# Orofacial function of persons having
# Ectodermal dysplasia

Report from questionnaires | 28 questionnaires

**Synonym** 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 occurance** 1-7 boys/100,000 born (X-linked HED)

**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.

**Oral 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.

**Oral 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.

**Sources**

The MHC database Rare diseases Dokumentation-Ågrenska Bergendal B. Orodental manifestations in ectodermal dysplasia-a review. Am J Med Genet A. 2014;164A:2465-2471.

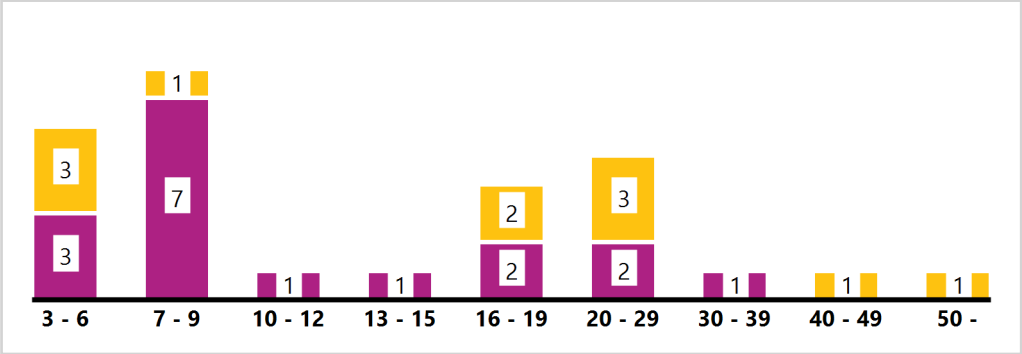
## Age distribution

Number: 28

Ages: 3 - 50

♂ (17)

♀ (11)




## Additional diagnoses

| Medical impairment | Yes | No | Missing data |
|---|---|---|---|
| **Inborn heart defect** | 0 | 28 | 0 |
| **Other cardiovascular disease** | 0 | 28 | 0 |
| **Epilepsy** | 0 | 28 | 0 |
| **Asthma** | 5 | 23 | 0 |
| **Need of respiratory support** | 0 | 28 | 0 |
| **Allergy** | 10 | 18 | 0 |

| Neuropsychiatric diagnosis | Yes | No | Missing data |
|---|---|---|---|
| **ADHD/ADD** | 0 | 28 | 0 |
| **Autism (Includes autism, Asperger syndrome and autistic traits)** | 0 | 25 | 3 |

| General disability | Yes | No | Missing data |
|---|---|---|---|
| **Intellectual disability** | 0 | 28 | 0 |
| **Motoric functional impairment** | 1 | 27 | 0 |
| **Visual impairment** | 7 | 19 | 2 |
| **Hearing loss** | 3 | 24 | 1 |
| **Communication difficulties** | 1 | 27 | 0 |

# About dental care and oral health

| Do you feel that you receive the dental care you need? | Number |
|---|---|
| **Yes, very much so** | 20 |
| **Yes, somewhat** | 6 |
| **No, not really** | 1 |
| **No, not at all** | 1 |
| **Total:** | **28** |

| How many times per year do you normally seek dental care? | Number |
|---|---|
| **Less than once per year** | 4 |
| **One time per year** | 5 |
| **Two times per year** | 3 |
| **Three or more times per year** | 16 |
| **Total:** | **28** |

| When were your teeth last X-rayed? | Number |
|---|---|
| **During the past two years** | 20 |
| **More than two years ago** | 5 |
| **Never had my teeth X-rayed** | 3 |
| **Total:** | **28** |

| Do you look after your teeth in a good way? | Number |
|---|---|
| **Yes, very much so** | 6 |
| **Yes, somewhat** | 22 |
| **Total:** | **28** |

| Who brushes your teeth? | Number |
|---|---|
| **I always brush myself** | 13 |
| **Someone else always helps me** | 7 |
| **Sometimes I brush myself** | 8 |
| **Total:** | **28** |

| How often are your teeth brushed? | Number |
|---|---|
| **Once per day** | 6 |
| **Two times per day** | 21 |
| **Three or more times per day** | 1 |
| **Total:** | **28** |

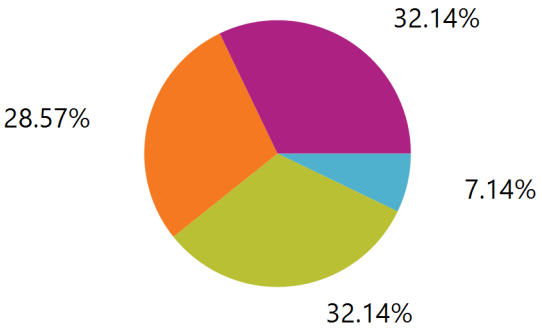
# About dental care and oral health

| | Yes | No | Missing data |
|---|---|---|---|
| **Does your mouth hurt?** | 7 | 20 | 1 |
| **Does your mouth feel dry?** | 10 | 15 | 3 |
| **Have you ever taken a serious hit to your permanent front teeth?** | 5 | 23 | 0 |
| **Do you feel that you have a divergent bite?** | 11 | 15 | 2 |
| **Have you had a brace?** | 11 | 16 | 1 |
| **Do you feel that you need orthodontics/a brace?** | 11 | 14 | 3 |

| Do you grind or press your teeth at night? | Number |
|---|---|
| **Never** | 18 |
| **Once or twice per week** | 6 |
| **Every night** | 3 |
| **Missing data** | 1 |
| **Total:** | **28** |

| Do you grind or press your teeth during the day? | Number |
|---|---|
| **Never** | 21 |
| **Once or twice per week** | 6 |
| **Every night** | 1 |
| **Total:** | **28** |

# About eating




| Do you have any problems with eating? | Number |
|---|---|
| Yes, very much so | 2 |
| Yes, somewhat | 9 |
| No, not really | 8 |
| No, not at all | 9 |
| Missing data | 0 |
| **Total:** | **28** |

| | Yes | No | Missing data |
|---|---|---|---|
| **Do you cough daily in connection with meals?** | 1 | 26 | 1 |
| **Do you gag daily in connection with meals?** | 0 | 27 | 1 |
| **Do you get acid reflux daily?** | 2 | 26 | 0 |
| **Do you throw up often (at least twice per week)?** | 0 | 27 | 1 |
| **Do you have a poor appetite?** | 5 | 22 | 1 |
| **Does it take a long time before you can swallow a mouthful?** | 6 | 17 | 5 |
| **Do you press your tongue forward when you swallow so that food ends up outside the mouth?** | 1 | 22 | 5 |
| **Do you find it difficult to chew, i.e. grind food using your molars?** | 10 | 15 | 3 |
| **Do you find it difficult to take food from the spoon using your lips?** | 1 | 24 | 3 |
| **Have you had problems with food and drink leaking out through the corners of your mouth?** | 0 | 22 | 6 |
| **Does food tend to remain in your mouth after meals?** | 5 | 20 | 3 |
| **Do you get nutrition in any other way than through your mouth?** | 1 | 27 | 0 |

# About drooling

| Do you drool? | Number |
|---|---|
| **Never drool** | 23 |
| **Drool sometimes – not every day** | 2 |
| **Drool often – every day** | 3 |
| **Missing data** | 0 |
| **Total:** | **28** |

| How much do you drool? | Number |
|---|---|
| **Moderate drooling, on lip and chin** | 5 |
| **Total:** | **5** |

| Is your drooling a problem for you? | Number |
|---|---|
| **No, not really** | 3 |
| **No, not at all** | 1 |
| **Missing data** | 1 |
| **Total:** | **5** |

| Is your drooling a problem for your family or people around you? | Number |
|---|---|
| **Yes, somewhat** | 1 |
| **No, not really** | 2 |
| **No, not at all** | 2 |
| **Total:** | **5** |