

Report on My Experience at the Medical University of Vienna through the ESHG Mentorship Programme

I am honoured to have been accepted to the ESHG Mentorship Programme, which provides an important platform for exchanging knowledge and building connections across different areas of human genetics, offering young clinicians and researchers a valuable opportunity to broaden their perspectives.

In this context, I had the chance to spend time at the Medical University of Vienna under the supervision of Dr. Paul Dremsek. During my stay, I was able to observe a wide range of laboratory workflows and genetic testing strategies running in parallel. This experience helped me connect my clinical background as a clinical geneticist with the technical aspects of genomic diagnostics that guide many of our everyday decisions.

Although my visit was relatively short, Dr. Dremsek and his colleagues ensured that it was both intensive and informative. I had the opportunity to observe various methodologies, from classical cytogenetic techniques to modern genomic platforms. This exposure strengthened my understanding of how different diagnostic tools can complement each other in complex cases, and these insights will support my ongoing work with unresolved rare genetic disorders at my home institution.

During the visit, I also gained practical experience with Optical Genome Mapping (OGM), a method not yet available in my centre. We performed OGM on a sample from our unresolved Epidermodysplasia Verruciformis patient and identified a multigenic, complex structural variant contributing to the disease mechanism. Observing how the Vienna team incorporates OGM into routine diagnostics allowed me to learn the workflow and interpretation steps in a concrete and structured way. Being involved in the clarification of a previously unsolved case through this collaboration was a valuable part of my time in Vienna.

I am grateful to Dr. Dremsek and his team for their warm welcome, generosity, and mentorship, as well as to the ESHG for making this experience possible.

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