

(age range 1.5-7.0 years median 7) a score was calculated range 0-70 median 35. EB subtypes included: 2 junctional EB, 3 EBS-generalised severe and 27 EB recessive dystrophic-generalised severe. A Score of ≥ 15 for growth section only and/or total score ≥ 40 were suggested to prompt further education and referral to surgeon *Conclusions:* The decision for gastrostomy placement can be a difficult one for health professionals to guide on and families to make. Few resources are available to assist with this. We believe annual assessment with this tool can contribute to discussions and potentially guide optimal time of insertion at EB centres where this procedure is supported. Further validation is planned to assess use as predictor of optimal time for gastrostomy referral, considering regional factors such as waiting-list times.

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RDEB-CSCC DERIVED INDUCED PLURIPOTENT CANCER CELLS FOR STUDYING CANCER INITIATION AND PROGRESSION

Joanna Jacków, Ryota Hayashi, Avina Rami, Zongyou Guo and Angela M. Christiano

Department of Dermatology, Columbia University, New York, NY

Introduction & objectives: The early onset of aggressive cutaneous squamous cell carcinoma (cSCC) and its rapid progression in recessive dystrophic epidermolysis bullosa (RDEB) leads to high mortality rates in these patients. Reprogramming differentiated cancer cells into induced pluripotent cancer cells (iPCCs) is a novel tool used to investigate cancer progression and understand the epigenetic transformation process during tumorigenesis. We hypothesized that if cSCC cells were converted to pluripotency and then allowed to differentiate back into cSCCs, they would undergo changes reminiscent with the stages of early-onset cancer. Due to the genetic and epigenetic barriers in cancer cells, the reprogramming efficiency is very low. *Materials & methods:* In this study, we used a humanized version of the single polycistronic lentiviral (LV) vector to reduce the number of viral integrations in cSCCs derived from RDEB patients to generate iPCCs from cSCCs. *Results:* Successful generation of iPCCs from RDEB-cSCC after infection with a single LV vector was demonstrated by epithelial-like morphology, colony-forming efficiency, the expression of pluripotency markers such as Sox2, Oct-4, SSEA-3 and TRA-1-60, and the ability of the cells to differentiate *in vitro*. Moreover, TaqMan assays of human stem cell pluripotency gene signatures of RDEB-cSCC-iPCCs were similar to WT-iPCCs generated from normal human fibroblasts demonstrating that the RDEB-cSCC-iPCCs correlated with the stem cell state. The RDEB-cSCC-iPCC undergo differentiation into RDEB-cSCC -cancer stem cells (CSC) and will be used to generate 3D skin constructs (SCs) and grafted onto immunocompetent mice. After tumors develop, they will be characterized and used as a model to test ruxolitinib for its *in vivo* activity to reverse the growth of a tumors formed by RDEB-cSCC-iPCCs. *Conclusions:* Taken together, the RDEB-cSCC-iPCC tumor model offers a valuable tool to study early RDEB-cSCC development and progression, as well as the development of novel therapeutic approaches

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PSYCHOSOCIAL ASPECTS OF EPIDERMOLYSIS BULLOSA AND QUALITY OF LIFE. A SYSTEMATIC REVIEW

Gudrun Salamon, Alexander Ruberl and Laura Maar

Sigmund Freud University, Vienna, Faculty of Psychology

Introduction & objectives: Epidermolysis bullosa (EB) is a group of rare diseases characterized by skin fragility. As indicated in previous research, EB has an enormous impact on all aspects of psychosocial life. Hence, quality of life is an important measure

in psychological as well as in clinical research. However, quality of life is highly influenced by individual perception. The same applies to the concept of wellbeing, which describes the individual balance between personal resources and challenges faced. Coping strategies are an individual's mental or behavioural response options to stress and to challenging situations. In EB, stress is caused by chronic pain, the need of intensive wound management and restrictions in everyday life. Additionally, these restrictions lead to social and emotional challenges. *Materials & methods:* In order to address all psychosocial aspects of EB and corresponding coping strategies thoroughly, we conducted a systematic review of literature. A set of exclusion and inclusion criteria led to a final selection of 38 papers presenting original work, comprising quantitative, qualitative, mixed methods and single case studies with the focus on EB patients or their families. *Results:* The following psychological aspects were described as helpful on an individual coping level: Control of life is associated with autonomy, which can be increased by active EB management and by the help of external support (like care assistance) when necessary; Containing the impact of EB relies on strategies for dealing with pain and for emotion regulation and is positively correlated with self-esteem and self-efficacy. The validation of the expertise for the own or a family member's EB case based on specific knowledge and personal experience in, e.g., helpful routines for wound care, is highly appreciated; Interaction with others is a great resource and easier when taking an active role in the exchange with health care professionals, with healthy people, with people in a similar/comparable condition, and with people with EB. Strategies for communicating the personal health situation are helpful in order to enhance knowledge and understanding of others; The attitude to live a close-to-normal life or at least close-to-normal moments is linked to decisions in favour of physical or social activities in spite of pain. It needs situational impact assessment and flexible decision-making. *Conclusions:* Quality of life and wellbeing are highly individual and vary widely within people living with EB. Our systematic review identifies and explores psychosocial aspects of EB and their corresponding coping strategies.

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ANTERIOR SEGMENT SPECTRAL DOMAIN OPTICAL COHERENCE TOMOGRAPHY IN EPIDERMOLYSIS BULLOSA

Nihaal Mehta, BA, Calvin Robbins, BA, Elizabeth Noh, BS, Nadia K. Waheed, MD, MPH, Vicki M. Chen, MD

Sigillum Universitatis Tuftensis Pax Ethux 1852, Children's Glaucoma foundation, Massachusetts Lions Eye Research Fund Inc, Research to Prevent Blindness, Floating Hospital for Children at Tufts Medical Centre

Introduction & objectives: As therapies for DEB make progress in skin, there remains a great distance between skin application and readiness for ophthalmic use. Unlike skin, the cornea is not easily biopsied, which limits objective assessment of disease modification in response to therapy. Anterior segment optical coherence tomography (AS-OCT) provides high-resolution, depth-resolved, cross-sectional and three-dimensional imaging of the cornea. We developed a non-invasive tool to assess EB-associated corneal pathology. *Materials & methods:* Patients were recruited during the 3-day long DEBRA care conference in Phoenix Arizona, USA in 2018. A 10-item "vision and abrasion symptoms" questionnaire was given to all subjects. Visual acuity (VA) was tested monocularly. AS-OCT images were acquired by a trained ophthalmic photographer on the Optovue RTVue-XR Avanti SD-OCT system (Optovue, Inc., Fremont, CA, USA). We examined the ability of corneal AS-OCT imaging to visualize EB-associated corneal lesions, as well as associations between OCT findings and clinical metrics including visual acuity, abrasion frequency,