



Orofacial function of persons having CHARGE syndrome

Report from observation charts

The survey comprises 46 observation charts.

Synonym: CHARGE association

Estimated occurrence: 5-12:100 000 live births.

Etiology: The symptoms appear during early foetal development and are most often caused by gene mutations or deletions on chromosome 8 (the CHD7-gene).

General symptoms:

- C** Coloboma (keyhole-shaped clefting of the eye)
- H** Heart defects of varying severity
- A** Atresia of the choanae (constriction of the passage between nose and throat)
- R** Retardation of growth and/or mental development
- G** Genital hypoplasia (underdeveloped sexual organs)
- E** Ear anomalies (abnormalities of the outer ear and/or hearing loss)

All children with Charge syndrome will not have all the symptoms. Symptoms may also differ regarding degree of severity.

Orofacial/odontological symptoms: Characteristic facial features are associated with the diagnosis. Facial palsy may occur, as well as cleft lip, jaw and palate. Cardiac malformations and malformations in the respiratory and gastrointestinal tract will often cause feeding impairment. Delayed speech and language are common due to a combination of causes such as hearing impairment, learning disability, cleft lip and palate or oral motor dysfunction. Some have difficulties with daytime teeth grinding or drooling.

Orofacial/odontological treatment:

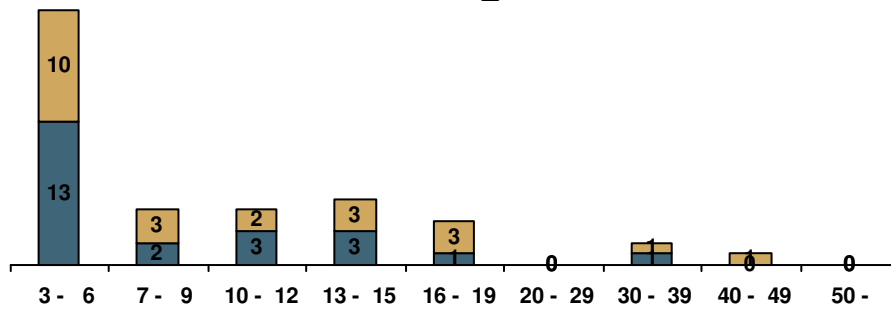
- In cases of craniofacial deformities, a specialist team will be needed for follow up and treatment.
- Early contact with dental services for intensified prophylactic care and oral hygiene information is essential.
- Regular check-ups of dental and jaw development. Orthodontist should be consulted when needed
- Tooth grinding should be followed up, and be managed with a splint when necessary.
- Feeding and swallowing difficulties are investigated and treated by a specialist team at the hospital or multidisciplinary treatment center.
- Training in oral motor skills in cases of eating disorders, speech difficulties and drooling may be relevant
- Communication skills training is frequently essential.
- When treating medically compromised patients always contact their doctors for medical advice (bleeding problems, heart diseases etc).

Source:

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.



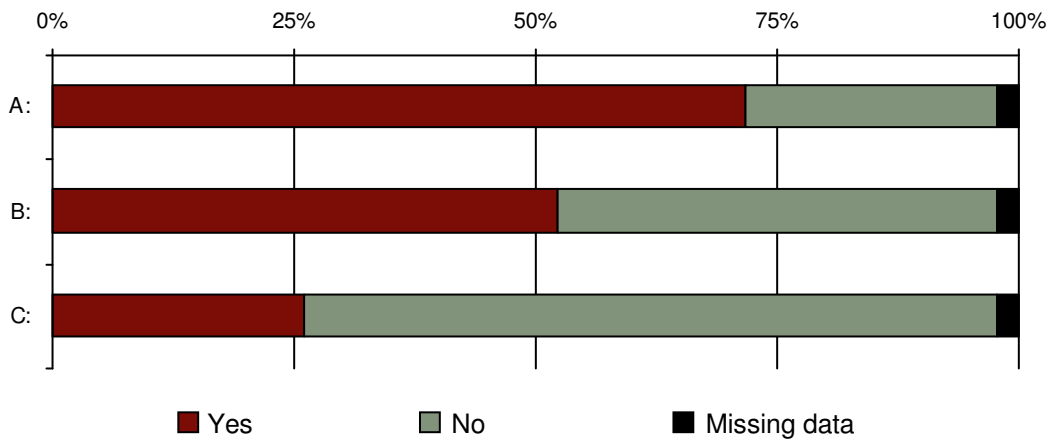
Age distribution



Number: 46
 Ages: 3 - 44
 Sex: M (23) F (23)

Overview

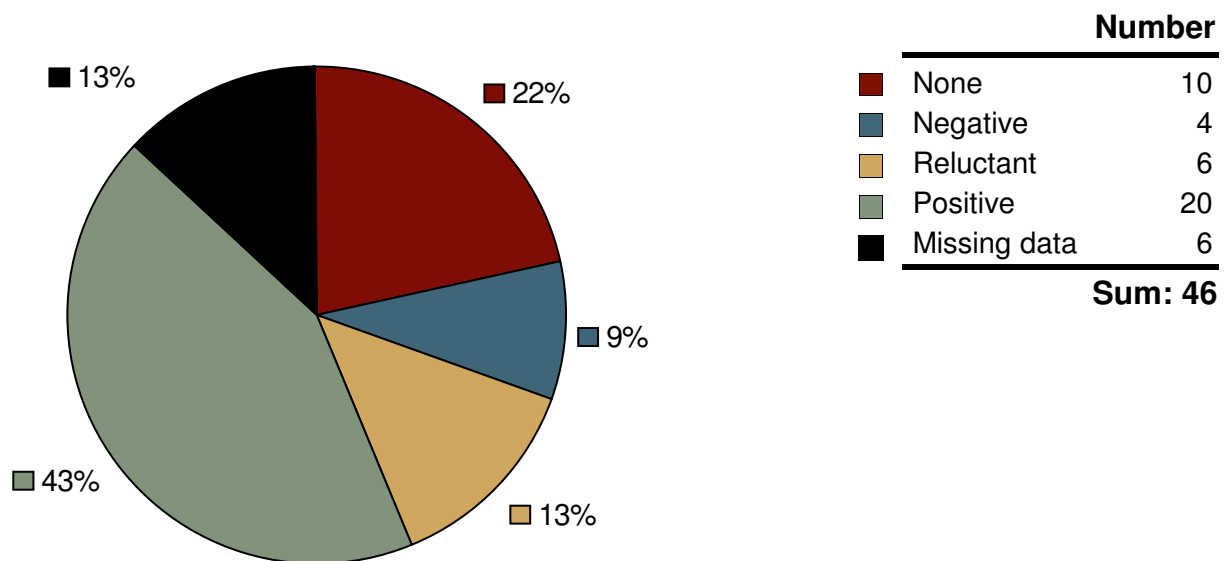
| | Yes | No | Missing data | N |
|--|-----|----|--------------|----|
| A: Incomprehensible speech/No speech | 33 | 12 | 1 | 46 |
| B: Eating and drinking difficulties ¹ | 24 | 21 | 1 | 46 |
| C: Profuse drooling, on clothes ¹ | 12 | 33 | 1 | 46 |



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 | 16 | 5 | | |
| Number of individuals with deft=0 | 13 | 4 | | |
| Mean | 0,7 | 0,8 | | |
| Standard deviation | 1,6 | 1,6 | | |
| Missing data | 7 | 5 | | |
| DMFT² | | | | |
| Examined | | 7 | 10 | 3 |
| Number of individuals with DMFT=0 | | 5 | 7 | 1 |
| Standard deviation | | 1,0 | 0,9 | 5,3 |
| Mean | | 0,6 | 0,5 | 6,3 |
| Missing data | | 3 | 0 | 0 |

1: Number of carious or filled deciduous teeth

2: Number of carious or filled permanent teeth

Occlusal relationship

| | Number |
|----------------|---------------|
| Neutral bite | 19 |
| Post normal | 11 |
| Pre normal | 10 |
| Missing data | 6 |
| Sum: 46 | |

Maximum jaw opening

Children younger
than 10 years

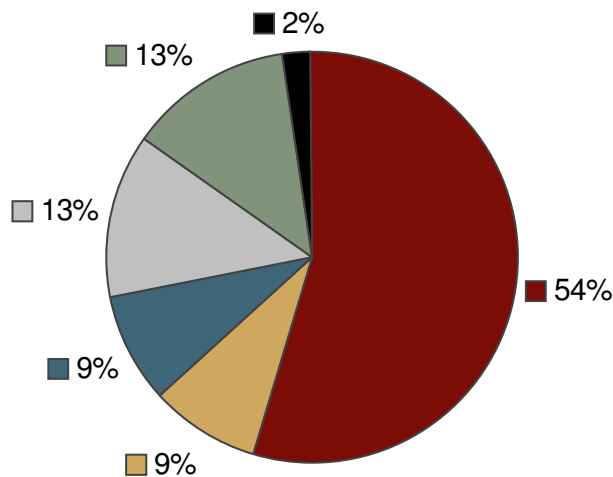
| | Number |
|----------------|---------------|
| - 20 | 0 |
| 21 - 30 | 2 |
| 31 - 40 | 7 |
| 41 - 50 | 0 |
| 51 - | 0 |
| Missing data | 19 |
| Sum: 28 | |

Children, 10 years or
older, and adults

| | Number |
|----------------|---------------|
| - 20 | 0 |
| 21 - 30 | 0 |
| 31 - 40 | 5 |
| 41 - 50 | 5 |
| 51 - | 1 |
| Missing data | 7 |
| Sum: 18 | |

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

Speech difficulty



| | Number |
|----------------------------|---------------|
| No speech | 25 |
| Very incomprehensible | 4 |
| Incomprehensible speech | 4 |
| Slightly indistinct speech | 6 |
| No problems | 6 |
| Missing data | 1 |
| Sum: 46 | |

| Clinical findings | Yes-answers | | | |
|--|-------------------|----------------------|-------------------------|-----------------|
| | Total N=46 (%) | Boys/Men N=23 (%) | Girls/Women N=23 (%) | Missing data |
| Cranio-facial abnormality | 20 (48) | 9 (45) | 11 (50) | 4 |
| Facial asymmetry | 19 (42) | 11 (48) | 8 (36) | 1 |
| Cleft lip and palate | 14 (31) | 8 (35) | 6 (27) | 1 |
| Facial palsy | 14 (33) | 5 (23) | 9 (43) | 3 |
| Impaired tongue motility | 12 (29) | 5 (22) | 7 (39) | 5 |
| Open mouth at rest | 11 (24) | 4 (17) | 7 (32) | 1 |
| Frontal open bite | 11 (28) | 7 (35) | 4 (21) | 7 |
| Narrow palate | 11 (29) | 5 (24) | 6 (35) | 8 |
| Over crowding | 11 (28) | 5 (23) | 6 (33) | 6 |
| Low muscle tone in lips | 9 (20) | 3 (13) | 6 (27) | 1 |
| High palate | 8 (22) | 3 (15) | 5 (29) | 9 |
| Low muscle tone in tongue | 6 (14) | 3 (14) | 3 (14) | 3 |
| Intra oral hypo-sensitivity | 4 (10) | 3 (14) | 1 (5) | 4 |
| Short tongue frenulum | 4 (9) | 3 (14) | 1 (5) | 3 |
| Deep bite with gingival contact | 3 (8) | 2 (10) | 1 (5) | 6 |
| Reduced stability in neck | 3 (7) | 3 (13) | 0 (0) | 1 |
| Low muscle tone in masticatory muscles | 3 (8) | 2 (10) | 1 (5) | 6 |