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## VIDEOS OF SURGICAL PROCEDURES IN DERMATOLOGY

# [Translated article] Skin Biopsy in a Patient With Suspected Epidermolysis Bullosa

## Biopsia de piel en un paciente con sospecha de epidermólisis ampollosa

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### Introduction

Epidermolysis bullosa (EB) comprises a group of genetic diseases characterized by extremely fragile skin. It is caused by mutations in genes coding for proteins in the epidermis, basement membrane, and/or dermis. The estimated worldwide incidence of EB ranges from 3.8 and 20.1 cases per million persons. While a genetic work-up is essential when attempting to confirm the diagnosis and the subtype of EB, the histology study is very useful during the initial approach to the patient, since, in addition to guiding the diagnosis, it makes it possible to rule out other, much more frequent diseases.

In order to perform an appropriate skin biopsy in a patient with suspected EB, several considerations must be taken into account ([supplementary material](#), [video](#)). It is important to be aware of such considerations, since the diagnostic usefulness of the sample will depend on them.<sup>1,2</sup>

### Description of the Technique

First, we must select a nonacral and blister-free area of skin. The area must be rubbed with a pencil eraser by rotating it 180 degrees on its axis from side to side. This should be done for at least 5–10 cycles until erythema appears, without producing clinically visible erosions or blisters. After about 30 minutes, samples can be taken following the usual approach in skin biopsy.

If possible, 3 skin samples should be obtained.

The first biopsy should be with a punch (4 mm), attempting to ensure that half of the sample contains erythematous skin after friction and the other half contains normal skin. The sample should be sent in formol for assessment with hematoxylin–eosin.

The second sample – for the immunofluorescence study – can be obtained using a punch measuring 3–4 mm. It should be sent in saline solution and frozen or in Michel transport medium. This sample should be sent as soon as possible to a reference center with experience in the diagnosis of EB, since standard panels do not contain all the antibodies necessary to diagnose this disease.

Lastly, a third, very small sample can be obtained with a punch measuring 2 mm and sent in glutaraldehyde for electron microscopy. This study makes it possible to determine

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the level at which the blister forms, as well as the presence or absence of structural proteins. Electron microscopy is more relevant when the genetic work-up and/or immunofluorescence do not yield conclusive results.<sup>1</sup>

### Indications

Patients with suspected EB.

### Complications

Excess rubbing with the eraser could generate a blister that would lead to a false positive. We should only rub until erythema appears, without damaging the skin where the biopsy specimen is taken from.

### Conclusions

EB comprises an uncommon group of diseases. Histology is a useful early diagnostic tool when assessing affected patients. It is important to use the correct approach in order to obtain samples that will prove useful to the pathologist during the diagnostic process.

### Conflicts of Interest

The authors declare that they have no conflicts of interest.

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### Appendix A. Supplementary Data

Supplementary data associated with this article can be found in the online version at [doi:10.1016/j.ad.2023.04.033](https://doi.org/10.1016/j.ad.2023.04.033).

### References

1. Lucky AW, Whalen J, Rowe S, Marathe KS, Gorell E. Diagnosis and care of the newborn with epidermolysis bullosa. *Neoreviews*. 2021;22:e438–51.
2. Hernández-Martín A, Torrelo A. Inherited epidermolysis bullosa: from diagnosis to reality. *Actas Dermosifiliogr*. 2010;101:495–505.