



Orofacial function of persons having Aicardi syndrome

Report from observation charts



25 observation charts

Synonym

ICD-10 Q04.0

Estimated occurrence Aicardi syndrome only affects females. Very rare.

General symptoms Characteristic symptoms are malformation of the colossal commissure and the retina (retina lacunae) as well as epilepsy. Between the ages of 3 and 6 months these girls develop epileptic seizures of the infantile spasm type. Later, different kinds of seizures commonly occur, and the epilepsy may be life-long. Various brain defects may occur, and give rise to different symptoms. Most individuals with Aicardi syndrome have severe mental retardation and never learn to speak or walk. Impaired vision is common and may be caused by different types of eye defects.

Oral symptoms Eating difficulties, drooling and tooth grinding are common. Poor muscle tonus of the orofacial musculature is also common, and tends to impact on the development of face and jaws. Thus occlusal (bite) anomalies are frequent, for example, post-normal occlusion and a large horizontal overjet. There is an increased risk of tooth cavities (caries) and gingivitis. Epileptic seizures may lead to an increased risk for dental trauma.

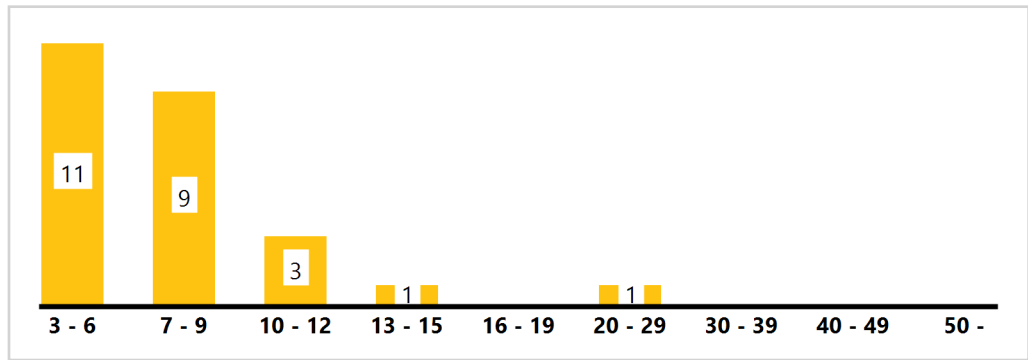
Oral treatment

- It is important that individuals with Aicardi syndrome receive dental preventive treatment, with extra prophylactic care and information on oral hygiene from an early age.
- Tooth grinding should be followed up, and possibly treated with a splint.
- 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 centre.
- Orofacial therapy and oral motor stimulation may be relevant.

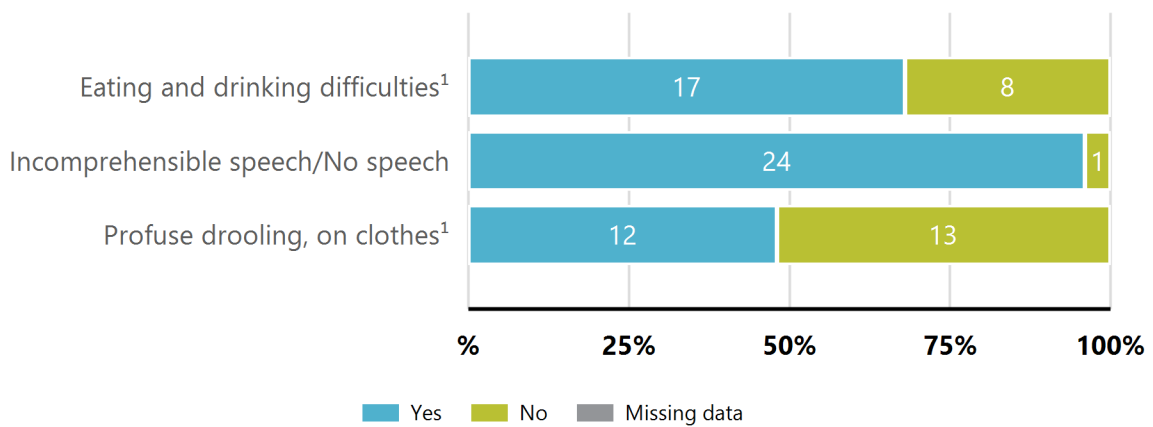
Sources The MHC database Rare diseases Dokumentation-Ågrenska

Age distribution

Count: 25
 Ages: 3 - 27
 ♂ (0)
 ♀ (25)



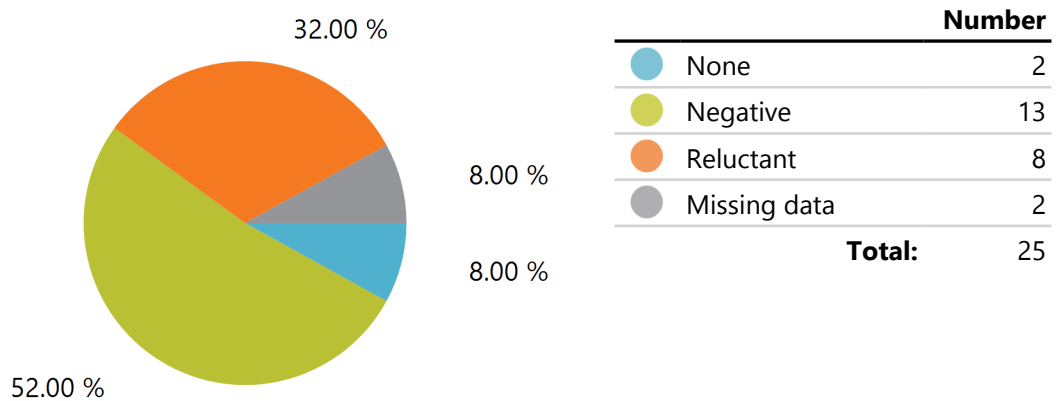
Summary



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	>19 years
deft Number of carious or filled deciduous teeth	Examined	11	12		
	Number of individuals with deft=0	9	11		
	Mean	2.0	0.1		
	Standard deviation	2.8	0.4		
	No data	6	5		
DMFT Number of carious or filled permanent teeth	Examined		12	1	1
	Number of individuals with DMFT=0		12	1	
	Mean		0.0		3.0
	Standard deviation		0.0		
	No data		5	1	0

Oral health index (indices)¹

	0	1	2	3	4	5	6	Missing data	Number
Calculus	1	0	0	0	0	0	0	24	12
Gingivitis	1	0	0	0	0	0	0	24	12
Plaque	1	0	0	0	0	0	0	24	12
Tooth wear	1	0	0	0	0	0	0	24	12

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	10
Underbite	0
Missing data	5
Total:	25

Maximum jaw opening

Children younger than 10 years

Children, 10 years or older, and adults

	Number
-20	0
21-30	0
31-40	3
41-50	0
51-	0
Missing data	17
Total:	20

	Number
-20	0
21-30	0
31-40	1
41-50	0
51-	0
Missing data	4
Total:	5

Profile¹

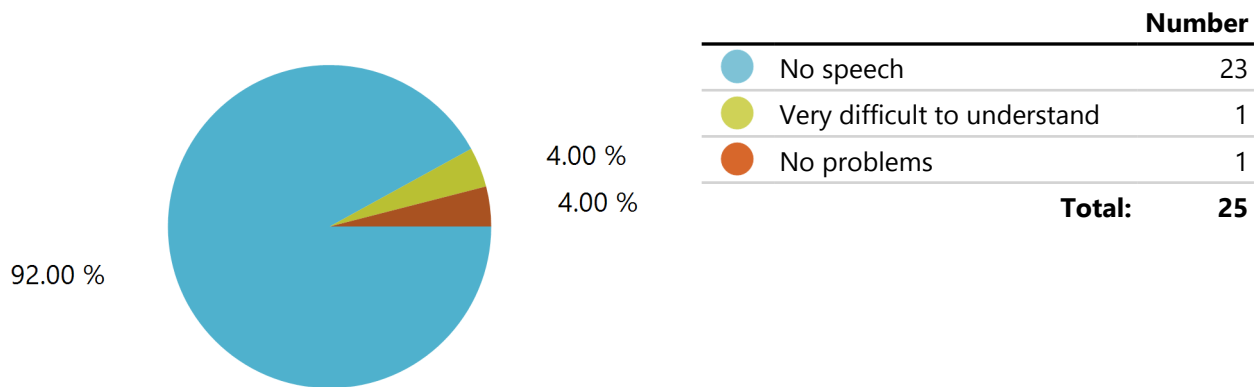
	Number
Normal	9
Convex	1
Concave	0
Missing data	2
Total:	12

Mandibular plane¹

	Number
Normal	6
Increased	0
Reduced	0
Missing data	6
Total:	12

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

Speech problems



Clinical findings

Number of yes-answers

	Total = 25 (%)	♂ = 0 (%)	♀ = 25 (%)	Missing Data
Low muscle tone in lips	22 (88)	#Error	22 (88)	0
Reduced stability in neck	21 (84)	#Error	21 (84)	0
Impaired tongue motility	18 (72)	#Error	18 (72)	3
Open mouth at rest	15 (60)	#Error	15 (60)	1
Low muscle tone in tongue	14 (56)	#Error	14 (56)	4
Facial asymmetry	7 (28)	#Error	7 (28)	1
Spacing	6 (24)	#Error	6 (24)	6
Frontal open bite	5 (20)	#Error	5 (20)	6
Deep bite with gingival contact	5 (20)	#Error	5 (20)	8
High palate	3 (12)	#Error	3 (12)	6
Low muscle tone in masticatory muscles	3 (12)	#Error	3 (12)	15
Over crowding	2 (8)	#Error	2 (8)	6