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## **Living with X-linked Hypohidrotic Ectodermal Dysplasia**

**Ectodermal Dysplasia Society** 108 Charlton Lane, Cheltenham, Glos. GL53 9EA. England.

Telephone +44 (0) 1242 261332, Email: [diana@ectodermaldysplasia.org](mailto:diana@ectodermaldysplasia.org), Website [www.ectodermaldysplasia.org](http://www.ectodermaldysplasia.org)

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Charles Darwin first documented the earliest 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 19<sup>th</sup> century it appears nobody understood the sex-linked inheritance and it wasn't until 1910 that this began to be understood.

Ectodermal Dysplasia (ED) is not a single disorder, but a group of closely related disorders known as the Ectodermal Dysplasias (EDs). More than 170 different syndromes (types) have been identified. The 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, lens or retina of the eye, parts of the inner ear, development of fingers and toes, nerves and other parts of the body.

All the information contained in this article concerns the x-linked Hypohidrotic type ED, that is where the number of sweat glands is reduced, but the advice given is fairly general and can be used or adapted for any other type.

## **Defining Ectodermal Dysplasia**

At our beginning we are composed of three layers of cells which, as we grow and develop, become the different parts of our bodies. We are concerned with the outer layer of cells only, which progress to become the skin, hair, nails, teeth, nerve cells, sweat glands, parts of the eye and ear and parts of some other organs. This outer layer is known as the ectoderm.

Although there are numerous disorders, which affect the ectoderm, one cannot properly be diagnosed as having ED unless two or more structures are affected, in any combination. Each combination represents a different syndrome within the ED group of which over 170 have been described.

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 to have affected children depend on the type of ED 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 ED 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.

## ***Dealing with ED***

Having been blessed with ED, despite not ordering it, you cannot send it back. The only course of action is to accept and deal with it. I hope that the following will help.

## ***Feeding***

Almost all babies with ED have early feeding problems, the worst of which is vomiting, projectile vomiting and choking. Feeding little and often can help as does not heating the feeds, give at room temperature instead. For breast feeding mothers it is advisable not to hold the baby too close to her body. Babies do grow out of these problems, but they can very easily grow into faddy eaters if care isn't taken. There is no reason why a normal diet cannot be followed even without teeth.

### ***Temperature Control***

One of the main factors of Hypohidrotic 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. If these children are not cooled down they become tired and lethargic, appear to be in a dream, not concentrating or listening. 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. Should this happen the child will appear to be in a dream, not concentrating or listening. It is important to train yourself to curb a very natural instinct to wrap your ED child warmly, (this does not mean keep him cold) and even harder, I think, to ignore Granny and well meaning friends and relations. Just keep feeling him, and judge for yourself. However, some individuals have found their ears go very red whilst their body remains pale when they are overheating. As your baby gets older the problem gets easier to deal with, although it will never go away.

One of the signs that a child's temperature is too high is their behaviour. They may become disruptive, disobedient, unco-operative 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 by the use of 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 ED children 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 (paracetamol suspension). Plenty of flannelling down with tepid water and the use of a fan will help reduce the temperature.

There are some things you can do to help (which would be of benefit to all not just individuals with ED) such as a refillable water spray bottle that produces a fine mist (available at most garden centres). The benefits of a damp T-shirt and dampened sun hat are incalculable. Portable fans are readily available and it is now possible to buy fans for the car which run off the cigarette lighter. A fan by their bed will help them settle more quickly at night. Frequent cool drinks help. If your child has overheated a tepid bath is needed and possibly paracetamol. 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: A large container of water and a flannel for sponging down, a fan if you have one, plenty of cool drinks (be prepared for lots of toilet stops on longer journeys) and it is a good idea to make use of sun blinds to shade the passengers in the back of the car. Always try and park in the shade. Cardboard windscreen 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 usually increased pigmentation around the eyes or on the elbows, palms and soles of the feet. The heavily pigmented skin around the eyes is 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 ever be used as these all have a drying effect and are irritating to the skin which may be sensitive. Frequent bathing keeps the skin moist. 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. If you are lucky and have a good relationship with your doctor you may find that he will be prepared to try some alternative treatments or refer you to someone who can help. If persistent problems occur an allergy test may be offered and if this is the case it is a good idea to go ahead with it. 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.

## ***Nasal Congestion***

This is a common problem and happens because the mucus membrane may be malformed. The mucous secretions are excessively thicker than normal, forming a crusty mass. Nasal infections are common and are usually accompanied by a foul odour. Clearing this mucous out is essential to prevent the odour and frequent nosebleeds. There are several things you can do. Steam inhalations are good. Older children can learn to wash out their noses and I know one younger child that does this but he is exceptional. For small babies the doctor can prescribe Minims which are saline water eye drops, but are packaged in such small quantities that they are brilliant for use as nose drops. A few drops a day is all that is needed to help them keep their nose moist and thereby make breathing and feeding easier. For small children the doctor can prescribe Steripods which are larger packages of saline water. You then need to purchase a hayfever type spray bottle from the chemist, pour the contents away and fill with saline water. Spraying their nose a few times a day will help keep it moist and help prevent nosebleeds. This type of bottle can be used by small children themselves.

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 are 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.

## ***Eyes***

Most people with ED have normal vision and appear to have no greater need for glasses than anyone else. However, visual problems in ED 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 ED, and the one on which a diagnosis is frequently made or proven, is the absence or malformation of teeth (Hypodontia). Teeth that are present are widely spaced, pointed and in some cases the enamel is defective and discoloured. Many babies with ED 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 2 ½ 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, so that the teeth appear more square, 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 each 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 visa versa before any treatment is commenced. 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 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 (providing they are well looked after) if there are no adult teeth behind them to dissolve the root and push them out. So hang on to whatever grows and take double care of them. If a dentist recommends tooth extraction, for whatever reason, seek a second opinion.

Some dental hospitals have special 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 is 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 is 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.

## ***Ears***

Hearing loss may occur due to hard impacted wax. Due to the underproduction of body fluids ear wax becomes very dry. Olive oil drops may help soften the wax, but sometimes this does not penetrate deep enough to prevent impaction, especially as the function of the ear canal does not move the wax out of the ear in a normal fashion. Syringing is often very painful as the water gets behind the wax, which is very dry and impacted, and presses on the eardrum. As the child may need the ear wax removed regularly, possibly every 6 months or so, it will be more comfortable to have an ear specialist extract the wax under a microscope.

## ***Throat***

Saliva is 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.

## ***Nails***

The nails may be poorly developed, small, thick or thin, brittle, discoloured, cracked, or ridged. In addition, they grow slowly, are 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 perhaps with something like Vaseline.

## ***Speech Problems***

Lack of teeth can cause a child to mispronounce certain sounds, likewise any hearing problems can cause difficulties with speech and some people have felt the need to resort to speech therapy. A dry mouth due to lack of saliva sometimes has an affect on speech, as does cold weather in the winter months. It is important to deal with speech imperfections as these may possibly have some effect on education.

## ***Relationships outside the Family***

It is essential to have a good relationship with your doctors and dentist. This will benefit everyone. There will be times when you will have a lot of appointments with various people who are all there to help. ED is uncommon and it is therefore important to remember that the professionals you see may have had very little experience of ED and will appreciate all the help and information you can provide. If you feel you have to be either defensive or aggressive you will gain nothing.

When your child is starting nursery or school for the first time make sure that all staff (including dinnertime staff) are aware of ED and its implications. This also applies when your child changes school. The ED Society is able to provide information to help you when talking to Head teachers and education authorities. Nurture your child's friendships so that their friends are used to their appearance, they will then be less self conscious if at all. Encourage them to join in every activity they want to try such as swimming, cubs/brownies, music groups, gymnastics, and football, in fact anything and everything you can. Confidence is a very valuable asset and ED will not prevent them from doing anything providing the right provisions have been made.

## ***Your relationship with your ED child***

It is very easy for an ED child to grow up with a poor self-image and this should be avoided at all costs. One way is to develop a tactile relationship. We should try to fit as many hugs into the day as possible, but try not to be over protective, our children need to be taught how to look after themselves and become independent. I believe it is important to encourage skills and interests. On paper ED sounds appalling but in reality it is not. If all of the symptoms are carefully and properly dealt with an absolutely normal lifestyle can be led to the full and therefore there is no need for pity at all. However, sometimes sympathy is desirable. Sympathy itself can be debilitating and therefore we should seek to encourage positive emotions. It is important always to stay positive.

## ***Other***

It should be said that not everything that affects an individual with ED is as a result of having ED and that it is possible to have other conditions or symptoms as well. For example, it is possible that some speech problems are attributable to ED but a stammer would be a separate problem.

This document has been rewritten from an article that was originally produced in 1993 by Moira Hargreaves. Moira has a son with ED and wrote the article with the help of Dr Miller, Consultant Paediatrician who has cared for her son since he was born, and whom she will never be able to thank enough, together with Dr Angus Clarke whose research into ED brought about the beginning of this support group in 1988, and his continued care and concern for affected families is deeply appreciated by us all.

Diana Perry  
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