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Report from International Observership programme awardee,
Nikolaos Marinakis from Athens, Greece to Nijmegen, the Netherlands

I am a motivated and progress-driven Molecular Biologist and Geneticist with an MSc in Clinical Biochemistry-Molecular Diagnostics and a PhD in Clinical Laboratory Medical Genetics. Since 2022, I work as a Postdoctoral fellow in the Laboratory of Medical Genetics, (LMG) School of Medicine, National & Kapodistrian University of Athens, directed by Prof. J. Traeger-Synodinos. LMG is the largest Academic Center for Medical Genetics in Greece, devoted in offering genetic diagnosis and investigating novel genetic mechanisms in rare disorders.

Being awarded a position in the ESHG International Observership programme for one of the best departments in Genomics research and diagnosis, namely that Genome Diagnostics, including the Laboratory of Genome Bioinformatics, (Prof. Christian Gilissen, leading) in Radboud University Medical Center in Nijmegen, the Netherlands, was therefore a great pleasure and an exciting opportunity for me to be trained by leader scientists and hopefully acquire expert skills in the field of Medical Genetics.

During this 4-week observership programme, I had the chance to go into routine lab workups, especially those concerning the pipelines followed towards the analysis of genomic data (Whole Exome Sequencing, Whole Genome Sequencing etc). This allowed me to familiarize with basic principles, applications and innovations implemented when analyzing exome and genome sequencing data, including variant interpretation, whereby enhancing new, and improving already acquired skills. Additionally, using the in-house bioinformatic pipeline of the department, I performed re-analysis of existing unsolved WES and WGS data from my current laboratory. I also got involved with a project focusing on the investigation of new candidate genes for autosomal recessive conditions, hopefully contributing towards the recognition of potential underlying genetic mechanisms in unsolved cases.

Further to the general training, the last week I attended the Nijmegen Summer school on medical genetics which included seminars on genomic diagnostics and research as well as lectures on WES and WGS applications, bioinformatic analysis, Long-reads sequencing, CNV analysis, Cancer genomics, Carrier screening, Optical Genome Mapping applications, analysis and best practices, RNA sequencing and Prenatal genetics.

All in all, it is my belief that this observership programme may be considered the beginning of a fruitful collaboration between the two labs, where the exchange of knowledge and expertise will become fundamental for the discovery of novel insights in rare genetic diseases.

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