Conference Abstract Submission for Biostatistics Research Day

Phenome-Wide Association Study to Determine the Effects of Cystic Fibrosis Modifier Genes in the UK Biobank Population.

Authors: Faizan Khalid Mohsin[1]; Lisa Strug[1,2], Ph.D; Naim Panjwani[2], M.Sc; and Zeynep Baskurt[2], Ph.D.

[1]Division of Biostatistics, Dalla Lana School of Public Health, University of Toronto, Toronto, ON.

[2]Program in Genetics and Genome Biology, Research Institute, The Hospital for Sick Children, Toronto, ON.

Objective: Our object is to determine the impact of having gene modifiers that increase severity of Cystic Fibrosis in people who do not have Cystic Fibrosis. We looked at three particular SNPs annotated to three genes of interest: SNP rs4077468 at SLC26A9 (Chromosome 1); SNP rs3788766 at SLC6A14 (Chromosome X); and SNP rs57221529 at SLC9A3 (Chromosome 5). **Method**: We used the UK Biobank data which has over 500,000 participants to conduct the study. After the QC steps approximately 264,000 unrelated individuals remained. We performed a PheWAS to find the associations between the three SNPs and disease phenotypes. **Results and Conclusion**: In the UKBiobank population we found that individuals with allele C at SNP rs57221529 of the gene SLC9A3 are associated with having 6.4% higher probability of developing Esophagitis, GERD and related disease (OR = 1.064, S.E. = 0.013, P-value = 1.79E-06, Cases = 19,687, Controls = 243,236). Further, males with allele G at SNP rs3788766 annotated to the gene SLC6A14 were associated with having 68% higher probability of developing Urinary Obstruction (OR = 1.68, S.E. = 0.127, P-value = 4.24E-05, Cases = 64, Controls = 117,334). No other statistically significant associations were found.

Acknowledgement: This research has been conducted using the UK Biobank Resource under Application Number 40946.