

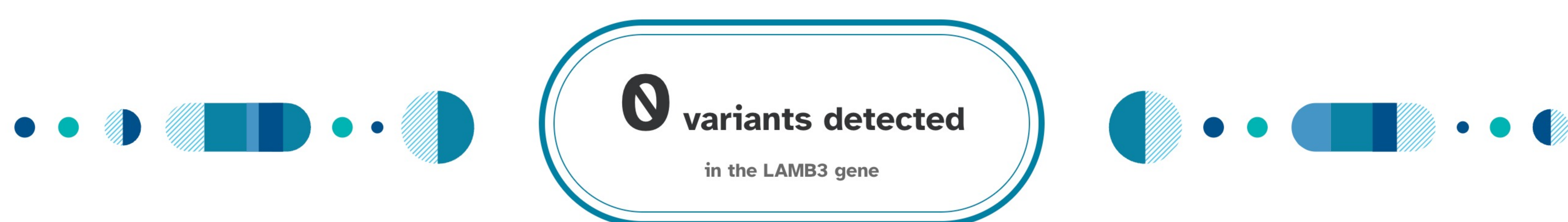
Herlitz Junctional Epidermolysis Bullosa (LAMB3-Related)

LAMB3-related JEB is a rare genetic disorder. The Herlitz form is characterized by severe blistering of the skin and mucous membranes and, typically, death in infancy. A person must have two variants in the LAMB3 gene in order to have this condition.

Overview Scientific Details

Jamie, you **do not have the variants** we tested.

You could still have a variant not covered by this test.



How To Use This Test

This test does not diagnose any health conditions.

Please talk to a healthcare professional if this condition runs in your family, you think you might have this condition, or you have any concerns about your results.

[Review the Carrier Status tutorial](#)
[See Scientific Details](#)

+ Intended Uses

- Tests for **multiple variants** in the LAMB3 gene associated with the severe, Herlitz form of JEB.
- To identify **carrier** status for LAMB3-related JEB.

- Limitations

- Does **not test** for all possible variants for the condition.
- Does **not report** if someone has two copies of a tested variant.
- Does **not include** variants in the LAMA3, COL17A1, or LAMC2 genes that are also related to JEB.

🌐 Important Ethnicities

- This test does **not** include the majority of LAMB3 variants that cause LAMB3-related JEB in any ethnicity.

You are likely not a carrier.

This result may be less relevant for you because the variants that cause LAMB3-related JEB are rarely found in people of your ethnicity.

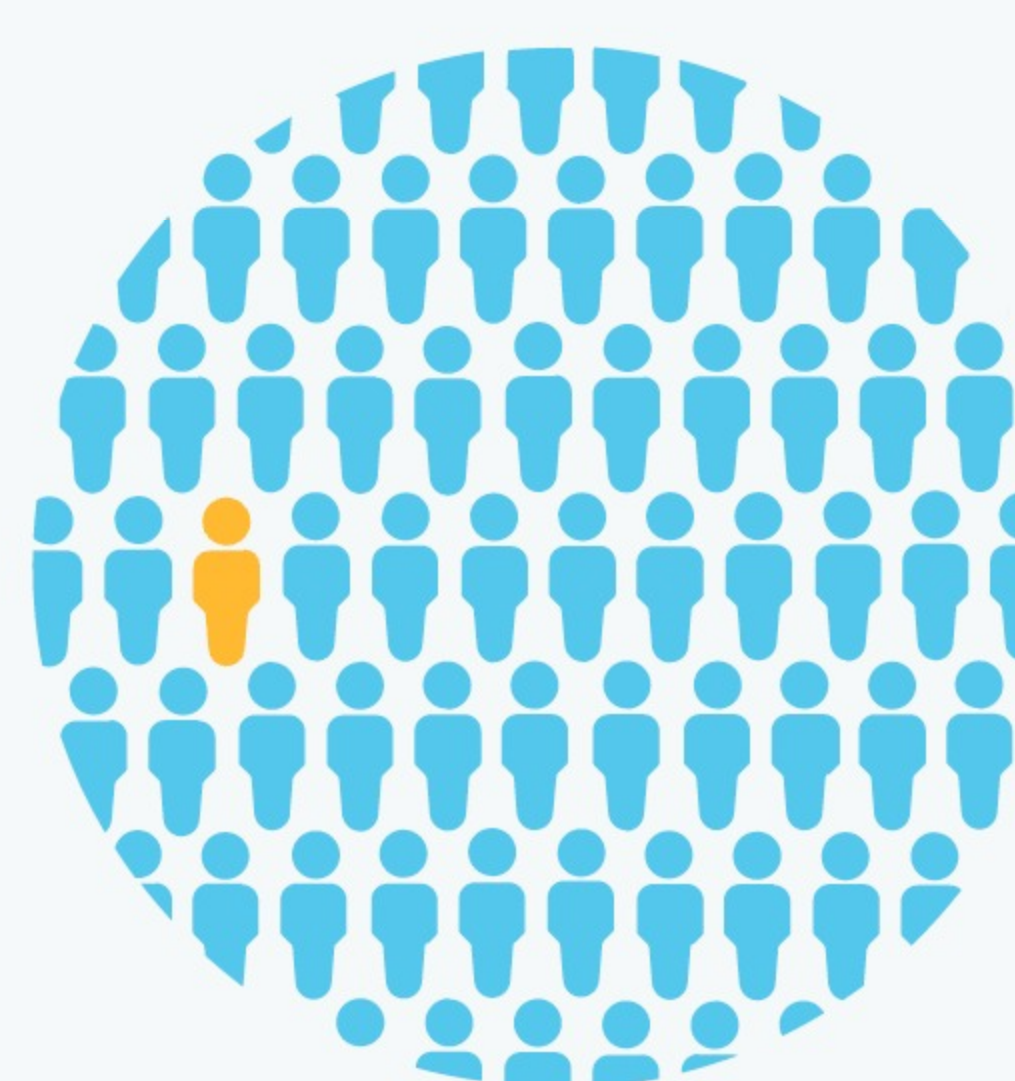


We ruled out the tested variants for LAMB3-related JEB.

These variants are very rare in all ethnicities.

You still have a chance of being a carrier for LAMB3-related JEB.

We cannot estimate your chances because this condition is rare and not well studied in your ethnicity.



About Herlitz Junctional Epidermolysis Bullosa (LAMB3-Related)

📅 When symptoms develop

Symptoms of Herlitz JEB are typically present at birth.

👥 Ethnicities most affected

This condition can affect people of all ethnicities.

🌡️ Typical signs and symptoms

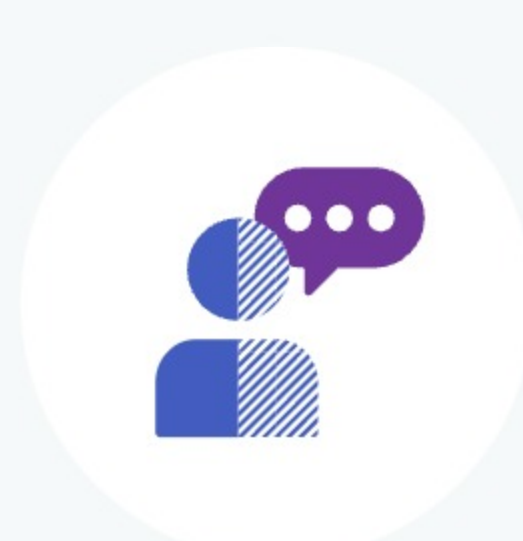
- Fragile skin and mucous membranes
- Severe blistering
- Recurrent infections
- Difficulty swallowing, speaking, and breathing

🏥 How it's treated

There is currently no known cure. Treatment focuses on protecting the skin, wound care, and managing infections and other complications.

Read more at: [Genetics Home Reference](#) [GeneReviews](#) [Mayo Clinic](#) [National Institutes of Health](#)

Consider talking to a healthcare professional if you are concerned about your results.



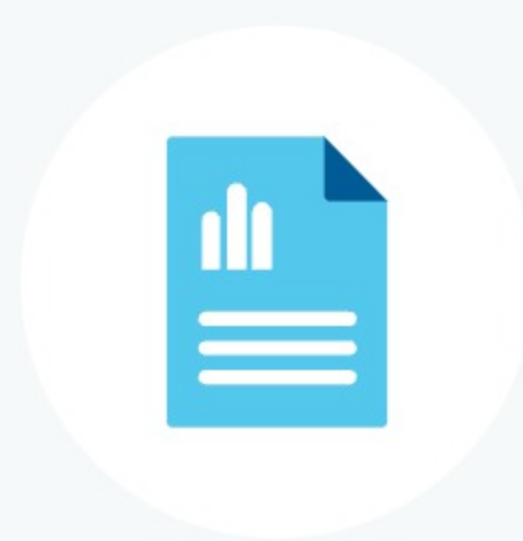
If you're starting a family, a genetic counselor can help you and your partner understand if additional testing might be appropriate.

Connect with a GC



Share your results with a healthcare professional.

Print report



Learn more about this condition and connect with support groups.

Learn more



Give the gift of DNA discovery.

Gift a kit

Refer friends, earn rewards.

Get reward

ANCESTRY

- Ancestry Overview
- All Ancestry Reports
- Ancestry Composition
- DNA Relatives
- Order Your DNA Book

HEALTH & TRAITS

- Health & Traits Overview
- All Health & Traits Reports
- My Health Action Plan
- Health Predisposition
- Pharmacogenetics
- Carrier Status
- Wellness
- Traits

RESEARCH

- Research Overview
- Surveys and Studies
- Edit Answers
- Publications

FAMILY & FRIENDS

- View all DNA Relatives
- Family Tree
- Your Connections
- GrandTree
- Advanced DNA Comparison

Herlitz Junctional Epidermolysis Bullosa (LAMB3-Related)

LAMB3-related JEB is a rare genetic disorder. The Herlitz form is characterized by severe blistering of the skin and mucous membranes and, typically, death in infancy. A person must have two variants in the LAMB3 gene in order to have this condition.

Overview Scientific Details

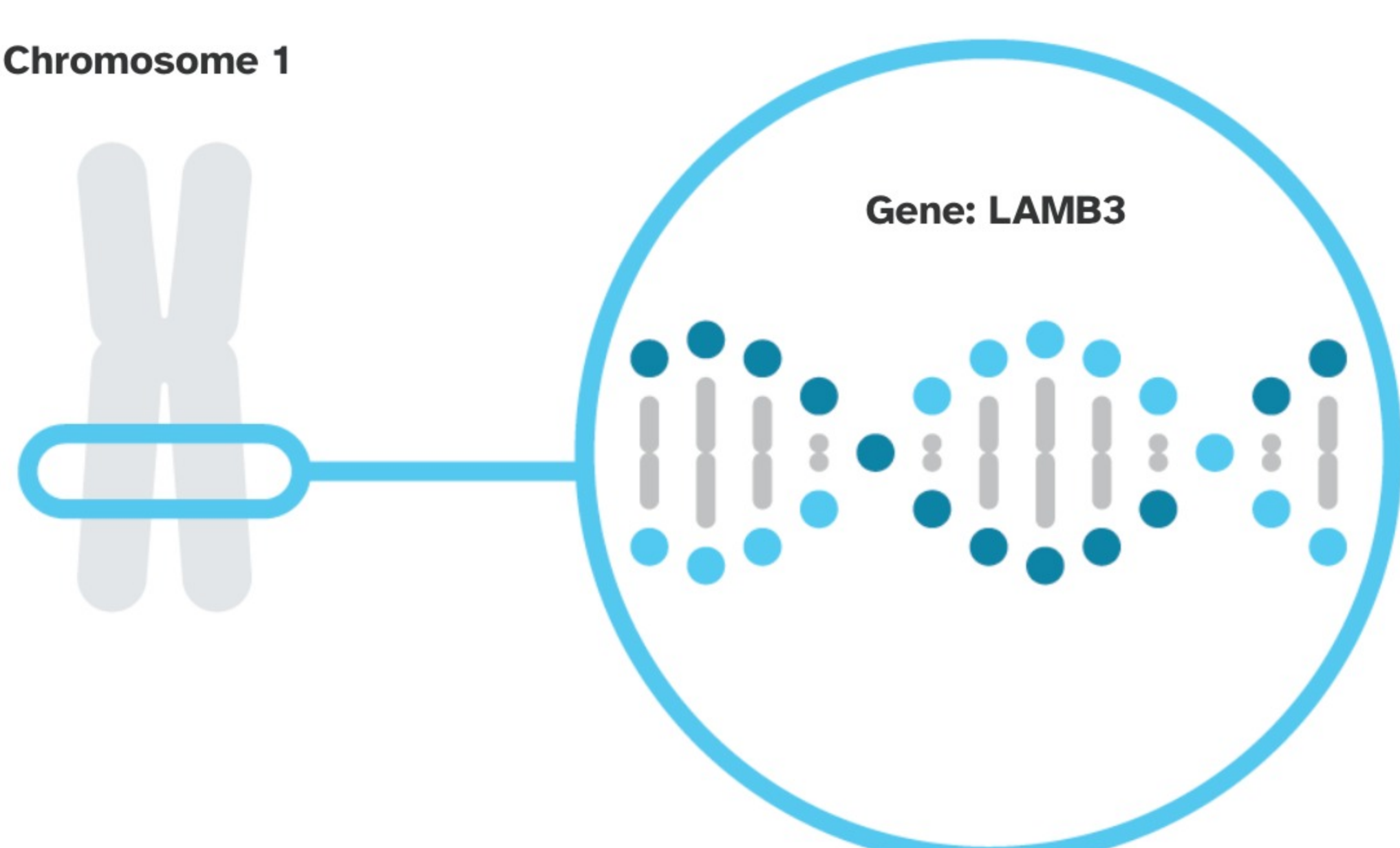
LAMB3-related JEB is caused by variants in the LAMB3 gene.

LAMB3




The LAMB3 gene contains instructions for making a part of a protein called laminin 332. This protein plays a role in the regulation of cell movement, growth, and how cells stick together. Certain variants in the LAMB3 gene disrupt this function, causing the skin and other tissues to be extremely fragile.

Read more at [Genetics Home Reference](#)

Chromosome 1



You have no variants detected by this test.

Variants Detected		View All Tested Markers	
Marker Tested	Your Genotype*	Additional Information	
R635X Gene: LAMB3 Marker: i5012672	G Typical copy from one of your parents 	G Typical copy from your other parent	<ul style="list-style-type: none"> Biological explanation Typical vs. variant DNA sequence(s) Percent of 23andMe customers with variant References [1, 2, 3, 4, 5, 6, 8, 9, 10, 12] ClinVar
R42X Gene: LAMB3 Marker: i5012669	G Typical copy from one of your parents 	G Typical copy from your other parent	<ul style="list-style-type: none"> Biological explanation Typical vs. variant DNA sequence(s) Percent of 23andMe customers with variant References [2, 5, 11] ClinVar
Q243X Gene: LAMB3 Marker: i5012671	G Typical copy from one of your parents 	G Typical copy from your other parent	<ul style="list-style-type: none"> Biological explanation Typical vs. variant DNA sequence(s) Percent of 23andMe customers with variant References [2, 5, 10, 11] ClinVar

*This test cannot distinguish which copy you received from which parent. This test also cannot determine whether multiple variants, if detected, were inherited from only one parent or from both parents. This may impact how these variants are passed down.

23andMe always reports genotypes based on the 'positive' strand of the human genome reference sequence (build 37). Other sources sometimes report genotypes using the opposite strand.

Test Interpretation

Post-test carrier risk for LAMB3-related JEB is the chance of still being a carrier for the condition if you do not have the variants tested. This chance depends on how common it is to be a carrier for LAMB3-related JEB and whether the variants we tested tend to be found in people of your ethnicity.

Because you do not have the variants we tested, your chances of still being a carrier are lower than for someone who has not been tested. However, we cannot provide an exact estimate because the information needed to calculate post-test carrier risk is not available for your ethnicity.

Test Details

Indications for Use

The 23andMe PGS Carrier Status Test for Herlitz Junctional Epidermolysis Bullosa (LAMB3-Related) is indicated for the detection of three variants in the LAMB3 gene. This test is intended to be used to determine carrier status for LAMB3-related JEB in adults, but cannot determine if a person has two copies of a tested variant.

Special Considerations

- This test does not include the majority of LAMB3 variants that cause LAMB3-related JEB in any ethnicity.
- There are currently no professional guidelines in the U.S. for carrier testing for this condition.

Test Performance Summary

Carrier Detection Rate & Relevant Ethnicities

The "carrier detection rate" is an estimate of the percentage of carriers for this condition that would be identified by this test. Carrier detection rate differs by ethnicity and is provided only where sufficient data is available.

General U.S. Population	48%	[11]
-------------------------	-----	--------

Analytical Performance

Accuracy was determined by comparing results from this test with results from sequencing. Greater than 99% of test results were correct. While unlikely, this test may provide false positive or false negative results. For more details on the analytical performance of this test, refer to the package insert.

Warnings and Limitations

- This test does not cover all variants that could cause this condition.*
- This test does not diagnose any health conditions.
- Positive results in individuals whose ethnicities are not commonly associated with this condition may be incorrect. Individuals in this situation should consider genetic counseling and follow-up testing.
- Share results with your healthcare professional for any medical purposes.
- If you are concerned about your results, consult with a healthcare professional.

See the [Package Insert](#) for more details on use and performance of this test.

* Variants not included in this test may be very rare, may not be available on our genotyping platform, or may not pass our testing standards.

References

- Floeth M et al. (1999). "Digenic junctional epidermolysis bullosa: mutations in COL17A1 and LAMB3 genes." *Am J Hum Genet.* 65(6):1530-7. ^
- Kivirikko S et al. (1996). "Mutational hotspots in the LAMB3 gene in the lethal (Herlitz) type of junctional epidermolysis bullosa." *Hum Mol Genet.* 5(2):231-7. ^
- Laimer M et al. (2010). "Herlitz junctional epidermolysis bullosa." *Dermatol Clin.* 28(1):55-60. ^
- Mühle C et al. (2005). "Novel and recurrent mutations in the Laminin-5 genes causing lethal junctional epidermolysis bullosa: molecular basis and clinical course of Herlitz disease." *Hum Genet.* 116(1-2):33-42. ^
- Nakano A et al. (2000). "Herlitz junctional epidermolysis bullosa: novel and recurrent mutations in the LAMB3 gene and the population carrier frequency." *J Invest Dermatol.* 115(3):493-8. ^
- Pasmooij AM et al. (2007). "Revertant mosaicism in junctional epidermolysis bullosa due to multiple correcting second-site mutations in LAMB3." *J Clin Invest.* 117(5):1240-8. ^
- Pfendner EG et al. (2008). "Junctional Epidermolysis Bullosa." [Updated 2014 Jan 2]. ^
- Posteraro P et al. (2004). "Laminin-5 mutational analysis in an Italian cohort of patients with junctional epidermolysis bullosa." *J Invest Dermatol.* 123(4):639-48. ^
- Pulkinen L et al. (1994). "A homozygous nonsense mutation in the beta 3 chain gene of laminin 5 (LAMB3) in Herlitz junctional epidermolysis bullosa." *Genomics.* 24(2):357-60. ^
- Pulkinen L et al. (1997). "Predominance of the recurrent mutation R635X in the LAMB3 gene in European patients with Herlitz junctional epidermolysis bullosa has implications for mutation detection strategy." *J Invest Dermatol.* 109(2):232-7. ^

See all references ^

Change Log

Your report may occasionally be updated based on new information. This Change Log describes updates and revisions to this report.

Date	Change
Aug. 10, 2016	Herlitz Junctional Epidermolysis Bullosa (LAMB3-Related) report created.



Give the gift of DNA discovery.

Gift a kit

Refer friends, earn rewards.

Get reward

ANCESTRY

- Ancestry Overview
- All Ancestry Reports
- Ancestry Composition
- DNA Relatives
- Order Your DNA Book

HEALTH & TRAITS

- Health & Traits Overview
- All Health & Traits Reports
- My Health Action Plan
- Health Predisposition
- Pharmacogenetics
- Carrier Status
- Wellness
- Traits

RESEARCH

- Research Overview
- Surveys and Studies
- Edit Answers
- Publications

FAMILY & FRIENDS

- View all DNA Relatives
- Family Tree
- Your Connections
- GrandTree
- Advanced DNA Comparison