ECTODERMAL DYSPLASIA FACT SHEET

What is Ectodermal Dysplasia?

Ectodermal Dysplasias comprise a diverse group of genetic disorders characterised by defects in the development or function of various ectodermal tissues, including hair, nails, teeth, skin, and glands. These disorders can affect additional parts of the body, such as the eyes, throat, skin, ears, and more. The term "ectoderm" refers to the earliest cells in a developing embryo that eventually give rise to teeth, hair, nails, and sweat glands, among other structures. The term "dysplasia" indicates a deviation from the typical pattern of growth. Therefore, Ectodermal Dysplasia is a descriptive term denoting structural changes in body parts developed from the ectoderm.

With over 180 different types of Ectodermal Dysplasias identified, each syndrome presents a unique combination of symptoms, ranging from mild to severe. The key features of these disorders must exhibit abnormal functioning for a condition to be classified as an Ectodermal Dysplasia. The specific abnormalities and affected areas vary among the different types of syndromes. Ectodermal Dysplasia is a result of a genetic mutation passed from parent to child. In some cases, the genetic mutation occurs spontaneously in the affected person.

Ectodermal Dysplasia Symptoms

Each type of Ectodermal Dysplasia usually involves a different combination of symptoms, which can range from mild to severe, such as:

- Absence or abnormality of hair growth.
- Absence or malformation of some or all teeth.
- Impairment in the development of many glands, especially sweat glands, but also salivary glands (make saliva), lacrimal glands (make tears), mucous glands and the breasts.
- Lack of the ability to sweat causing overheating.
- Too little production of tears and other protective secretions of the eyes. This can make them sensitive and even painful.
- Reduced production of mucus in the airways, that leads to chest infections and – in those exposed too often to smoke or dust – to chronic lung damage (emphysema).
- Impairment or loss of hearing.
- Nasal blockage due to a build-up of secretions.
- Frequent infections due to immune system deficiencies and, in some cases, the inability to keep bacteria from entering the body through cracked or eroded skin.
- Less effective barrier properties of the skin, airways and gut leading to infections and to allergies (e.g. asthma, eczema and hay-fever).
- Respiratory problems: not only asthma and chest infections but also, in those who smoke or are exposed to dust, a severe form of chronic chest disease (may be diagnosed as emphysema or as pneumoconiosis).
- Absence or malformation of some fingers or toes.
- Cleft lip and/or palate.
- Irregular skin pigmentation.

In addition to the above individuals affected by Ectodermal Dysplasia may have:

- Sensitivity to light.
- A lack of breast development.
- Psychological challenges due to changes in physical appearance.

Individuals affected by Ectodermal Dysplasia may face a lifetime of special needs. These can include:

- Dentures at a young age with frequent adjustments and replacements.
- Osseo integrated dental implants.
- Special diets to meet dental/nutritional needs.
- Air-conditioned environments.
- Wigs to conceal the lack of hair and scalp conditions.
- Creams or devices to protect from direct sunlight.
- Respiratory therapies for asthma and infections.

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