



Orofacial function of persons having Aicardi syndrome

Report from questionnaires



13 questionnaires

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

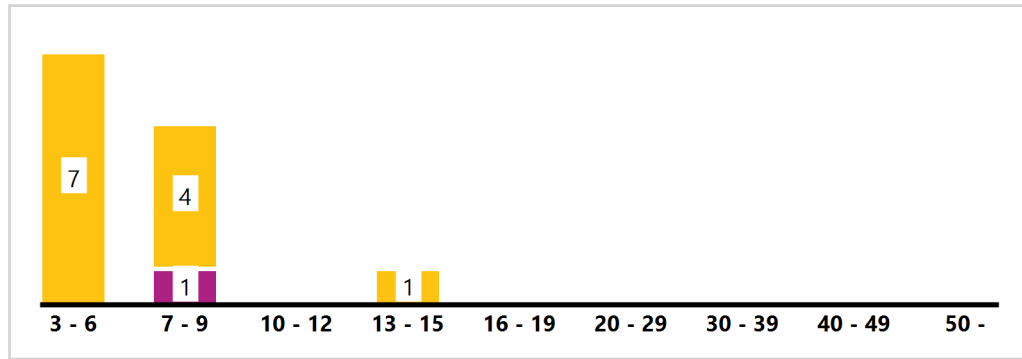
Age distribution

Number: 13

Ages: 3 - 13

♂ (1)

♀ (12)



Additional diagnoses

Medical impairment	Yes	No	Missing data
Inborn heart defect	0	13	0
Other cardiovascular disease	0	13	0
Epilepsy	12	0	1
Asthma	2	10	1
Need of respiratory support	3	9	1
Allergy	1	11	1

Neuropsychiatric diagnosis	Yes	No	Missing data
ADHD/ADD	0	12	1
Autism (Includes autism, Asperger syndrome and autistic traits)	2	9	2

General disability	Yes	No	Missing data
Intellectual disability	0	1	12
Motoric functional impairment	2	0	11
Visual impairment	5	1	7
Hearing loss	1	11	1
Communication difficulties	0	0	13

About dental care and oral health

Do you feel that you receive the dental care you need?	Number
Yes, very much so	10
Yes, somewhat	2
No, not really	1
Total:	13

How many times per year do you normally seek dental care?	Number
Less than once per year	1
One time per year	3
Two times per year	4
Three or more times per year	5
Total:	13

When were your teeth last X-rayed?	Number
During the past two years	4
Never had my teeth X-rayed	8
Missing data	1
Total:	13

Do you look after your teeth in a good way?	Number
Yes, very much so	6
Yes, somewhat	6
Missing data	1
Total:	13

Who brushes your teeth?	Number
Someone else always helps me	13
Total:	13

How often are your teeth brushed?	Number
Once per day	2
Two times per day	9
Three or more times per day	2
Total:	13

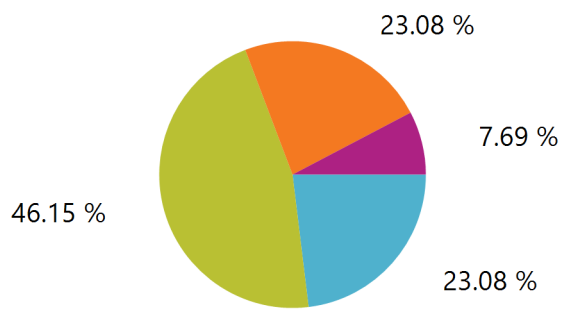
About dental care and oral health

	Yes	No	Missing data
Does your mouth hurt?	1	12	0
Does your mouth feel dry?	1	10	2
Have you ever taken a serious hit to your permanent front teeth?	3	10	0
Do you feel that you have a divergent bite?	5	6	2
Have you had a brace?	0	13	0
Do you feel that you need orthodontics/a brace?	3	9	1

Do you grind or press your teeth at night?	Number
Never	7
Once or twice per week	3
Every night	2
Missing data	1
Total:	13

Do you grind or press your teeth during the day?	Number
Never	7
Once or twice per week	4
Every night	2
Total:	13

About eating



Do you have any problems with eating?

Response	Number
Yes, very much so	3
Yes, somewhat	6
No, not really	3
No, not at all	1
Missing data	0
Total:	13

	Yes	No	Missing data
Do you cough daily in connection with meals?	3	10	0
Do you gag daily in connection with meals?	0	13	0
Do you get acid reflux daily?	0	11	2
Do you throw up often (at least twice per week)?	0	13	0
Do you have a poor appetite?	2	11	0
Does it take a long time before you can swallow a mouthful?	2	10	1
Do you press your tongue forward when you swallow so that food ends up outside the mouth?	3	9	1
Do you find it difficult to chew, i.e. grind food using your molars?	7	6	0
Do you find it difficult to take food from the spoon using your lips?	7	6	0
Have you had problems with food and drink leaking out through the corners of your mouth?	5	7	1
Does food tend to remain in your mouth after meals?	3	9	1
Do you get nutrition in any other way than through your mouth?	6	6	1

About drooling

Do you drool?	Number
Never drool	4
Drool sometimes – not every day	5
Drool often – every day	3
Constant drooling	1
Missing data	0
Total:	13

How much do you drool?	Number
Slight drooling, only on the lips	2
Moderate drooling, on lip and chin	6
Profuse drooling	1
Total:	9

Is your drooling a problem for you?	Number
Yes, somewhat	2
No, not really	3
No, not at all	4
Total:	9

Is your drooling a problem for your family or people around you?	Number
Yes, somewhat	2
No, not really	4
No, not at all	3
Total:	9