

Dear ESHG Education Committee,

My name is Emre Akbaş, and I am a resident doctor in the Department of Medical Genetics at Eskişehir Osmangazi University, Türkiye. I have a special interest in dysmorphology, rare diseases, bioinformatics, and artificial intelligence applications in medical genetics. In 2025, I received the Dian Donnai and Jill Clayton-Smith Award at the ESHG Conference for my presentation on an unknown case of craniofacial dysmorphism with normal neuromotor development in a large family. This case later became the starting point of an important research project for me.

Thanks to the ESHG Observership Programme, I had the opportunity to spend four weeks in Germany in May and June, before the ESHG Conference. During this period, I visited the Institute for Genomic Statistics and Bioinformatics at University Hospital Bonn under the supervision of Prof. Dr. Peter Krawitz. I also had the opportunity to discuss our linkage analysis with Prof. Dr. Michael Nothnagel at the University of Cologne, Department of Statistical Genetics and Bioinformatics.

The main focus of my visit was a family in which we suspect a novel autosomal dominant craniofacial syndrome. The affected individuals have a recognizable craniofacial dysmorphism that segregates through four generations. Routine genetic analyses performed at our own center, including karyotype analysis, chromosomal microarray, and whole-exome sequencing, had not provided a clear molecular diagnosis. We collected peripheral blood samples from affected and unaffected relatives and generated genome-wide SNP genotype data using the Infinium Global Screening Array v3.0. The aim was to use these data for linkage analysis and to define a candidate genomic region shared by affected family members.

During my stay in Bonn, I had the chance to see how rare disease research can be approached from clinical observation to advanced genomic analysis. I learned more about linkage analysis, long-read sequencing, whole-genome sequencing, and the interpretation of complex genetic data. I also observed how artificial intelligence can support the analysis of genetic diseases, especially through tools and projects such as GestaltMatcher and Bone2Gene. It was very inspiring to see how clinical dysmorphology, patient photographs, genomic data, statistics, and artificial intelligence can be combined in the same research environment.

My discussions with Prof. Michael Nothnagel helped me understand the logic and limitations of linkage analysis more clearly. These discussions were especially useful because linkage analysis is not routinely performed in my department, but it is highly relevant for our family project and my residency thesis.

After this observership, I had the opportunity to present our progress again at the ESHG Conference 2026 in Gothenburg, in the Dysmorphology Interactive Sessions, with the title “Chasing a new syndrome: Updates since ESHG Conference 2025 and what’s next?”. In this presentation, I shared the clinical updates, the linkage analysis work, and the ongoing steps with the participants. This was a very meaningful continuation of the work that started after my first ESHG presentation.

This observership was a unique and very valuable experience for me. It broadened my scientific perspective and showed me the importance of international collaboration for unsolved rare disease cases. The knowledge and experience I gained will directly support my medical genetics residency thesis and my future academic work. Our collaboration with Prof. Krawitz will also continue in the next phase, especially for functional studies related to our candidate findings.

I am deeply grateful to the ESHG Education Committee for giving me this exceptional opportunity. I would like to express my sincere thanks to Prof. Peter Krawitz for welcoming me to his institute, for his guidance, and for his generous support of our project. I am also very thankful to Prof. Michael Nothnagel for sharing his expertise and experience in linkage analysis. Finally, I would like to thank all members of the teams in Bonn and Cologne for their kindness, time, and hospitality during my stay in Germany. This experience will remain an important milestone in my scientific and professional development.

Sincerely,

Emre Akbas, MD