Epilepsy, affecting 1 % of world population is a complex disease with multiple dimensions. The most important contributor of the disease is genetic predisposition in a large spectrum. While some epilepsy types are pure genetic some other epilepsies arise from acquired defects like brain tumors or brain malformations. As epilepsy genetics research group we aim to discover the genetic or genomic variations underlying the epilepsy etiology, which later on supports patient diagnosis and identification of novel treatment strategies. For this purpose we apply main genetic research approaches including single gene and genomic analyses.

Genomic Research Lab

Over the last decade the implementation of high throughput technologies had a great impact on genetics research. Next Generation Sequencing technology allows sequencing of human genome in single run with specialized equipment. On the other hand, requirement of middle throughput data by diagnostic laboratories are met by massive parallel sequencing devices. While genome sequencing identifies point mutations and small scale indels, larger genomic abnormalities like copy number variations are detected by array comparative hybridization analysis, which shows genomic scale insertion and deletions in variable resolution.

Genomic Research Lab is equipped with a GS Junior Sequencer, which is a middle scale NGS device and Nimblegen MS200 microarray scanner which is used in array CGH analyses. Besides these devices, the laboratory also has an Agilent Bioanalyzer for on chip electrophoresis, Thermofischer Nanodrop for nanoscale spectrophotometry and all other equipment required for NGS and array-CGH analyses.
Current Research Projects:

- **ARX Gene Screening in Epileptic Encephalopathy patients**
  - MSc Study of Hande Özünlü

- **SCN1A gene Screening in Dravet Syndrome and Epileptic Encephalopathy patients**
  - Sunay Usluer and Aslı Gündoğdu

- **KCNQ2 gene screening**

- **Whole Exome Analysis of Patients with Benign Familial Neonatal Seizures**
  - Postdoctoral Research by Sunay Usluer

- **Gene Identification in a Large Pedigree with Genetic Epilepsy with Febrile Seizures Plus Phenotype**
  - Postdoctoral Research by Sunay Usluer
Collaborations

**Nationwide:** Departments of Neurology and Pediatric Neurology in academic institutions and hospitals in Turkey.

**International:** EPICURE Consortium, EuroEPINOMICS/RES-CRP (Rare Epilepsy Research, Collaborative Research Project)

Selected Publications:

- **Sunay Usluer, Canan Aykut-Bingol, Berrin Aktekin, Kadriye Agan, N. Berfu Akbas, S. Hande Çağlayan** “Mir137 VNTR Expansion As A Risk Factor For GEFS+ Phenotype In A Large Kindred” (in preparation)
- **Seda Salar*, Sunay Usluer*, Özlem Yalçın Çapan, Bülent Kara, Cihan Meral, Uluç Yis, Mutluay Arslan, Rüdvan Akin, Ashlı Gündoğdu Eken, S. Hande Çağlayan** “Exome Sequencing Of The SCN1A Gene In 21 Turkish Patients” (in preperation. * Equal Contribution)
- **Sunay Usluer, Melek Asli Kayserili, Uluç Yis, Costin Leu, Thomas Sander, S. Hande Çağlayan** “Whole genome linkage and whole exome sequence analyses in a multiplex BFIS (Benign Familial Infantile Seizures) family reveal a synonymous change affecting splicing efficiency of SCN1B mRNA as the major causative variant” (Submitted to Epilepsy Research)
- **Julian Schubert,…. EuroEPINOMICS RES consortium (…Hande Çağlayan…), … Holger Lerche (2014), Mutations in STX1B, Encoding a Presynaptic Protein, Cause Fever-Associated Epilepsy Syndromes, Nature Genetics, published online 2 November 2014; doi:10.1038/ng.3130.
- **A Suls, …. C Meral, HS Çağlayan, …I Helbig, the EuroEPINOMICS RES Consortium (2013) De novo loss of function mutations in CHD2 cause a fever-sensitive myoclonic epileptic encephalopathy sharing features with Dravet syndrome, American Journal of Human Genetics, 93:967-975."


