# Abstract Submission for Biostatistics Research Day

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

Authors: Faizan Khalid Mohsin1; Professor Lisa Strug1,2, Ph.D; Naim Panjwani2, M.Sc; and Zeynep Baskurt2, Ph.D.

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

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

**Objective**: Our object is to determine the impact of having the gene variants that increase severity of the Cystic Fibrosis disease in people who do not have Cystic Fibrosis. We looked at three particular SNP’s for three genes of interest: SNP rs4077468 – gene SLC26A9 (Chromosome 1); SNP rs3788766 – gene SLC6A14 (Chromosome X); and SNP rs57221529 – gene SLC9A3 (Chromosome 5). **Method**: We used the UKBiobank data which has over 500,000 registered individuals 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 SNP’s 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 of 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.