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## Ectodermal dysplasia

### What is ectodermal dysplasia?

Ectodermal dysplasia is a large group of inherited disorders characterised by a primary defect in hair, teeth, nails or sweat gland function, in addition to another abnormality in a tissue of ectodermal origin, e.g. ears, eyes, lips, mucous membranes of the mouth or nose, central nervous system.

The ectoderm is the outermost layer of cells in embryonic development and contributes to the formation of many parts of the body including all those described above. Ectodermal dysplasia occurs when the ectoderm of certain areas fails to develop normally. All ectodermal dysplasias are present from birth and are non-progressive.

### Classification of ectodermal dysplasia

Currently there are about 150 different types of ectodermal dysplasias. In an attempt to classify these, different subgroups are created according to the presence or absence of the four primary ectodermal dysplasia (ED) defects:

- ED1: Trichodysplasia (hair dysplasia)
- ED2: Dental dysplasia
- ED3: Onychodysplasia (nail dysplasia)
- ED4: Dyshidrosis (sweat gland dysplasia)

Based on the above, the 150 different types of ectodermal dysplasias are categorised into one of the following subgroups made up from the primary ED defects:

- Subgroup 1-2-3-4
- Subgroup 1-2-3
- Subgroup 1-2-4
- Subgroup 1-2
- Subgroup 1-3
- Subgroup 1-4
- Subgroup 2-3-4
- Subgroup 2-3
- Subgroup 2-4
- Subgroup 3
- Subgroup 4

The most common ectodermal dysplasias are hypohidrotic (anhidrotic) ED which falls under subgroup 1-2-3-4 and hydrotic ED which comes under subgroup 1-2-3.

The three most recognised ectodermal dysplasia syndromes fall into the subgroup 1-2-3-4, as they show features from all four of the primary ED defects. They are:

- Ectrodactyly-ED-clefting syndrome
- Rapp-Hodgkin hypohidrotic ED
- Ankyloblepharon, ectodermal defects, cleft lip/palate (AEC) or Hay-Wells syndrome

## What is the cause of ectodermal dysplasia?

The different types of ectodermal dysplasia are caused by the mutation or deletion of certain genes located on different chromosomes. Because ectodermal dysplasias are caused by a genetic defect they may be inherited or passed on down the family line. In some cases, they can occur in people without a family history of the condition, in which case a de novo mutation has occurred.

## What are the signs and symptoms of ectodermal dysplasia?

The signs and symptoms of ectodermal dysplasia differ markedly between the different types of the condition and depend on the structures that are affected. Signs and symptoms are not usually apparent in newborns and may not be picked up till infancy or childhood.

### Anhidrotic ectodermal dysplasia



Ectodermal dysplasia typically affects the four organs primarily involved in the following ways:

Affected organ	Features
Hair	<ul style="list-style-type: none"> <li>Scalp and body hair may be thin, sparse, and light in colour</li> <li>Hair may be coarse, excessively brittle, curly or even twisted</li> </ul>
Nails	<ul style="list-style-type: none"> <li>Fingernails and toenails may be thick, abnormally shaped, discoloured, ridged, slow growing, or brittle</li> <li>Sometimes nails may be absent</li> <li>Cuticles may be prone to infection</li> </ul>
Teeth	<ul style="list-style-type: none"> <li>Abnormal tooth development resulting in missing teeth or growth of teeth that are peg-shaped or pointed</li> <li>Tooth enamel is also defective</li> <li>Dental treatment is necessary and children as young as 2 years may need dentures</li> </ul>
Sweat glands	<ul style="list-style-type: none"> <li>Eccrine sweat glands may be absent or sparse so that sweat glands function abnormally or not at all</li> <li>Without normal sweat production, the body cannot regulate temperature properly</li> <li>Children may experience recurrent high fever that may lead to seizures and neurological problems</li> <li>Overheating is a common problem, particularly in warmer climates</li> </ul>

Other signs and symptoms include:

- Lightly pigmented skin, in some cases red or brown pigment may be present. Skin can be thick over the palms and soles and is prone to cracking, bleeding and infection.
- Skin may be dry and is prone to rashes and infection.
- Dry eyes occur due to lack of tears. Cataracts and visual defects may also occur.
- Abnormal ear development may cause hearing problems.
- Cleft palate/lip.
- Missing fingers or toes (digits).
- Respiratory infections due to lack of normal protective secretions of the mouth and nose.
- Foul smelling nasal discharge from chronic nasal infections.
- Lack of breast development.

## What is the treatment for ectodermal dysplasia?

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There is no specific treatment for ectodermal dysplasia. Management of the condition is by treating the various symptoms. Patients often need to be treated by a team of doctors and dentists, rather than a sole practitioner.

- Patients with abnormal or no sweat gland function should live in cooler climates or in places with air conditioning at home, school and work. Cooling water baths or sprays may be useful in maintaining a normal body temperature.
- Artificial tears can be used to prevent damage to the cornea in patients with defective tear production. Saline sprays can also be helpful.
- Saline irrigation of the nasal mucosa may help to remove purulent debris and prevent infection.
- Early dental evaluation and intervention is essential.
- Surgical procedures such as repairing a cleft palate may lessen facial deformities and improve speech.
- Wigs may be worn to improve the appearance of patients with little or no hair.

Most people with ectodermal dysplasia can lead a full and productive life once they understand how to manage their condition. Special attention must be paid to children if sweating and mucous production abnormalities are present. Recurrent high fevers may lead to seizures and neurological problems.

### Related information

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#### References:

- Book: Textbook of Dermatology. Ed Rook A, Wilkinson DS, Ebling FJB, Champion RH, Burton JL. Fourth edition. Blackwell Scientific Publications.

#### On DermNet NZ:

#### Other websites:

- [Ectodermal Dysplasia](#) – emedicine dermatology, the online textbook
- [National Foundation for Ectodermal Dysplasia](#)
- [Ectodermal Dysplasia Society](#)

#### Books about skin diseases:

See the [DermNet NZ bookstore](#)

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If you have any concerns with your skin or its treatment, see a [dermatologist](#) for advice.

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