

20th May, 2024

Dear Committee Members,

I would like to express my gratitude for the wonderful opportunity offered by the ESHG and the chance to be one of the 5 young geneticists granted this year with a fellowship within the ESHG Observership Programme. My choice was to apply for a clinical observership at the Clinical Genetics Department from the Manchester Rare Conditions Center, under the supervision of Professor Siddharth Banka.

During the programme, I participated in clinical consultations in pediatric and adult care liaising with patients and specialists from various fields such as neurogenetics, cardiogenetics, oncogenetics, renal genetics and metabolic diseases. My favorite clinics were the developmental delay clinic and chromatin disorders clinics as I had the opportunity to interact with newly diagnosed patients who had been identified by international research collaborative projects such as DDD study or 100,000 Genomes Project.

I also learned about several laboratory protocols and technologies such as RNA sequencing and DNA Methylation Arrays, as well as specific guidelines used for gene variant interpretation and classification. I was introduced to the research activities carried out in the center such as disease-gene discovery workflows and genotype-phenotype studies.

During the 4 weeks, I participated in various activities held across the department such as weekly departmental meetings, Multi-Disciplinary-Team (MDT) meetings, laboratory meetings, and educational seminars. I also had the chance to interact with other young doctors and scientists involved in studying human genetics and we established connections for possible collaborations in the future.

Taken all together, I would like to thank Prof. Banka for his wonderful hospitality, as he ensured I have a full and diverse time schedule, so I could take advantage of all the facilities available in the department.

Overall, the Observership Programme was an excellent educational experience. I was encouraged to engage in numerous scientific activities, carried out by world-renowned geneticists, in a public healthcare setting. I believe that participating in the programme, based in a prestigious genomic center, is a must for every young geneticist's academic journey.

Gratefully yours,

Teodora Barbarii

