Published on *My46* (https://www.my46.org) 01/20/2022

My46 Trait Profile

Hypohidrotic Ectodermal Dysplasia

Other Names: Anhidrotic Ectodermal Dysplasia, Christ-Siemens-Touraine syndrome

Hypohidrotic ectodermal dysplasia is a genetic condition characterized by abnormalities of the sweat gland, teeth, skin, and hair. It is caused by mutations in the EDA, EDAR, EDARADD, and WNT10A genes.

Characteristics of Hypohidrotic Ectodermal Dysplasia

Hypohidrotic Ectodermal Dysplasia (HED) is a rare genetic condition characterized by a reduced ability to sweat (hypohidrosis), missing teeth (hypodontia), and fine sparse hair (hypotrichosis). Individuals with HED share a similar facial appearance with thin, dark skin beneath the eye with extra folds or wrinkles, a depressed “saddle” nose, small narrow jaw, and small pointed teeth. There are a number of additional features of HED including dry eyes, eczema, asthma and dry mucous membranes in the mouth and nose. Individuals with HED are at risk for serious medical complications such as abnormally high body temperature (hyperthermia) and unexplained fever. Many of these complications are often associated with their inability to sweat. They are also at risk for respiratory infections because of their reduced mucous secretions.

Diagnosis/Testing

Changes or mutations in the EDA, EDAR, EDARADD, and WNT10A genes are most commonly associated with HED. These genes tell the body to make proteins that are needed early in life (before birth and shortly after) for the normal development of sweat glands, teeth, hair, skin, and other mucous glands.

Management/Surveillance

Individuals with HED cannot sweat effectively. Therefore, it is very important that their temperature is controlled, especially early in infancy, to avoid hyperthermia. There are many ways to help keep cool, including the use of spray bottles and cooling vests. It is important for school-aged children to have access to air-conditioned classrooms. Medical management often includes skin care, dental management (dentures, caps and implants), as well as the management of upper respiratory tract infections and asthma.

Due to the challenges of temperature regulation, individuals with HED often react poorly to drastic changes in temperature. Because of asthma and upper respiratory tract infections, circulated air may need to be filtered. Nutrition awareness is important as dietary issues have been reported and associated with poor growth. Many families have reported the need for psychosocial support due to different facial characteristics, the need for dentures from an early age, thinning hair in adolescence and bullying in school.

Mode of inheritance

HED may be inherited in one of three patterns of inheritance: X-linked recessive, autosomal recessive and autosomal dominant.

X-linked recessive inheritance:

The majority of HED is caused by changes in the EDA gene located on the X chromosome (X-linked HED or XLHED). An X-linked recessive pattern means that in a person with two X chromosomes (most females), both copies of

a gene (i.e., one on each X chromosome) must have a change or mutation whereas in a person with one X chromosome (most males), only one copy of a gene must have a mutation to be affected. A female with a mutation in one copy of a gene on the X chromosome is said to be a carrier of XLHED.

Autosomal recessive and autosomal dominant inheritance:

Autosomal recessive inheritance and autosomal dominant inheritance accounts for a small proportion of HED. Mutations in the EDAR, EDARADD, and WNT10A genes follow these two inheritance patterns.

Autosomal recessive inheritance pattern means that an individual has to inherit two mutations (i.e., one from each parent) to be affected. If both parents are carriers of a mutation they have a 1 in 4 (25%) chance with each pregnancy of having a child with the condition.

Autosomal dominant inheritance pattern means that inheriting one mutation is enough for an individual to be affected with HED. The mutation can be inherited from an affected parent or it can occur brand new (de novo) in an affected child.

Risk to family members

The risk to family members depends on the pattern of inheritance.

X-linked recessive inheritance:

If a father is affected with XLHED, his daughters will be carriers of XLHED and his sons will be unaffected. If a mother is a carrier of XLHED, each daughter has a 1 in 2 chance (i.e., 50%) of being a carrier and each son has a 1 in 2 chance (i.e., 50%) of being affected with XLHED.

Autosomal dominant inheritance:

The risk to family members depends on whether or not the individual with HED has a parent affected with HED. If a parent also has the condition, the risk of having a child with HED is 50% with each pregnancy. If a parent does not have HED, the risk of other siblings being affected is very low.

Autosomal recessive inheritance:

Parents of a child with HED are carriers of HED. If a sibling of a child with HED is unaffected, he/she has a 2 in 3 (66%) chance of being a carrier of HED.

Special considerations

Carriers of XLHED may experience mild symptoms such as thin hair, reduced sweating, one or more missing teeth, and sometimes have difficulty breastfeeding. While carriers of XLHED may not exhibit full symptoms, they can pass the gene change on to their children.

Resources

Genetics Home Reference: Hypohidrotic ectodermal dysplasia
http://ghr.nlm.nih.gov/condition/hypohidrotic-ectodermal-dysplasia

XLHED Network
http://www.xlhednetwork.com/

Have the Conversation (XLHED specific information regarding planning a family)
http://www.havetheconvo.com/

National Foundation for Ectodermal Dysplasias
http://nfed.org/

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Created: 04/2013

Updated: mm/yyyy

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