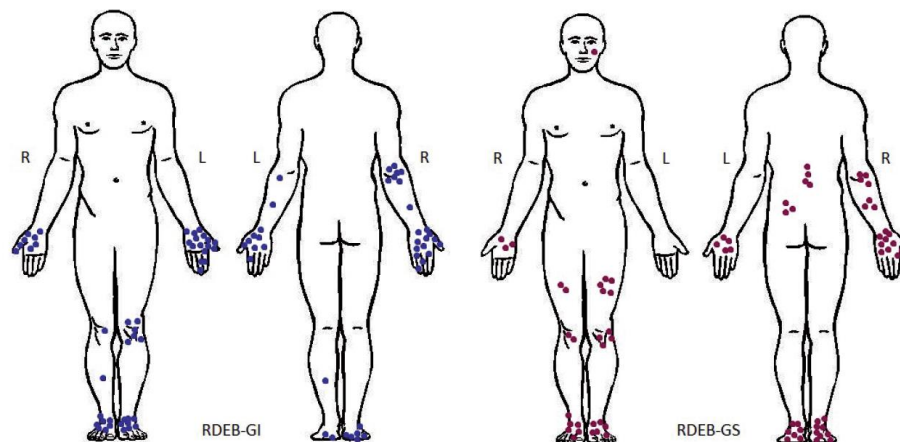


Cancer in Epidermolysis Bullosa

What is it? The most frequent cancer that affects patients with epidermolysis bullosa (EB) is called Squamous Cell Carcinoma. This cancer is so called because it occurs in tissues that are said to be “squamous”, or more plainly put, tissues that form a barrier between an individual and their environment such as the skin. This cancer can also occur in other parts of the body such as the mouth, the esophagus, the upper airways of the lung, and the cervix, but cancer does not usually affect these parts of the body in EB.

Who is at risk? Patients with recessive dystrophic EB (RDEB) are at most risk of developing squamous cell carcinoma, or SCC for short. There are also reports of increased risk of SCC in patients with junctional EB and Kindler.

Where are they found? SCC are found in the skin of EB patients and most frequently on the hands and feet. The diagram below is taken from a study from Australia showing where 161 SCC have occurred in 17 RDEB patients.



Distribution of 161 primary SCC in 17 RDEB patients from Australia (reproduced from Kim et al., 2018. *Acta Derm Venereol* 98: 70-76). RDEB-GI = generalized intermediate, RDEB-GS = generalized severe.

What are the treatment options? Current guidelines published in 2015 report that surgery is the main treatment option. Radiotherapy or chemotherapy are also available but the location of the cancer and the well-being of the patient are important to consider when thinking about these options. In September 2018 the drug Libtayo (also known as cemiplimab) was approved for the treatment of advanced squamous cell carcinoma of the skin and can be used to treat SCC in EB patients. It is reported that one patient with RDEB SCC has shown a good response to Libtayo.

What is the long term prospect? Unfortunately SCC in patients with RDEB are very aggressive and long term prospects, measured by 5-year survival, are not very good. However, there are exceptions and some RDEB patients with cancer do live longer. For all types of cancer, EB and non EB, early detection improves survival.

What are new treatment options in development or clinical trials? Currently there is a clinical trial running in Philadelphia and in Europe to test an experimental drug called “Rigosertib”. Rigosertib is a targeted therapy, not a chemotherapy, and has a very mild side effect profile. One patient has completed the trial in Europe and remains cancer free. The drug is available in pill or infusion form. Patients with RDEB who have SCC that has not responded to standard treatment such as surgery may contact neda.nikbakht@jefferson.edu to inquire about enrollment.