

Report on ESHG Mentorship at Radboud UMC, Nijmegen

I am deeply grateful for the opportunity provided by the ESHG International Mentorship Programme, which allowed me to spend a week at Radboud University Medical Center in Nijmegen under the guidance of Prof. Dr. Christian Gilissen. This mentorship provided a unique and invaluable experience, enabling me to engage with leading experts in genetic technologies and patient diagnostics and to gain insights into the application of some of the most advanced technologies in the field, including long-read sequencing and optical genome mapping.

Throughout my stay, Prof. Dr. Christian Gilissen and the staff at Radboud UMC were exceptionally welcoming and had organized a wide-ranging interactive program focused on various aspects of the genomic diagnostic workflow. During my time there, we discussed different genomic diagnostic strategies and tools, including exome and genome sequencing, chromosomal microarrays, liquid biopsies, and the sophisticated bioinformatics infrastructure that supports these analyses. I was also introduced to some of the specifics of next-generation sequencing in cancer diagnostics, a topic with which I have previously had little opportunity to engage.

I am particularly grateful for the opportunity to delve into the bioinformatic workflows for whole-exome sequencing (WES) and whole-genome sequencing (WGS) and how structural variants can be identified using these types of tests, something I would like to help implement in my home country. Additionally, I had the chance to engage in detailed discussions on the complexities of RNA sequencing, particularly on the types of reference data required for this type of analysis, and how to subsequently interpret the obtained results. These skills will be instrumental in enhancing the care I can provide to patients in my own country.

I wish to extend my deepest thanks to the European Society of Human Genetics for granting me this mentorship opportunity. I am also profoundly grateful to Prof. Dr. Christian Gilissen and the team at Radboud UMC for their warm hospitality, expert guidance, and generous sharing of their extensive knowledge and experience. Thanks to them, this mentorship has been a pivotal step in my professional development, and I am deeply inspired to apply the lessons learned to advance genetic diagnostics in my home country of Bulgaria.