A Family Guide

to the Ectodermal Dysplasias

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NATIONAL FOUNDATION FOR
ECTODERMAL DYSPLASIAS

OUR MISSION

Our mission is to empower and connect people touched by ectodermal dysplasias through education, support and research. For more information, visit us online at www.nfed.org.

DEDICATION

This booklet is dedicated to three past members of the National Foundation for Ectodermal Dysplasias Scientific Advisory Council.

L. Stefan Levin, DDS, MSD, whose wisdom and humor enriched our lives.

Nancy Esterly, MD, who continues to set an unparalleled example of excellence.

Virginia P. Sybert, MD, who is the quintessential pediatric dermatologist.

1st Revision 2000, 2nd Revision 2002,
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5th Revision 2015

This book was authored by the National Foundation for Ectodermal Dysplasias Scientific Advisory Board.

INTRODUCTION

The ectodermal dysplasias are a group of inherited disorders that involve defects of the hair, nails, teeth, and sweat glands. Other parts of the body may be affected also. The many ectodermal dysplasias are recognized by the combinations of physical features that an affected person has and the way(s) in which they are inherited.

SPECIAL NOTE

Information on management is suggested in this booklet is meant to be a guide to what might be expected or recommended by medical professionals who are consulted by our families. The reader should understand that these statements are generalizations about the many and diverse ectodermal dysplasias, and therefore no absolute recommendations are made or implied. Trust the clinicians whom you consult to do the right things for each individual, but also be an advocate for yourself or your child. Your medical professional may appreciate a copy of this booklet to increase their awareness of the ectodermal dysplasias.

The mention of any brand name drugs in this booklet is presented as example only, intended as guides to groups or classes of products, is not either inclusive or restrictive, and must not be interpreted as endorsement of a specific product or brand or company by the National Foundation for Ectodermal Dysplasias (NFED) or its Scientific Advisory Council.

MORE INFORMATION

Medical and dental treatment for people affected by ectodermal dysplasias may be quite complicated. Dental treatment is particularly important since it is critical for a normal diet, facial appearance, speech, and emotional development. This booklet provides helpful information for anyone involved in decisions about dental care for people affected by ectodermal dysplasias.

For more information on care associated with the ectodermal dysplasias, please visit our website (www.nfed.org) or contact us at 618-566-2020.

DEFINITION OF ECTODERMAL DYSPLASIA

The ectodermal dysplasias are a group of genetic (gene-based) disorders in which there are abnormalities of the structures that come from the ectoderm.

What Is The Ectoderm?

Before a developing fetus is large enough to be seen, a layer of cells covers the outside of the body. This layer of cells is called the ectoderm. Typically, the hair, nails, teeth, and/or sweat glands derive from this layer of the developing embryo and thus any of these derived cells and tissues may be affected in the ectodermal dysplasias. In some cases, problems also may be noted with parts of the eye, ear, or other organs and body features which develop from the ectoderm.

What Does Dysplasia Mean?

Dysplasia means literally “abnormal tissue growth”, that is, that a part of the body is not formed well. Thus, there is an inherent abnormality in the way a particular part of the body is put together. A single part of the body may be abnormal or more than one part may be abnormal simultaneously. If a disorder consistently involves more than one part of the body, it is called a syndrome.

Combinations of Ectodermal Abnormalities

By convention, the ectodermal dysplasias are really syndromes.

While there may be abnormalities (dysplasias) of the hair alone or teeth alone, for example, these are not usually considered to be ectodermal dysplasias because only one structure is affected. A disorder is considered to be an ectodermal dysplasia only when more than one ectodermal structure is affected.

- Pure ectodermal dysplasias involve abnormalities of structures derived from the ectoderm only.

- Complex ectodermal dysplasias involve abnormalities of other structures that are formed by other cell layers, such as the bone.

- Finally, ectodermal structures may be involved in a secondary way in some syndromes, such as missing teeth in people with cleft lip and palate. The latter disorders are not really EDs but are closely related disorders. The reason for emphasizing how the ectodermal dysplasias are classified is so that everyone understands how terminology is used and diagnoses are made.

Throughout this booklet, the term ectodermal dysplasias is applied to pure ectodermal dysplasias and complex ectodermal dysplasias. There are more than 180 ectodermal dysplasias, each one recognized and named for the specific combination of abnormalities shown in affected individuals. The pattern of these features is important when a physician tries to make a formal diagnosis.

In some types, teeth may be missing and nails may be defective, while in another type there may be missing teeth, inability to sweat, and sparse hair. Still another may have hearing loss and defects of the enamel of the teeth. Each combination of abnormalities may be a distinct ectodermal dysplasias and therefore has a unique name (often derived from the name of the person who first described it accurately), and is inherited in specific way(s).

WHAT ARE GENETIC FACTORS?

The body is composed of billions of cells. Each cell has within it a small central structure called the nucleus. Within the nucleus are chromosomes, which are small thread-like strands of hereditary material (DNA). The chromosomes, in turn, are composed of genes. It is easy to understand the concept of genes by thinking of a cell as a jewelry box and the nucleus as one of the drawers in the box. The chromosomes, then, can be considered as necklaces in the drawer, and the genes as the individual beads on each necklace.

Ectodermal dysplasias are caused by alterations in genes. Altered genes may be inherited from a parent, or normal genes may become altered (mutate) at the time of egg or sperm formation or after fertilization. It is important to remember that a person cannot choose or modify the genes that he or she has, and that conception and events of pregnancy by and large do not change the genes. Thus, parents who have a child affected by an ectodermal dysplasia should not think that they did anything to cause the condition and cannot blame themselves or take credit for its occurrence.

The chances for parents to have affected children depend on the inheritance pattern of the type of ectodermal dysplasia that affects the family. The diagram on the next page highlights these chances but cannot explain them well enough to cover all cases. A family affected by an ectodermal dysplasia should consult with a geneticist or a genetic counselor for a better understanding of the specific ectodermal dysplasia and for information about the spectrum of the specific condition and the probabilities or risks that the ectodermal dysplasia will occur in any future pregnancy.

NEW MUTATION

Generally, when a mutation has occurred, there is little chance that it will occur in another child of the same parents.

The affected child may transmit the gene, however.

AUTOSOMAL DOMINANT

When the ectoderemal dysplasia is an autosomal dominant trait, the parent who is affected has a single copy of the abnormal gene and may pass it on to his or her children.

Regardless of the gender of the parents or the child, there is a 50% chance for each child to receive the abnormal gene. All children who receive it will be affected.

AUTOSOMAL RECESSIVE

When there is an autosomal recessive trait, each parent is unaffected.

The parents are said to be carriers. They each have a single copy of the abnormal gene and the chance for them to have an affected child is 1 in 4.

1/4 get the abnormal gene from both and are affected. 2/4 get 1 copy each & are carriers. 1/4 inherits a normal gene from each and is not affected.

X-LINKED RECESSIVE

If a woman is a carrier of an X-linked recessive disorder, there is a 50% chance each male child will receive the abnormal gene and be affected and a 50% chance that each female will be a carrier (like the mother).

If a man has the abnormal gene, he is affected and will pass the gene on to all his daughters. The daughters will be carriers. Since the gene is on the X chromosome, sons will not be affected because they receive the man's Y chromosome.

HOW SKIN CAN BE AFFECTED BY ECTODERMAL DYSPLASIAS

The skin may be pale and typically appears thin in some types of ectodermal dysplasias. The surface blood vessels may be prominent. Individuals affected by ectodermal dysplasia may be prone to sunburn but can tolerate outdoor activities with routine use of commonly available sun protection products. There is often darkening around the eyes or on the elbows, palms, and soles. The skin around the eyes may be lax, and that on the palms and soles may be thick.

Eczema flare-ups are common in certain types of ectodermal dysplasia.

The skin may be dry, scaly, and easily irritated, particularly in the winter. Oil and sweat glands may be poorly developed or absent. Often, the skin of a newborn is like that of a "post-mature" baby with more than usual redness and peeling. This clue can lead to early diagnosis of ectodermal dysplasia. Diaper rashes may be frequent and persisting through infancy. Some individuals are prone to eczema.

Protect Infants under the age of six months with clothing. Skin care in older children and adults should include application of sunscreen with outdoor activities. Generally, sunscreens with a moisturizing cream or lotion base are better tolerated than those with alcohol based gels or sprays. The most effective products are labeled "highly water-resistant" and provide "broad spectrum" including UVA and UVB as well as an SPF of at least 30, and UVA and UVB protection. Look for zinc oxide or titanium dioxide as the active ingredient. Reapply the sunscreen every two hours after swimming or if exposure is for prolonged periods. Consider these examples of sunscreens: Banana Boat Kids Sunblock Spray Lotion SPF 48, BlueLizard Baby, Elta Block, Neutorgena UVA/ UVB Sunblock Lotion and Ombrelle Sunscreen Lotion Extreme.

Daily or every other day bathing will help moisten the skin to prevent overgrowth of germs. Cleansing products should be used sparingly and limited to areas that need cleaning (e.g., diaper area and skin folds). Gentle products include superfatted bar soaps such as Basis, Dove, Oilatum, or liquid cleansers such as Aguunil or Cetaphil Gentle Cleansing Lotion or Neutrogena Non Drying Lotion.

Immediately after bathing, gently pat the skin and cover with a moisturizer. In general, an ointment is more effective and more soothing than a cream and a cream more effective and soothing than a lotion. However, children affected by ectodermal dysplasias may not tolerate ointments and prefer less occlusive creams or lotions, especially during summer months.

Avoid heavily fragranced or colored moisturizers. The safest and most effective moisturizers are plain petroleum jelly (Vaseline) or mineral oil. Other moisturizers that work well for some individuals are Aqua Glycolic Hand and Body Lotion, Aquaphor, Cetaphil, Complex 15, Curel, Eucerin, Fougera Hydrophilic Ointment, Lubriderm, Nutraderm and Purpose.

Most diaper rashes improve with discontinuation of irritating products, including diaper wipes. If redness and scaling are mild, treatment may include changing diapers frequently and applying a protective barrier of generic zinc oxide ointment during each diaper change. Avoid commercial diaper wipes on the rash; substitute a soft cloth or paper towel or cotton squares moistened with water or mineral oil. If this treatment is ineffective, a physician should evaluate and treat the rash.

Most people with ectodermal dysplasia do not experience frequent skin infections. However, in one condition affecting some young children affected by Ankloblepharon Ectodermal Dysplasia - Clefting (AEC), the scalp may become red, weepy, crusty and somewhat swollen. This eruption requires care by a physician in order to relieve discomfort, prevent secondary infection and minimize scarring.

HOW SWEATING CAN BE AFFECTED BY ECTODERMAL DYSPLASIAS

Diminished or absent sweating is a common problem in some ectodermal dysplasias. The sweat glands may be absent, reduced in number, or may not function normally. Reduced sweating may result in very high fevers because the body regulates its temperature by sweating. Often, the first clue that the sweat glands are absent or are not functioning normally is an elevated temperature.

Swimming is often a favorite cooling activity for individuals who don't perspire.

Elevations in body temperature are often caused by high environmental temperatures, excessive physical activity, or heavy clothing. When the body temperature is elevated, the skin feels dry or hot and may be flushed or pale.

Prevention is the best treatment for elevated body temperature. This is especially important for parents to remember since infants and small children cannot complain about the heat. Because of the climate in some parts of the world, the home, school, or place of employment should ideally be air-conditioned. Individuals affected by ectodermal dysplasias easily learn the extent of heat that they can tolerate.

Individuals affected by ectodermal dysplasias may participate in almost any activity including most sports. But, individuals must take precautions in hot environments if their body temperature may be elevated by strenuous activity. Such precautions include wearing wet T-shirts, drinking extra fluids, having immediate access to a cool environment, or using a cooling device.

When body temperature is elevated, a bath or a sponge bath with lukewarm water helps to reduce the fever. Drinking fluids also helps. A persistent high fever, however, always should be evaluated by a physician, since it may be caused by an infection or may cause dehydration. You must tell the physician about the ectodermal dysplasia.

HOW HAIR CAN BE AFFECTED BY ECTODERMAL DYSPLASIAS

Hair in ectodermal dysplasias can be sparse and thin.

The scalp hair may be absent, sparse, fine, lightly pigmented, or abnormal in texture. The hair may also be fragile and unruly, sticking out in all directions and difficult to comb. The hair is dry because the oil glands are absent or poorly developed.

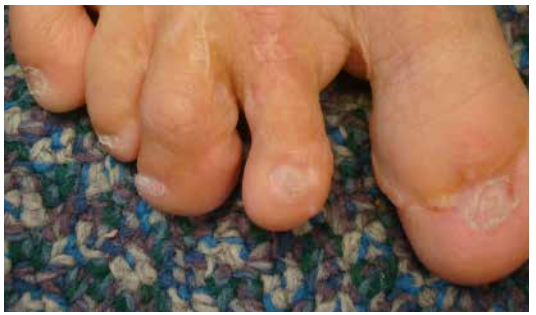
Some defects of the hair are obvious at birth, while others may not be noted until later in life. Hair growth in some types of ectodermal dysplasia is slow and haircuts are not needed as often as in unaffected family members. After puberty, hair growth improves for some individuals. Early male pattern baldness is common in some ectodermal dysplasias.

The eyebrows, eyelashes, and other body hair may also be absent or sparse, but beard growth in males is usually normal.

There are no treatments that can change the structure of the hair. Care is directed toward preventing damage to the hair shafts. Gentle shampoos, such as Neutogena or Purpose, are good for washing hair. Mild dandruff shampoos, containing selenium sulfide (e.g., Sebulex), salicylic acid (e.g., T-Sal), or pyrithione zinc (e.g., Head & Shoulders) may help excessive scaling of the scalp. In some people, however, these shampoos may increase the dryness. Protein-coating shampoos such as Daragen may give extra body to the hair shafts, and cream rinses, such as DHS Cream Rinse or Ionil Cream Rinse, may increase manageability. A gentle body wave permanent may be tried but cannot be tolerated by everyone.

Careful cutting and styling of the hair improves appearance, but if the hair is sparse, a well-fitting, age- appropriate wig may be used. Remember that wigs do not look natural until they are thinned and styled. Information about where such wigs may be purchased is available from dermatologists or the NFED.

HOW NAILS CAN BE AFFECTED BY ECTODERMAL DYSPLASIAS



Fingernails can have dark patches and be slowing growing. Toenails can also be affected.

The nails in some types of ectodermal dysplasia may be poorly developed, small, thick or thin, brittle, discolored, cracked, abnormally curved, or ridged. In addition, they may grow slowly or shed periodically and may develop light spots, lines, or patches. The nails and surrounding cuticle area may become infected by fungus, yeast or bacteria. If this occurs, nails may become thick or discolored, and the area may develop a bad odor, or become swollen and tender.

Like the hair, nails are made up of dead protein. Medications will treat secondary infections but will not correct the inborn nail defects. Lubricants such as petroleum jelly or lanolin can be massaged into the nails to reduce some of the dryness, brittleness, or cracking. Keep the nails short and trimmed smoothly. File thick nails can be filed with the fine pumice used by beauticians for pedicures. Pumice can be purchased from most beauty supply companies. Thick, crumbly nails may indicate an infection and should be examined by a physician. This is especially true if the skin around the nail is swollen, red, tender, or if it drains a yellow colored material. Synthetic nails (sculptured nails) are available. Not everyone can use these products, because they may cause further damage to nails or

allergic reactions. If sculptured nails are considered, a technician experienced in the proper technique to apply them should be consulted.

HOW TEETH CAN BE AFFECTED BY ECTODERMAL DYSPLASIAS

Teeth may be widely spaced a part and need bonding to correct conical shape.

Some or all teeth may be delayed in eruption or fail to erupt. Teeth that do erupt may be widely spaced, tapered or malformed. In individuals with some ectodermal dysplasias, the enamel (outer layer of the teeth) is defective and there may be an excessive number of cavities. When teeth are missing, the jawbones in which they normally are held do not develop well. This may lead to an "old-age" appearance of the face. Some females who are carriers for the types of ectodermal dysplasias that affect males much more severely (the genetically X-linked types) may have small jaws and front teeth that are narrower than normal.

Partial or even complete dentures can be made to enhance function and replace missing teeth. Success with such dentures depends on cooperation. But, children as young as two years of age have used them successfully, particularly if the parents and other relatives properly motivate the child is properly motivated. A dentist with a caring attitude and interest in working with children is essential. During a child's periods of rapid growth, dentures must be replaced periodically.

The psychological and physical advantages of having teeth are considerable. Orthodontics and bridgework are useful when the teeth are widely spaced or malformed. If the enamel layer is defective, a variety of protective and restorative procedures is available. Whatever the treatment, make an effort to minimize dental decay and save as many natural teeth as possible. Initiate and maintain scrupulously an attentive program of decay prevention (regular check-ups, cleaning, X-rays, sealants, and fluoride treatment).

In some forms of ectodermal dysplasia, the saliva production (a.k.a., spit) may be reduced leading to dry- mouth (xerostomia) which may put the individual at increased risk for tooth decay. It is recommended to have the dentist carefully check the teeth and apply preventive treatments (e.g., fluoride varnish) as needed. Extra care in maintaining oral hygiene is critical, even if the affected individual has only primary or baby teeth. It is important to maintain these teeth as long as possible to allow for normal speech, appearance and chewing function. These teeth also maintain what bone is formed upon the formation of these primary teeth.

We convened a panel of dental professionals with expertise in treating the complex oral health needs of individuals affected by ectodermal dysplasia to develop a consensus on guidelines for appropriate oral care for these individuals. The professionals wrote *The Parameters of Oral Health Care for Individuals Affected by Ectodermal Dysplasias*, a resource for clinicians and families as to age appropriate care. The guidelines document the medical aspects of the oral health manifestations of individuals affected by ectodermal dysplasia and the appropriate prophylactic and management plans. Medical and dental professionals and families can receive a copy of this document by contacting the NFED.

HOW THE EARS, NOSE, AND THROAT CAN BE AFFECTED BY ECTODERMAL DYSPLASIA

The underproduction of body fluids in some types of ectodermal dysplasia leads to several problems. Saliva may be sparse, causing problems with tasting, lubricating, chewing, and swallowing foods. The mucous secretions of the nose may be excessively thick, forming a crusty mass. These secretions are common and are usually accompanied by a foul odor. A hoarse, raspy voice is also common. Respiratory infections may be common. Ear wax may become impacted. Hearing loss may occur either from wax impaction or damage to the hearing nerve.

Individuals with some types of ectodermal dysplasias may experience hearing loss and need to wear hearing aids.

A deficiency of saliva can be counteracted with large quantities of fluids during and between meals. Saliva substitutes, such as Moistir, Salivart, and Xero-lube, may also be beneficial. Humidification (central and room humidifiers) may help to prevent nasal crusting. Saline nasal drops may be used as often as necessary. You can make these solutions can be homemade (one teaspoon of salt in one quart of water) or commercial products, such as Alkolal, Ayr, and Ocean are available. If a crusty mass in the nose persists, you may attempt gentle mechanical removal. Humidification often helps the respiratory problems.

Individuals with abnormal speech should undergo a thorough speech and hearing assessment. This is frequently available through the local school system. In addition, speech therapy may be needed to assure normal language development.

HOW EYES CAN BE AFFECTED BY ECTODERMAL DYSPLASIAS

In most individuals affected by ectodermal dysplasia, eye and vision problems are no different than those in the general population. any eye problems relate to the decreased tear supply or to blepharitis (smoldering infections in the oil glands of the eye lashes). However, an ophthalmologist (a physician with specific training in disorders of vision and the eye) should perform a complete medical eye examination if vision appears to be abnormal. Cataracts, corneal scarring, infections, and retinal changes may cause visual problems in some of the ectodermal dysplasias. An ophthalmologist must evaluate, diagnose, and treat these problems.

Dry eye can make eyes sensitive to light.

Tearing, especially the natural tears present in normal daily living (called “basal tear production”) may be reduced, leading to dryness and irritation of the eyes and thus to sensitivity to sunlight. In some uncommon ectodermal dysplasias, cloudy corneas or cataracts may be part of the syndrome.

Individuals with reduced tearing can use artificial tears, such as BionTears, Liquifilm, Optive, Refresh, which can be purchased at drug stores without a prescription. They may be used as frequently as needed to resolve the complaints of dryness or irritation. Some of these are available in formulations and packaging without preservatives andmay “sting” less, but are substantially more expensive than the versions with preservatives because of the costs of special packaging. If parents suspect that a child has either cloudy corneas or cataracts, they should consult an ophthalmologist promptly for evaluation.

HOW ECTODERMAL DYSPLASIAS CAN AFFECT GROWTH

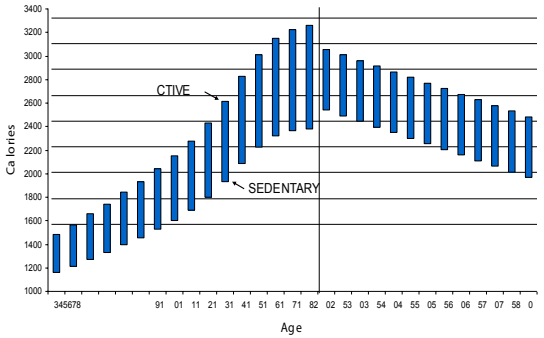
Individuals with Goltz syndrome tend to be smaller in stature.

Growth abnormalities are common in children with ectodermal dysplasia. Individuals affected by some types of ectodermal dysplasia are shorter than average early in life. Other problems in some types of ectodermal dysplasias include a prominent forehead, flat or depressed bridge of the nose, and breast development in females.

Weight deficits are present at an early age and persist throughout adolescence. Height deficits are seen primarily in children with ectodermal dysplasias other than HED. Parents should monitor their child's height and weight gain each time they visit the pediatrician's office. The pediatrician generally plots the child's height and weight measurements on standardized growth charts prepared by the National Center for Health Statistics. These growth charts serve as the barometer for assessing the health, growth patterns, and nutritional status of all children, including those with ectodermal dysplasias.

Good nutrition is essential to maintain normal growth in children affected by ectodermal dysplasia. Although chewing difficulties may impair their eating capabilities, cooking or blenderizing can modify food textures to facilitate better nutrition. The use of dentures at an early age also promotes good nutritional health. Adequate oral intake of dietary energy (carbohydrates and fats), protein, vitamins, and minerals is essential for normal growth. The best way for these children to develop good eating habits is to follow the recommendations of the U.S. Department of Agriculture's (USDA) food guidance system, "ChooseMyPlate", for children (http://www. choosemyplate.gov).

ChooseMyPlate.gov provides practical information on formulating healthier diets. It features messages on balancing calories and changing the types of food consumed (figure above). A well-balanced diet for children affected by ectodermal dysplasias includes daily servings from all the food groups in amounts sufficient to meet their energy and nutrient needs for normal growth.



The best way to assess the adequacy of the child's diet is to determine food portion sizes based on his or her energy needs. A child's daily energy need is estimated from his or her age and level of physical activity. Once energy needs have been determined, the child's pattern of food consumption and serving sizes can be derived using the USDA website.

For example, a 10-year-old boy who is physically active for 30 to 60 minutes daily has an energy need of approximately 1800 kcal per day. Based on this information, he should eat approximately 6 oz of grains, 2.5 cups of vegetables, 1.5 cups of fruits, 3 cups

of milk, and 5 oz of meat or beans. Children with ectodermal dysplasias who have growth deficits need to consume larger food portion sizes and may use more discretionary foods to restore height and weight deficits. If an affected child's appetite or weight gain is persistently poor, seek advice from a physician. Additional diagnostic studies and therapeutic interventions may be warranted.

Women's Concerns

Results from a women's study funded by the NFED has also shown that there are specific women's issues associated with ectodermal dysplasias.

HED females reported:

- Congenital absence of breast (14%)
- Congenital absence (3.4%) or hypoplasia (30%) of nipple
- Absent, sparse or thin pubic, axillary and body hair (62%)
- Absent or decreased sweating (85%)
- Infant requiring fever evaluation in the newborn period (11%)
- Chronically painful intercourse (40%)
- Infertility (26%) – 20% in the general population

ALLERGIES AND ECTODERMAL DYSPLASIAS

Atopy (allergic reactions) and primary immunodeficiencies may occur more frequently in children with ectodermal dysplasias. They have higher rates of asthma, rhinitis symptoms, and eczema compared with unaffected children. The prevalence of physician-diagnosed food allergies and primary immunodeficiencies also exceed known rates in the general pediatric population. A combination of genetic and environmental factors in ectodermal dysplasias may contribute to breaches of skin and mucosal barriers, permitting enhanced transmission and sensitization to irritants, allergens, and pathogens.

THE PSYCHOSOCIAL IMPACT OF ECTODERMAL DYSPLASIA

Individuals affected by ectodermal dysplasias have a wide range of mental abilities, as one would expect in the general population. Mental retardation is not a feature of most ectodermal dysplasias. Nonetheless, they may see themselves or their peers may see them as less intelligent or different because of their physical appearance. The earlier sections on teeth, hair, and skin describe things that may help this perceptual issue.

There is little published information about emotional adjustment or development. Ectodermal dysplasia may cause stress for the entire family. While such stress is an extra burden, it can also be a source for emotional growth for everyone concerned. Parents can help their children to maximize their adjustment through supportive emotional environment, by encouraging the special skills that they possess and by helping them to lead essentially normal lives.

Most children affected by ectodermal dysplasia should be placed in a regular classroom, although parents should inform teachers of their special needs. In general, individuals can succeed in any career that they choose and may be limited only by physical considerations, such as the need to avoid hot workplaces.

Beyond this, the person affected by ectodermal dysplasia has access to the usual range of jobs from an airline pilot and physician, to computer programmer and farmer.

ECTODERMAL DYSPLASIAS RESEARCH

Families play an important role in advancing our understanding of the ectodermal dysplasias when they participate in studies.

We provide small grants to initiate research to benefit individuals and families affected by ectodermal dysplasias. Funding is determined yearly based on the dollars available. We have funded projects that focus on the surface characteristics of the skin, hair, and nails, the dental treatments that have managed successfully the oral health features, and the behavioral aspects of the ectodermal dysplasias.

In addition, the Foundation has also funded projects that focus on the pathogenesis, molecular and biochemical pathways, diagnosis, or management of the ectodermal dysplasias or that expands the understanding of the ectodermal dysplasias or improves their care.

The NFED Research Program's goals are to provide support to efforts that:

- Advance the understanding of the genetic involvement of the ectodermal dysplasias and expand the understanding of the ectodermal dysplasias;
- Develop new or improved approaches for diagnosing ectodermal dysplasia;

- Advance the understanding of the characteristics of the skin, hair, and nails in the ectodermal
- dysplasias, the types of dental treatments that successfully manage the features of ectodermal dysplasia, and psychosocial aspects of ectodermal dysplasia;
- Develop research-based information resources;
- Communicate research-based information to increase public awareness; and
- Transfer knowledge to health care providers.

TYPES OF ECTODERMAL DYSPLASIA

The Table below summarizes many ectodermal dysplasias. Since the Table does not emphasize that severity may differ greatly among affected people and that some conditions may be inherited in more than one way, view it as a limited resource.

Ectodermal dysplasias involve combinations of abnormalities of two or more ectodermal structures. International experts are currently reviewing this definition of ectodermal dysplasias and it could be revised to be more detailed in the near future. Some of the many combinations are shown in the Table at the end of this section. A few examples from the Table will illustrate the complexity of the ectodermal dysplasias.

One of the first recognized types of ectodermal dysplasia was HED, originally called anhidrotic ectodermal dysplasia or Christ-Siemens-Touraine syndrome. Individuals with this condition have sparse, lightly pigmented hair, absent or sparse brows and lashes, and a reduced number of teeth. They do not sweat normally. The few teeth that are present are widely spaced and pointed, often

with thin enamel. The skin is excessively dry, and other bodily secretions are diminished. Other ectodermal structures are normal. Any one of these abnormalities may be mild or severe. The physical appearance, then, varies from person to person and family to family.

Another type of ectodermal dysplasia, the tricho-dental syndrome, shows only sparse hair and reduction of the number of teeth. In still another ectodermal dysplasia, tooth and nail syndrome, there are fewer than the normal numbers of teeth and slow growing, flat nails. Because of such complexity, a team of knowledgeable medical professionals including geneticists, dermatologists, dentists, ophthalmologists, and otolaryngoloigsts, must be involved in the evaluation of an individual affected by an ectodermal dysplasia to ensure a correct diagnosis, accurate prognosis, and genetic counseling.

These children all are affected by ectodermal dysplasia but have three different types: Goltz syndrome, hyphidrotic ectodermal dysplasia and ankyloblepharon-ectodermal dysplasia-clefing (AEC) syndrome.

AD = autosomal dominant
AR = autosomal recessive
XLD = X-linked dominant
XL = X-linked

Type	Most Obvious Features	Inheritance
Ankyloblepharon-ectodermal dysplasia-clefting syndrome	Sparse hair, missing teeth, defective nails, cleft lip-palate, eyelid adhesions, skin erosions	AD
Clouston syndrome (hidrotic)	Sparse dry hair, defective nails	AD
Ectrodactyly-ectodermal dysplasia-clefting (EEC) syndrome	Sparse hair, missing teeth, cleft lip-palate, missing digits	AD
Focal dermal hypoplasia (Goltz)	Missing teeth, decreased sweat, sparse hair, asymmetric skull, skeleton	XL
Hypohidrotic ectodermal dysplasia	Decreased sweating, sparse hair, missing teeth	AD, AR, XL
Marshall ectodermal dysplasia	Missing teeth, decreased sweating, cataracts, hearing loss	AD
Pachyonychia congenita	Dry hair, teeth at birth, thick nails, moist palms	AD
Robinson ectodermal dysplasia	Missing teeth, defective nails, hearing loss	AD
Tooth and nail syndrome	Thin hair, missing teeth, slow nail growth	AD
Tricho-dento-osseous syndrome	Defective tooth enamel, abnormal tooth roots, curly hair	AD
Tricho-dental syndrome	Thin hair, missing teeth	AD
Witkop syndrome	Defective tooth enamel, defective nails, decreased	AR

GLOSSARY OF TERMS

Alopecia: absence of hair from areas where it is present normally

Anhidrosis: total absence of functioning sweat glands

Anodontia: congenital absence of all teeth

Atrophic rhinitis: a chronic form of nasal inflammation marked by wasting of the mucous membranes (of the nose)

Autosome: one of the non-sex determining chromosomes

Autosomal dominant: when a single copy of an altered gene on an autosome causes a recognizable disorder

Autosomal recessive: when two copies of an altered gene, one on each member of a pair of autosomes, occur (and no normal copy is present) causing a recognizable disorder

Carrier: an individual who has a single copy of an autosomal recessive gene and females with a single copy of an X-linked recessive gene

Cataract: any change in the normal transparency of the lens of the eye

Chromosome: one of a set of thread-like structures, in the nucleus structure that contains the hereditary material (DNA)

Conjunctivitis: an inflammation of the inner lining of the eyelid or surface of the white of the eye

Corneal clouding (corneal opacity): cloudiness of the "watch crystal" of the front surface of the eye

Ectoderm: the outermost covering of the embryo

Ectrodactyly: congenital absence of one or more of the central fingers or toes

Eczema: a superficial inflammation of the skin

Enamel: the white, hard substance that covers the surface of the crown of a tooth

Epidermis: the outermost layer of skin

Gene: The smallest unit of heredity

Genetic: pertaining to the genes, or to disorders that are capable of being inherited

Geneticist: a physician who specializes in diagnosis and counseling of families affected by genetic conditions

Hereditary: genetically transmitted (in a family)

Hyperhidrosis: excessive perspiration

Hyperpigmentation: excessive pigmentation

Hyperkeratosis: thickening of the skin

Hypodontia: congenital absence of some, but not all, of the teeth

Hypohidrosis: diminished perspiration

Hypotrichosis: presence of less than normal amount of hair

Keratitis sicca: dryness of the surface of the eye usually the result of diminished tear production

Mammary: pertaining to the breast

Mandible: the lower jaw

Maxilla: the upper jaw

Microdontia: abnormally small teeth

Mosaic: a pattern made of numerous small pieces (in genetics, the presence of two or more distinct cell lines in a single individual)

Mucus: the thick liquid covering a moist membrane

Mutation: change or alteration, for example as in an alteration of a normal gene to an abnormal form

Neonatal desquamation of skin: peeling of skin shortly after birth

Nucleus: a small body within a cell that contains the chromosomes

Occluded nasolacrimal sacs: blockage of the ducts that drain tears from the lids to the nose

Onychodysplasia: malformation of the nails

Ophthalmologist: a physician who specializes in diseases of the eyes and visual systems

Oral cleft: an opening through the lip (cleft lip) or through the roof of the mouth (cleft palate)

Orthodontist: a dentist who specializes in correcting malocclusions

Pediatric Dentist: a dentist trained to manage children and young patients with special health care needs

Photophobia: discomfort in bright light

Prosthodontist: a dentist who specializes in dentures

Syndrome: the repetitive co-existence of two or more abnormalities

X-linked: pertaining to genes on the X (female sex) chromosome

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