



Orofacial function of persons having Rett syndrome

Report from observation charts

The survey comprises 83 observation charts.

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.

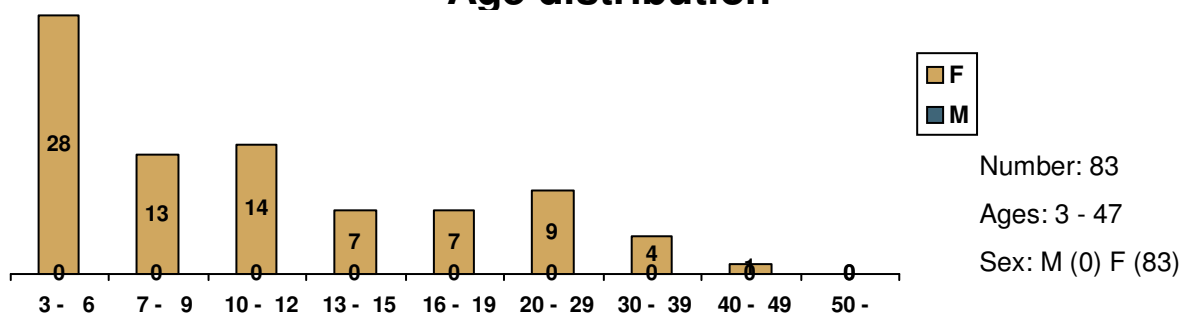
Sources:

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.

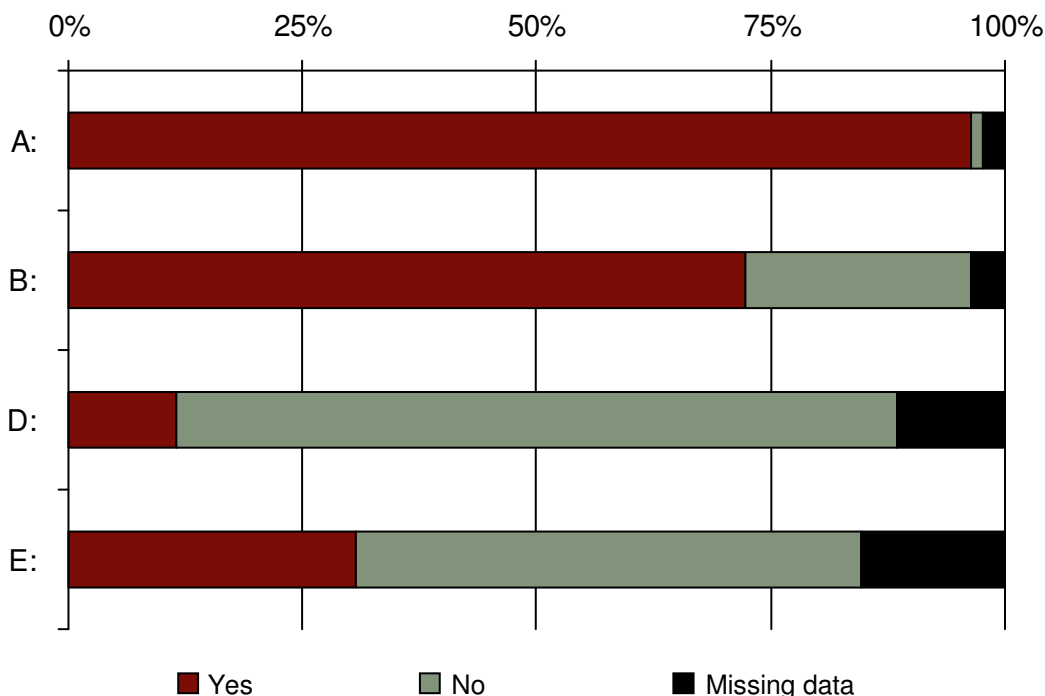
The Documentation from the Ågrenska Center.

Age distribution



Overview

	Yes	No	Missing data	N
A: Incomprehensible speech/No speech	80	1	2	83
B: Eating and drinking difficulties ¹	60	20	3	83
C: Profuse drooling, on clothes ¹	60	20	3	83
D: Breathing difficulties ^{1 2}	3	20	3	26
E: Grinding every day ^{1 2}	8	14	4	26



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

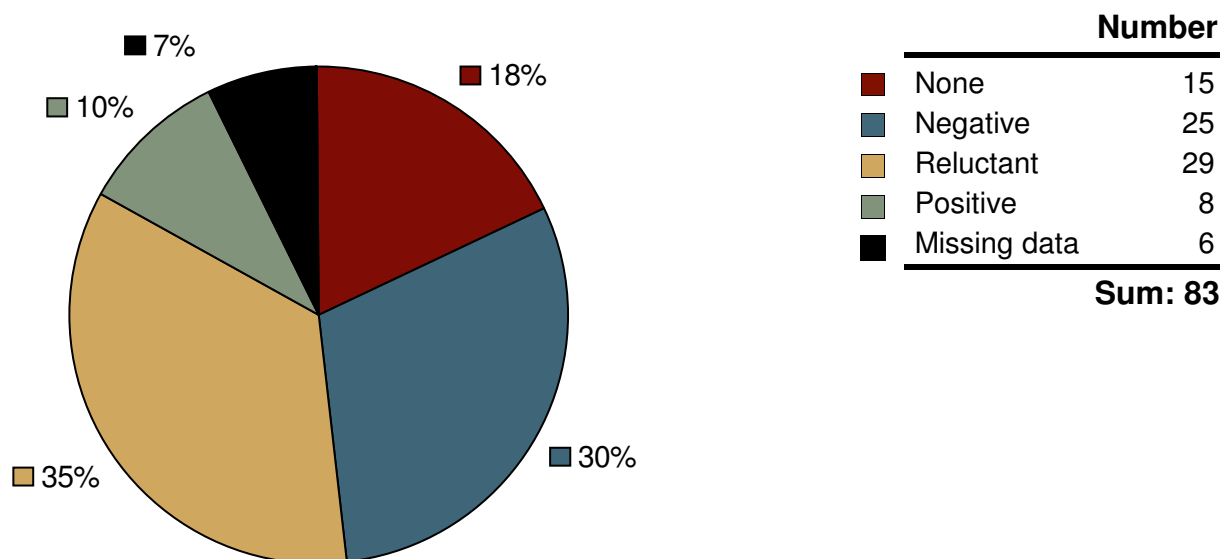
Oral health index (indices)¹

		0	1	2	3	4	5	6	Missing data	N
Calc	Calculus	7	1	0	0	0	0	0	18	26
GI	Gingivitis	4	1	3	0	0	0	0	18	26
Plaq	Coating	3	2	1	1	1	0	0	18	26
Toot	Tooth wear	2	3	5	2				14	26

- 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



Caries

	3-6 years	7-12 years	13-19 years	Adults
deft¹				
Examined	20	17		
Number of individuals with deft=0	20	15		
Mean	0,0	0,5		
Standard deviation	0,0	1,9		
Missing data	8	10		
DMFT²				
Examined		20	11	13
Number of individuals with DMFT=0		19	9	9
Standard deviation		0,2	2,3	3,8
Mean		0,1	0,8	2,3
Missing data		7	3	1

1: Number of carious or filled deciduous teeth

2: Number of carious or filled permanent teeth

Occlusal relationship

	Number
Neutral bite	38
Post normal	21
Pre normal	7
Missing data	17
<hr/>	
	Sum: 83

Maximum jaw opening

Children younger than 10 years

	Number
- 20	0
21 - 30	0
31 - 40	4
41 - 50	1
51 -	0
Missing data	36
<hr/>	
	Sum: 41

Children, 10 years or older, and adults

	Number
- 20	0
21 - 30	0
31 - 40	4
41 - 50	2
51 -	1
Missing data	35
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	Sum: 42

Profile¹

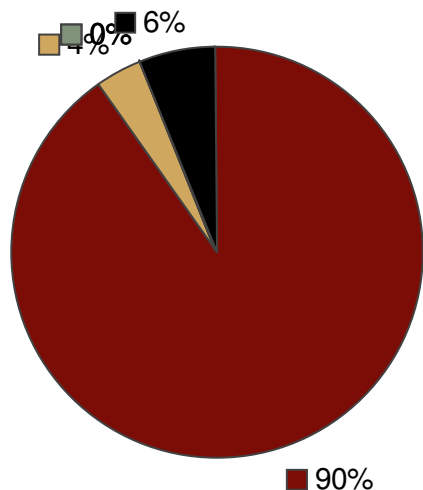
	Number
Normal	24
Convex	0
Concave	0
Missing data	2
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	Sum: 26

Mandibular plane¹

	Number
Normal	24
Increased	0
Reduced	0
Missing data	2
<hr/>	
	Sum: 26

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

Speech difficulty



	Number
■ No speech	75
■ Very incomprehensible	3
■ Incomprehensible speech	0
■ Slightly indistinct speech	0
■ No problems	0
■ Missing data	5
Sum: 83	

Clinical findings	Yes-answers			
	Total N=83 (%)	Boys/Men N=0 (%)	Girls/Women N=83 (%)	Missing data
Impaired tongue motility	56 (81)	0 ()	56 (81)	14
Reduced stability in neck	47 (59)	0 ()	47 (59)	4
Open mouth at rest	46 (58)	0 ()	46 (58)	4
Low muscle tone in lips	43 (54)	0 ()	43 (54)	3
High muscle tone in masticatory muscles	25 (47)	0 ()	25 (47)	30
M mentalis overactive	24 (32)	0 ()	24 (32)	7
Spacing	20 (27)	0 ()	20 (27)	8
High palate	19 (25)	0 ()	19 (25)	8
Narrow palate	17 (23)	0 ()	17 (23)	8
Frontal open bite	15 (20)	0 ()	15 (20)	8
Over crowding	15 (20)	0 ()	15 (20)	7
Low muscle tone in tongue	15 (22)	0 ()	15 (22)	14
Deep bite with gingival contact	8 (11)	0 ()	8 (11)	12
Facial asymmetry	8 (10)	0 ()	8 (10)	0
High muscle tone in lips	6 (8)	0 ()	6 (8)	5
Gingival hyperplasia	4 (6)	0 ()	4 (6)	15
Intra oral hypo-sensitivity	4 (6)	0 ()	4 (6)	17
Macroglossia	3 (4)	0 ()	3 (4)	6