



Orofacial function of persons having Ektodermal dysplasia

Report from questionnaires

The survey comprises 24 questionnaires.

Sub-groups: There are 186 (July 2013) different types of ectodermal dysplasia (ED), of which hypohidrotic ectodermal dysplasia (HED) is the most common. Of these, the X-linked variety (Christ-Siemens-Touraine syndrome) is the most common, and makes up about 80–90 % of all ED cases.

ICD-10: Q82.4

Estimated prevalence: 1–7 boys:100,000 born (X-linked HED)

Cause: HED is caused by mutations in *EDA* (X-linked HED), *EDAR*, *EDARADD* and *WNT10A*.

General symptoms: The ED diagnosis is made when the development of two out of the four ectodermal structures hair, nails, teeth and sweat glands is abnormal. The lack or reduced function of sweat glands make the natural regulation of body temperature more difficult. The skin is often dry, and the hair is often thin and light in colour, and may be of abnormal structure. Nails can be thin and brittle. Other possible symptoms are dry mucous membranes, dry eyes, and thick, sticky earwax. Low body weight (small amounts of subcutaneous fat) in relation to height is common. Girls/women with X-linked HED have less severe symptoms.

Orofacial/odontological symptoms: A characteristic appearance is associated with the diagnosis. Tooth buds for primary teeth and permanent teeth are completely or partly lacking. The lack of tooth buds results in reduced jawbone growth in the areas where genes are lacking. Milk teeth appear later than normal. The teeth may be of abnormal shape. If many teeth are missing in the upper jaw, the palate is often flat, and the occlusal height low. Dry mouth is common if salivary glands have reduced function or are lacking completely. Dry mucous membranes in the mouth and throat and the lack of teeth can make chewing and swallowing difficult, and can affect speech somewhat. Vomiting and gastrointestinal problems are common in smaller children.

Orofacial/odontological treatment:

- In connection with narcosis, the reduced heat regulation and fragile mucous membranes in the nose, mouth and throat should be taken into account.
- Children suffering from ED should be referred to specialist dental care for follow-up and treatment early on. The treatment plan often requires collaboration between specialists in paediatric dentistry, orthodontics, prosthodontics and oral surgery.
- Teeth should be x-rayed before the child reaches school age.
- It may be appropriate to try out some form of tooth replacement or to rebuild the teeth as early as at the age of three or four.
- The production of saliva should be investigated as early as possible. In cases of dry mouth, dentistry often requires increased levels of care.
- Any speech difficulties are investigated and treated by a speech therapist.

Source

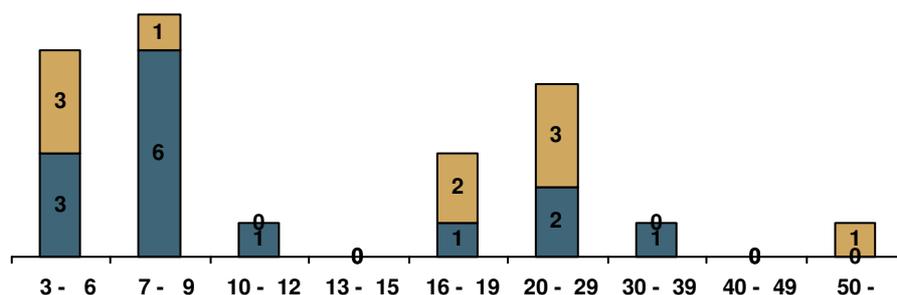
The rare disease database of the Swedish National Board of Health and Welfare.

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

The Documentation from the Ågrenska Center.

Bergendal B. Orofacial manifestations in ectodermal dysplasia-a review. Am J Med Genet A. 2014;164A:2465-2471.

Age distribution



Number: 24

Ages: 3 - 50

Sex: M (14) F (10)

Additional diagnoses

Medical impairment	Yes	No	Missing data
Inborn heart defect	0	24	0
Other cardiovascular disease	0	24	0
Epilepsy	0	24	0
Asthma	4	20	0
Need of respiratory support	0	24	0
Allergy	9	15	0

Neuropsychiatric diagnosis	Yes	No	Missing data
ADHD/ADD	0	24	0
Autistic syndrome	0	24	0
Autistic traits	0	24	0
Asperger's syndrome	0	24	0
Tourette's syndrome	0	24	0

General disability	Yes	No	Missing data
Intellectual disability	0	24	0
Mobility dysfunction	1	23	0
Impaired vision	7	16	1
Impaired hearing	3	20	1
Difficulty in communicating	1	23	0

About dental care and oral health

Do you feel that you receive the dental care you need?

	Number
Yes, very much so	18
Yes, some what	5
No, not really	1
No, not at all	0
Missing data	0
Sum:	24

How many times per year do you normally seek dental care?

	Number
Three or more times per year	14
Twice per year	3
Once per year	4
Less than once per year	3
Missing data	0
Sum:	24

When were your teeth last X-rayed?

	Number
During the past two years	16
More than two years ago	5
Never had my teeth X-rayed	3
Missing data	0
Sum:	24

Do you look after your teeth in a good way?

	Number
Yes, very much so	5
Yes, some what	19
No, not really	0
No, not at all	0
Missing data	0
Sum:	24

Who brushes your teeth?

	Number
I always brush myself	10
Sometimes I brush myself	7
Someone else always helps me	7
Missing data	0
Sum:	24

How often are your teeth brushed?

	Number
Three or more times per day	0
Twice per day	18
Once per day	6
Not every day	0
Missing data	0
Sum:	24

About dental care and oral health

	Yes	No	Missing data
Does your mouth hurt?	5	18	1
Does your mouth feel dry?	9	14	1
Have you ever taken a serious hit to your permanent front teeth?	3	21	0
Do you feel that you have a divergent bite?	8	14	2
Have you had a brace?	8	15	1
Do you feel that you need orthodontics/a brace?	9	12	3

Do you grind or press your teeth at night?

	Number
Never	17
Once or twice per week	5
Every night	1
Missing data	1
Sum:	24

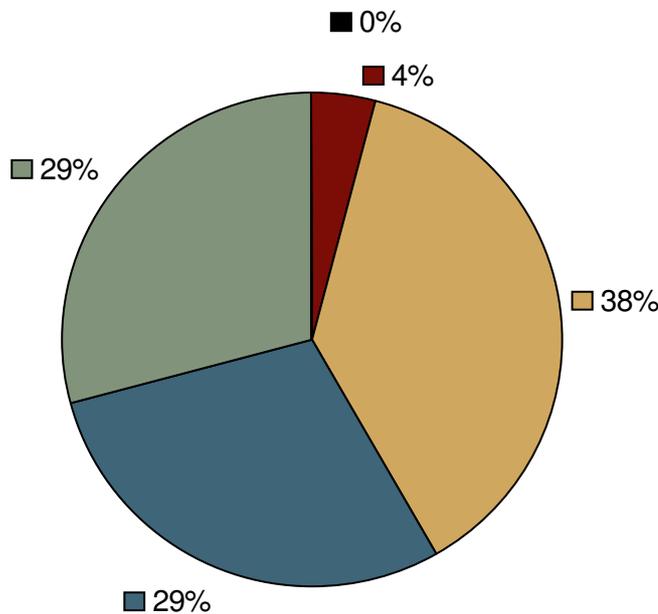
Do you grind or press your teeth during the day?

	Number
Never	19
Once or twice per week	5
Every day	0
Missing data	0
Sum:	24



About eating

Do you have any problems with eating?



	Number
Yes, very much so	1
Yes, some what	9
No, not really	7
No, not at all	7
Missing data	0
Sum:	24

	Yes	No	Missing data
Do you cough daily in connection with meals?	0	23	1
Do you gag daily in connection with meals?	0	23	1
Do you get acid reflux daily?	1	23	0
Do you throw up often (at least twice per week)?	0	23	1
Do you have a poor appetite?	3	20	1
Does it take a long time before you can swallow a mouthful?	6	16	2
Do you press your tongue forward when you swallow so that food ends up outside the mouth?	1	21	2
Do you find it difficult to chew, i.e. grind food using your molars?	9	12	3
Do you find it difficult to take food from the spoon using your lips?	1	20	3
Have you had problems with food and drink leaking out through the corners of your mouth?	0	21	3
Does food tend to remain in your mouth after meals?	4	17	3
Do you get nutrition in any other way than through your mouth?	1	23	0

About drooling

Do you drool?

	Number
Never drool	20
Drool sometimes – not every day	2
Drool often – every day	2
Constant drooling	0
Missing data	0
Sum:	24

How much do you drool?

	Number
Slight drooling, only on the lips	0
Moderate drooling, on lip and chin	4
Profuse drooling, on clothes	0
Very profuse drooling, on hands/objects	0
Missing data	0
Sum:	4

Is your drooling a problem for you?

	Number
Yes, very much so	0
Yes, some what	0
No, not really	2
No, not at all	1
Missing data	1
Sum:	4

Is your drooling a problem for your family or people around you?

	Number
Yes, very much so	0
Yes, some what	1
No, not really	2
No, not at all	1
Missing data	0
Sum:	4