Health Awareness

Know Your Family's Risk Of A Rare Genetic Disorder And The Resources Available To Help You

(NAPSA)—There may be a rare genetic disorder in your family that you don't even know about, but the good news is that a genetic counselor can provide the information, guidance and support to help you understand the chance of passing the condition to your children and options to help your child manage the condition.

The Disease

The condition is called hypohidrotic ectodermal dysplasia (HED)—also known as anhidrotic ectodermal dysplasia and Christ-Siemens-Touraine syndrome. It's estimated that at least one in 5,000 to 10,000 newborns has it. Seventy-five to 85 percent of boys with HED have the X-linked recessive form called XLHED.

The Signs

Common symptoms of XLHED include a reduced ability to sweat, because babies affected by XLHED have no, or fewer than normal, sweat glands or they don't sweat sufficiently. People with the disorder are missing or often have pointed teeth (hypodontia). They experience sparse and slow-growing hair on the head and body and also have drier than normal linings or membranes in the nose and mouth. In addition, breathing problems such as asthma, increased respiratory tract infections (colds, pneumonias, etc.) and scaly skin are common.

How It's Diagnosed

A change in a gene called EDA may result in XLHED. This gene controls the production of a protein (a molecule the body needs for growth) that starts the development of skin, hair, nails, teeth, and sweat glands. However, when the gene is changed, there is not enough protein for typical development of these tissues and organs. XLHED is often diagnosed after birth or in early childhood based on several physical features. Diagnosis can also be made through family history and genetic testing.

Some families may be able to help cure a difficult, inherited disease.

How It's Inherited

XLHED is caused by a change in a gene that is passed from parents to children on the X chromosome. Chromosomes are "messengers" that carry "instructions" (genes) to the body in cells. Women have two X's; men have an X and a Y. A girl gets an X from each parent, a boy gets an X from his mom and a Y from his dad. Since the gene is only on the X chromosome, the daughters of a man affected by XLHED will all be carriers and his sons will not be affected. A woman with XLHED, or who is a carrier, may have some symptoms, but because women have two X chromosomes, these may be less severe. There's a 50 percent chance a woman with XLHED will pass the gene to a son who will be affected by XLHED and a 50 percent chance her daughter will be a carrier.

Genetic Counseling

It's possible for prospective parents to be tested to see if they're carriers, and this is most often done when there is a known family history of XLHED. Understanding the testing options available can be challenging. Genetic counseling is a discussion between a patient or family and a health care provider trained in genetics and communicating genetic and medical information to families. Families that talk to a genetic counselor gain a better understanding of the condition, their family history, test results, clinical trial opportunities and potential treatment options. Free genetic counseling services are available to families that have or suspect they have XLHED. The counseling is from InformedDNA, a phone-based, confidential, genetic counseling service. Experienced, board-certified genetic counselors are available at (617) 758-4300. You just make an appointment that works for your schedule.

Knowledge Is Power

Understanding your family's experience with XLHED can help identify other family members who may develop the condition or have the chance to have a child affected by it. There are currently no specific treatments available for XLHED; however, there are ongoing clinical trials of a new drug that could potentially treat the symptoms of XLHED. Furthermore, doctors and patients have found ways to manage the challenges of living with the disorder. Research options are advancing every day and this information may determine if you or your family member is eligible to participate in ongoing clinical trials. Free genetic counseling services provided by InformedDNA are funded by Edimer Pharmaceuticals. The information you provide during the genetic counseling session will be strictly confidential and will not be shared with the pharmaceutical company. Using the service in no way obligates you to participate in genetic testing or any clinical trials.

Learn More

To hear from families that have experience with the disorder and to access additional resources and advice, you can go to the National Foundation for Ectodermal Dysplasias' website, www.NFED.org, and visit the XLHED Network's online community at www.xlhednetwork.com. To learn about clinical trials, visit www.clinicaltrials.gov and search for Edimer.