ASHI MEMBER SPOTLIGHT

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How long have you been an ASHI member? 4 years

When and where was your first ASHI meeting? My first ASHI meeting was the 2019 ASHI/BANFF Joint Scientific Meeting in Pittsburgh. I was pleasantly surprised that our poster "A novel algorithm for Killer-cell immunoglobulin-like receptor (KIR) copy number imputation by KIR imputation tool (KIBAG) and consolidation of population specific KIR references in HLA & KIR imputation network (HKimpNet)" received the President's Choice award.

What made you decide to work in HLA? I have been trained as a genetic specialist and have been involved in several Genome-Wide Association Studies (GWAS) under the supervision of Prof. Katsushi Tokunaga. The first GWAS study that I was involved in was narcolepsy with cataplexy in which almost 100% of the Japanese patients carry HLA-DRB1*15:01-DQB1*06:02 haplotype; this is shown in the Manhattan plot of the GWAS as a very strong peak in the HLA region. At the time, no method was available to convert the SNP markers in the GWAS to HLA alleles association.

As a PhD student, I have decided to focus my research on converting the SNPs markers to HLA alleles by evaluating available tools and improving the references for specific populations. It turned out to be a success and researchers around the world are performing HLA imputation as part of the routine of GWAS analysis. Next, we will focus our attention on another immune polymorphic gene, KIR, and developed a highly accurate KIR imputation system.

We are also actively building an HKimpNet where we provide computational resources and imputation references for free to the research communities. Besides that, our team is also working to promote the use of NGS HLA genotyping by working closely with the IMGT to register novel alleles detected in the Japanese populations.

Most rewarding aspect: The HLA & KIR imputation tools (HIBAG&KIBAG) that we developed have helped to identify novel HLA alleles that are associated with diseases and reducing the cost of HLA genotyping of all the subjects involved in these studies. We are still working to improve the accuracy of our imputation system to the level of possible use in clinical settings.

Fun fact: I enjoy outdoor activities and hopefully, after the COVID-19 pandemic, we can still enjoy nature as we used to.