Deep Learning-Aided Diagnosis of Autoimmune Blistering Diseases

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Autoimmune blistering diseases (AIBDs) are rare, chronic disorders of the skin and mucous membranes, with a broad set of clinical manifestations. Considering that 1) diagnosis of AIBDs is a challenging task, owing to their rarity and heterogeneous clinical features, and 2) misdiagnoses are common, and the resulting diagnostic delay is a major factor in a high mortality rate, prognosis stands to benefit greatly from the development of a computer-aided diagnostic tool for AIBDs. Artificial intelligence (AI) research into rare skin diseases like AIBDs is severely underrepresented, due to a variety of factors, foremost sparse, variable imaging data. A study by Julia S. et al. finds that, as of 2020, there exists no machine learning studies on rare skin diseases, despite the demonstrated success of AI in the field of dermatology. Whereas previous research has primarily looked to improve performance through extensive data collection, this approach remains tedious and impractical for rarer, under-documented skin diseases. This study proposes a novel approach in the development of a deep learning based diagnostic aid for AIBDs. Leveraging visual similarities with pre-existing repositories, we demonstrate classification of AIBDs using techniques such as transfer learning and data augmentation over a convolutional neural network (CNN), using a three-loop training process that combines feature extraction and fine-tuning. Our final model retains an accuracy nearly on par with dermatologists' diagnostic accuracy on more common skin cancers. Given our model's efficacy despite low amounts of training data, this approach holds the potential to benefit clinical diagnoses of AIBDs. Furthermore, our approach can be extrapolated to the diagnosis of other clinically similar rare diseases.

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