



Orofacial function of persons having Cystinosis

Report from observation charts



14 observation charts

Synonym

ICD-10 E72.0A

Estimated occurrence 1:160,000 - 200,000 live births.

General symptoms

There are three forms of the condition: Nephropathic cystinosis (also called infantile), intermediate cystinosis (a juvenile form) and the third form, nonnephropathic cystinosis occurs exclusively in adults. The children are born healthy and appear generally normal during the first months. Impaired kidney function eventually leads to severe kidney failure. Excessive urination and thirst commonly lead to evaluation and diagnosis. Kidney damage may result in the need for dialysis or kidney transplantation. The electrolyte balance is affected. The eyes and vision may be affected by storage of cystine crystals in the cornea and retina. Growth disorders are common as well as weak muscles. The function of the thyroid gland may be impaired (hypothyroidism) and puberty may be delayed and/or stop. In non-nephropathic cystinosis the only symptoms are from the eyes. Today, cystinosis may be treated with medications (Cysteamine®, Cystagon). Treatment may also be required in the form of thyroid hormones, growth hormones and sex hormones as well as ensuring that the child receives adequate amounts of fluids, electrolytes and nutrition. Many have very high energy requirements, which means that they need to eat a lot and often.

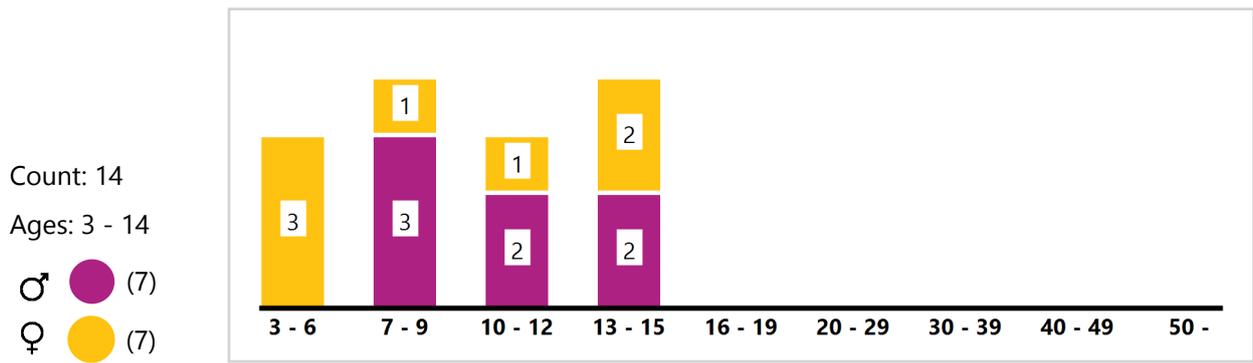
Oral symptoms Sucking, chewing and swallowing difficulties occur. Enteral feeding is common and sometimes necessary to ensure that the child's nutritional requirements are met. Sometimes white spots on the dental enamel can be seen.

Oral treatment

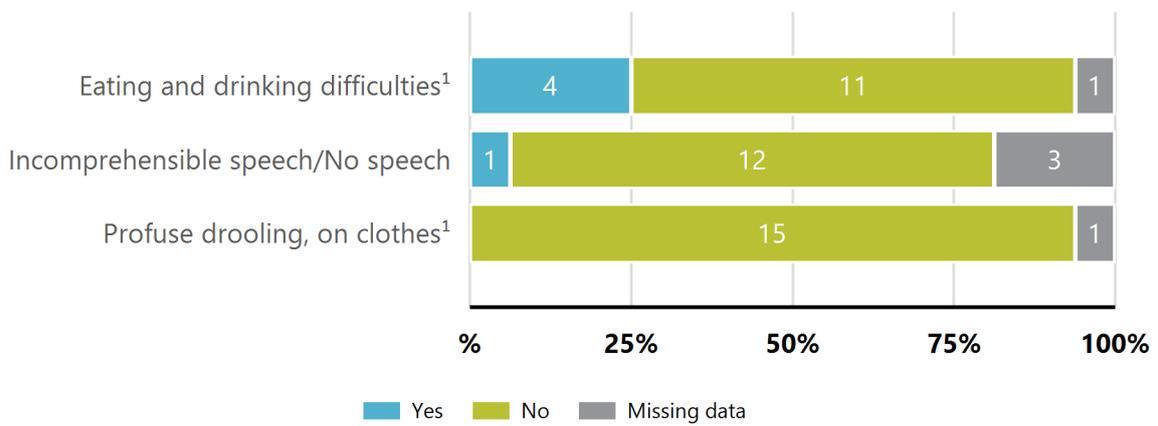
- Early contact with dental services for intensified prophylactic care and oral hygiene information is essential.
- Dental development is checked up by dentist who is familiar with deviating growth pattern and as need arises consult orthodontist.
- Feeding and swallowing difficulties are investigated and treated by a specialist team at the hospital or multidisciplinary treatment center.

Sources The MHC database Rare diseases Dokumentation-Ågrenska

Age distribution



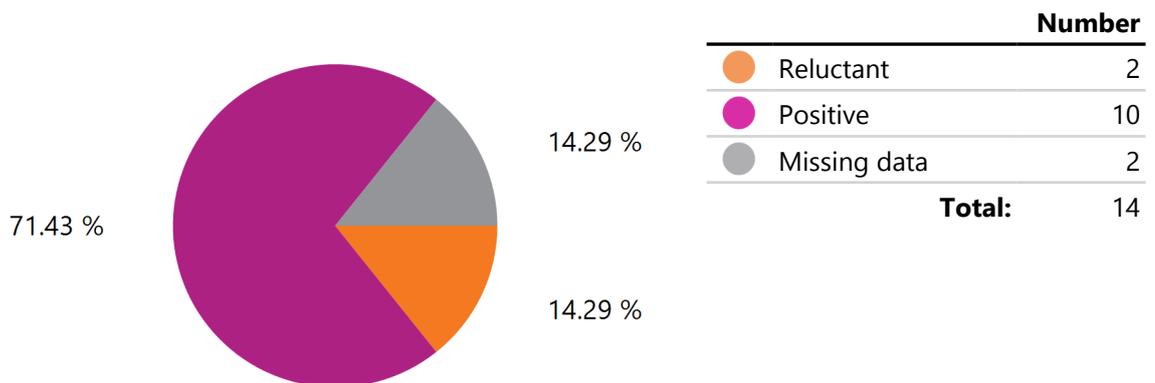
Summary



Certain caution should be observed when interpreting the chart because the number of individuals is less than 100.

¹ Reported via questionnaire

Acceptance of dental examination



Oral health

Caries

		3-6 years	7-12 years	13-19 years
deft	Examined	3	7	
Number of carious or filled deciduous teeth	Number of individuals with deft=0	3	6	
	Mean	0.0	0.3	
	Standard deviation	0.0	0.5	
	No data	0	3	
DMFT	Examined		7	4
Number of carious or filled permanent teeth	Number of individuals with DMFT=0		6	3
	Mean		0.3	1.5
	Standard deviation		0.5	2.1
	No data		3	2

Oral health index (indices)¹

	0	1	2	3	4	5	6	Missing data	Number
Calculus	3	1	0	0	0	0	0	10	5
Gingivitis	3	0	0	1	0	0	0	10	5
Plaque	2	1	0	0	0	0	1	10	5
Tooth wear	2	0	0	2	0	0	0	10	5

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	10
Overbite	0
Underbite	3
Missing data	1
Total:	14

Maximum jaw opening

Children younger than 10 years

Children, 10 years or older, and adults

	Number
-20	0
21-30	0
31-40	1
41-50	3
51-	0
Missing data	3
Total:	7

	Number
-20	0
21-30	0
31-40	2
41-50	3
51-	1
Missing data	1
Total:	7

Profile¹

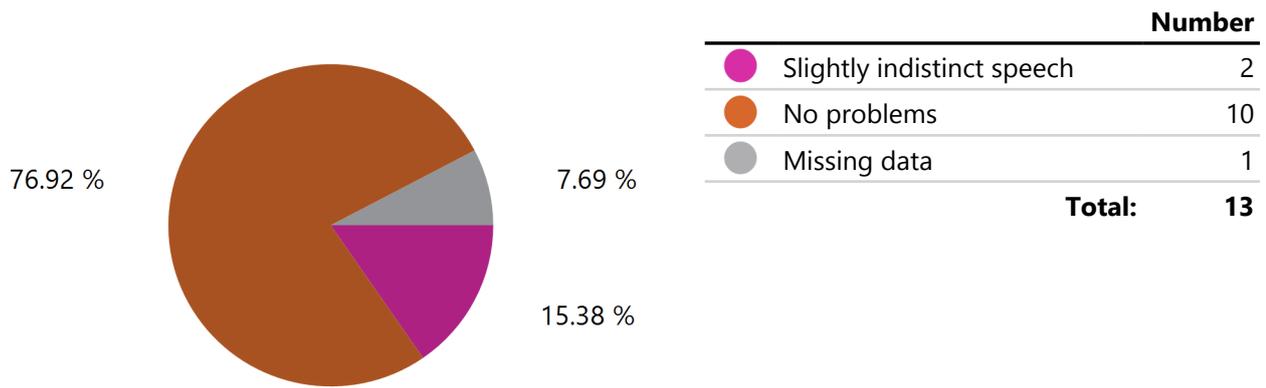
	Number
Normal	3
Convex	0
Concave	0
Missing data	1
Total:	4

Mandibular plane¹

	Number
Normal	3
Increased	1
Reduced	0
Missing data	1
Total:	5

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

Speech problems



Clinical findings

Number of yes-answers

	Total = 14 (%)	♂ = 7 (%)	♀ = 7 (%)	Missing Data
Open mouth at rest	2 (14)	1 (14)	1 (14)	0
Over crowding	2 (14)	2 (29)	0 (0)	1
	1 (7)	1 (14)	0 (0)	0