

Report on my experience - Observership Programme 2023 (in Prof. Alexandre Reymond's lab in Le Genopode, Lausanne, Switzerland)

First of all, I would like to say that I am MD Specialist in Medical Genetics, PhD student from Bucharest, Romania. I gladly received the Observership Programme to visit Alexandre Reymond's lab in Lausanne, Switzerland. The lab is in the Center for Integrative Genomics, University of Lausanne (UNIL), the Genopode Building being the host institution.

During my 4 weeks stay, I participated in the ongoing project to identify novel genes for Mendelian disorders with an associated intellectual disability phenotype, from consanguineous families from Pakistan, Iran and Tunisia. I also took part in the interpretation of variants from the raw data obtained from the Whole Exome Sequencing (WES) but from the researcher's point of view, being able to look at candidate variants and novel, undescribed genes. This was a very precious point of view for me as a clinician and MD who interprets and reports mostly known/established variants for given phenotypes.

Moreover, for novel and possibly pathogenic variants I took part in the process of knockdown orthologs of the candidate genes in zebrafish to establish an animal model. In order to do that, I performed the "wet lab" work alongside a postdoc and a technician from the lab, to whom I am deeply thankful. I also visited the Center's zebrafish facility which was an amazing experience.

Apart from being a unique professional experience it was also a cultural one, giving me the opportunity to visit Lausanne, and the surrounding areas. I visited The Natural History Museum of Lausanne, Zoo of Les Marécottes and Saas Balen mountain village where I got the chance to witness the transhumance of cows from the high alpine region to the lowlands.

I am very thankful I had this opportunity to participate in the Observership Programme and as a member of the ESHG-Y community I strongly encourage young members to apply for it!