Dear Committee Members,

I would like to express my sincere gratitude to the European Society of Human Genetics (ESHG) for granting me the opportunity to be one of the five young geneticists selected this year for the prestigious ESHG Observership Programme. I had the privilege of spending four weeks (11th Aug-11th Sept, 2025) at the Manchester Centre for Genomic Medicine, University of Manchester, United Kingdom, under the supervision of Professor Siddharth Banka.

During this observership, I worked closely with the genomic diagnostics laboratory team, where I strengthened my expertise in DNA methylation–based diagnostic approaches for rare diseases, including both laboratory procedures and clinical interpretation strategies. I was introduced to laboratory and data analysis workflow for advanced technologies such as optical genome mapping and long-read sequencing. In addition, I gained exposure to innovative methodologies being developed by the Development team, attended multidisciplinary team meetings, and participated in seminars organized by the Manchester Rare Conditions Centre (MRCC). These activities also provided me with a comprehensive understanding of clinical laboratory operations, diagnostic workflows, and reporting practices.

This experience was extremely enriching both professionally and personally. Beyond acquiring technical knowledge, I gained insight into collaborative clinical decision-making, translational research approaches, and the working culture of a leading genomic medicine center. I believe such exposure is highly beneficial for early-career researchers, and I strongly encourage other young scientists to apply for this programme to broaden their perspectives and enhance their skills.

I wish to extend my deepest appreciation to Professor Siddharth Banka and Professor David Gokhale for facilitating this observership, and to their team members for their generosity, guidance, and support throughout my stay.

The expertise and perspectives I have gained will be directly applied to my ongoing research on rare genetic disorders in India, particularly in developing diagnostic approaches and fostering collaborative studies. I am confident that the knowledge acquired during this observership will significantly contribute to my academic growth and my future contributions to the field of rare disease genomics.

Regards

Purvi Majethia