

Do your patients have fragile skin? Blistering? Wounds? Scarring? Nail abnormalities? It may not be what you think it is. It could be **dystrophic epidermolysis bullosa (DEB)—**genetic testing** is recommended by guidelines for accurate diagnosis.**



What is DEB?

DEB, a form of epidermolysis bullosa (EB), is a **serious genetic blistering disorder**. It is caused by mutations in the *COL7A1* gene, resulting in lack of functional collagen VII driving fragile skin, mucosa, and epithelial linings¹

DEB can have serious risks and complications

DEB patients require proactive management and care due to an increased risk of serious consequences, such as aggressive squamous cell carcinoma (SCC), internal complications, and early mortality. **An early and accurate DEB diagnosis is critical**²⁻⁷

A DEB diagnosis can be missed or delayed

Diagnosis of DEB is often based on symptoms alone. This can lead to a missed or delayed DEB diagnosis because of symptom overlap across EB types and other non-EB blistering conditions⁸⁻¹⁰

Guidelines specify that genetic testing is always recommended for the diagnosis of EB type.^{9,11}

Diagnose DEB with simple genetic testing at **no charge through the Decode DEB™ program.**

REFERENCES: 1. Fortuna G, Aria M, Cepeda-Valdes R, Trevino MGM, Salas-Alanis JC. Pain in patients with dystrophic epidermolysis bullosa: association with anxiety and depression. *Psychiatry Investig.* 2017;14(6):746-753. doi:10.4306/pi.2017.14.6.746 2. Eichstadt S, Tang JY, Solis DC, et al. From clinical phenotype to genotypic modelling: incidence and prevalence of recessive dystrophic epidermolysis bullosa (RDEB). *Clin Cosmet Investig Dermatol.* 2019;12:933-942. doi:10.2147/CCID.S232547 3. EB in depth. Debra of America. Accessed June 3, 2022. <https://www.debra.org/about-eb/eb-depth> 4. Christiano AM, Crollick J, Pincus S, Uitto J. Squamous cell carcinoma in a family with dominant dystrophic epidermolysis bullosa: a molecular genetic study. *Exp Dermatol.* 1999;8(2):146-152. doi:10.1111/j.1600-0625.1999.tb00364.x 5. Tabor A, Pergolizzi JV Jr, Marti G, Harmon J, Cohen B, Lequang JA. Raising awareness among healthcare providers about epidermolysis bullosa and advancing toward a cure. *J Clin Aesthet Dermatol.* 2017;10(5):36-48. 6. Montaudié H, Chiaverini C, Sbidian E, Charlesworth A, Lacour J-P. Inherited epidermolysis bullosa and squamous cell carcinoma: a systematic review of 117 cases. *Orphanet J Rare Dis.* 2016;11(1):117. doi:10.1186/s13023-016-0489-9 7. Dystrophic epidermolysis bullosa. Genetic and Rare Diseases Information Center. Accessed June 3, 2022. <https://rarediseases.info.nih.gov/diseases/2150/dystrophic-epidermolysis> 8. Bruckner AL, Losow M, Wisk J, et al. The challenges of living with and managing epidermolysis bullosa: insights from patients and caregivers. *Orphanet J Rare Dis.* 2020;15(1):1. doi:10.1186/s13023-019-1279-y 9. Has C, Liu L, Bolling MC, et al. Clinical practice guidelines for laboratory diagnosis of epidermolysis bullosa. *Br J Dermatol.* 2020;182(3):574-592. doi:10.1111/bjd.18128 10. Has C, Bauer JW, Bodemer C, et al. Consensus reclassification of inherited epidermolysis bullosa and other disorders with skin fragility. *Br J Dermatol.* 2020;183:614-627. doi:10.1111/bjd.18921 and Appendix S1. doi:10.1111/bjd.18921 11. Feinstein JA, Jambal P, Peoples K, et al. Assessment of the timing of milestone clinical events in patients with epidermolysis bullosa from North America. *JAMA Dermatol.* 2019;155(2):196-203.

Image sources: (Image 1): Reprinted from *Orphanet Journal of Rare Diseases*, 5, Fine JD. [Inherited epidermolysis bullosa](#). 2010;5(1):12:1-17. Copyright [2010]. Licensed under the [Creative Commons Attribution 2.0 Generic License](#). (Image 2): Reprinted with permission. (Image 3): Reprinted from *Journal of Tissue Viability*, 30, Wong T-W, Yang C-C, Hsu C-K, Liu C-H, Yu-Yun Lee J. Transplantation of autologous single hair units heals chronic wounds in autosomal recessive dystrophic epidermolysis bullosa: a proof-of-concept study. 2021;30(1):36-41. Copyright [2021], with permission from Elsevier. (Image 4): Reprinted from *JAAD International*, 2, Rogers CL, Gibson M, Kern JS, et al. A comparison study of outcome measures for epidermolysis bullosa: Epidermolysis Bullosa Disease Activity and Scarring Index (EBDASI) and the Instrument for Scoring Clinical Outcomes of Research for Epidermolysis Bullosa (iscorEB). 2021;2:134-152. Copyright [2021], with permission from Elsevier.

Decode DEB™ provides simple, no-charge genetic testing to eligible patients through GeneDx



Cost can often be a barrier to genetic testing, but the cost of a missed or delayed diagnosis is too high.

The Decode DEB genetic test panel was developed to identify genes associated with DEB or conditions with similar phenotypes to DEB to help provide an accurate diagnosis.

Conditions	Genes involved
Dystrophic EB (DEB)	<i>COL7A1</i>
EB Simplex (EBS)	<i>CD151, DST, EXPH5, KLHL24, KRT5, KRT14, PLEC</i>
Junctional EB (JEB)	<i>COL17A1, ITGA3, ITGA6, ITGB4, LAMA3, LAMB3, LAMC2</i>
Kindler EB (KEB)	<i>FERMT1 (KIND1)</i>
Peeling Skin Syndrome	<i>CDSN, CHST8, CSTA, SERPINB8, TGM5</i>
Palmoplantar Keratoderma	<i>DSG1, DSP, JUP</i>
Epidermolytic Ichthyosis and Ectodermal Dysplasia-Skin Fragility Syndrome	<i>KRT1, KRT10, PKP1</i>

Patients are eligible if they meet all 3 criteria:



Have symptoms consistent with EB



Have not had previous genetic testing for EB



Are residents of the United States or Puerto Rico

How Decode DEB™* works

1 For new accounts only

Set up a GeneDx account

Contact GeneDx and provide NPI:

- Phone: (888) 729-1206
- Email: support@GeneDx.com

GeneDx will send notification with an account number within approximately 2 days

2 Order sample collection kit and fill out Test Requisition Form (TRF)

Through Decode DEB™ by:

- Phone: (888) 729-1206
- Email: DecodeDEB@genedx.com
- Visit: www.genedx.com/decode-deb

3 Collect sample

- Collect a blood or buccal sample
- Label samples as directed

4 Send sample and completed TRF to GeneDx

- Send sample and completed TRF to GeneDx (prepaid shipping label will be in kit)
- TRF and sample do not have to be submitted together
- TRF can also be submitted electronically to DecodeDEB@genedx.com
- To avoid delays, ensure TRF is fully completed

5 Receive results

- Lab will email or fax results to ordering clinician within 4 weeks of receiving the sample and fully completed TRF

Know it's DEB with a simple, no-charge genetic test. Visit DEBFacts.com/hcp to learn more or call 1-844-5-KRYSTAL.



*The Decode DEB™ program was created to provide access to genetic testing for patients who have been diagnosed with or suspected of having epidermolysis bullosa (EB) and have not had genetic testing. Decode DEB™ is open to all US residents, including residents of Puerto Rico. Testing is available to eligible patients free of charge. No patients, healthcare professionals, or payers, including government payers, are billed for this program. Krystal Biotech does not receive any data that can identify individual patients. Krystal Biotech covers the cost of laboratory testing only—excludes office visits, copays, sample collection, and any other related costs. Krystal reserves the right to amend, suspend, or terminate this program without notice.