# Orofacial function of persons having
# Fragile X syndrome
## Report from observation charts

The survey comprises 93 observation charts.

**Estimated occurrence:** 1:5000 boys and 1:4000 girls have the genetic mutation that causes Fragile X syndrome. All boys, but only 20% of the girls, who have the mutation also have the symptoms.

**Etiology**: An unstable DNA segment on the long arm of the X chromosome. Girls almost always have less severe symptoms, since females have two X chromosomes.

**General symptoms:** Delayed psychomotor development. Varying degrees of intellectual disability. Many of these children have concentration problems and poor endurance. There may be some autism spectrum or autism-like features, and there is an increased frequency of epilepsy. There may be a cardiac defect, mitrialis valve prolapse, usually unproblematic.

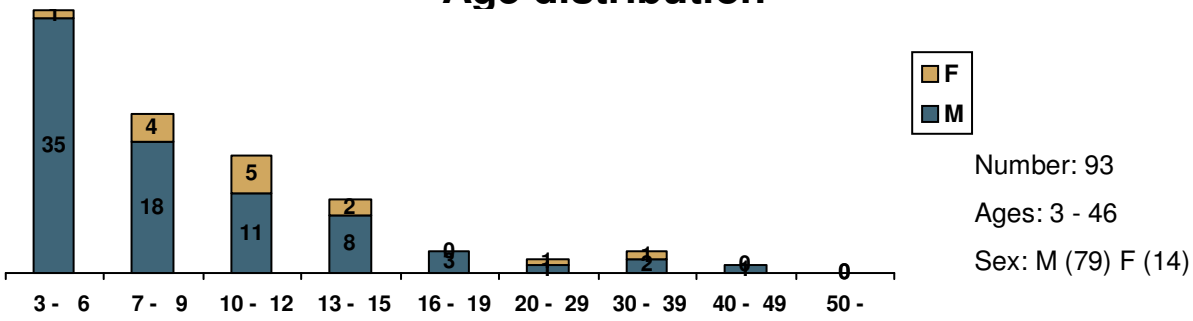
**Orofacial/odontological symptoms:** Characteristic facial features are associated with the diagnosis. These features are less pronounced in children. Many have malocclusions. Speech, language and communication difficulties are frequent, as are eating and drooling problems. Speech is often rapid. Mouth hypersensitivity may occur, and be problematic in terms, for instance, of feeding and tooth brushing.

**Orofacial/odontological treatment:**

- Early contact with dental services for intensified prophylactic and oral hygiene information is essential.
- Regular check-ups of dental and jaw development. Orthodontist should be consulted when needed.
- Orofacial therapy and oral motor skills training in cases of difficulties with eating, speech or drooling.
- Speech, language and communication training are often required.
- When treating medically compromised patients always contact their doctors for medical advice.

**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.
The Documentation from the Ågrenska Center.

## Age distribution



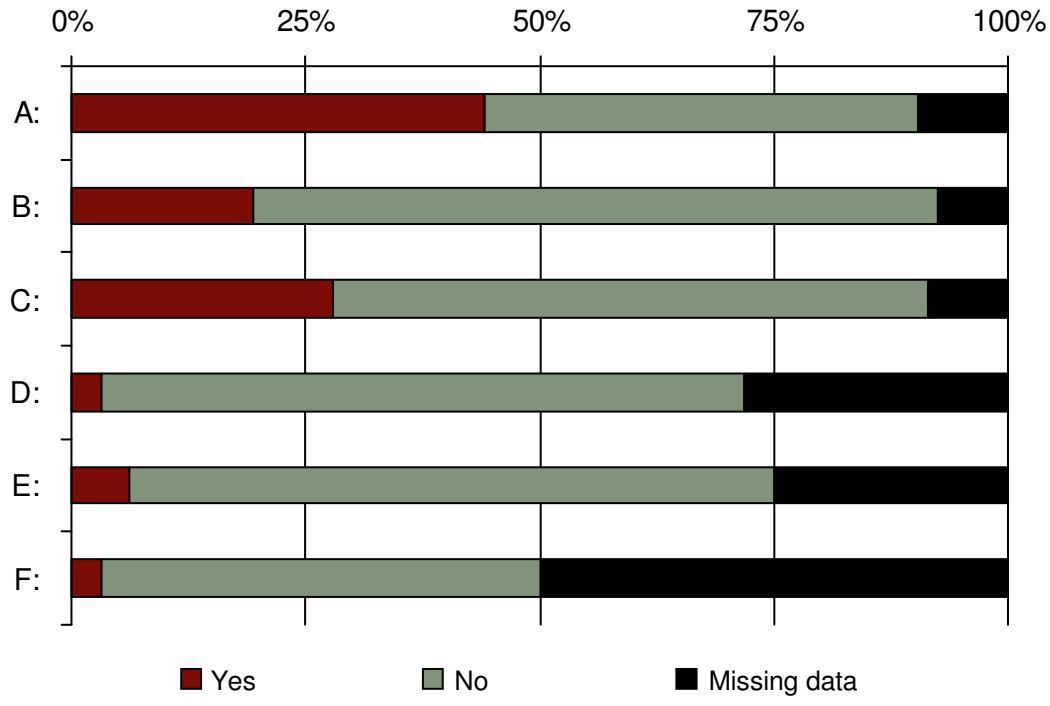

Number: 93

Ages: 3 - 46

Sex: M (79) F (14)

## Overview

| | | Yes | No | Missing data | N |
|---|---|---|---|---|---|
| A: | Incomprehensible speech/No speech | 41 | 43 | 9 | 93 |
| B: | Eating and drinking difficulties[1] | 18 | 68 | 7 | 93 |
| C: | Profuse drooling, on clothes[1] | 26 | 59 | 8 | 93 |
| D: | Breathing difficulties[1] [2] | 1 | 22 | 9 | 32 |
| E: | Grinding every day[1] [2] | 2 | 22 | 8 | 32 |
| F: | Severe malocclusions[2] | 1 | 15 | 16 | 32 |




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

1: Compiled using questionnaire

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

# Oral health

## Oral health index (indices)[1]

| | | 0 | 1 | 2 | 3 | 4 | 5 | 6 | Missing data | N |
|---|---|---|---|---|---|---|---|---|---|---|
| Calc | Calculus | 12 | 3 | 1 | 0 | 0 | 0 | 0 | 16 | 32 |
| GI | Gingivitis | 10 | 1 | 2 | 1 | 1 | 0 | 1 | 16 | 32 |
| Plaq | Coating | 10 | 2 | 1 | 2 | 0 | 0 | 1 | 16 | 32 |
| Toot | Tooth wear | 14 | 1 | 1 | 0 | | | | 16 | 32 |

C Calculus index is based on the presence of visible calculus on the buccal surface of 6 index teeth. 0 indicates that there is no calculus at all, 6 indicates calculus on all index teeth.

GI Gingivitis index is based on the presence of visible gingivitis on the buccal surface of 6 index teeth. 0 indicates that there is no bleeding, 6 indicates bleeding on all index teeth.

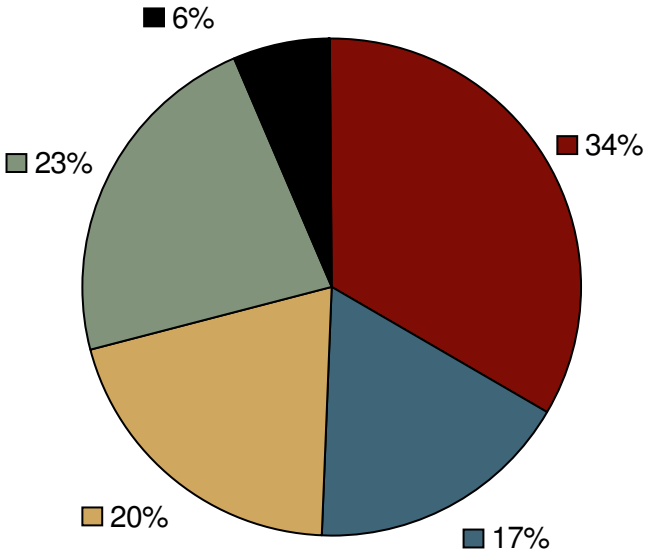
PI Plaque index is based on the presence of visible plaque on the buccal surface of 6 index teeth. 0 indicates that there is no plaque, 6 indicates plaque on all index teeth.

To Tooth wear index is a weighted summary of the degree of tooth wear on 6 different segments. Tooth wear is only evaluated in the permanent dentition, not in the primary teeth. The final index score is based on the degree of tooth wear found in most segments.

0: No tooth wear or minor wear of enamel in either of the segments
1: Marked tooth wear of the enamel, possibly exceeding into dentin
2: tooth wear in the dentine reaching up to 1/3 of the tooth crown
3: Tooth wear in the dentine reaching up to more than 1/3 of the tooth crown. If 3 is given in any segment then SI is 3.

1: Oral health index (indices) was (were) introduced in the observations in 2008

## Acceptance of dental examination




| | Number |
|---|---|
| None | 31 |
| Negative | 16 |
| Reluctant | 19 |
| Positive | 21 |
| Missing data | 6 |
| **Sum:** | **93** |

## Caries

| | 3-6 years | 7-12 years | 13-19 years | Adults |
|---|---|---|---|---|
| **deft[1]** | | | | |
| Examined | 21 | 18 | | |
| Number of individuals with deft=0 | 21 | 13 | | |
| Mean | 0,0 | 0,8 | | |
| Standard deviation | 0,0 | 1,6 | | |
| Missing data | 15 | 20 | | |
| **DMFT[2]** | | | | |
| Examined | | 19 | 10 | 6 |
| Number of individuals with DMFT=0 | | 16 | 7 | 2 |
| Standard deviation | | 0,8 | 2,1 | 6,4 |
| Mean | | 0,3 | 1,3 | 5,7 |
| Missing data | | 19 | 3 | 0 |

1: Number of carious or filled deciduous teeth

2: Number of carious or filled permanent teeth

## Occlusal relationship

| | Number |
|---|---|
| Neutral bite | 52 |
| Post normal | 9 |
| Pre normal | 8 |
| Missing data | 24 |
| | **Sum: 93** |

## Maximum jaw opening

Children younger than 10 years

| | Number |
|---|---|
| - 20 | 0 |
| 21 - 30 | 0 |
| 31 - 40 | 12 |
| 41 - 50 | 10 |
| 51 - | 0 |
| Missing data | 36 |
| | **Sum: 58** |

Children, 10 years or older, and adults

| | Number |
|---|---|
| - 20 | 0 |
| 21 - 30 | 0 |
| 31 - 40 | 4 |
| 41 - 50 | 10 |
| 51 - | 5 |
| Missing data | 16 |
| | **Sum: 35** |

## Profile[1]

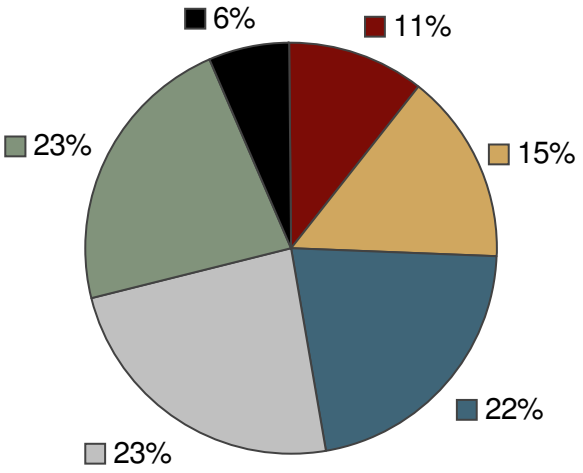
| | Number |
|---|---|
| Normal | 20 |
| Convex | 1 |
| Concave | 0 |
| Missing data | 11 |
| | **Sum: 32** |

## Mandibular plane[1]

| | Number |
|---|---|
| Normal | 9 |
| Increased | 2 |
| Reduced | 1 |
| Missing data | 20 |
| | **Sum: 32** |

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

## Speech difficulty




| | Number |
|---|---|
| No speech | 10 |
| Very incomprehensible | 14 |
| Incomprehensible speech | 20 |
| Slightly indistinct speech | 22 |
| No problems | 21 |
| Missing data | 6 |
| **Sum:** | **93** |

| **Clinical findings** | Yes-answers Total N=93 (%) | Boys/Men N=79 (%) | Girls/Women N=14 (%) | Missing data |
|---|---|---|---|---|
| Open mouth at rest | **45** (49) | 40 (51) | 5 (36) | 1 |
| High palate | **30** (38) | 29 (43) | 1 (8) | 13 |
| Low muscle tone in lips | **27** (30) | 25 (33) | 2 (14) | 3 |
| Impaired tongue motility | **25** (32) | 23 (35) | 2 (15) | 15 |
| Narrow palate | **25** (34) | 25 (40) | 0 () | 20 |
| Over crowding | **14** (19) | 9 (14) | 5 (42) | 18 |
| Intra oral hypo-sensitivity | **11** (16) | 11 (19) | 0 () | 23 |
| Frontal open bite | **11** (15) | 10 (17) | 1 (8) | 21 |
| Spacing | **10** (14) | 9 (15) | 1 (8) | 20 |
| Low muscle tone in tongue | **8** (9) | 6 (8) | 2 (15) | 7 |
| Low muscle tone in masticatory muscles | **8** (12) | 7 (13) | 1 (9) | 26 |
| M mentalis overactive | **6** (7) | 4 (5) | 2 (14) | 5 |
| Facial asymmetry | **5** (6) | 5 (7) | 0 () | 4 |
| Mucous membrane changes | **4** (6) | 4 (7) | 0 () | 24 |
| Short tongue frenulum | **3** (4) | 3 (4) | 0 () | 12 |