

1: This variable was introduced in version 2 (2008) of the Observation chart.

Speech difficulty



	Number
No speech	6
Very incomprehensible	1
Incomprehensible speech	0
Slightly indistinct speech	3
No problems	8
Missing data	0
Sum: 18	

Clinical findings	Yes-answers			
	Total N=18 (%)	Boys/Men N=9 (%)	Girls/Women N=9 (%)	Missing data
Facial asymmetry	14 (78)	8 (89)	6 (67)	0
Cranio-facial abnormality	13 (76)	8 (100)	5 (56)	1
Frontal open bite	7 (39)	5 (56)	2 (22)	0
Over crowding	7 (39)	2 (22)	5 (56)	0
Facial palsy	6 (33)	5 (56)	1 (11)	0
Open mouth at rest	6 (33)	4 (44)	2 (22)	0
Impaired tongue motility	6 (33)	4 (44)	2 (22)	0
High palate	4 (22)	2 (22)	2 (22)	0
Narrow palate	4 (22)	1 (11)	3 (33)	0
M mentalis overactive	3 (17)	2 (22)	1 (11)	0
Reduced opening capacity	3 (17)	2 (22)	1 (11)	0