

Child Health Research CIO

CHILD HEALTH RESEARCH CHARITABLE INCORPORATED ORGANISATION (CHR CIO) PROGRESS FORM – VACATION STUDENT - 2018

Student's name:	Emily Conibear
Primary supervisor:	Dr Anastasia Petrova
Subsidiary supervisor:	Prof Waseem Qasim
Project title(s):	CRISPR/Cas9 gene editing for Recessive Dystrophic Epidermolysis Bullosa

Summary

What are you trying to do in this studentship?

Collagen 7 is the key protein secreted by skin cells involved in anchoring the different skin layers together. Without a functioning copy of collagen, the skin loses its integrity and individuals suffer from dystrophic epidermolysis bullosa (DEB). There are numerous hotspots for the mutations and the particular mutation I am looking at is where a C base has been replaced with a T. This change prematurely stops collagen 7 production, creating a dysfunctional protein. My aim is to use a novel gene engineering tool, xCAS ABE base editor, to change the T back to a C, allowing the correct function of collagen. The base editor will be used to treat a patient's cells in a guided fashion.

Why is this research important?

DEB is a debilitating skin disease caused by mutations in collagen 7. Although it is rare, there is currently no cure and affected individuals suffer greatly. Symptoms include severe blistering from loss of skin integrity leading to non-healing wounds and scars that affect simple day-to-day tasks. Moreover, the risk of developing fatal skin cancer is abnormally high. Not only are the symptoms devastating, the costs of symptomatic treatments are extortionate due to the vast number of bandages needed by sufferers daily.

Value of Your Experience

My experience in this studentship has been incredible as I have learnt a huge array of laboratory skills that I never would have whilst being at university. Being able to actually perform the different techniques and apply them to real life cutting edge research allowed me to understand the principles behind the techniques a lot better, whilst intensifying my passion for research. Not only did I learn the skills necessary for my project, I was also exposed to a number of techniques from other people's projects which allowed me to widen my knowledge even further.

I would personally like to give a huge thanks to Waseem's III team at ICH, with a special mention to Anastasia Petrova and Gaetano Naso, for guiding me through my studentship. They were very patient, explained things extremely well and the whole process is an experience I will not forget. I would definitely recommend this studentship to anyone with a love for science.