



Orofacial function of persons having Joubert syndrome

Report from observation charts

The survey comprises 13 observation charts.

Synonym: Disorders related to Joubert syndrome

Estimated prevalence: 5:100 000 are born with changes in the cerebellum and ataxia

Cause: Changes in one of 13 different (known) genes. The most common is autosomally recessive inheritance.

General symptoms: Children with Joubert syndrome have a deviating breathing pattern in infancy, low muscle tension, unsteady and fitful movement pattern (ataxia) and eye jitters (nystagmus). Visual impairments (retinal dystrophy) and strabismus can be present, and some have drooping eyelids (blepharoptosis). Developmental disabilities and behavioural changes have been described. Changes to the kidneys leading to renal insufficiency can occur.

Orofacial/odontological symptoms: The low muscle tension and breathing difficulties may bring eating difficulties – especially in infancy. The child doesn't manage to suckle and often doesn't swallow correctly. Difficulties in manoeuvring the tongue by will are common, but can't be explained by paralysis or muscle weakness (dyspraxia). Oral motor problems often affect speech and eating abilities and can cause drooling. Speech can be jerky and unclear due to ataxia. Sensitivity to touch in the mouth is common and can make tooth brushing difficult. Other risk factors for oral hygiene are difficulties working for oral/dental care, difficulties eating, as well as reduced own ability to clean the mouth after a meal due to oral motor limitations. Many have a habit of biting fingers and hands. Benign knots in the tongue are related to the diagnosis.

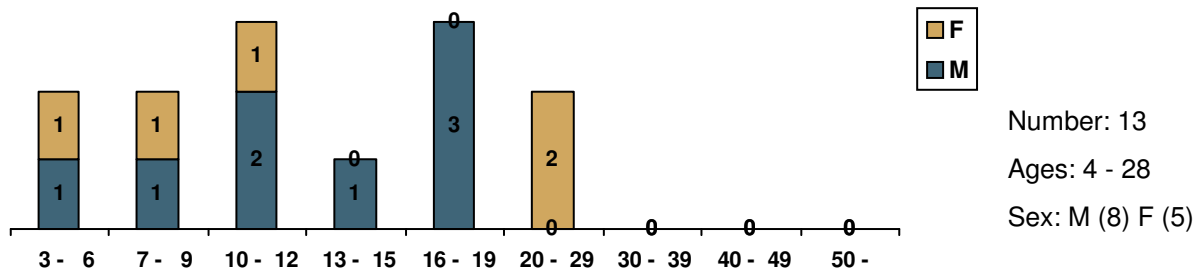
Orofacial/odontological treatment:

- It is important to have contact with a child dental specialist (paedodontist) early on for strengthened preventive care and oral health information.
- Tooth and bite development is to be monitored. If there is any divergence, an orthodontic specialist (orthodontist) is to be consulted early on to plan any orthodontic treatment.
- Oral motor training and stimulation is often urgently needed in cases with eating difficulties, speech difficulties and drooling.
- Speech, language and communication difficulties are evaluated and treated by a speech therapist.

Source

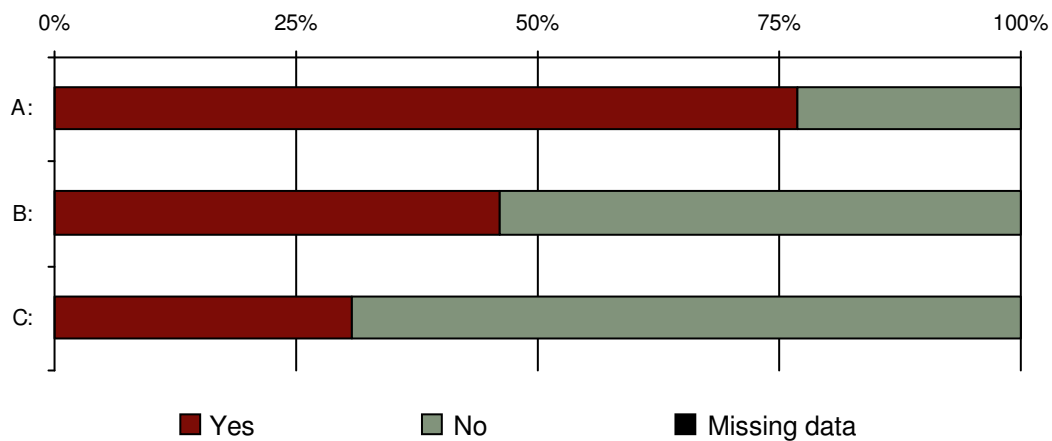
The rare disease database of the Swedish National Board of Health and Welfare.
The MHC database - The Mun-H-Center database on oral health and orofacial function in rare diseases.
The Documentation from the Ågrenska Center.

Age distribution



Overview

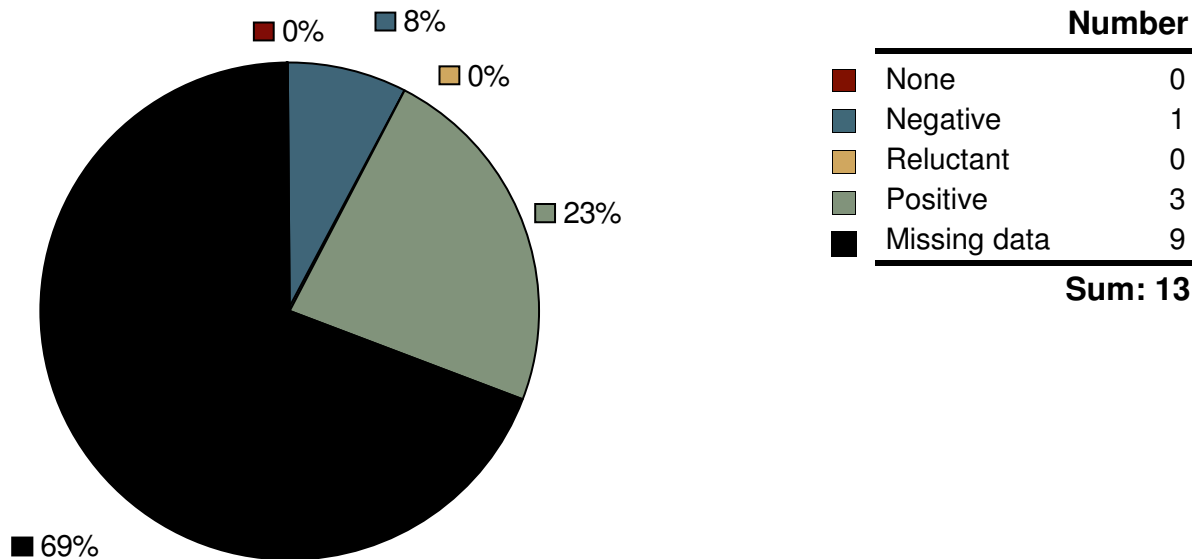
	Yes	No	Missing data	N
A: Incomprehensible speech/No speech	10	3	0	13
B: Eating and drinking difficulties ¹	6	7	0	13
C: Profuse drooling, on clothes ¹	4	9	0	13



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

¹: Compiled using questionnaire

Acceptance of dental examination



Caries

	<u>3-6 years</u>	<u>7-12 years</u>	<u>13-19 years</u>	<u>Adults</u>
deft¹				
Examined	1	2		
Number of individuals with deft=0	1	1		
Mean	0,0	3,0		
Standard deviation	0,0	3,0		
Missing data	1	3		
DMFT²				
Examined		2	3	1
Number of individuals with DMFT=0		2	2	0
Standard deviation		0,0	0,9	0,0
Mean		0,0	0,7	10,0
Missing data		3	1	1

1: Number of carious or filled deciduous teeth

2: Number of carious or filled permanent teeth

Occlusal relationship

	Number
Neutral bite	6
Post normal	2
Pre normal	1
Missing data	4
Sum: 13	

Maximum jaw opening

Children younger
than 10 years

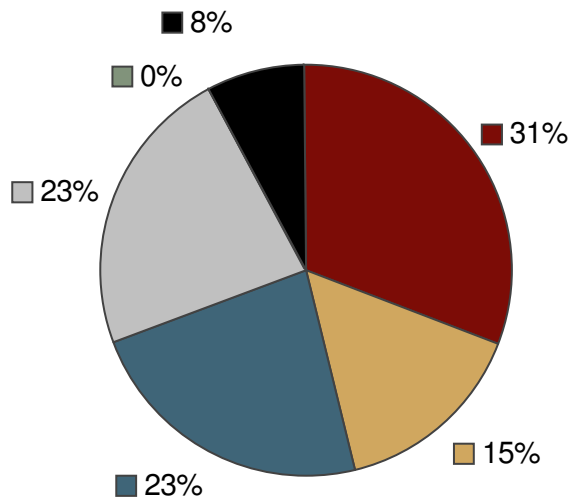
	Number
- 20	0
21 - 30	0
31 - 40	0
41 - 50	0
51 -	1
Missing data	3
Sum: 4	

Children, 10 years or
older, and adults

	Number
- 20	0
21 - 30	1
31 - 40	1
41 - 50	4
51 -	1
Missing data	2
Sum: 9	

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

Speech difficulty



	Number
No speech	4
Very incomprehensible	2
Incomprehensible speech	3
Slightly indistinct speech	3
No problems	0
Missing data	1
Sum: 13	

Clinical findings	Yes-answers			
	Total N=13 (%)	Boys/Men N=8 (%)	Girls/Women N=5 (%)	Missing data
Impaired tongue motility	10 (77)	7 (88)	3 (60)	0
Open mouth at rest	6 (50)	5 (63)	1 (25)	1
Low muscle tone in lips	6 (50)	4 (50)	2 (50)	1
Low muscle tone in tongue	5 (42)	4 (57)	1 (20)	1
Mucous membrane changes	5 (50)	2 (29)	3 (100)	3
Reduced stability in neck	4 (31)	3 (38)	1 (20)	0
Narrow palate	4 (33)	2 (29)	2 (40)	1
M mentalis overactive	3 (25)	3 (38)	0 ()	1
Frontal open bite	3 (30)	1 (20)	2 (40)	3