Orofacial function of persons having Tuberous sclerosis

Report from observation charts

The survey comprises 70 observation charts.

Estimated occurence: 10:100 000 live births.

Etiology: Tubular sclerosis is caused by a genetic mutation, which, in turn, disrupts normal cellular development in the bodily organs. Although the genetic mutation is congenital, it often occurs as a spontaneous mutation. The pattern of heredity is an autosomal dominant genetic trait. At least two different genes, one on chromosome 9 and one on chromosome 16, have been found to give rise to this mutation.

General symptoms: The anomalies may be found in one or more organs, and may be minor or major. Organs that may be affected include the brain, kidneys, heart, eyes, lungs, nails and skin. The most common symptoms are skin abnormalities, and the most discernible functional disabilities include epilepsy, mental retardation and autism or autistic traits. Hyperactivity is a serious problem in nearly half of all individuals with severe tubular sclerosis. Impulse control disabilities and temper tantrums are also very frequent symptoms.

Orofacial symptoms: Enamel aberrations in the form of pitting are frequent. Gum fibromas may occur, particularly in the region of the front teeth. There is a risk of bone cyst formation in the jaw. Speech- and language development is often delayed, and some of these children never acquire speech. Eating difficulties and drooling are common.

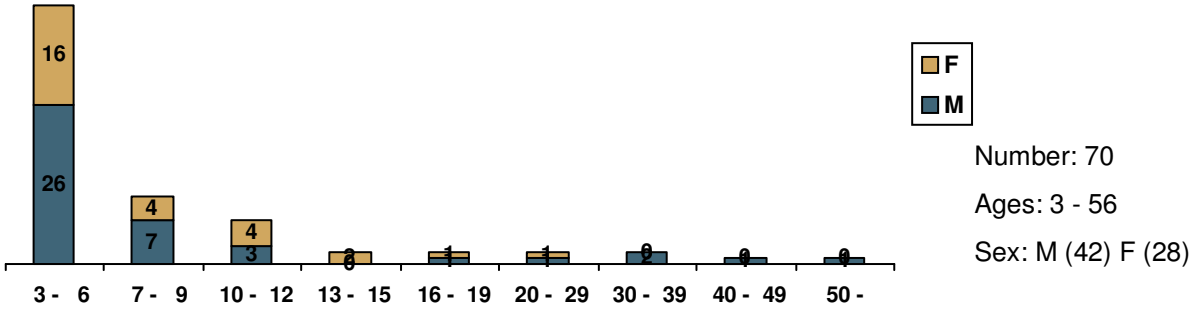
Orofacial/ odontological treatment:

- Early contact with dental services for intensified prophylactic care and oral hygiene information is essential.
- Enamel pitting seldom requires treatment but may increase the risk for caries.
- Because of the risk of bone cyst formation in the jaw, radiographic evaluation is recommended by the age of 6 to 7 years or earlier if asymmetry, asymptomatic swelling or delayed or abnormal tooth eruption sequence is evident.
- Orofacial therapy and oral motor skills training and stimulation in cases of difficulties with eating, speech or drooling may be relevant.
- Speech, language and communication training are often required.
- Feeding and swallowing difficulties are investigated and treated by a specialist team at the hospital or multidisciplinary treatment centre.

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.
Teng et al. Dermatological and Dental Aspects of the 2012 International Tuberous Sclerosis Complex Consensus Statements. JAMA Dermatology 2014; 150:1095-1101.

Age distribution



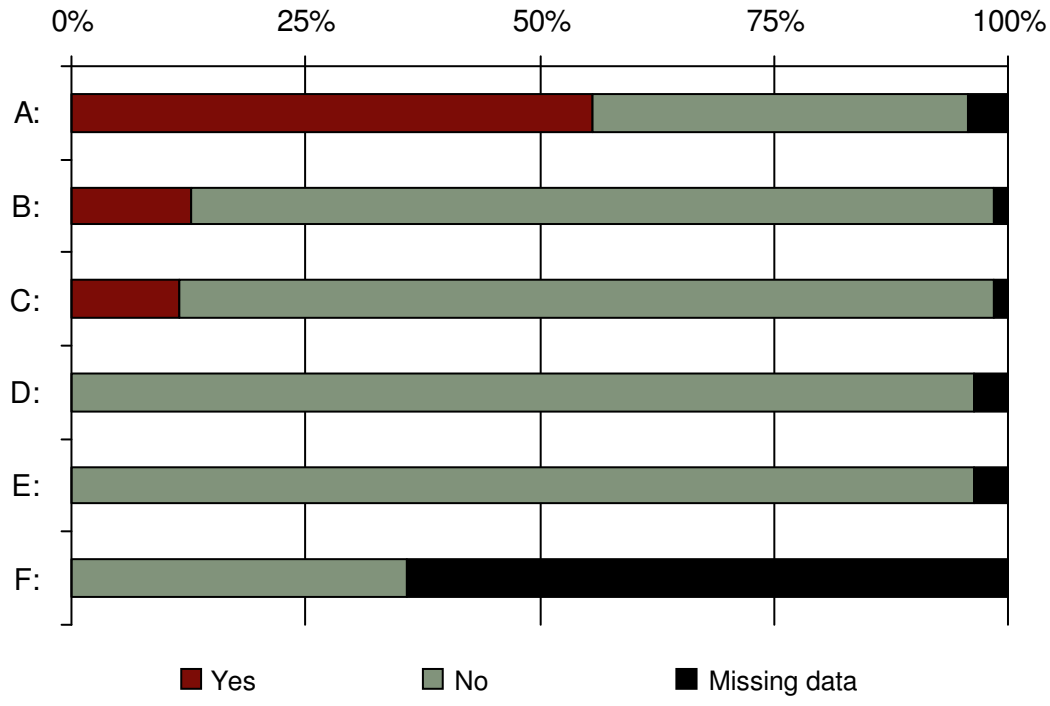
Number: 70

Ages: 3 - 56

Sex: M (42) F (28)

Overview

		Yes	No	Missing data	N
A:	Incomprehensible speech/No speech	39	28	3	70
B:	Eating and drinking difficulties[1]	9	60	1	70
C:	Profuse drooling, on clothes[1]	8	61	1	70
D:	Breathing difficulties[1] [2]	0	27	1	28
E:	Grinding every day[1] [2]	0	27	1	28
F:	Severe malocclusions[2]	0	10	18	28



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	7	0	0	0	0	0	0	21	28
GI	Gingivitis	5	2	0	0	0	0	0	21	28
Plaq	Coating	5	1	1	0	1	0	0	20	28
Toot	Tooth wear	8	0	0	0				20	28

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.

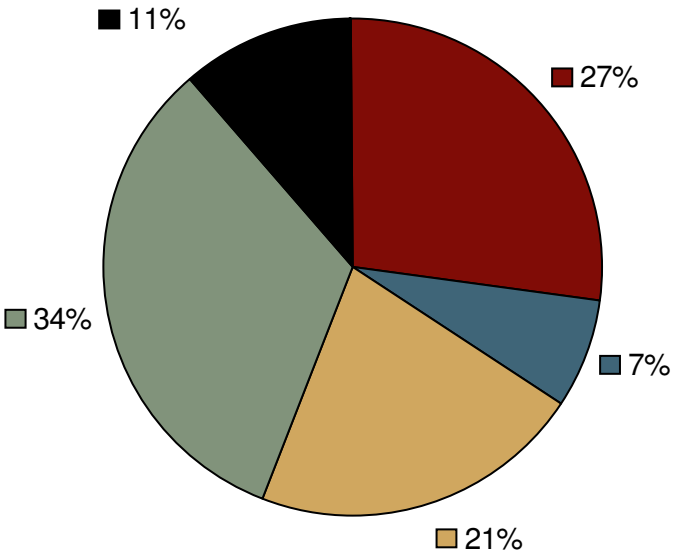
PI Plaque index is based on the presence of visible plaque on the buccal surface of 6 index teeth. 0 indicates that there is no plaque, 6 indicates plaque on all index teeth.

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	19
Negative	5
Reluctant	15
Positive	23
Missing data	8
Sum:	**70**

Caries

	3-6 years	7-12 years	13-19 years	Adults
deft[1]				
Examined	25	13		
Number of individuals with deft=0	23	11		
Mean	0,2	0,2		
Standard deviation	0,9	0,6		
Missing data	17	5		
DMFT[2]				
Examined		13	4	6
Number of individuals with DMFT=0		12	0	1
Standard deviation		1,1	3,7	3,0
Mean		0,3	5,0	3,5
Missing data		5	0	0

1: Number of carious or filled deciduous teeth

2: Number of carious or filled permanent teeth

Occlusal relationship

	Number
Neutral bite	42
Post normal	12
Pre normal	3
Missing data	13
	Sum: 70

Maximum jaw opening

Children younger than 10 years

	Number
- 20	0
21 - 30	0
31 - 40	10
41 - 50	9
51 -	0
Missing data	34
	Sum: 53

Children, 10 years or older, and adults

	Number
- 20	0
21 - 30	0
31 - 40	3
41 - 50	3
51 -	1
Missing data	10
	Sum: 17

Profile[1]

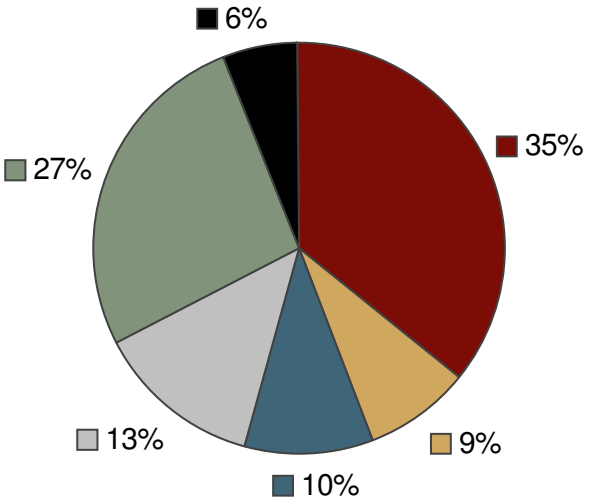
	Number
Normal	16
Convex	5
Concave	0
Missing data	7
	Sum: 28

Mandibular plane[1]

	Number
Normal	13
Increased	0
Reduced	3
Missing data	12
	Sum: 28

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

Speech difficulty



	Number
No speech	25
Very incomprehensible	6
Incomprehensible speech	7
Slightly indistinct speech	9
No problems	19
Missing data	4

Sum: 70

Clinical findings	Yes-answers Total N=70 (%)	Boys/Men N=42 (%)	Girls/Women N=28 (%)	Missing data
Open mouth at rest	**23** (33)	12 (29)	11 (39)	0
Frontal open bite	**17** (29)	10 (29)	7 (29)	11
Low muscle tone in lips	**10** (14)	5 (12)	5 (18)	0
Spacing	**8** (15)	4 (12)	4 (19)	15
M mentalis overactive	**7** (10)	3 (7)	4 (14)	0
Low muscle tone in tongue	**6** (10)	3 (8)	3 (12)	8
Impaired tongue motility	**6** (12)	2 (7)	4 (17)	18
Intra oral hypo-sensitivity	**5** (9)	2 (6)	3 (13)	13
Over crowding	**4** (7)	3 (9)	1 (5)	15
Gingival hyperplasia	**4** (8)	1 (3)	3 (16)	19
Facial asymmetry	**3** (4)	2 (5)	1 (4)	1
Mucous membrane changes	**3** (6)	3 (10)	0 ()	22