madridge CONFERENCES
International
Translational and Regenerative Medicine Conference
April 25-27, 2018 Rome, Italy

Gene Discovery in a Neurogenetic Cohort

Wilhemina Koomson*, Adife Ercan-Sencicek, Katsuhito Yasuno and **Murat Gunel**
Yale University, USA

We analyzed 1,505 patients with neurological/neurodevelopmental disorders of varying severity including brain structural malformations from a network of 28 clinical centers in Turkey, specifically ascertaining consanguineous cases. Next generation sequencing technologies, specifically whole-exome sequencing (WES), led to identification of disease causing variants in these cases. Our study considered various modes of inheritance, focusing on recessive forms of the disease as well as de novo variation, especially for non-consanguineous cases. We have identified several pathogenic mutations falling within a Homozygous by descent (HBD) segment in consanguineous families. With an improved filtering strategy we were able to identify multiple independent coding mutations or copy number variations (CNV), suggesting novel disease-causing genes have been identified. We prioritized these genes, as well as other strong candidate genes, based on known biological function, molecular interaction and Weighted Gene Co-expression Network Analysis (WGCNA). We identify gene co-expression module profiles of the new candidate genes, correlating them with the spatial and temporal expression patterns (during brain development) of known phenotype concordant disease genes.

Biography:

Wilhemina Koomson is a 3rd year PhD candidate in the Department of Genetics at Yale University. Her research incorporates bioinformatics in studying Neurological and Neurodevelopmental disorders. She completed her undergraduate work at Princeton University majoring in Molecular Biology.