

Child Health Research CIO

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

Student's name:	Louise Mathilde Bresson
Academic Programme:	Genetics & Genomic Medicine
Project title(s):	Testing the potential efficacy of translational read-through inducing drugs for mitochondrial diseases

1. Lay Summary

What are you trying to do in this studentship?

To fulfil its role as the powerhouse of the cell the mitochondria needs to import large amounts of protein produced by molecular machines called ribosomes. Ribosomes synthesise proteins from a template called mRNA which is a temporary copy of the DNA. However, mutations in the DNA, which carries on as mutations in the mRNA, can cause the ribosome to stop pre-emptively and create malfunctioning proteins. I am trying to see if read-through-promoting drugs, which should allow the ribosome to ignore the mutations, can help increase the concentration of functioning protein in the mitochondria.

Why is this research important?

Mitochondrial disorders are a very heterogeneous group of disorders thought to affect around 1:5000 of the population. A subset of these disorders involves early-onset severe forms of mitochondrial diseases most commonly due to nuclear mutations with a high mortality and morbidity rate. Children affected by these disorders often suffer from seizures, poor growth and cognitive disabilities. As there is no cure for mitochondrial diseases and symptomatic treatment is often limited, research into novel therapies is of utmost importance.

2. Value of Your Experience

The studentship feels like an authentic introduction into what a research career is like and I would therefore recommend it to anyone considering taking on a PhD. Working with the mitochondrial research unit, I got to experience laboratory work, scientific writing and the clinical importance of mitochondrial research.

The lab work involved several techniques that were new to me as well as the chance to hone my skills with more familiar techniques. I practised many transferrable skills as much of the laboratory work consisted of very standard procedures including cell culture, cell pellet isolation, RNA and DNA extraction, qPCR and western blotting. Working independently in the lab also gave me the chance to improve my ability to follow written protocols and the chance to improve my techniques through trial-and-error.

The mitochondrial unit made me feel like an integrated member of their research group, not just expecting me to show up 9 to 5 and do the days' tasks. In addition to my research, I got the opportunity to attend the Lily Foundation family weekend to meet the families affected by mitochondrial diseases, attended a scientific conference, and had the chance to write a review article for the group.