



Orofacial function of persons having Tuberous sclerosis

Report from questionnaires

The survey comprises 28 questionnaires.

Estimated occurrence: 1:100 000 live births.

Etiology: Tuberous 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 tuberous 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

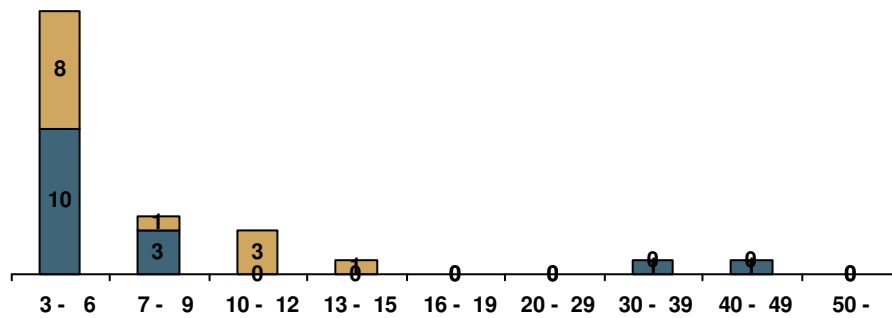
The rare disease database of the Swedish National Board of Health and Welfare.

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: 28

Ages: 3 - 46

Sex: M (15) F (13)

Additional diagnoses

Medical impairment	Yes	No	Missing data
Inborn heart defect	6	22	0
Other cardiovascular disease	2	26	0
Epilepsy	22	5	1
Asthma	5	23	0
Need of respiratory support	0	28	0
Allergy	5	22	1

Neuropsychiatric diagnosis	Yes	No	Missing data
ADHD/ADD	3	23	2
Autistic syndrome	9	16	3
Autistic traits	13	8	7
Asperger's syndrome	0	26	2
Tourette's syndrome	0	27	1

General disability	Yes	No	Missing data
Intellectual disability	6	7	15
Mobility dysfunction	10	12	6
Impaired vision	3	22	3
Impaired hearing	0	28	0
Difficulty in communicating	6	5	17

About dental care and oral health

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

	Number
Yes, very much so	13
Yes, some what	13
No, not really	0
No, not at all	2
Missing data	0
Sum:	28

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

	Number
Three or more times per year	7
Twice per year	5
Once per year	11
Less than once per year	5
Missing data	0
Sum:	28

When were your teeth last X-rayed?

	Number
During the past two years	11
More than two years ago	2
Never had my teeth X-rayed	15
Missing data	0
Sum:	28

Do you look after your teeth in a good way?

	Number
Yes, very much so	15
Yes, some what	12
No, not really	1
No, not at all	0
Missing data	0
Sum:	28

Who brushes your teeth?

	Number
I always brush myself	2
Sometimes I brush myself	6
Someone else always helps me	20
Missing data	0
Sum:	28

How often are your teeth brushed?

	Number
Three or more times per day	1
Twice per day	20
Once per day	6
Not every day	1
Missing data	0
Sum:	28

About dental care and oral health

	Yes	No	Missing data
Does your mouth hurt?	0	20	8
Does your mouth feel dry?	0	20	8
Have you ever taken a serious hit to your permanent front teeth?	5	22	1
Do you feel that you have a divergent bite?	5	21	2
Have you had a brace?	0	28	0
Do you feel that you need orthodontics/a brace?	1	20	7

Do you grind or press your teeth at night?

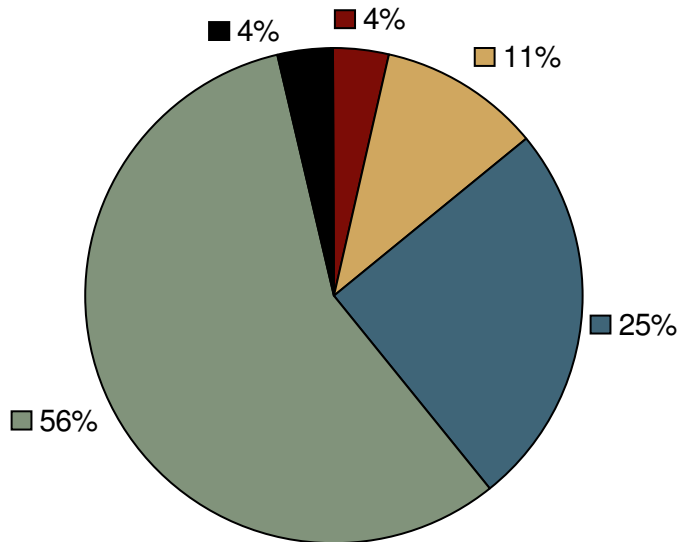
	Number
Never	15
Once or twice per week	10
Every night	3
Missing data	0
Sum:	28

Do you grind or press your teeth during the day?

	Number
Never	18
Once or twice per week	9
Every day	0
Missing data	1
Sum:	28

About eating

Do you have any problems with eating?



	Number
Yes, very much so	1
Yes, some what	3
No, not really	7
No, not at all	16
Missing data	1
Sum:	28

	Yes	No	Missing data
Do you cough daily in connection with meals?	4	24	0
Do you gag daily in connection with meals?	0	28	0
Do you get acid reflux daily?	0	24	4
Do you throw up often (at least twice per week)?	0	28	0
Do you have a poor appetite?	3	24	1
Does it take a long time before you can swallow a mouthful?	1	27	0
Do you press your tongue forward when you swallow so that food ends up outside the mouth?	0	28	0
Do you find it difficult to chew, i.e. grind food using your molars?	2	26	0
Do you find it difficult to take food from the spoon using your lips?	2	26	0
Have you had problems with food and drink leaking out through the corners of your mouth?	0	28	0
Does food tend to remain in your mouth after meals?	0	28	0
Do you get nutrition in any other way than through your mouth?	0	28	0

About drooling

Do you drool?

	Number
Never drool	18
Drool sometimes – not every day	8
Drool often – every day	2
Constant drooling	0
Missing data	0
Sum:	28

How much do you drool?

	Number
Slight drooling, only on the lips	4
Moderate drooling, on lip and chin	6
Profuse drooling, on clothes	0
Very profuse drooling, on hands/objects	0
Missing data	0
Sum:	10

Is your drooling a problem for you?

	Number
Yes, very much so	0
Yes, some what	0
No, not really	6
No, not at all	4
Missing data	0
Sum:	10

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	5
No, not at all	5
Missing data	0
Sum:	10