Natural History of Wounds in Patients with Recessive Dystrophic Epidermolysis Bullosa

Daniel C. Solis, BA; Jaron Nazaroff, BS; Yana Dutt-Singkh, MD; Sara Choi, BA; Melissa Barriga, MSc; Shufeng Li, MS; Vanessa Rangel-Miller, BA; M. Peter Marinkovich, MD; Jean Y. Tang MD, PhD

1. Stanford University School of Medicine, Department of Dermatology, Stanford, CA, USA
2. Invitae, San Francisco, CA
3. Veterans Affairs Medical Center, Palo Alto, California, USA

Corresponding author: Jean Y. Tang MD PhD, Department of Dermatology, Stanford University School of Medicine, 450 Broadway St, Pavilion C, 2nd Floor - MC5334, Redwood City, CA 94063. Phone: (650) 721-7190, Fax: (650) 721-3476, E-mail: tangy@stanford.edu

Understanding the natural history of wounds in patients with recessive dystrophic epidermolysis bullosa (RDEB) is essential for assessing efficacy of clinical trials. A cross-sectional study, utilizing patient questionnaires to evaluate chronic open wounds, defined as areas unhealed for ≥ 12 weeks and recurrent wounds, defined as areas that heal but easily open. A total of 1463 wounds were characterized in 128 RDEB patients; 52% pediatric, 51% male; 39% (155/393) of chronic open wounds were ≥ 40 cm² and 71% (63/89) remained unhealed for ≥ 4 years. In contrast, 92% (954/1033) of recurrent wounds healed in ≤ 3 weeks; 93% (101/109) opened again in ≤ 12 weeks; 49% (519/1057) of recurrent wounds were ≥ 20 cm² and 72% (74/103) of recurrent wounds were present for ≥ 4 years. On multivariate regression analysis, patients with anemia were 2.9 fold more likely to have larger recurrent wounds [odds ratio (OR) 2.89, 95% CI (1.01-8.26), P = 0.047]. Limitations include selection bias and recall bias. Chronic open wounds and recurrent wounds in RDEB patients are associated with significant morbidity and protracted time course. This natural history study sets a baseline from which to assess wound healing in future prospective clinical studies.

References