



GENETICS AND GENODERMATOSES

IMMUNE CELL INFILTRATION IN CUTANEOUS SQUAMOUS CELL CARCINOMAS IN RECESSIVE DYSTROPHIC EPIDERMOLYSIS BULLOSA: COMPARISON WITH NON-RDEB CUTANEOUS SQUAMOUS CELL CARCINOMAS

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Background: Recessive dystrophic epidermolysis bullosa (RDEB) is a highly disabling genodermatosis characterized by skin and mucosal fragility and blistering.

Cutaneous squamous cell carcinoma (SCC) is one of the most devastating complications of Recessive Dystrophic Epidermolysis Bullosa (RDEB) with a high morbidity and mortality rate. Patients with RDEB were reported to have up to a 70-fold higher risk of developing SCC than that of unaffected individuals in the US. Immune cells play a role in cancer evolution.

Objective: The aim of our study is to evaluate immuno-histological difference between cSCC in RDEB with primary cSCC and secondary cSCC (developed by burn and radiotherapy scars). The immuno-histological evaluation was performed also in skin biopsies in RDEB non-neoplastic skin.

Materials and Methods: Consecutives biopsies of cSCC taken by 5 RDEB patients were analysed. As controls we analysed 5 primary cSCC, 5 secondary cSCC and 5 skin hyperplasia in RDEB.

Results: We found a significant reduction in immune infiltration in RDEB compared to controls. In particular we found a reduction in CD3+, CD4+, CD8+, CD20+, CD68+. No difference was found in size, histology, grading, number of mitosis and EGFR expression between the different groups.

Conclusions: Our data shows a reduction in immune cell peritumoral infiltration. Considering the well-known evolution of cSCC in RDEB as well as the youngest age at diagnosis, we could assume that immune dysfunction can lead the cSCC aggressivity in these patients.

