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My46 Trait Profile

Dystrophic Epidermolysis Bullosa

Epidermolysis Bullosa describes a large group of genetic conditions that cause blistering of the skin after very little, or no injury. Dystrophic Epidermolysis Bullosa is caused by mutations in the COL7A1 gene that makes the type 7 collagen protein.

Characteristics of Dystrophic Epidermolysis Bullosa

Fragile skin characterizes all types of Epidermolysis Bullosa. Mutations in genes that code for proteins used to anchor skin layers cause all four main types of Epidermolysis Bullosa: Epidermolysis Bullosa Simplex (EBS; see trait profile), Dystrophic Epidermolysis Bullosa (DEB), Junctional Epidermolysis Bullosa (JEB; see trait profile), and Kindler syndrome. The type of Epidermolysis Bullosa depends on which layer of skin is affected. In EBS the epidermis, or outer-most layer is affected. In DEB, the lower layer is affected. Junctional EB (JEB) causes fragility of the lamina lucida, the layer between the dermis and epidermis. Kindler syndrome is another very rare type of Epidermolysis Bullosa that affects all three layers. Epidermolysis Bullosa can appear very different between types, and between people with the same type.

In most cases of DEB, multiple blisters or open areas without a layer of skin are seen at birth. Unlike in other types of EB, scarring of healed blisters is often seen in DEB. Milia may form on healed areas. Nails may become misshapen, lost easily, and may not regrow. Blistering or erosions can affect the mouth and esophagus, causing difficulty eating and swallowing. Dominantly inherited DEB is usually milder, while recessively inherited DEB tends to be more severe (See “Inheritance”). The palms, soles, knees and elbows may be the only affected areas in individuals with mild DEB. Children with severe DEB are more likely to have difficulty with growing, iron deficiency, and maintaining fluids in the body while fighting infections. Progressive scarring in severe DEB eventually prevents movement of fingers and toes, which then fuse together. People with severe recessively inherited DEB have a high chance of developing an aggressive form of squamous cell carcinoma, a type of skin cancer. In recessively inherited DEB, early death is unfortunately common. DEB does not affect intellectual abilities. However, DEB can be emotionally, socially and financially difficult for families.

Diagnosis/Testing

A diagnosis of DEB can be made by a dermatologist by looking at a skin sample under an electron microscope. Diagnosis can be confirmed by genetic testing for a change or mutation in the COL7A1 gene. This gene makes the type 7 collagen protein that helps to strengthen the skin. Mutations in this gene do not allow the protein to work normally, thus causing the features seen in DEB.

Management/Surveillance

Management of DEB varies by severity. The use of soft clothing and shoes may help minimize blisters or erosions. Newborns with EBS need special attention to diaper fastening, how they are held, and how they are carried in baby carriers. For instance, picking up a child with severe DEB under the arms may cause the skin to shear. Completely preventing blisters is not possible. Once they form, blisters should be lanced and drained to prevent them from becoming larger. Ointments for the skin, vinegar or small amounts of bleach in a bath, or oral antibiotics may be needed to prevent or heal infections. Bandaging helps shield open areas from infection and assist healing. Various non-adhesive

dressings are available commercially to cover open wounds and provide padding. Band-Aids, hospital tape or other adhesives can tear the skin or cause blisters at the edges. Specialized dressings may be needed over nearly all the limbs and trunk area. Properly wrapping hands between fingers can help delay fusing. In severe DEB, pain management needs to be addressed. Heat and humidity can worsen blistering, or the itching under bandages. Air conditioning is necessary in hot or mixed climates. Poorly fitted or rough shoes and clothing should be avoided when possible. The level of acceptable activity will be different for each individual.

Tightening of the esophagus due to scar tissue may be treated with a dilation procedure at a specialty hospital.

Physical and occupational therapy are helpful to develop strength. Because the skin is constantly trying to repair itself, maintain fluid balance, and fight infections, nutritional supplements may be necessary.

Mode of inheritance

DEB may be inherited in one of two patterns of inheritance: autosomal dominant and autosomal recessive. They are explained below.

Autosomal dominant inheritance:

DEB is inherited in an autosomal dominant pattern. This means inheriting one mutation is enough for an individual to be affected and show signs of DEB. The mutation can be inherited from an affected parent or it can occur brand new (de novo) in an affected child.

Autosomal recessive inheritance:

DEB can also be inherited in an autosomal recessive pattern. This means that an individual has to inherit two mutations (i.e., one from each parent) to be affected with DEB. If both parents are carriers of a mutation, they have a 1 in 4 (25%) chance with each pregnancy of having a child with DEB. Recessive DEB tends to be more severe than dominantly inherited DEB.

Risk to family members

The risk to family members depends on the inheritance pattern.

Autosomal dominant inheritance:

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

Autosomal recessive inheritance:

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

Special considerations

None

Resources

Genetics Home Reference: Dystrophic Epidermolysis Bullosa
http://ghr.nlm.nih.gov/condition/dystrophic-epidermolysis-bullosa
DebRA of America, Inc.
http://www.debra.org/dystrophic
ebnurse
http://ebnurse.org/index.php?id=1
NIAMS: Epidermolysis Bullosa
http://www.niams.nih.gov/Health_Info/Epidermolysis_Bullosa/
EBCare Registry
https://ebcare.patientcrossroads.org

References

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