MEDICAL STUDENT PRIZE WINNER:

# ANEESH SHARMA

**PROJECT:** Determining the Association Between SNPs in the Disease Modifier Genes PCSK7, PNPLA3, TM6SF2 and MBOAT7 and the Development of Significant Liver Injury in Hereditary Haemochromatosis

*'It is a great honour for me to have been chosen as one of the winners of the Dr Falk Core Medical Student prize. Over the past year I have learnt a great deal about the importance of scientific research in medicine and the opportunities available to medical students who wish to also pursue a career in research. I now count myself as one of these students and winning this prize has only strengthened my desire to follow such a path.*

*'Holding such a prestigious award will be highly beneficial when it comes to applying for further research-based opportunities, particularly in the field of gastroenterology which is an area that I have become increasingly interested in.*

*'Finally, this award is a testament to the brilliant support and guidance I have been fortunate enough to receive throughout my iBSc research year.'*

*Aneesh Sharma has just completed an intercalated BSc in Clinical Sciences at University College London Institute of Molecular Psychiatry. He will return to take up his 4th year medical studies at UCL in September.*

Hereditary Haemochromatosis (HH) is a common inherited disorder in which individuals absorb too much iron from their diet. Iron, though essential, is toxic in excess and may accumulate in and damage several organs. This may manifest as complications such as diabetes, heart failure and liver disease. HH is diagnosed by finding mutations in certain genes, however not everyone with these mutations suffer from complications and when they do, it may only be specific types. This suggests that there may be other factors which affect the risk of suffering from such complications in these patients. One of these may be mutations in other 'disease modifier' genes.

The aim of my project is to identify these genes specifically in relation to liver disease. Focusing on mutations in four candidate genes, I am comparing the frequency of these mutations in HH patients with liver disease vs those without, using data from several European countries.

It is expected that the mutation frequency will be higher in those with liver disease suggesting that having these mutations may increase the risk of a HH patient suffering from this complication. If this is the case, similar genetic mutations may be found for other complications and allow clinicians to predict which complications a patient may develop based on the mutations they have. Consequently, this would enable a more targeted approach to treating or even preventing these complications from occurring.

This project really appealed to me as it has taken lab-based work (which has taught me many fundamental and useful scientific skills and techniques) and applied it to a context of understanding how genes influence our health. This is an area that has always fascinated me. Moreover, the project's clinical significance as outlined above, has opened my eyes to the importance of the research-clinical based interaction in medicine.

## *Mr Sharma's Supervisor Dr Niamh O'Brien comments:*

'Aneesh is a hardworking and motivated student who, prior to starting his project, had no practical laboratory experience. He has excelled in learning the wide variety of techniques needed for his research project and is highly self-motivated. He is a pleasure to work with and is committed to furthering the knowledge of the genetics of haemochromatosis and how certain genetic factors influence the development of liver cirrhosis.'