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| Project Reference | T1/65 |
| Project Title | **Understanding the role of mucin genetic variation in lung disease** |
| Theme(s) | Theme 1: Genomics for drug development & pharmacogenetics |
| Supervisors | **Prof Ed Hollox (University of Leicester)** [**ejh33@leicester.ac.uk**](mailto:ejh33@leicester.ac.uk)  Prof Matthew Loose (University of Nottingham)  Dr Katherine Fawcett (University of Leicester) |
| Department | Genetics and Genome Biology |
| Project Summary | Understanding how genetic variation contributes to lung diseases is important in understanding how the disease develops and identifying potential molecular targets for therapy. It is known that genetic variation of mucin genes (which encode proteins that form part of the mucus) are important in determining the risk to lung diseases such as COVID-19 severity, idiopathic pulmonary fibrosis, chronic obstructive pulmonary disease and asthma. How this genetic variation leads to increased disease risk is not known.  Mucin genes have a variable number tandem repeat which encodes a very variable length of the protein. This genetic variation has been very difficult to measure until now, thanks to long read sequencing using nanopore technology. This project will involve a supervisory team of Dr Hollox, an expert in structural genomic variation such as tandem repeats, Prof Loose, a pioneer of nanopore sequencing, and Dr Fawcett, an expert in the genetics of lung disease. By using new long read sequencing of genomes, this project will determine and characterise the variation at several disease-associated mucin genes and investigate the role of that variation in lung disease. |