



Orofacial function of persons having Rett syndrome

Report from questionnaires

The survey comprises 28 questionnaires.

Estimated occurrence: 10:100 000 girls/women.

Etiology: Not entirely clarified. A mutation in the MECP2 gene on the X-chromosome (Xq28) can be identified in 80 percent. The gene mutation affects the development of the nervous system.

General symptoms: Symptoms present around the age of 1 to 2 years. The symptoms appear in four stages: Early Onset Phase (I), Rapid Destructive Phase (II), Plateau Phase (III) and Late Motor Deterioration Phase (IV). The syndrome includes inability to perform voluntary movements (apraxia), epilepsy, motor and intellectual impairment and difficulties with the regulation of breathing, blood pressure and pulse. Scoliosis is common. Repetitive hand or hand and mouth movements are characteristic for the diagnosis.

Orofacial/odontological symptoms: Severe oral motor impairment resulting in eating problems, drooling, and difficulties with oral hygiene. Most of these girls do not learn to speak. The majority have severe tooth grinding, resulting in tooth wear, and many also have involuntary tongue movements. Overbite and an open bite in the area of the front teeth are more common than in the general population.

Orofacial/ odontological treatment:

- Early contact with dental services for intensified prophylactic care and oral hygiene information is essential.
- Tooth grinding should be followed up, and be managed with a splint when necessary.
- Regular check-ups of dental and jaw development. Orthodontist should be consulted when needed
- Feeding and swallowing difficulties are investigated and treated by a specialist team at the hospital or multidisciplinary treatment center.
- Communication skills training is frequently essential.
- Oral motor training and stimulation may be relevant.

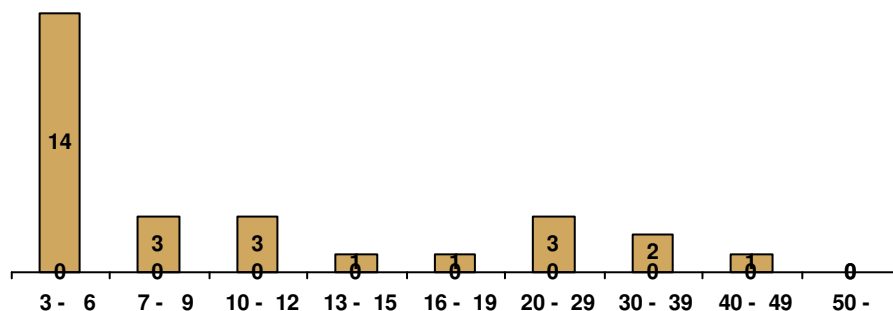
Sources:

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: 28

Ages: 3 - 47

Sex: M (0) F (28)

Additional diagnoses

Medical impairment	Yes	No	Missing data
Inborn heart defect	1	27	0
Other cardiovascular disease	1	27	0
Epilepsy	22	5	1
Asthma	0	28	0
Need of respiratory support	2	26	0
Allergy	3	25	0

Neuropsychiatric diagnosis	Yes	No	Missing data
ADHD/ADD	0	28	0
Autistic syndrome	4	22	2
Autistic traits	8	18	2
Asperger's syndrome	0	28	0
Tourette's syndrome	0	28	0

General disability	Yes	No	Missing data
Intellectual disability	1	1	26
Mobility dysfunction	0	0	28
Impaired vision	5	17	6
Impaired hearing	0	28	0
Difficulty in communicating	0	0	28

About dental care and oral health

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

	Number
Yes, very much so	16
Yes, some what	10
No, not really	2
No, not at all	0
Missing data	0
Sum:	28

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

	Number
Three or more times per year	15
Twice per year	4
Once per year	6
Less than once per year	3
Missing data	0
Sum:	28

When were your teeth last X-rayed?

	Number
During the past two years	5
More than two years ago	3
Never had my teeth X-rayed	14
Missing data	6
Sum:	28

Do you look after your teeth in a good way?

	Number
Yes, very much so	13
Yes, some what	12
No, not really	3
No, not at all	0
Missing data	0
Sum:	28

Who brushes your teeth?

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

How often are your teeth brushed?

	Number
Three or more times per day	1
Twice per day	24
Once per day	3
Not every day	0
Missing data	0
Sum:	28

About dental care and oral health

	Yes	No	Missing data
Does your mouth hurt?	1	16	11
Does your mouth feel dry?	1	16	11
Have you ever taken a serious hit to your permanent front teeth?	1	26	1
Do you feel that you have a divergent bite?	5	21	2
Have you had a brace?	1	27	0
Do you feel that you need orthodontics/a brace?	3	22	3

Do you grind or press your teeth at night?

	Number
Never	11
Once or twice per week	10
Every night	4
Missing data	3
Sum:	28

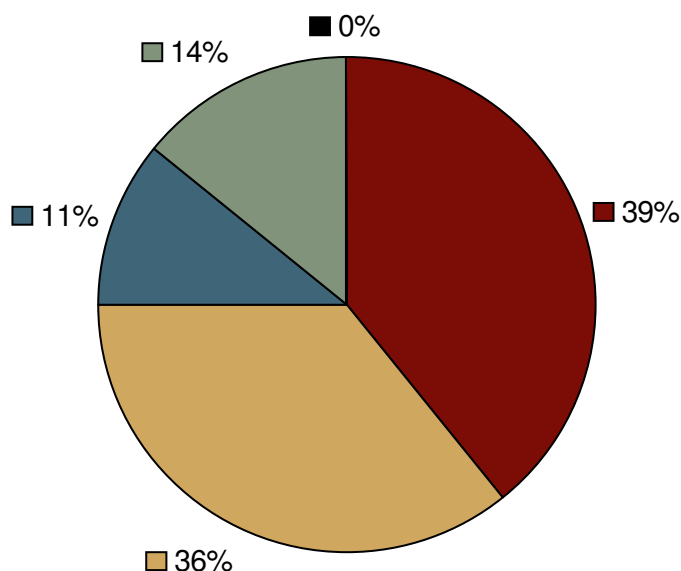
Do you grind or press your teeth during the day?

	Number
Never	5
Once or twice per week	9
Every day	12
Missing data	2
Sum:	28



About eating

Do you have any problems with eating?



	Number
Yes, very much so	11
Yes, some what	10
No, not really	3
No, not at all	4
Missing data	0
Sum:	28

	Yes	No	Missing data
Do you cough daily in connection with meals?	6	20	2
Do you gag daily in connection with meals?	3	23	2
Do you get acid reflux daily?	3	19	6
Do you throw up often (at least twice per week)?	4	22	2
Do you have a poor appetite?	5	20	3
Does it take a long time before you can swallow a mouthful?	10	12	6
Do you press your tongue forward when you swallow so that food ends up outside the mouth?	7	16	5
Do you find it difficult to chew, i.e. grind food using your molars?	8	14	6
Do you find it difficult to take food from the spoon using your lips?	7	17	4
Have you had problems with food and drink leaking out through the corners of your mouth?	21	4	3
Does food tend to remain in your mouth after meals?	12	12	4
Do you get nutrition in any other way than through your mouth?	11	17	0

About drooling

Do you drool?

	Number
Never drool	4
Drool sometimes – not every day	6
Drool often – every day	12
Constant drooling	6
Missing data	0
Sum:	28

How much do you drool?

	Number
Slight drooling, only on the lips	1
Moderate drooling, on lip and chin	11
Profuse drooling, on clothes	6
Very profuse drooling, on hands/objects	6
Missing data	0
Sum:	24

Is your drooling a problem for you?

	Number
Yes, very much so	7
Yes, some what	7
No, not really	8
No, not at all	2
Missing data	0
Sum:	24

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

	Number
Yes, very much so	5
Yes, some what	6
No, not really	9
No, not at all	4
Missing data	0
Sum:	24