GENETICS AND GENODERMATOSES

INHERITED EPIDERMOLYSIS BULLOSA: DESCRIPTION OF CLINICAL AND SUBCLINICAL MORPHOLOGICAL FEATURES WITH OPTICAL COHERENCE TOMOGRAPHY

B De Pace [1] - C Fiorentini [1] - S Ciardo [1] - J Chester [1] - S Kaleci [1] - T Veltri [1] - M De Luca [2] - G Pellacani [1]

University Of Modena And Reggio Emilia, Dermatology Unit, Modena, Italy [1] - Center For Regenerative Medicine "stefano Ferrari", University Of Modena And Reggio Emilia, Department Of Life Sciences, Modena, Italy [2]

Introduction: Inherited epidermolysis bullosa (EB), is characterized by recurrent skin and mucosal blistering and are classified according to the level of blister cleavage, as EB simplex (EBS), junctional EB (JEB) and dystrophic EB (DEB). Punch biopsy is essential for the gold standard immunofluorescence antigen mapping (IFM) diagnosis. However multiple punch biopsies, which can cause iatrogenic trauma, are required in the current grafting follow-up procedures to detect eventual subclinical blisters, erosions or epidermal detachment. Optical coherence tomography (OCT), an in-vivo non-invasive imaging technique may guide biopsy location and assist in patient follow-up.

Objective: To identify morphological features and the level of cleavage for the different types of EB blisters evidenced by the optical coherence tomography (OCT), and to assess whether subclinical disease involvement in perilesional skin is revealed with OCT assessment.

Materials and Methods: A consecutive series of 13 patients with clinically evident EB were assessed with OCT between October 2017 and April 2018.

Results: A total of 17 blisters and 51 perilesional skin sites (at 1 cm and at 5 cm from the blister margins) were evaluated. EB blisters' levels of cleavage (intra or sub-epidermal), blister content, epidermal aspect and perilesional skin features were evidenced with OCT for all EB types. Subclinical blisters were evidenced in 41% (14/34) of the perilesional areas (all located 1cm from blister margin) and were not associated with blister type.

Conclusion: OCT allows the identification of morphological features, the level of cleavage and blister content of EB blisters in cross-sectional views. As OCT reveals in-vivo skin architectural changes induced by EB, the detection of a targeted donor site free from subclinical disease, required for gene therapy, may reduce inappropriate invasive biopsies

throughout the post-grafting follow-up.