Orofacial function of persons having

Osteogenesis imperfecta

Report from observation charts

The survey comprises 23 observation charts.

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.

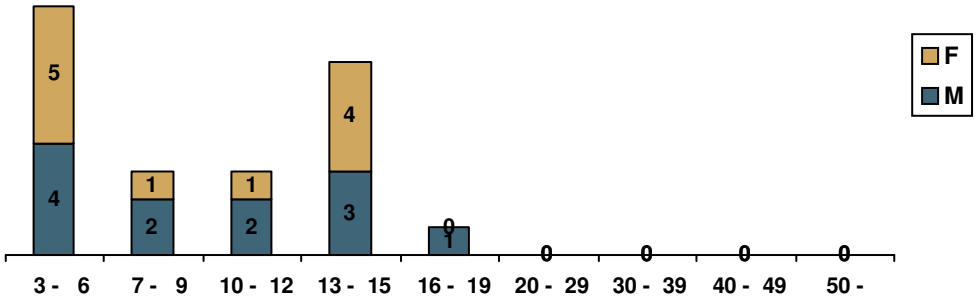
Aplasia of one or more permanent teeth is common (some permanent teeth are missing).
X-rays sometimes reveal elongated pulp chambers. The upper jaw is usually small causing malocclusion with prenormal occlusion (underbite)

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



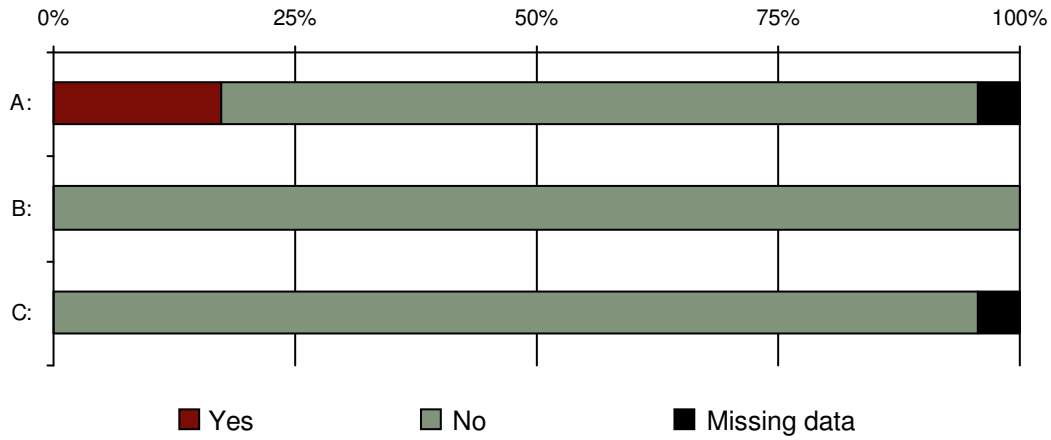
Number: 23

Ages: 3 - 18

Sex: M (12) F (11)

Overview

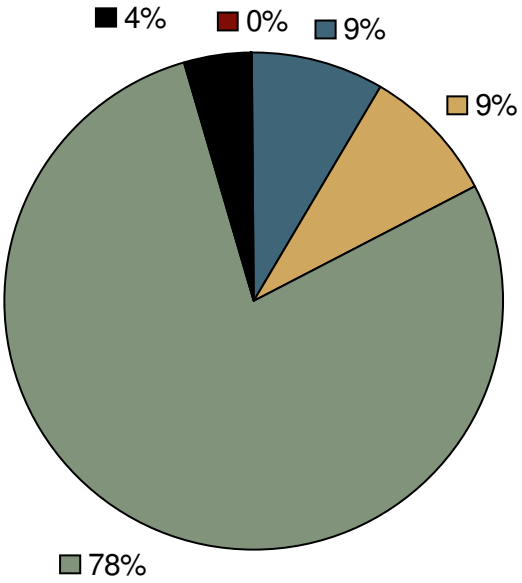
		Yes	No	Missing data	N
A:	Incomprehensible speech/No speech	4	18	1	23
B:	Eating and drinking difficulties[1]	0	23	0	23
C:	Profuse drooling, on clothes[1]	0	22	1	23



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

1: Compiled using questionnaire

Acceptance of dental examination



	Number
None	0
Negative	2
Reluctant	2
Positive	18
Missing data	1
Sum:	**23**

Caries

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

1: Number of carious or filled deciduous teeth

2: Number of carious or filled permanent teeth

Occlusal relationship

	Number
Neutral bite	15
Post normal	3
Pre normal	4
Missing data	1
	Sum: 23

Maximum jaw opening

Children younger than 10 years

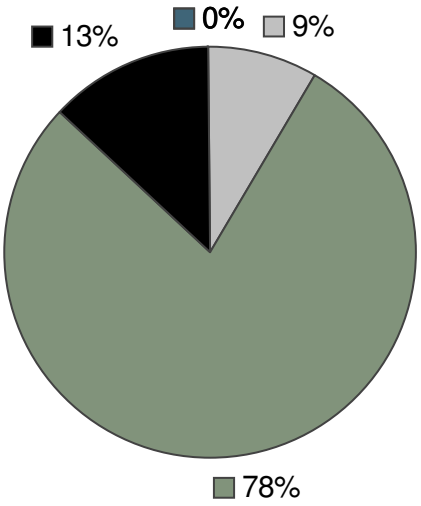
	Number
- 20	0
21 - 30	1
31 - 40	2
41 - 50	5
51 -	1
Missing data	3
	Sum: 12

Children, 10 years or older, and adults

	Number
- 20	0
21 - 30	0
31 - 40	1
41 - 50	8
51 -	1
Missing data	1
	Sum: 11

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

Speech difficulty



	Number
No speech	0
Very incomprehensible	0
Incomprehensible speech	0
Slightly indistinct speech	2
No problems	18
Missing data	3

Sum: 23

Clinical findings	Yes-answers Total N=23 (%)	Boys/Men N=12 (%)	Girls/Women N=11 (%)	Missing data
Frontal open bite	**4** (17)	2 (17)	2 (18)	0
Spacing	**3** (13)	1 (8)	2 (18)	0