Nigel Clarke Bursary Award - 2021

By Shuxiang Goh

Thank you for the generous bursary in consecutive years. 2020 and 2021 has been unique years for travel due to COVID. Nonetheless, the bursary has allowed me to present my research findings, via online platforms, at national and international conferences (HGSA, ESHG, ASHG), as well as to undertake a PhD in this topic.

My topic revolves around the mathematics of estimating penetrance for neurosusceptibility loci. With the encouragement of awards such as this, I have been able to improve upon the standard formula significantly to provide (preliminary) results for more accurate penetrance estimates. Some neurosusceptiblity loci have revised penetrance estimates that are either 0% or close to 0%, which is a novel finding.

An accurate formula for penetrance also enables the conversion of odds ratios to penetrance. This has not previously been possible because a correct formula for penetrance had not been published. Converting odds ratios to penetrance has a wide range of applications, as penetrance is more clinically-intuitive. Preliminary results show that for some conditions with odds ratios as high as 20 (or even 100), penetrance can still be <1%. I am hoping that I will be able to develop ways to help clinicians make sense of odds ratios and to encourage future publications using the concept of penetrance (where appropriate) in preference to odds ratios.

The next extension would be in polygenic risk, which relies on combining multiple odds ratios in various ways, and seeing if these can be converted into penetrance. Current methods employ complex (and sometimes conflicting or different) statistical techniques to do so and it would be interesting to see if the method I have developed, which is mathematically simpler, can lead to similar results.

I once again thank the Board members of this bursary for the honour of this award, the confidence it provides for young researchers and for helping kickstart my career in this direction.

Yours sincerely,

Shuxiang Goh