



Orofacial function of persons having Marfan syndrome

Report from observation charts

The survey comprises 42 observation charts.

Estimated occurrence: 10-20:100 000 inhabitants.

Aetiology: Marfan syndrome is caused by a genetic mutation, usually on chromosome 15. This gene is responsible for production of fibrillin, a protein essential to the formation of connective tissue. It is inherited as an autosomal dominant trait. Spontaneous mutations are also known to occur.

General symptoms: Individuals with Marfan syndrome are often extremely tall. They may have problems associated with various body parts. Most common are muscular and skeletal problems, joint laxity, dilatation (widening) of aorta, mitral valve problems, vision impairment and some times pulmonary disorders.

Orofacial/odontological symptoms: Common symptoms include a high, sometimes narrow palate, and lack of space for teeth in the jaws. Snoring and an increased risk of sleep apnea may occur. The temporomandibular joint is often affected, and there may be instability of the joint-capsules. Tooth grinding implies extra pressure on the jaw, which may increase the joint problems. For the same reason, the patient may have difficulty opening his or her mouth wide and for a long time, as may be necessary, for example, at dental appointments.

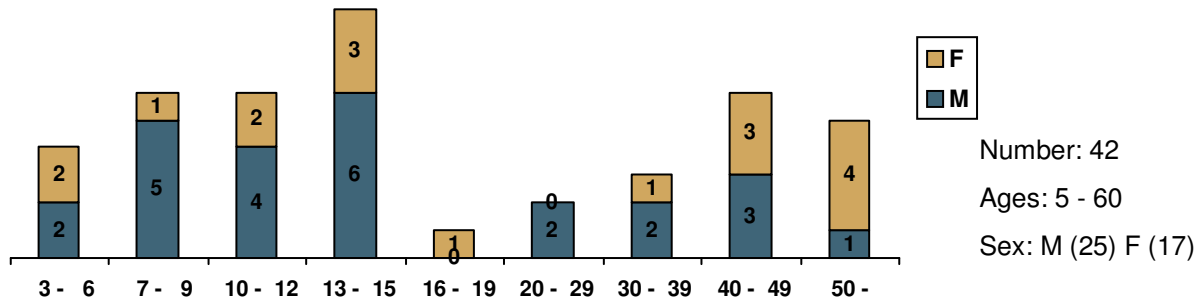
Orofacial/odontological treatment:

- Extra prophylactic dental care may be relevant in order to prevent the need for dental treatment that would be very demanding for these patients because of their jaw problems. In some patients there is a risk of endocarditis if oral bacteria gain access to the blood stream. Improved oral and especially gingival health reduces this risk.
- Regular check-ups of dental and jaw development. Orthodontist should be consulted when needed.
- In cases of temporomandibular joints disorders, this should be investigated and appropriate treatment thereafter prescribed.
- Problems associated with snoring and sleep apnea should be followed up by a physician.
- When treating medically compromised patients always contact their doctors for medical advice.

Sources:

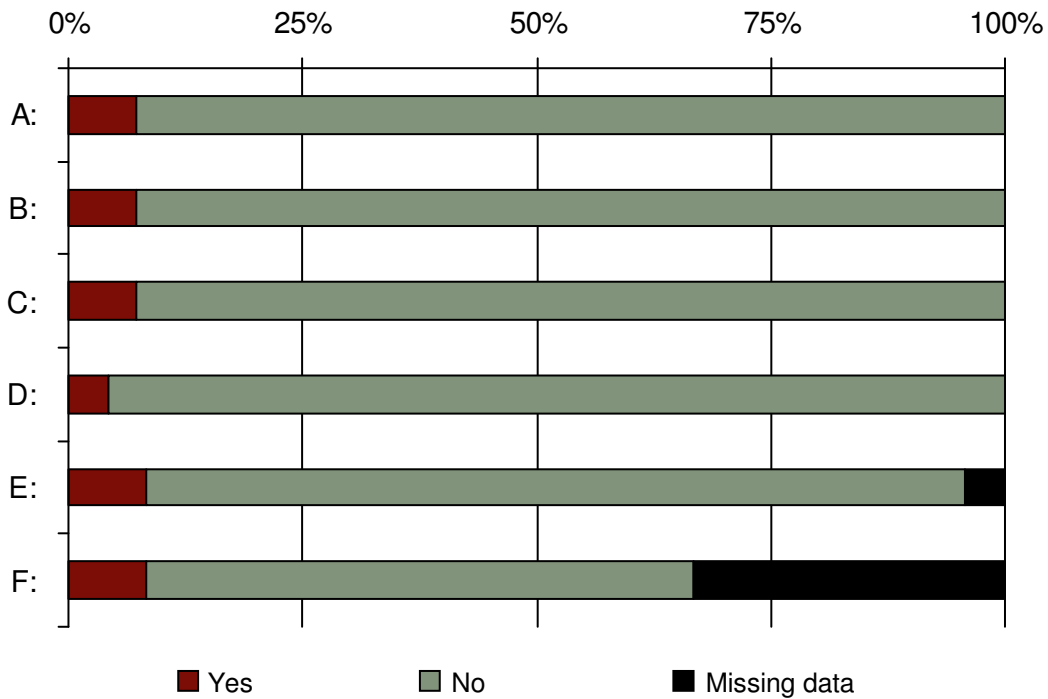
The rare disease database of the Swedish National Board of Health and Welfare.
The MHC database - The Mun-H-Center database on oral health and orofacial function in rare diseases.
The Documentation from the Ågrenska Center.

Age distribution



Overview

	Yes	No	Missing data	N
A: Incomprehensible speech/No speech	3	39	0	42
B: Eating and drinking difficulties ¹	3	39	0	42
C: Profuse drooling, on clothes ¹	3	39	0	42
D: Breathing difficulties ^{1 2}	1	23	0	24
E: Grinding every day ^{1 2}	2	21	1	24
F: Severe malocclusions ²	2	14	8	24



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

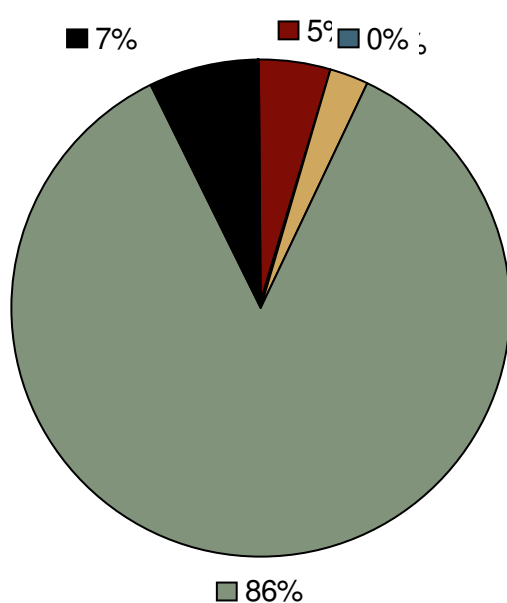
Oral health index (indices)¹

		0	1	2	3	4	5	6	Missing data	N
Calc	Calculus	17	2	0	0	0	0	0	5	24
GI	Gingivitis	19	2	0	0	0	0	0	3	24
Plaq	Coating	12	1	5	0	0	0	1	5	24
Toot	Tooth wear	13	8	1	0				2	24

- 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	2
Negative	0
Reluctant	1
Positive	36
Missing data	3
Sum:	42

Caries

	3-6 years	7-12 years	13-19 years	Adults
deft¹				
Examined	2	8		
Number of individuals with deft=0	1	6		
Mean	2,5	0,4		
Standard deviation	2,5	0,7		
Missing data	2	4		
DMFT²				
Examined		9	7	14
Number of individuals with DMFT=0		8	1	1
Standard deviation		0,3	1,4	3,1
Mean		0,1	1,9	3,2
Missing data		3	3	2

1: Number of carious or filled deciduous teeth

2: Number of carious or filled permanent teeth

Occlusal relationship

	Number
Neutral bite	26
Post normal	10
Pre normal	5
Missing data	1
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	Sum: 42

Maximum jaw opening

Children younger than 10 years

	Number
- 20	0
21 - 30	0
31 - 40	4
41 - 50	4
51 -	0
Missing data	2
<hr/>	
	Sum: 10

Children, 10 years or older, and adults

	Number
- 20	0
21 - 30	0
31 - 40	8
41 - 50	15
51 -	6
Missing data	3
<hr/>	
	Sum: 32

Profile¹

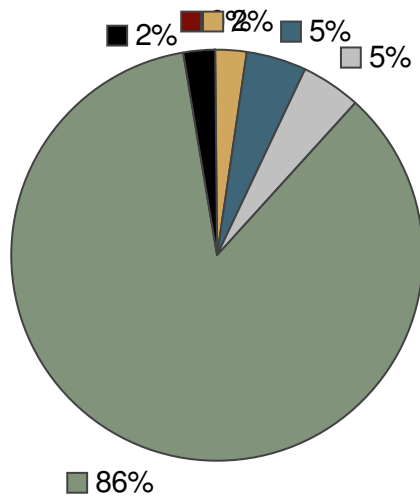
	Number
Normal	22
Convex	1
Concave	0
Missing data	1
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	Sum: 24

Mandibular plane¹

	Number
Normal	16
Increased	2
Reduced	0
Missing data	6
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	Sum: 24

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

Speech difficulty



	Number
No speech	0
Very incomprehensible	1
Incomprehensible speech	2
Slightly indistinct speech	2
No problems	36
Missing data	1
Sum: 42	

Clinical findings	Yes-answers			
	Total N=42 (%)	Boys/Men N=25 (%)	Girls/Women N=17 (%)	Missing data
High palate	27 (64)	18 (72)	9 (53)	0
Open mouth at rest	17 (40)	12 (48)	5 (29)	0
Narrow palate	16 (38)	9 (36)	7 (41)	0
Over crowding	14 (33)	9 (36)	5 (29)	0
M mentalis overactive	6 (15)	6 (26)	0 ()	3
Low muscle tone in lips	6 (14)	6 (24)	0 ()	0
Reduced opening capacity	6 (14)	3 (12)	3 (18)	0
Low muscle tone in masticatory muscles	4 (10)	3 (12)	1 (6)	0
Frontal open bite	4 (10)	4 (16)	0 ()	0
Mucous membrane changes	3 (7)	2 (8)	1 (6)	0

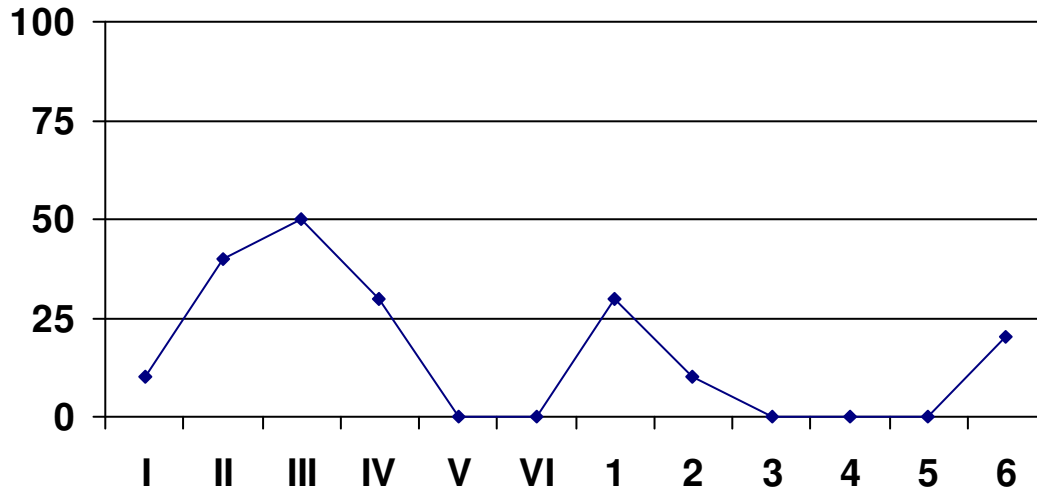
NOT-S

Total mean score: 1,90

Number: 10

Ages: 5 - 56

Sex: M (6) F (4)



NOT-S interview	Number	%
I : Sensory function	1	10,0
II : Breathing	4	40,0
III : Habits	5	50,0
IV : Chewing and swallowing	3	30,0
V : Drooling	0	,0
VI : Dry mouth	0	,0

NOT-S examination	Number	%
1 : Face at rest	3	30,0
2 : Nose breathing	1	10,0
3 : Facial expression	0	,0
4 : Masticatory muscle and jaw function	0	,0
5 : Oral motor function	0	,0
6 : Speech	2	20,0