



# Orofacial function of persons having Sanfilippo syndrome

## Report from observation charts

The survey comprises 14 observation charts.

**Synonym:** MPS III (mucopolysaccharide type III)

**Estimated occurrence:** The prevalence is unknown in Sweden; however, it is estimated at 1-2 cases per million inhabitants, i.e. about 10-15 live births.

**Etiology:** The syndrome is caused by a mutation in the genes coding for four different enzymes that are involved in the breakdown of the mucopolysaccharide heparan sulfatase. Normally the breakdown occurs in the cell lysosome; however, in Sanfilippo syndrome, the mucopolysaccharides accumulate in the cells. This results in progressive damage to different bodily tissues and organs. Sanfilippo syndrome is inherited autosomal recessively.

**General symptoms:** In Sanfilippo syndrome, mucopolysaccharides are stored primarily in the nerve system. There are four different enzyme deficiencies that cause Sanfilippo syndrome and hence the syndrome is classified as type A, B, C or D. Type A is considered the most severe form, type B consists of both a milder and more severe form, and type C is considered to lie between types A and B in severity. Type D is very uncommon. The children are born healthy and develop normally up to between 2 and 6 years of age, after which developmental disorders, such as delayed speech and language development, and autistic traits appear. Extreme hyperactivity is common. Thereafter, a progressive deterioration occurs. Gradually mental retardation becomes apparent. In the next phase, balance worsens, as well as the ability to walk and mental capabilities. Epilepsy may occur. Skeletal deformities and stiffness of the joints may occur. Respiratory tract infections are common. Ear infections may cause hearing impairments. Abdominal/bowel problems are common. The accumulation of heparan sulfate inhibits blood coagulation and thus afflicted persons have increased bleeding times.

**Orofacial/odontological symptoms:** Neurologic impairment may affect oral motor function, and speech development. Eating, drinking and swallowing difficulties are common. Poor muscle tonus of the lips and orofacial musculature may cause drooling. Bite, teeth and jaw may be expected to grow and develop normally.

### Orofacial/odontological treatment:

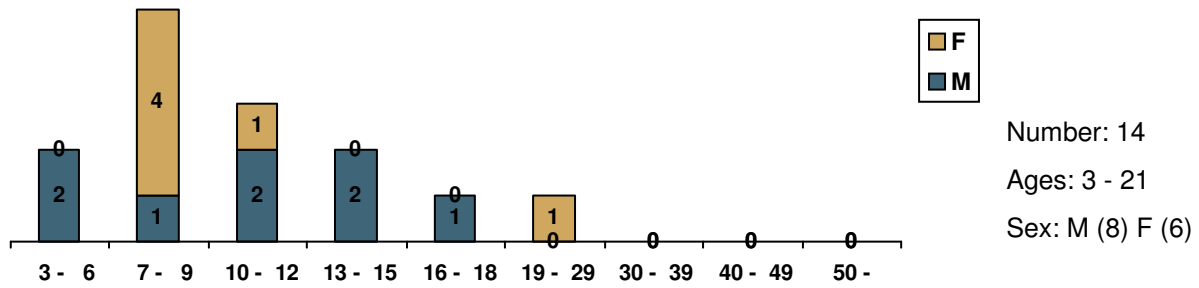
- Early contact with dental services for evaluation and treatment planning is important. Prevention is important in their dental care. These patients require extra prophylactic care and should preferably be treated by the same dental personnel at each visit.
- Risk exists for impaired coagulation and long bleeding times. This risk should be examined and, if needed, be treated prior to impending tooth extraction or operations in the oral cavity.
- Orofacial therapy and oral motor skills training and stimulation in cases of difficulties with eating, speech or drooling may be relevant.
- Speech, language and communication training are often required.
- Feeding and swallowing difficulties are investigated and treated by a specialist team at the hospital or multidisciplinary treatment center.

### Sources:

The rare disease database of the Swedish National Board of Health and Welfare.

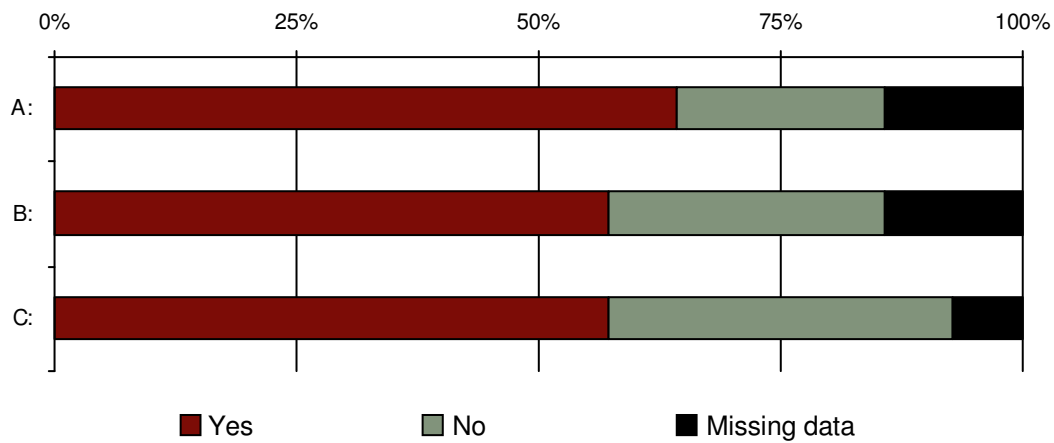
The MHC database - The Mun-H-Center database on oral health and orofacial function in

### Age distribution



### Overview

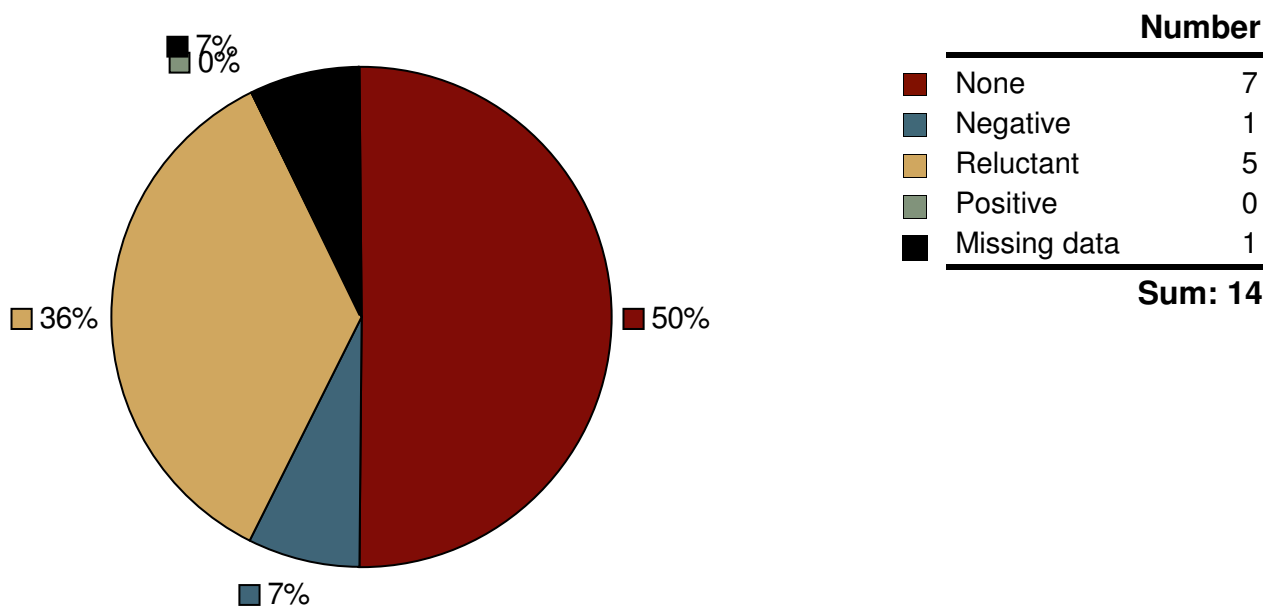
	Yes	No	Missing data	N
A: Incomprehensible speech/No speech	9	3	2	14
B: Eating and drinking difficulties <sup>1</sup>	8	4	2	14
C: Abundant drooling <sup>1</sup>	8	5	1	14



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

<sup>1</sup>: Compiled using questionnaire

## Acceptance of dental examination



## Caries

	3-6 years	7-12 years	13-19 years	Adults
<b>deft<sup>1</sup></b>				
Examined	1	3		
Number of individuals with deft=0	1	3		
Mean	0,0	0,0		
Standard deviation	0,0	0,0		
Missing data	1	5		
<b>DMFT<sup>2</sup></b>				
Examined		5	3	1
Number of individuals with DMFT=0		4	1	0
Standard deviation		0,4	1,2	
Mean		0,2	1,3	
Missing data		3	0	0

1: Number of carious or filled deciduous teeth

2: Number of carious or filled permanent teeth

## Occlusal relationshi

	<u>Number</u>
Neutral bite	6
Post normal	0
Pre normal	2
Missing data	6
	<b>Sum: 14</b>

## Maximum jaw opening

Children younger  
than 10 years

	<u>Number</u>
- 20	0
21 - 30	0
31 - 40	1
41 - 50	0
51 -	0
Missing data	6
	<b>Sum: 7</b>

Children, 10 years or older,  
and adults

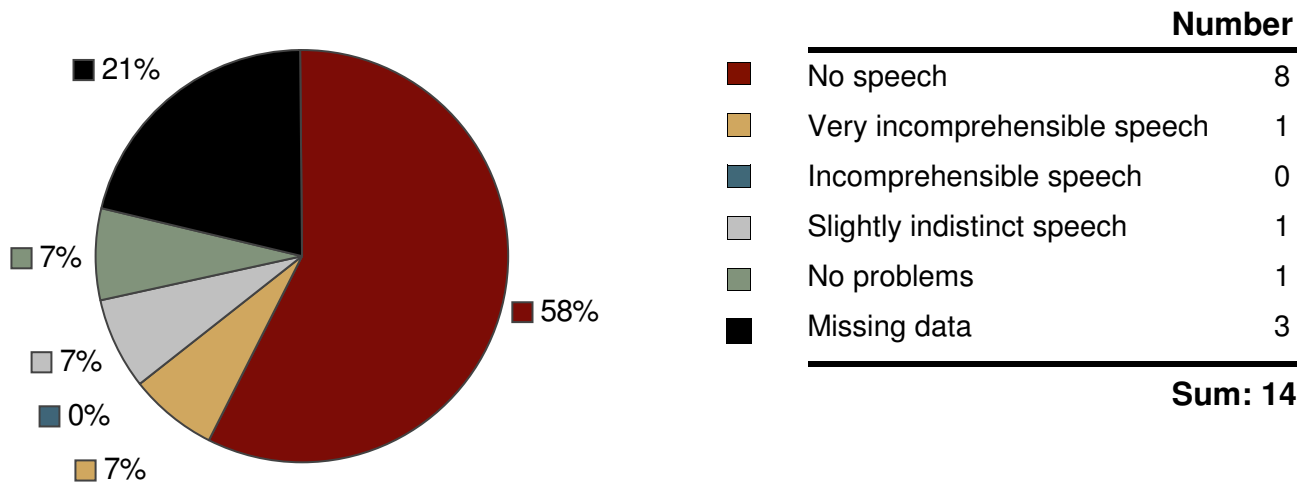
	<u>Number</u>
- 20	0
21 - 30	0
31 - 40	2
41 - 50	1
51 -	1
Missing data	3
	<b>Sum: 7</b>

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

## Orofacial function

### Speech difficulty



Clinical findings	Yes-answers			
	Total N=14 (%)	Boys/Men N=8 (%)	Girls/Women N=6 (%)	Missing data
Low muscle tone in lips	8 (67)	5 (83)	3 (50)	2
Impaired tongue motility	8 (80)	4 (67)	4 (100)	4
Open mouth at rest	7 (64)	4 (80)	3 (50)	3
Low muscle tone in tongue	7 (78)	3 (60)	4 (100)	5
M mentalis overactive	4 (36)	3 (50)	1 (20)	3
Low muscle tone in masticatory muscles	3 (30)	2 (33)	1 (25)	4
Reduced stability in neck	1 (8)	0 (0)	1 (20)	2
Cleft lip and palate	1 (8)	0 (0)	1 (17)	2
Facial palsy	1 (8)	1 (14)	0 (0)	2
High muscle tone in tongue	1 (10)	1 (17)	0 (0)	4
Intra oral hypo-sensitivity	1 (10)	1 (17)	0 (0)	4
Deep bite with gingival contact	1 (11)	0 (0)	1 (25)	5