What is Ectodermal Dysplasia?

Ectodermal Dysplasia (ED) is not a single disorder, but a group of closely related conditions of which more than 150 different syndromes have been identified. The Ectodermal Dysplasias (EDs) are genetic disorders affecting the development or function of the teeth, hair, nails and sweat glands. Depending on the particular syndrome ED can also affect the skin, the lens or retina of the eye, parts of the inner ear, the development of fingers and toes, the nerves and other parts of the body.

Each syndrome 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
- Inability to perspire, which causes overheating
- Impairment or loss of hearing or vision
- Frequent infections due to immune system deficiencies or, in some cases, the inability of cracked or eroded skin to keep out disease-causing bacteria
- Absence or malformation of some fingers or toes
- Cleft lip and/or palate
- Irregular skin pigmentation.

In addition to the above they may have:

- Sensitivity to light
- Respiratory problems
- A lack of breast development
- A host of other challenges

Individuals affected by ED face a lifetime of special needs which may include:

- Dentures at a young age with frequent adjustments and replacements
- Special diets to meet dental/nutritional needs
- Air conditioned environments
- Wigs to conceal hair and scalp conditions
- Carrier identification testing
- Protective devices from direct sunlight
- Osseointegrated dental implants
- Respiratory therapies

The inheritance patterns are variable according to the specific type of ED. Patterns include spontaneous mutations, autosomal dominant, autosomal recessive, X-linked dominant and X-linked recessive. When questions of a diagnosis exist, the expertise of a geneticist or other doctor with experience with the EDs is strongly recommended.

It is important to remember that not all individuals affected by the EDs will have physical features that fit the description of a specific syndrome. There may be a great deal of variation in the physical appearance of the same type of ED from one affected person to the next. It is also conceivable for a person to have a type of ED that has not been described yet. Nonetheless, the EDs share certain features, an understanding of which makes it possible to appreciate the ramifications for most affected individuals and allows everyone involved to respond appropriately to the individual's needs.