Towards a minimally invasive method for diagnosing aggressive SCCs in 'butterfly disease'.

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Background & objectives

Death from malignant squamous cell carcinoma (SCC) is increasingly becoming a health problem world-wide. In those patients with underlying health issues, treatment and diagnosis can be particularly problematic. 'Butterfly disease' [aka Epidermolysis bullosa (EB)] is a rare genetic condition, linked to defects in the extracellular matrix. A particularly, debilitating form of the disease, Recessive Dystrophic EB (RDEB) is linked to mutations in collagen (COLVII) and is associated with susceptibility to wounding, infections, as well as a range of myocardial disorders. If patients live long enough, they develop an aggressive and lethal form of SCC. In addition, to the lack of agreed upon molecular markers of malignant SCCs, the high levels of wounding and risk of infection for EB patients makes regular biopsy collection, tumor monitoring and assessment of the efficacy of any treatment challenging.

Methods

In collaboration with the Wally Lab, EB House, Salzburg, Austria, the team from the Mellick Lab, Ingham Institute for Applied Medical Research, University of New South Wales, Australia have been working towards identifying markers of malignant SCCs, and adapting current filtration based circulating tumor cell (CTC) isolation protocols (e.g. ScreenCell® Cyto Kit) that can be applied to the development of a minimally invasive tool for tracking early cancer spread, and clinical course in EB patients.

Results

We have identified several novel robust markers of malignant SCCs, including small noncoding RNAs (e.g. miR-10b), and successfully adapted previously developed methods in combined *in situ hybridization (ISH)*-immunohistochemistry (IHC) and CTC filtration/isolation protocols to identify putative circulating tumor cells (CTCs) in the blood of cancer patients.

Conclusion

While the use of the liquid biopsy in cancer treatment has great potential to enhance patient care, there remains a need for the continued development of diagnostic tools that can be applied to ‘at risk’ groups with specific needs, such as EB patients. We believe that our work has begun to reveal the potential of CTC analysis to address these needs, with the broader aim of making the liquid biopsy more accessible to clinicians and patients at point of care.