



Orofacial function of persons having Galactosemia

Report from questionnaires

The survey comprises 12 questionnaires.

ICD-10: E74.2

Estimated prevalence: 1:100,000 live births.

Cause: Galactosemia is an umbrella term for a group of hereditary diseases involving deficiency of one of the enzymes that are needed to convert the sugar galactose to glucose in the liver. The most common type is GALT, galactose-1-phosphate uridylyltransferase, often called classic galactosemia. GALT deficiency can lead to damage to the liver, kidneys, central nervous system, the crystalline lens, and the ovaries.

General symptoms: Children generally fall ill within the first two weeks of life, with unspecific symptoms like muscle weakness, difficulties sucking, and vomiting. Jaundice is a sign that the functions of the liver are affected. Liver enlargement, increased bleeding tendency, low blood sugar, clouding of the crystalline lens, and severe general infection can also emerge.

Orofacial/odontological symptoms: There is an increased risk of caries, healing ability can be impaired, and the tendency to bleed somewhat increased. Mild enamel changes, a narrow, high palate, a tendency towards a so-called frontal open bite, and low muscle tension in the lips have also been observed. Some degree of jaw hypermobility, difficulties chewing, and some effect on speech can be present. Many children suffering from galactosemia have language difficulties, and over half have difficulties with their speech motor skills. Difficulties eating are also common.

Orofacial/odontological treatment:

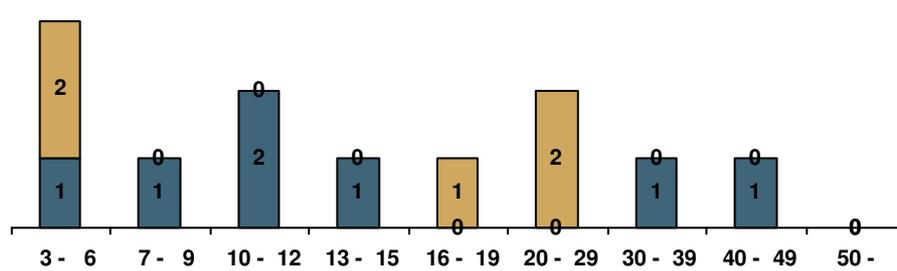
- Early contact with dental services for intensified prophylactic care and oral hygiene information is essential.
- An increased tendency to bleed may result in complications when teeth are extracted.
- Speech and language difficulties should be treated by a speech therapist.

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.

Age distribution



Number: 12

Ages: 4 - 48

Sex: M (7) F (5)

Additional diagnoses

Medical impairment	Yes	No	Missing data
Inborn heart defect	0	12	0
Other cardiovascular disease	0	11	1
Epilepsy	0	12	0
Asthma	0	12	0
Need of respiratory support	0	12	0
Allergy	2	9	1

Neuropsychiatric diagnosis	Yes	No	Missing data
ADHD/ADD	0	12	0
Autistic syndrome	0	12	0
Autistic traits	0	12	0
Asperger's syndrome	0	12	0
Tourette's syndrome	0	12	0

General disability	Yes	No	Missing data
Intellectual disability	1	10	1
Mobility dysfunction	3	9	0
Impaired vision	3	8	1
Impaired hearing	0	12	0
Difficulty in communicating	6	6	0

About dental care and oral health

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

	Number
Yes, very much so	6
Yes, some what	3
No, not really	3
No, not at all	0
Missing data	0
Sum:	12

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

	Number
Three or more times per year	1
Twice per year	0
Once per year	7
Less than once per year	4
Missing data	0
Sum:	12

When were your teeth last X-rayed?

	Number
During the past two years	6
More than two years ago	3
Never had my teeth X-rayed	3
Missing data	0
Sum:	12

Do you look after your teeth in a good way?

	Number
Yes, very much so	4
Yes, some what	5
No, not really	3
No, not at all	0
Missing data	0
Sum:	12

Who brushes your teeth?

	Number
I always brush myself	7
Sometimes I brush myself	2
Someone else always helps me	3
Missing data	0
Sum:	12

How often are your teeth brushed?

	Number
Three or more times per day	0
Twice per day	11
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	8	1
Does your mouth feel dry?	2	8	2
Have you ever taken a serious hit to your permanent front teeth?	5	7	0
Do you feel that you have a divergent bite?	3	9	0
Have you had a brace?	4	8	0
Do you feel that you need orthodontics/a brace?	2	10	0

Do you grind or press your teeth at night?

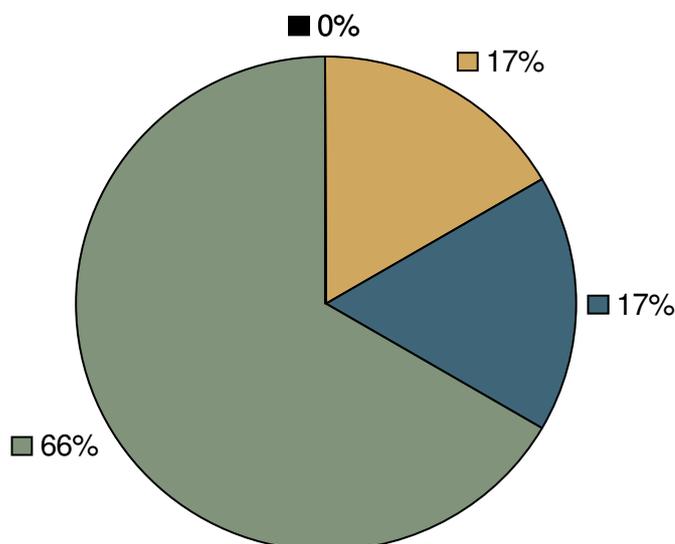
	Number
Never	10
Once or twice per week	1
Every night	0
Missing data	1
Sum:	12

Do you grind or press your teeth during the day?

	Number
Never	10
Once or twice per week	1
Every day	0
Missing data	1
Sum:	12

About eating

Do you have any problems with eating?



	Number
Yes, very much so	0
Yes, some what	2
No, not really	2
No, not at all	8
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?	1	10	1
Do you get acid reflux daily?	0	11	1
Do you throw up often (at least twice per week)?	0	12	0
Do you have a poor appetite?	2	9	1
Does it take a long time before you can swallow a mouthful?	1	10	1
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?	1	11	0
Do you find it difficult to take food from the spoon using your lips?	0	12	0
Have you had problems with food and drink leaking out through the corners of your mouth?	0	12	0
Does food tend to remain in your mouth after meals?	0	12	0
Do you get nutrition in any other way than through your mouth?	0	12	0

About drooling

Do you drool?

	Number
Never drool	9
Drool sometimes – not every day	2
Drool often – every day	1
Constant drooling	0
Missing data	0
Sum:	12

How much do you drool?

	Number
Slight drooling, only on the lips	3
Moderate drooling, on lip and chin	0
Profuse drooling, on clothes	0
Very profuse drooling, on hands/objects	0
Missing data	0
Sum:	3

Is your drooling a problem for you?

	Number
Yes, very much so	0
Yes, some what	0
No, not really	1
No, not at all	2
Missing data	0
Sum:	3

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

	Number
Yes, very much so	0
Yes, some what	1
No, not really	0
No, not at all	2
Missing data	0
Sum:	3