CHILD HEALTH RESEARCH CHARITABLE INCORPORATED ORGANISATION (CHR CIO)
PROGRESS FORM – VACATION STUDENTS

Student's name:	Samantha Wallworth
Academic Programme:	Developmental Biology & Cancer Programme
Project title(s):	Characterising a novel mouse model of Non-Ketotic Hyperglycinemia.

1. Lay Summary

What are you trying to do in this studentship?

This studentship focussed on an inherited metabolic disease called Non-Ketotic Hyperglycinemia (NKH) in which patients suffer severe neurological problems, including epilepsy. NKH is caused by mutations in the gene that codes for glycine decarboxylase (GLDC) which is a protein that acts to metabolise a small molecule, glycine. A hallmark of NKH is unusually high concentrations of glycine in the body, since a lack of GLDC permits the build-up of excess glycine.

I am studying mice which carry a genetic mutation that changes the mouse version of GLDC and models a mutation that is found in NKH patients. I am studying the brain to ask whether there are changes in the structure or cellular composition. In parallel, I am asking whether the specific protein change caused by the GLDC mutation results in a less stable protein.

Why is this research important?

NKH is a debilitating paediatric disorder, in which affected infants experience several devastating and progressive symptoms including seizures, hypotonia, cognitive impairment, and difficulty breathing. As high as 30% of neonates with severe NKH die shortly after birth. Therefore, it is vital that a comprehensive mouse model is characterised in order to investigate and understand NKH in humans at a deeper level, to eventually optimise the clinical outcome for diagnosed patients.

2. Value of Your Experience

Please comment on the value of your experience undertaking this CHR CIO vacation studentship.

While this was my first experience in a real working laboratory, it has been of immense value. I have developed countless practical skills of which I may not have been afforded the opportunity to do so at university, and been introduced to several new techniques too. Being able to put everything I have learnt on my degree programme into practice has been invaluable in giving me incredible insight into a career in research at such frontiers alongside a group of dedicated academics.

Additionally, this experience has shown me how creative and flexible a career in science can be. The fact that there are often so many answers to one problem allows you to think outside the box and develop your own solutions. This concept has instilled a huge amount of confidence within myself, and my ability in the laboratory.

I would like to personally thank Nick Greene's group at ICH, especially Sandra Castro who has been a huge part of my whole experience. Her patience and fantastic ability to explain complex concepts have been absolutely instrumental in my learning.