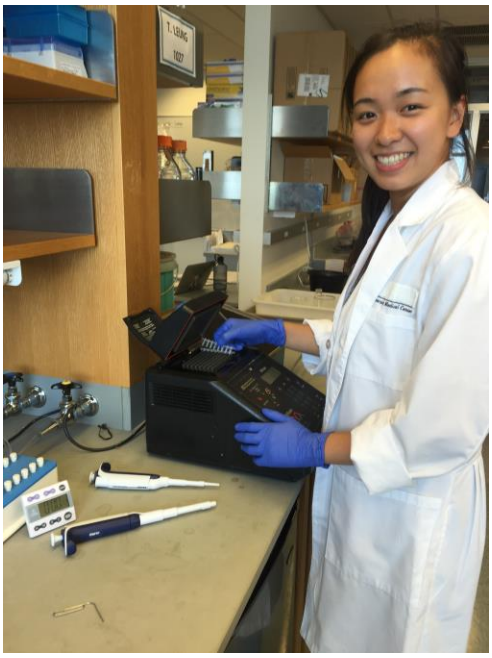

Investigating Adermatoglyphia and the Importance of SMARCAD1 Protein in Skin
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Advisor: Thomas Leung

The Leung Laboratory in the Department of Dermatology at the Perelman School of Medicine focuses its research on mammalian tissue regeneration and the mechanisms behind wound healing in mice and humans. This past summer, I worked with Dr. Thomas Leung on a project that investigated an extremely rare genetic disorder called adermatoglyphia, which results in the absence of fingerprints in people affected by it with no other known side effects. It is caused by a point mutation in the DNA that disrupts the SMARCAD1 gene, and thereby inhibits the expression of SMARCAD1 protein. My goal was to compare tissue grown with SMARCAD1 knockdown keratinocytes and wild type keratinocytes in an attempt to learn more about the characteristics of this little known disorder.

To accomplish this, I started by transfecting 293T cells to begin lentivirus production, which I then used to infect wild type keratinocytes. This process aims to create keratinocytes that are unable to produce SMARCAD1 protein, just like those in people affected by adermatoglyphia. I verified that I had indeed produced SMARCAD1 knockdown keratinocytes through two methods. First, after my transfection of 293T cells, I checked GFP (green fluorescent protein) expression in my control sample using fluorescent microscopy; if the control cells fluoresced, I would know that my transfection worked in my experimental samples as well. Second, I ran a western blot to ensure that no band came up at 117 kDa, the molecular weight of SMARCAD1 protein. After I was able to verify that I had successfully produced SMARCAD1 knockdown keratinocytes, I grew three-dimensional organotypic cultures by seeding both knockdown keratinocytes and wild type keratinocytes (as a control and basis of comparison) on acellular matrices. After one week, I harvested these samples and submitted them to be stained and turned into slides so

that I can look at them under a microscope and examine the differences between SMARCAD1 knockdown keratinocyte tissue and wild type keratinocyte tissue.

This summer research experience has given me a truly immersive experience in the world of research, a fundamental part of every scientist and engineer's training. I learned how to perform transfections, western blots, and various important tissue culture techniques as well as how to create and maintain organotypic cultures, all of which are skills that will prove indispensible to my future in research and medicine. Although this project is not yet complete, I look forward to continue working with Dr. Leung during the academic year.