# Oral Poster Presentation

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

Hello Everyone! My name is Faizan Khalid Mohsin. Today, I will be talking about my research at The Hospital for Sick Children supervised by Lisa Strug. My research was on Cystic Fibrosis which is a genetic disease that affects lung function as well as the digestive system. However, the severity of Cystic Fibrosis is affected by variation in present in certain genes. These genes are called modifier genes. My research focused on studying 3 SNPs of three different modifier genes. In particular, we focused on how these modifier genes affect the general population, that is, people who do not have cystic fibrosis (; in other words, how modifier genes are associated with any other diseases in a cystic fibrosis free population).

To conduct this study, we used the UK Biobank dataset which has over 500,000 participants from all over the UK. It has their genetic data as well as their health records. We tested over 1,500 diseases using phenome-wide association and, after correcting for multiple testing, found two statistically significant associations. First, we found that a particular variation in one of the modifier genes was associated with 6.4% higher probability of having Esophagitis, acid reflex, and other related diseases. Secondly, we found that, in men, another modifier gene for a particular variation, that is for a certain SNP, was associated with 68% higher probability of having Urinary Obstruction disease.

In summary, we studied the affect of cystic fibrosis modifier genes on the general public at large who do not have cystic fibrosis. Further, we found that the variations of two modifier genes were associated with higher probability of developing Esophagitis, acid reflex, and urinary obstruction disease. In conclusion, our research can help identify causes of disease in the general population and can be used to further understand modifier genes and what implications they have on the health of the general public.

My name is Faizan Khalid Mohsin and thank you for listening.