Junctional Epidermolysis Bullosa, *LAMB3*-Related

NxGen MDx

What Your Results Mean

Test results indicate that you are a carrier of junctional epidermolysis bullosa, *LAMB3*-related (JEB). Carriers are not expected to show symptoms. You and your partner or donor would both have to be carriers of JEB for there to be an increased chance to have a child with symptoms; this is known as autosomal recessive inheritance. Carrier testing of your partner or donor is recommended in addition to consultation with a genetic counselor for a more detailed risk assessment.

Since this is an inherited gene change, this information may be helpful to share with family members as it may impact their family planning.

Recommended Next Steps

Carrier testing of your partner or donor is recommended in addition to consultation with a genetic counselor for a more detailed risk assessment. If both you and your partner or donor are carriers for junctional epidermolysis bullosa, *LAMB3*-related, each of your children has a 1 in 4 (25%) chance to have the condition.

Junctional Epidermolysis Bullosa, *LAMB3*-Related Explained

What is Junctional Epidermolysis Bullosa, *LAMB3*-Related?

Junctional epidermolysis bullosa, *LAMB3*-related (JEB) is an inherited condition in which a subunit of laminin-5 is defective. Laminin-5 is responsible for holding layers of skin together, and when it is non-functional, the skin is very fragile and blisters easily. Affected individuals have blistering over large areas of their bodies including the skin, the mouth, and the digestive tract beginning in birth or infancy. Individuals with JEB are very susceptible to infections as blisters scar and bleed profusely. They may also have difficulties with eating and digestion. Fused fingers and toes and joint deformities that restrict movement may also be present.

Prognosis

Prognosis is poor as most affected individuals do not live longer than one year.

Treatment

Treatment is symptomatic. Some treatment strategies include protecting the skin through bandaging, preventing and treating infections, and maintaining good nutrition and hydration.

Resources

Dystrophic Epidermolysis Bullosa Research Association of America
http://www.debra.org/aboutdebra
Genetics Home Reference
https://ghr.nlm.nih.gov/condition/junctional-epidermolysis-bullosa
National Society of Genetic Counselors
https://www.nsgc.org/

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