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My46 Trait Profile

Junctional Epidermolysis Bullosa

Epidermolysis Bullosa describes a large group of genetic conditions that cause blistering of the skin after very little, or no injury. Junctional Epidermolysis Bullosa is caused by mutations in the LAMB3, COL17A1, LAMC2, and LAMA3 genes each of which makes proteins involved in strengthening the skin.

Characteristics of Junctional 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; see trait profile), Junctional Epidermolysis Bullosa (JEB), 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 EB that affects all three layers. Epidermolysis Bullosa can appear very different between types, and between people with the same type.

In nearly all cases of JEB, blisters or open areas without a layer of skin are seen at birth. In severe cases of JEB, large areas of skin may have open wounds or erosions that cannot fully heal. Granular tissue often appears on healing areas, and is particularly fragile. Blisters can form in the mouth, airway, and esophagus. Affected babies may have trouble keeping electrolytes in balance, and difficulty fighting infections. Unlike other types of Epidermolysis Bullosa, thin or pitted tooth enamel is common in JEB. Other physical features may include misshapen nails, mottled skin pigmentation after healing, and thin or nearly absent hair. Additional features include swallowing problems, pain, and trouble meeting nutritional needs. In very severe cases, newborns do not survive the first year of life. JEB does not affect intellectual abilities. However, JEB can be emotionally, socially and financially difficult for families.

Diagnosis/Testing

A diagnosis of JEB 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 LAMB3, COL17A1, LAMC2 or KAMA3 genes. These genes make proteins that help to strengthen the skin by attaching the outer most layer to the underlying layer to the outer layer of the skin. Mutations in any one of these genes do not allow the protein to work normally, thus causing the features seen in JEB.

Management/Surveillance

Management of JEB 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 JEB 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. In severe JEB, pain management needs to be addressed. Heat and humidity can worsen blistering, or the

itching under bandages. Air conditioning is essential in hot or mixed climates. Poorly fitting, or rough shoes and clothing should be avoided. The level of acceptable activity will be different for each individual.

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

JEB is 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 JEB. If both parents are carriers of a mutation, they have a 1 in 4 (25%) chance with each pregnancy of having a child with JEB.

Risk to family members

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

Special considerations

None

Resources

Genetics Home Reference: Junctional Epidermolysis Bullosa
http://ghr.nlm.nih.gov/condition/junctional-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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Pfendner EG, Lucky AW. (Updated 22 February 2008). Junctional Epidermolysis Bullosa. In: GeneReviews at GeneTests Medical Genetics Information Resource (database online). Copyright, University of Washington, Seattle. 1997-2013. Available at http://www.ncbi.nlm.nih.gov/books/NBK1125/. [Accessed 10/09/2013].

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