



Orofacial function of persons having Congenital muscular dystrophies

Report from observation charts

The survey comprises 18 observation charts.

Subclasses: Congenital muscular dystrophy with laminin $\alpha 2$ deficiency, Ullrich congenital muscular dystrophy, Integrin α deficiency, Walker-Warburg syndrome, Muscle-eye-brain disease, Fukuyama congenital muscular dystrophy, Congenital muscular dystrophy 1C and 1D, LMNA-related congenital muscular dystrophy, Congenital muscular dystrophy with rigid spine.

Estimated occurrence: 3:100 000 live births.

Etiology: Autosomal recessive inheritance. Muscle function is affected by the deficiency or absence of certain proteins.

General symptoms: In congenital muscular dystrophy there is a congenital or early-onset muscle weakness and delayed motor development. Muscle weakness can lead to joint stiffness and curvature of the spine. Breathing is often weakened. The degree of muscle weakness varies greatly between individuals and different muscle groups may be more or less affected depending on the diagnosis. Muscle strength does not deteriorate as a rule, but the strain on the muscles increases as the child grows. Other organs aside from the muscles may also be affected, such as the heart, eyes and brain. Epilepsy is relatively common. Mental retardation and delayed language development may occur.

Orofacial / odontological symptoms: Weakening of the orofacial muscles can affect the growth of the jaws and lead to malocclusion, usually open bite. There is also an increased risk of developing stiffness of the temporomandibular joints, limiting the ability to open the mouth wide. Reduced muscle tone and the impacts on general health often result in sucking difficulties in infants. If muscle disease affects the orofacial muscles there is a risk for eating difficulties, speech difficulties and drooling. Oral motor development is sometimes delayed.

Orofacial/odontological treatment:

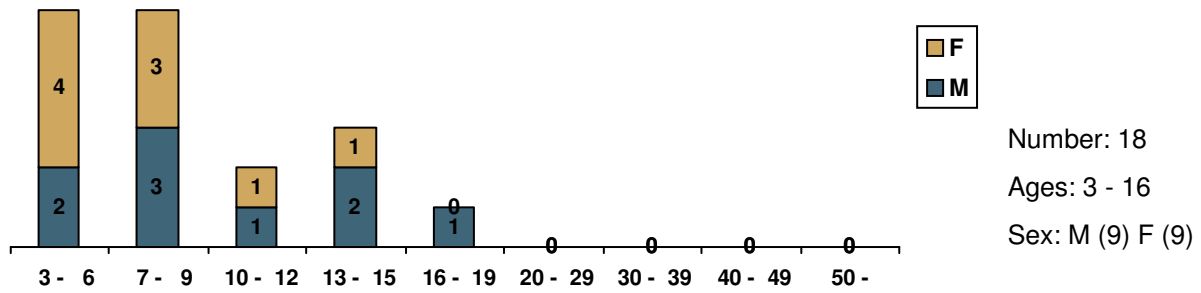
- Early contact with dental services for intensified prophylactic care and oral hygiene information is essential
- Regular check-ups of dental and jaw development. Orthodontist should be consulted when needed.
- When the jaws do not open properly, the function of the jaw joint should be investigated, and appropriate treatment thereafter prescribed.
- Feeding and swallowing difficulties are investigated and treated by a specialist team at the hospital or multidisciplinary treatment centre.
- Speech and language difficulties should be treated by a speech therapist.
- Training in oral motor skills and low intensive exercises in cases of eating disorders and drooling may be necessary.

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.

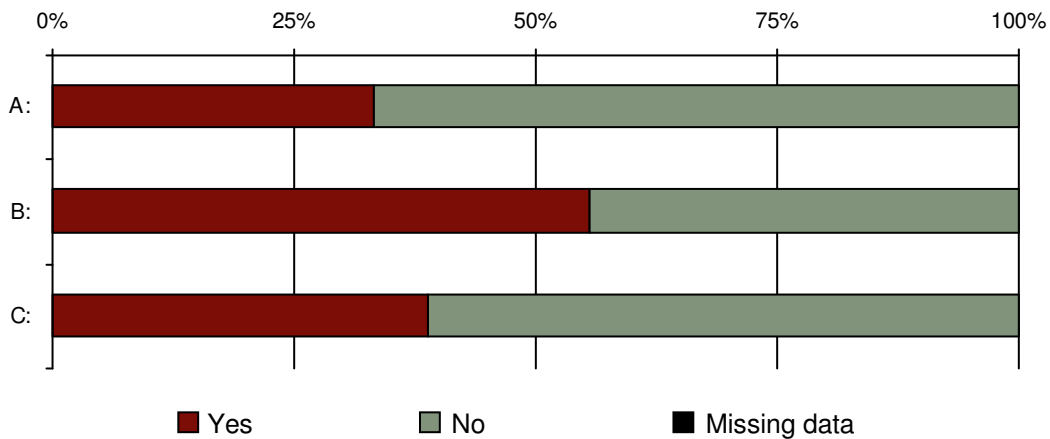


Age distribution



Overview

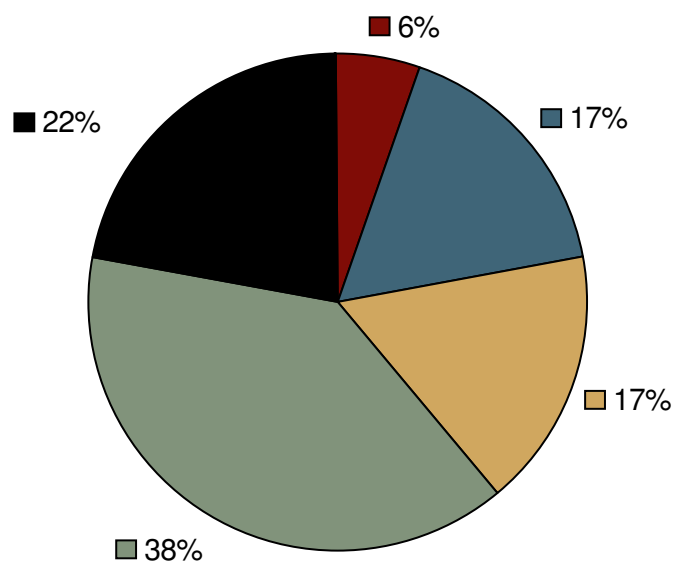
	Yes	No	Missing data	N
A: Incomprehensible speech/No speech	6	12	0	18
B: Eating and drinking difficulties ¹	10	8	0	18
C: Profuse drooling, on clothes ¹	7	11	0	18



Note that the diagram is based upon less than 100 individuals.

¹: Compiled using questionnaire

Acceptance of dental examination



	Number
None	1
Negative	3
Reluctant	3
Positive	7
Missing data	4
Sum:	18

Caries

	3-6 years	7-12 years	13-19 years	Adults
deft¹				
Examined	6	4		
Number of individuals with deft=0	5	3		
Mean	0,5	1,0		
Standard deviation	1,1	1,7		
Missing data	0	4		
DMFT²				
Examined		3	3	0
Number of individuals with DMFT=0		3	3	
Standard deviation		0,0	0,0	
Mean		0,0	0,0	
Missing data		5	1	

1: Number of carious or filled deciduous teeth

2: Number of carious or filled permanent teeth

Occlusal relationship

	Number
Neutral bite	7
Post normal	1
Pre normal	8
Missing data	2
Sum: 18	

Maximum jaw opening

Children younger
than 10 years

	Number
- 20	0
21 - 30	4
31 - 40	3
41 - 50	2
51 -	0
Missing data	3
Sum: 12	

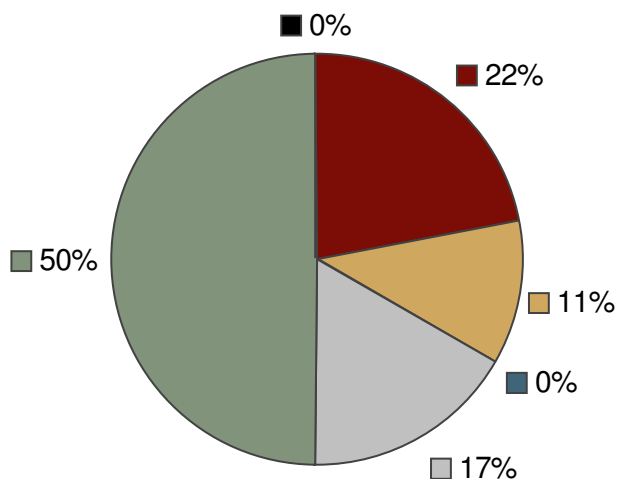
Children, 10 years or
older, and adults

	Number
- 20	0
21 - 30	2
31 - 40	2
41 - 50	1
51 -	0
Missing data	1
Sum: 6	

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



Speech difficulty



	Number
No speech	4
Very incomprehensible	2
Incomprehensible speech	0
Slightly indistinct speech	3
No problems	9
Missing data	0
Sum: 18	

Clinical findings	Yes-answers			
	Total N=18 (%)	Boys/Men N=9 (%)	Girls/Women N=9 (%)	Missing data
Open mouth at rest	14 (78)	6 (67)	8 (89)	0
Low muscle tone in lips	12 (67)	4 (44)	8 (89)	0
Reduced stability in neck	11 (65)	5 (63)	6 (67)	1
Low muscle tone in masticatory muscles	11 (69)	5 (63)	6 (75)	2
Frontal open bite	11 (61)	5 (56)	6 (67)	0
Low muscle tone in tongue	8 (44)	3 (33)	5 (56)	0
High palate	7 (41)	3 (33)	4 (50)	1
M mentalis overactive	6 (35)	4 (44)	2 (25)	1
Reduced opening capacity	5 (36)	2 (29)	3 (43)	4
Narrow palate	4 (25)	2 (22)	2 (29)	2
Over crowding	4 (27)	0 ()	4 (57)	3
Mucous membrane changes	3 (21)	2 (25)	1 (17)	4

Included diagnoses and their distribution

<i>Diagnosis</i>	<i>Number</i>	<i>Sex</i>		<i>Ages</i>	
		<i>M</i>	<i>F</i>	<i>Min</i>	<i>Max</i>
Congenital muscular dystrophy	17	9	8	3	16
Muscle Eye Brain syndrome	1	0	1	13	13