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Short Communication

^{18}F-FDG PET in a Patient with Epidermolysis Bullosa and Multifocal Squamous Cell Carcinoma

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Epidermolysis Bullosa (EB) has been assigned to a heterogenous group of rare chronic hereditary diseases usually observed at birth. It is characterized by extremely fragile skin and mucous membranes resulting in blistering and subsequent scarring following minor trauma [1]. The incidence ratio of individuals with EB in the overall population ranges from 1:50,000 to 1:500,000, depending on the type of inheritance mode (autosomal dominant or recessive forms) and the severity of the disorder. Mild forms of the illness are more frequently observed than severe forms, with both sexes being equally affected [2,3].

Cutaneous Squamous Cell Carcinoma (SCC) is the most common malignancy seen in Recessive Dystrophic Epidermolysis Bullosa (RDEB), which is one of the severe forms of EB presenting at birth. The risk of carcinoma increases with age and starts in teenage years [4]. Assessment for SCC in patients with EB can be difficult, because the appearances are often similar to the typical chronic ulceration, scarring and crusting widely seen in RDEB. Therefore, there should be a low threshold to biopsy chronic nonhealing ulcers and hyperkeratotic nodules or plaques.

We present a 33-year-old lady with congenital dystrophic epidermolysis bullosa who developed multifocal squamous cell carcinomas leading to an above-elbow-amputation of her left upper extremity. She was admitted to our institution for small bowel obstruction and bacteremia. A month later an ^{18}F-FDG PET CT was performed to evaluate the extent of SCC and to determine further management. The PET scan showed multifocal cutaneous and subcutaneous FDG-avid lesions with pulmonary nodules, left subpectoral, axillary, and internal mammary lymphadenopathy which were highly worrisome for metastatic deposits. 28 days after her PET CT, the patient died in our institution while she was switched to hospice/supportive care.

Clinical evaluation of SCC may be challenging in patients with EB because of the underlying skin conditions and manifestations. As in our patient's case ^{18}F-FDG PET CT was used to define the areas at highest risk to be cancerous in nature as well as the extent of disease. It not only depicted the areas of neoplastic involvement, but also determined the significant depth of infiltration of the disease in certain areas that were not distinguishable through clinical exam for example left breast, left axilla, and chest wall. It also provided a good staging assessment.

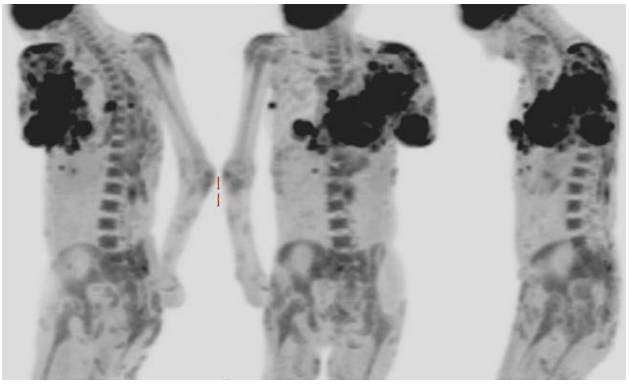

Figure 1: ^{18}F-FDG PET scan images showing intense uptake in several cutaneous lesions in the chest and the remainder of the body as well as deep infiltration of the entire subcutaneous tissues.

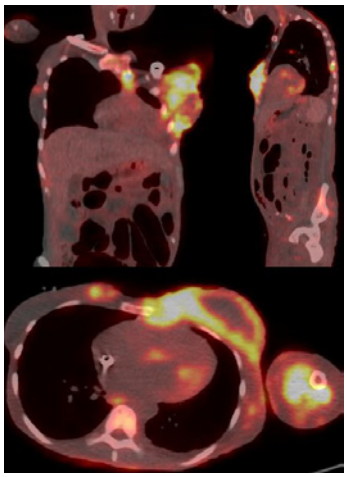

Figure 2: Fusion ^{18}F-FDG PET-CT images in coronal, sagittal and axial views respectively showing the deep subcutaneous and muscular tumor infiltration.

Conclusion

Epidermolysis bullosa is a rare chronic disease involving the skin and mucosal membranes with an increased lifetime risk of developing skin cancer. It is essential to diagnose neoplastic transformation in order to start appropriate treatment in a timely manner. ^{18}F-FDG PET scans if used judiciously and early enough may play a role in screening patients with highly suspicious skin lesions or an already established diagnosis of SCC. It may also help in detecting locoregional and distant extent of disease in the aggressive forms of this pathology.

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