



Orofacial function of persons having Rett syndrome

Report from observation charts



110 observation charts

Synonym

ICD-10 F84.2

Estimated occurrence 10:100,000 girls/women.

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.

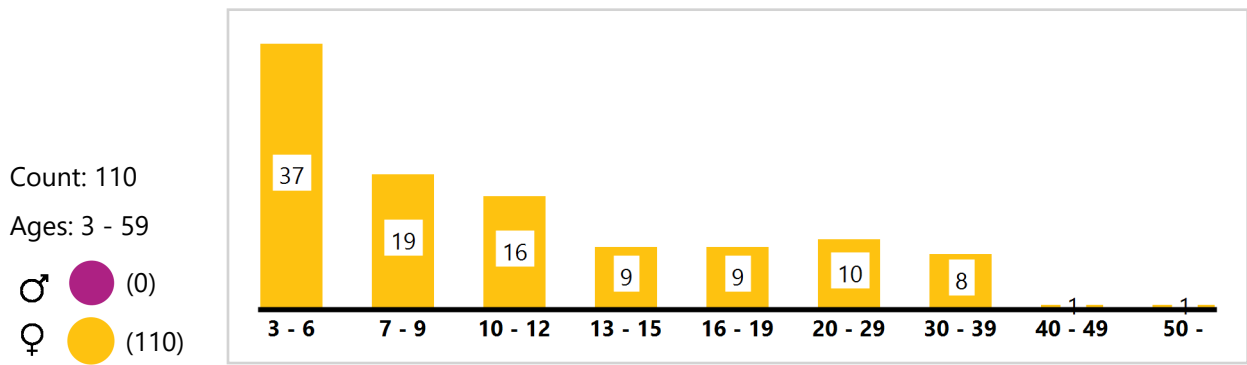
Oral symptoms Severe oral motor impairment resulting in eating problems, drooling, and difficulties with oral hygiene. Most of these girls do not learn to speak. The majority have severe tooth grinding, resulting in tooth wear, and many also have involuntary tongue movements. Overbite and an open bite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

Oral treatment

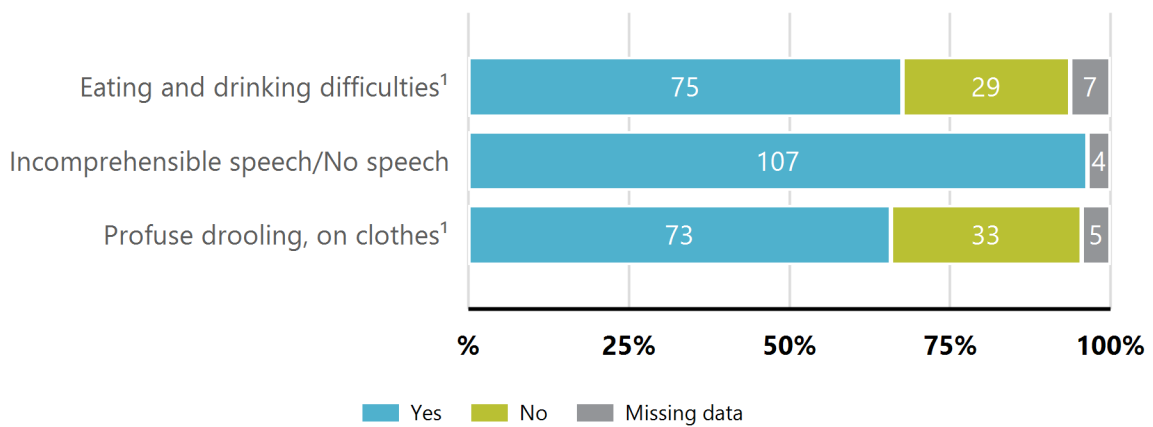
- Early contact with dental services for intensified prophylactic care and oral hygiene information is essential.
- Tooth grinding should be followed up, and be managed with a splint when necessary.
- Regular check-ups of dental and jaw development. Orthodontist should be consulted when needed.
- Feeding 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 MHC database Rare diseases Dokumentation-Ågrenska

Age distribution

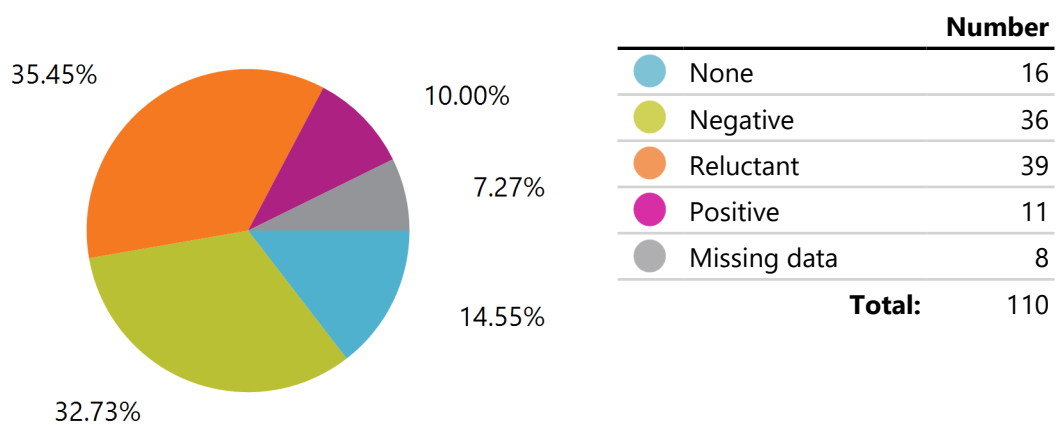


Summary



¹ Reported via questionnaire

Acceptance of dental examination



Oral health

Caries

		3-6 years	7-12 years	13-19 years	>19 years
deft	Examined	37	35		
Number of carious or filled deciduous teeth	Number of individuals with deft=0	37	32		
	Mean	0.0	0.6		
	Standard deviation	0.0	1.9		
	No data	14	18		
DMFT	Examined		35	18	20
Number of carious or filled permanent teeth	Number of individuals with DMFT=0		34	16	16
	Mean		0.1	0.8	1.9
	Standard deviation		0.2	2.3	3.6
	No data		15	6	4

Oral health index (indices)¹

	0	1	2	3	4	5	6	Missing data	Number
Calculus	12	3	1	0	1	1	1	91	56
Gingivitis	9	3	5	1	1	1	0	90	56
Plaque	10	2	2	3	1	0	0	92	56
Tooth wear	6	12	11	4	0	0	0	77	56

CALCULUS

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.

GINGIVITIS

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.

PLAQUE

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.

TOOTH WEAR

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

Occlusal relationship

	Number
Neutral bite	52
Overbite	26
Underbite	7
Missing data	25
Total:	110

Maximum jaw opening

Children younger than 10 years

Children, 10 years or older, and adults

	Number
-20	0
21-30	0
31-40	5
41-50	0
51-	0
Missing data	51
Total:	56

	Number
-20	0
21-30	0
31-40	4
41-50	2
51-	1
Missing data	48
Total:	55

Profile¹

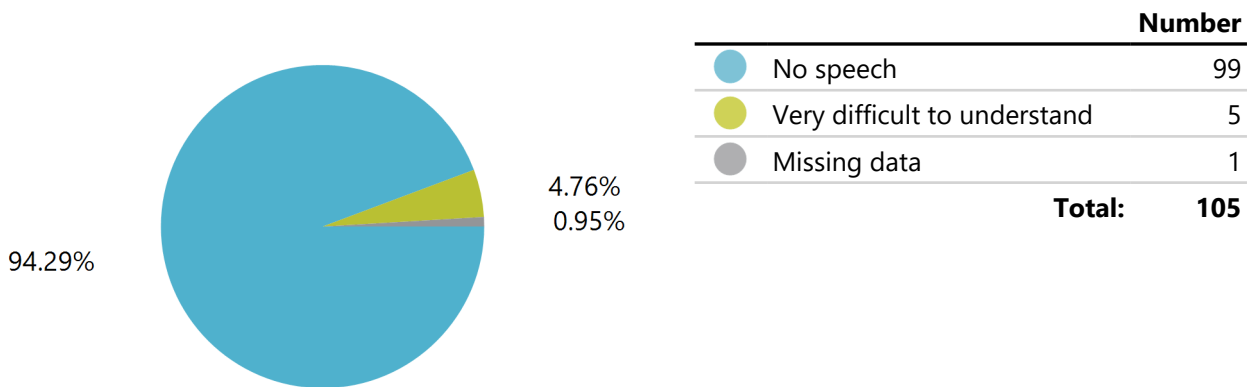
	Number
Normal	34
Convex	6
Concave	2
Missing data	4
Total:	46

Mandibular plane¹

	Number
Normal	44
Increased	1
Reduced	3
Missing data	8
Total:	56

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

Speech problems



Clinical findings

Number of yes-answers

	Total = 110 (%)	♂ = 0 (%)	♀ = 110 (%)	Missing Data
Impaired tongue motility	76 (69)	#Error	76 (69)	17
Open mouth at rest	70 (64)	#Error	70 (64)	5
Low muscle tone in lips	62 (56)	#Error	62 (56)	5
Reduced stability in neck	60 (55)	#Error	60 (55)	7
High palate	38 (35)	#Error	38 (35)	10
Spacing	31 (28)	#Error	31 (28)	13
Narrow palate	24 (22)	#Error	24 (22)	9
Over crowding	23 (21)	#Error	23 (21)	9
Low muscle tone in tongue	17 (15)	#Error	17 (15)	17
Frontal open bite	16 (15)	#Error	16 (15)	13
Deep bite with gingival contact	11 (10)	#Error	11 (10)	21
Facial asymmetry	8 (7)	#Error	8 (7)	2
Low muscle tone in masticatory muscles	6 (5)	#Error	6 (5)	32
High muscle tone in tongue	5 (5)	#Error	5 (5)	21
High muscle tone in lips	4 (4)	#Error	4 (4)	6
Short tongue frenulum	3 (3)	#Error	3 (3)	21
Reduced opening capacity	1 (1)	#Error	1 (1)	38

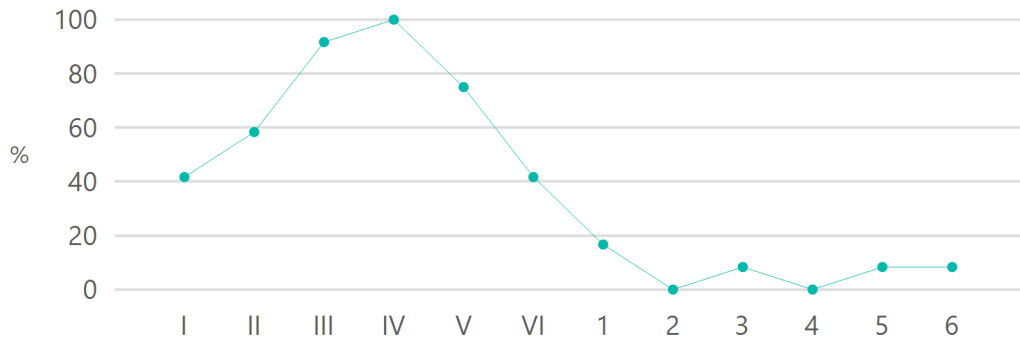
NOT-S

Total mean score:
4.50

Number: 12
Ages: 4 - 35

♂ (0)

♀ (12)



NOT-S interview

		Count	%
I	Sensory function	5	41.7%
II	Breathing	7	58.3%
III	Habits	11	91.7%
IV	Chewing and swallowing	12	100.0%
V	 Drooling	9	75.0%
VI	Dry mouth	5	41.7%

NOT-S examination

		Count	%
1	Face at rest	2	16.7%
2	Nose breathing	0	0.0%
3	Facial expression	1	8.3%
4	Masticatory muscle and jaw function	0	0.0%
5	Oral motor function	1	8.3%
6	Speech	1	8.3%

The report is based on data from the MHC database - the Mun-H-Center database on oral health and orofacial function in rare diseases. Data was collected by dentists and speech-language pathologists using the Orofacial Observation Chart.