



Orofacial function of persons having Osteogenesis imperfecta

Report from questionnaires

The survey comprises 12 questionnaires.

Estimated occurrence: 5:100 000 live births.

Etiology: The protein "collagen", that acts to reinforce the bone tissue, is defective. OI is caused by a mutation in the collagen gene on chromosome 7 or 17. The mutations differ, which result in varying degrees of fragility of the bones.

General symptoms: Type I – IV, from minor to severe symptoms. Moderate to severe bone fragility may cause multiple fractures, back pain and scoliosis. Other symptoms that may occur include discoloration of the sclera, hearing impairment, loose joints, instability of the cervical spine, cardiovascular problems, hyperthermia – "sweating", bleeding diathesis, varying degrees of shortness of stature, etc.

Orofacial/odontological symptoms:

The most common tooth development disorder is Dentinogenesis Imperfecta (DI).

DI is characterized by:

- Tooth discoloration (light blue to dark brown with a transparent glaze).
- Dentin is softer than normal.
- Enamel "splinters" from the soft dentin.
- The soft dentin causes the teeth to wear down rapidly, especially the primary teeth.

Aplasia of one or more permanent teeth is common (some permanent teeth are missing). X-rays sometimes reveal elongated pulp chambers. The upper jaw is usually small causing malocclusion with prenatal occlusion (underbite)

Orofacial/Odontological treatment:

- Early collaboration with specialists in child dentistry and orthodontics
- Steel crowns on six-year-molars, in combination with long-term temporary dental filling therapy, reduce the risk for extensive tooth wear on the crossbite side.
- Plan early corrective surgical and prosthetic treatment to improve functional and esthetic occlusion.
- Increased risk associated with anesthesia due to instability of the cervical spine.

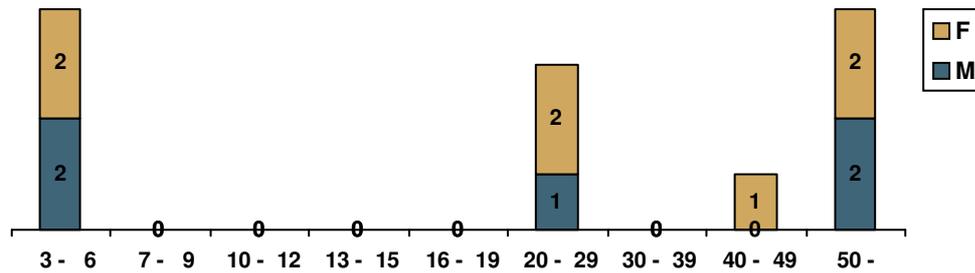
Sources

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.

The Documentation from the Ågrenska Center.

Age distribution



Number: 12

Ages: 3 - 58

Sex: M (5) F (7)

Additional diagnoses

Medical impairment	Yes	No	Missing data
Inborn heart defect	0	12	0
Other cardiovascular disease	0	12	0
Epilepsy	0	12	0
Asthma	0	12	0
Need of respiratory support	0	12	0
Allergy	4	8	0

Neuropsychiatric diagnosis	Yes	No	Missing data
ADHD/ADD	0	11	1
Autistic syndrome	0	11	1
Autistic traits	0	11	1
Asperger's syndrome	0	11	1
Tourette's syndrome	0	11	1

General disability	Yes	No	Missing data
Intellectual disability	0	11	1
Mobility dysfunction	3	7	2
Impaired vision	2	7	3
Impaired hearing	0	8	4
Difficulty in communicating	1	11	0

About dental care and oral health

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

	Number
Yes, very much so	7
Yes, some what	2
No, not really	1
No, not at all	1
Missing data	1
Sum:	12

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

	Number
Three or more times per year	2
Twice per year	3
Once per year	5
Less than once per year	1
Missing data	1
Sum:	12

When were your teeth last X-rayed?

	Number
During the past two years	8
More than two years ago	1
Never had my teeth X-rayed	1
Missing data	2
Sum:	12

Do you look after your teeth in a good way?

	Number
Yes, very much so	8
Yes, some what	4
No, not really	0
No, not at all	0
Missing data	0
Sum:	12

Who brushes your teeth?

	Number
I always brush myself	9
Sometimes I brush myself	0
Someone else always helps me	3
Missing data	0
Sum:	12

How often are your teeth brushed?

	Number
Three or more times per day	2
Twice per day	9
Once per day	1
Not every day	0
Missing data	0
Sum:	12

About dental care and oral health

	Yes	No	Missing data
Does your mouth hurt?	3	9	0
Does your mouth feel dry?	0	12	0
Have you ever taken a serious hit to your permanent front teeth?	1	11	0
Do you feel that you have a divergent bite?	6	6	0
Have you had a brace?	1	11	0
Do you feel that you need orthodontics/a brace?	4	8	0

Do you grind or press your teeth at night?

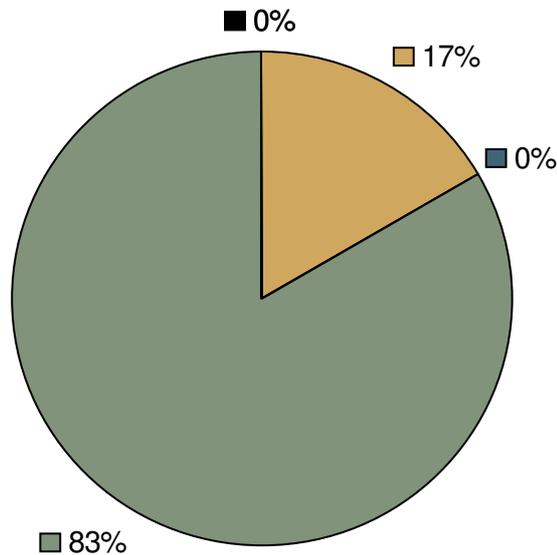
	Number
Never	8
Once or twice per week	3
Every night	1
Missing data	0
Sum:	12

Do you grind or press your teeth during the day?

	Number
Never	9
Once or twice per week	3
Every day	0
Missing data	0
Sum:	12

About eating

Do you have any problems with eating?



	Number
Yes, very much so	0
Yes, some what	2
No, not really	0
No, not at all	10
Missing data	0
Sum:	12

	Yes	No	Missing data
Do you cough daily in connection with meals?	0	12	0
Do you gag daily in connection with meals?	0	12	0
Do you get acid reflux daily?	0	12	0
Do you throw up often (at least twice per week)?	0	12	0
Do you have a poor appetite?	0	12	0
Does it take a long time before you can swallow a mouthful?	1	11	0
Do you press your tongue forward when you swallow so that food ends up outside the mouth?	0	12	0
Do you find it difficult to chew, i.e. grind food using your molars?	2	8	2
Do you find it difficult to take food from the spoon using your lips?	0	11	1
Have you had problems with food and drink leaking out through the corners of your mouth?	0	11	1
Does food tend to remain in your mouth after meals?	0	11	1
Do you get nutrition in any other way than through your mouth?	0	12	0

About drooling

Do you drool?

	Number
Never drool	10
Drool sometimes – not every day	2
Drool often – every day	0
Constant drooling	0
Missing data	0
Sum:	12

How much do you drool?

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

Is your drooling a problem for you?

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

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

	Number
Yes, very much so	0
Yes, some what	0
No, not really	1
No, not at all	1
Missing data	0
Sum:	2