



Orofacial function of persons having Fragile X syndrome Report from questionnaires

The survey comprises 41 questionnaires.

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, mitralis 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.

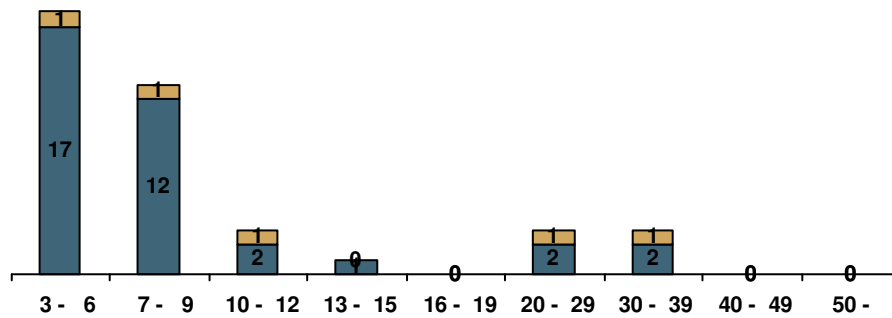
Orofacial/odontological treatment:

- 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: 41

Ages: 3 - 38

Sex: M (36) F (5)

Additional diagnoses

Medical impairment	Yes	No	Missing data
Inborn heart defect	0	39	2
Other cardiovascular disease	0	40	1
Epilepsy	0	38	3
Asthma	1	38	2
Need of respiratory support	0	40	1
Allergy	8	31	2

Neuropsychiatric diagnosis	Yes	No	Missing data
ADHD/ADD	9	29	3
Autistic syndrome	9	29	3
Autistic traits	23	12	6
Asperger's syndrome	1	39	1
Tourette's syndrome	0	40	1

General disability	Yes	No	Missing data
Intellectual disability	13	11	17
Mobility dysfunction	16	18	7
Impaired vision	5	32	4
Impaired hearing	0	40	1
Difficulty in communicating	11	8	22

About dental care and oral health

Do you feel that you receive the dental care you need?

	Number
Yes, very much so	23
Yes, some what	14
No, not really	2
No, not at all	2
Missing data	0
Sum:	41

How many times per year do you normally seek dental care?

	Number
Three or more times per year	4
Twice per year	8
Once per year	16
Less than once per year	8
Missing data	5
Sum:	41

When were your teeth last X-rayed?

	Number
During the past two years	14
More than two years ago	4
Never had my teeth X-rayed	23
Missing data	0
Sum:	41

Do you look after your teeth in a good way?

	Number
Yes, very much so	12
Yes, some what	27
No, not really	1
No, not at all	0
Missing data	1
Sum:	41

Who brushes your teeth?

	Number
I always brush myself	6
Sometimes I brush myself	11
Someone else always helps me	24
Missing data	0
Sum:	41

How often are your teeth brushed?

	Number
Three or more times per day	1
Twice per day	34
Once per day	5
Not every day	1
Missing data	0
Sum:	41

About dental care and oral health

	Yes	No	Missing data
Does your mouth hurt?	3	34	4
Does your mouth feel dry?	2	30	9
Have you ever taken a serious hit to your permanent front teeth?	5	35	1
Do you feel that you have a divergent bite?	3	34	4
Have you had a brace?	2	38	1
Do you feel that you need orthodontics/a brace?	2	33	6

Do you grind or press your teeth at night?

	Number
Never	32
Once or twice per week	5
Every night	1
Missing data	3
Sum:	41

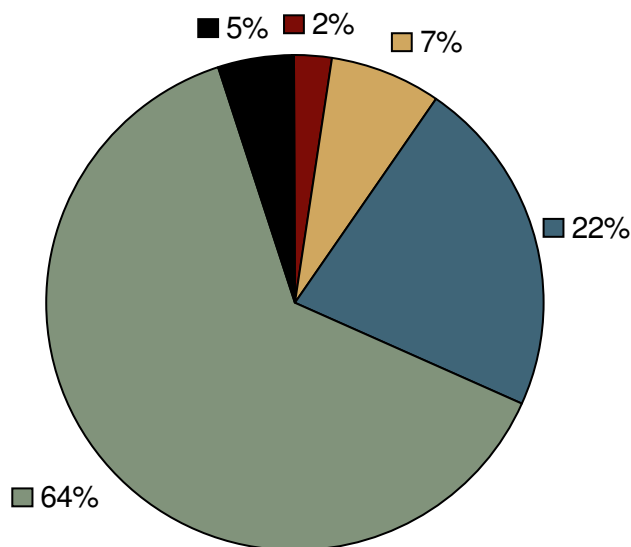
Do you grind or press your teeth during the day?

	Number
Never	37
Once or twice per week	0
Every day	4
Missing data	0
Sum:	41



About eating

Do you have any problems with eating?



	Number
Yes, very much so	1
Yes, some what	3
No, not really	9
No, not at all	26
Missing data	2
Sum:	41

	Yes	No	Missing data
Do you cough daily in connection with meals?	1	38	2
Do you gag daily in connection with meals?	0	41	0
Do you get acid reflux daily?	0	37	4
Do you throw up often (at least twice per week)?	2	39	0
Do you have a poor appetite?	3	36	2
Does it take a long time before you can swallow a mouthful?	2	39	0
Do you press your tongue forward when you swallow so that food ends up outside the mouth?	1	40	0
Do you find it difficult to chew, i.e. grind food using your molars?	2	33	6
Do you find it difficult to take food from the spoon using your lips?	5	35	1
Have you had problems with food and drink leaking out through the corners of your mouth?	3	37	1
Does food tend to remain in your mouth after meals?	1	39	1
Do you get nutrition in any other way than through your mouth?	0	41	0

About drooling

Do you drool?

	Number
Never drool	24
Drool sometimes – not every day	10
Drool often – every day	6
Constant drooling	0
Missing data	1
Sum:	41

How much do you drool?

	Number
Slight drooling, only on the lips	7
Moderate drooling, on lip and chin	6
Profuse drooling, on clothes	2
Very profuse drooling, on hands/objects	0
Missing data	1
Sum:	16

Is your drooling a problem for you?

	Number
Yes, very much so	1
Yes, some what	3
No, not really	6
No, not at all	5
Missing data	1
Sum:	16

Is your drooling a problem for your family or people around you?

	Number
Yes, very much so	1
Yes, some what	3
No, not really	6
No, not at all	5
Missing data	1
Sum:	16