GENETICS AND GENODERMATOSES

RECESSIVE DYSTROPHIC EPIDERMOLYSIS BULLOSA AND BASAL CELL CARCINOMA: A CASE REPORT

R De Oliveira Boabaid [(1)] - N Conceição Fernandes [(1)]

Hospital Universitario Clementino Fraga Filho/universidade Federal Do Rio De Janeiro, Sector Of Dermatology, Rio De Janeiro, Brazil [(1)]

Background: Epidermolysis bullosa (EB) is a rare genetic blistering disorder which is characterized by skin fragility and recurrent blistering of the cutaneous and mucous membranes following minimal trauma. Recessive dystrophic epidermolysis bullosa (RDEB) is the most severe form of EB, caused by mutations in the collagen VII gene COL7A1. Patients with RDEB have up to a 50 fold increased incidence of cutaneous squamous cell carcinoma (SCC) which is highly aggressive with early metastatic spread. It occurs most commonly in chronic non healing ulcers over bony prominences on limbs. Basal cell carcinoma (BCC) is seen almost exclusively in epidermolysis bullosa simplex and arises in sun exposed skin sites in 100% of patients. The cumulative risk of BCC is arising in EB patients before age 60 and to this day there are no published data on the prevalence of BCC in EB or the risk of its development.

Observation: A 37-year-old male diagnosed as RDEB since infancy presented after age 20 two SCCs on chronic ulcers over bony prominences on limbs and three BCCs on sun exposed areas of arms and hand. The tumors were treated primarily by wide excision.

Key message: To present knowledge there is not yet a clear mechanism that triggers the development of SCC and BCC in EB cases. As the risk of BCC is quite low in RDEB subtype we can speculate that our pacient has not enough avoided sun exposure and traumas during his life-time.