



Understanding Ectodermal Dysplasia

Living with Ectodermal Dysplasia

Ectodermal Dysplasia isn't something anyone chooses. But while we can't change the diagnosis, we *can* choose how we face it. Acceptance takes time, and managing the condition is a journey — but you're not alone. We hope the information that follows offers some support and guidance along the way.

What is Ectodermal Dysplasia (ED)?

Ectodermal Dysplasia is not a single disorder, but a group of closely related disorders known as the Ectodermal Dysplasias. More than 170 different syndromes (types) have been identified. The Ectodermal Dysplasias are genetic disorders affecting the development or function of the teeth, hair, nails and sweat glands. Depending on the particular syndrome, Ectodermal Dysplasia can also affect the skin, the eyes, the ears, development of fingers and toes, nerves and other parts of the body.

Ectodermal Dysplasias are caused by altered genes. The altered genes may be inherited, or normal genes may become defective (mutate) at the time of conception. The chances for parents with Ectodermal Dysplasia to have affected children depend on the type of Ectodermal Dysplasia that exists in the family.

It is important to remember that a person cannot choose or modify the genes that he or she has, and that events of pregnancy do not change the genes. Thus, parents who have a child with Ectodermal Dysplasia should not think that they did anything to cause the defective gene and should not blame themselves for its existence.

Genetic counselling is available for families.

Did you Know?

Charles Darwin **first documented** an accessible account of Ectodermal Dysplasia in English in the 1840s after he had received some correspondence from a Medical Officer in the Indian army. The Officer had observed families in Punjab whose grandfathers and grandsons were affected by the lack of hair and sweat glands, which is a major hazard when working out in the fields in India. These people survived the heat by tipping buckets of water over each other. There may possibly be earlier accounts than Darwin's, but these do not appear to have been documented. However, in the mid-19th century it appears nobody understood the sex-linked inheritance and it wasn't until 1910 that this began to be understood.

All the information contained in this article concerns X-linked Hypohidrotic Ectodermal Dysplasia (XLHED), a condition characterised by a reduced number of sweat glands. However, the advice provided is general enough to be applicable or adaptable to other types of Ectodermal Dysplasia syndromes.

Temperature Control

One of the main factors of Ectodermal Dysplasia is the lack of temperature control. Children with this condition have an inability to sweat due to absent or reduced sweat glands and therefore overheat at any time of year, either from atmospheric temperatures or an impending infection. This can be dangerous particularly to the very young. There can also be problems in the winter months in maintaining adequate body temperature, i.e. they get too cold and have difficulty in warming up. Whether overheating or underheating (hypothermia) the child may become tired, lethargic, appear to be in a dream, unable to concentrate or listen, become disruptive or unco-operative. They must be cooled down or warmed up. One sign for some individuals is that their ears go very red whilst the body remains pale when they are overheating. As your child gets older the problem gets easier to deal with, although it will never go away.

Keeping Your Baby Comfortable - One of the challenges of caring for a baby with Ectodermal Dysplasia is learning to balance warmth and safety. It's natural to want to keep your baby wrapped up and cosy — but for babies with ED, especially those who struggle to sweat, overheating can be dangerous.

This doesn't mean your baby should be kept cold, but it *does* mean being mindful not to overdress them. It can be hard to ignore advice from well-meaning friends or relatives who urge you to "bundle them up," but trust your instincts — you know

your baby best. Learning what feels right for your child will become second nature with time.

One of the signs that a child's temperature is too high is their behaviour. They may become disruptive, disobedient, uncooperative or fractious. A child with this condition will refuse to go outside in the sunshine or insist on going to the cloakroom a million and one times a day. They are not being awkward or trying to get out of doing something, they are naturally trying to keep cool, either in the shade or using water. They **must** be given access to water at all times, day and night.

A fan will help tremendously in winter as well as in summer. It must be remembered that children with Ectodermal Dysplasia suffer from inside temperatures in the winter months due to central heating, as much as outside temperatures in the summer months. Should the child have a temperature due to an infection, the usual Calpol (paracetamol suspension) will reduce the fever, but care should be taken as the internal core temperature may continue to rise after the use of Calpol. Plenty of flannelling down with tepid water and the use of a fan will help reduce the temperature.

The benefits of a damp T-shirt and dampened hat are incalculable even in the winter indoors when the central heating is on; a refillable water spray bottle that produces a fine mist is great for spraying on the child's head, arms and legs; portable fans are readily available to purchase and it is now possible to buy fans for the car which run off the cigarette lighter; a fan by their bed at night will help them settle more quickly and frequent cool drinks also help. If your child has overheated, a tepid bath (not cold) is advisable and possibly Calpol. A persistent fever however must always be referred to your doctor in case of infection.

Travelling by car or public transport (unless air conditioned) should be avoided in hot weather if at all possible, but if absolutely necessary the following precautions are essential: ensure you have a large container of water and a flannel for sponging down, a fan if you have one and plenty of cool drinks (be prepared for lots of toilet stops on longer journeys). It is a good idea to make use of sun blinds to shade the passengers in the back of the cars. Always try and park in the shade. Cardboard windscreens covers for when your car is stationary are also extremely helpful.

Skin

This may be pale and transparent with veins clearly visible and there is often increased pigmentation around the eyes or on the elbows, palms and soles of the

feet. The heavily pigmented skin around the eyes may be wrinkled and that on the palms and soles may be thick. Very dry skin is without exception a problem. There are a wide range of products available to help prevent and treat dry skin, but it may be best to avoid very greasy products in hot weather. Neither soap nor any perfumed bath additives, including baby bath products, should be used as these all have a drying effect and may cause irritation to the sensitive skin. Always pat dry and apply cream immediately. In a large number of cases eczema is a problem and for some it is extreme. This must be treated by your doctor, or he may refer you to a dermatologist. If persistent problems occur an allergy test may be offered. You should never try alternative remedies yourself without getting good professional advice. During the summer months' individuals with fair skin should use a high factor sun block at all times, preferably one for sensitive skin. Cotton clothes next to the skin and cotton bedclothes are advisable.

Feeding

Almost all babies with Ectodermal Dysplasia have early feeding problems, the worst of which are vomiting, projectile vomiting, choking and fractiousness due to overheating. Please see our leaflet "Neonates and Infants with Ectodermal Dysplasia". Many children manage a normal diet even without teeth, but others find many foods difficult to swallow, particularly dry food; this is possibly caused by lack of saliva; therefore, all foods should be moist.

Nasal Congestion

This is a common problem and happens because the mucous glands may be absent or reduced in number leading to abnormal function of the linings of the nose, sinuses, Eustachian tubes and throat. The mucous secretions may be much thicker than normal, forming a crusty mass. Nasal infections are common and are usually accompanied by a foul odour. Clearing this mucus out is essential to prevent the odour and frequent nosebleeds. There are several things you can do to help; steam inhalations are good; older children can learn to wash out their noses, but many children don't like doing this; saline water drops or spray every day is all that is needed to help them keep their nose moist. If you cannot get saline water in a spray bottle from your doctor a Chemist can supply a spray bottle that is easy enough for a child to use on their own, and the doctor can then prescribe Steripods which are pods of saline water that you can use to replenish the spray bottle or use as drops.

It is possible to buy or have your doctor prescribe nose drops, but vasoconstrictors e.g. ephedrine should not be used long term. However, the long-term use of saline water drops/spray is fine.

Respiration

The linings of the nose, larynx, trachea and lungs are moistened by various glands, some of which may be absent, reduced in number or may not function normally. The underproduction of respiratory fluids together with constant nasal congestion, are a cause of very frequent infections for which antibiotics are often necessary. Croup is a common problem in younger children and asthma is a problem common to individuals of all ages. Both should be referred to your doctor without delay.

Hoarseness of the voice is usually due to laryngeal dryness and can be alleviated using a humidifier, especially in the bedroom throughout the night. In addition, nodules on the vocal cords have been reported. These may improve with speech therapy, but in some cases, they need to be removed.

Eyes

Most people with Ectodermal Dysplasia have normal vision and appear to have no greater need for glasses than anyone else. However, visual problems in Ectodermal Dysplasia may be caused by lack of tears, infections (conjunctivitis), corneal scars, cataracts and retinal changes. Dry eyes are a common problem, and artificial tears may be necessary. Crusting of the eyes can also be a problem and if this happens bathe them gently with a tepid saline solution. Sensitivity to light (photophobia) all year round can be painful for dry eyes; sunglasses or tinted glasses will help or try to stay in the shade. Irritation to the eye may be caused by eyelashes which grow in an abnormal direction; this can be confirmed by an Optician and treated by an eye specialist.

Teeth

One of the main characteristics of Ectodermal Dysplasia, and the one on which a diagnosis is frequently made or proven, is the absence or malformation of teeth (Hypodontia). Teeth that are present may be widely spaced, pointed, and in some cases the enamel is defective and discoloured. Many babies with Ectodermal Dysplasia do not have any teeth erupt until they are around two years of age.

For cosmetic, psychological and social purposes teeth are desirable and they are also fairly useful if you like crusty bread! Great leaps and bounds are being made every day in dentistry and children as young as 2½ are now given dentures. Should your child have pointed upper front teeth, it is very easy for the dentist to build these up with an adhesive tooth-coloured material (dental composite) to make them appear squarer, before he/she begins school. Even if there are still some gaps, having square teeth as opposed to pointed/conical teeth will help prevent any name-calling and bullying by their peers, and will give them a happy smile.

Older patients may be considered for implants or bridgework, but implants are not always suitable for every individual, and it is necessary to seek advice from an implant specialist.

The dentist's chair may become a place where your child will spend a lot of time. It is important for the dentist to get to know your child and vice versa before any treatment commences.

Make sure your dentist is prepared to spend time with your child to build up a rapport, to explain about the room, tools, how the chair works, what he is going to do and most importantly, to have some fun.

It is important to remember that whatever teeth erupt, they become precious; a tooth, no matter what shape or size, can be used by your dentist. Baby teeth, especially canine (eye) teeth, may last for many decades if there are no adult teeth behind them to dissolve the root and push them out. So, hang on to whatever grows and take extra care of them. If a dentist recommends tooth extraction, for whatever reason, seek a second opinion.

Some dental hospitals have specialist teams to help patients with Hypodontia, which can provide a coordinated treatment plan involving all the necessary experts.

Ask your dentist about being referred to one, if this facility is available in your area.

Hair

The scalp hair may be absent, sparse, fine, lightly pigmented (very blond), or abnormal in texture. The hair may also be fragile and unruly, stick out in all directions, get extremely knotted and difficult to comb. The hair may be dry because the oil glands are absent or poorly developed. As shampoo will dry the hair it may be better to wash with bath oil as this will help the hair lie down and help when tackling knots.

Throat

Saliva may be sparse, causing problems with chewing, tasting and swallowing foods. It is advisable to avoid dry foods and always have a drink to hand when eating. A hoarse, raspy voice is common.

Ears

Hearing loss may occur due to hard impacted wax possibly caused by narrowness of the ear canal or the underproduction of body fluids leading to the ear wax becoming very dry. Olive oil drops may help soften the wax, but sometimes this does not penetrate deeply enough to prevent the wax becoming impacted,

especially as the function of the ear canal does not move the wax out of the ear in a normal fashion. Syringing is often extremely painful as the water gets behind the wax and presses on the eardrum. As the child may need the ear wax removed regularly, possibly every 6 months or so, it may be better not to accept syringing, but to have an ear specialist extract the wax under a microscope.

Nails

The nails may be poorly developed, small, thick or thin, brittle, discoloured, cracked, or ridged. In addition, they may grow slowly, shed periodically, and develop light spots, lines or patches. It is also possible for nails to become infected by fungus or yeast and to have a bad odour. Treatment will be needed. Generally, nails should be kept short and moisturised daily.

Speech Problems

Absent, widely spaced or misshapen teeth may cause a child to mispronounce certain sounds, likewise any hearing problems may cause difficulties with speech and speech therapy may be necessary. A dry mouth due to lack of saliva may also have an effect on speech, as does cold weather in the winter months and dryness in the air any time of year. Some individuals with Ectodermal Dysplasia have experienced weak muscles in the mouth and throat which may also cause speech problems and in some Ectodermal Dysplasia syndromes a cleft palate is present. It is important to seek medical help to deal with speech imperfections as these may possibly have some effect on education.

Relationships outside the Family

There will be times when you will have a lot of appointments with various people who are all there to help. It is essential to have a good relationship with your doctors and dentist as this will benefit everyone. Ectodermal Dysplasia is very rare, and it is therefore important to remember that the professionals you see may have had very little experience of Ectodermal Dysplasia and will appreciate all the help and information you can provide.

When your child is starting nursery or school for the first time or changing school it is important that all staff (including dinnertime staff) are aware of Ectodermal Dysplasia and its implications. The ED Society can provide information, such as a tailored health care plan, to help you when talking to Head teachers and Education Authorities. Encourage your child to join activities such as swimming, cubs/brownies, gymnastics and football; in fact, anything and everything they want to. Ectodermal Dysplasia should not prevent them from doing anything providing the right provisions have been made.

Supporting your child with Ectodermal Dysplasia

It's natural to worry when your child is diagnosed with Ectodermal Dysplasia, but with love and support, they can grow up confident and resilient. Children with ED may sometimes struggle with self-esteem, so it's important to nurture a close, caring relationship.

At the same time, it's important to encourage independence. Teaching your child how to care for themselves and explore their interests will help them build confidence and a sense of normalcy.

Though the diagnosis can feel overwhelming, remember that with the right care and support, children with ED can live full, happy lives.

Focus on positive feelings, strengths, and possibilities. While sympathy is natural, too much can sometimes be limiting. Staying hopeful and positive will help both you and your child face the journey ahead with courage.



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