SOX7: Novel Autistic Gene Identified Through Analysis of Multiomics and Human Brain Organoids

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Autism spectrum disorder (ASD), a disorder that affects brain development and can cause problems with social interaction and communication, is thought to be caused by both genetic and non-genetic factors. Early intervention can critically improve the quality of life for people with ASD. This project used multi-omics data and bioinformatics tools to identify novel ASD-associated genes, which help diagnose ASD early in childhood and even before birth. This is followed by generating human brain organoids derived from human induced pluripotent stem cells (hiPSCs) for autism modeling and drug screening. I applied a bioinformatics adaptive test to analyze ASD data from two genome-wide association studies (2019 Data: 18,381 ASD cases and 27,969 controls [Discovery Data]; 2017 Data: 7,387 ASD cases and 8,567 controls [Replication Data]), and further analyzed gene expression data (GSE30573: 3 ASD cases and 3 controls). Five genes significantly associated with ASD in the ASD 2019 data were identified, with one, SOX7 gene (p=2.22x10^-7 in 2019 data; p=0.0009 in 2017 data), being replicated in the ASD 2017 data having different levels of gene expression in people with and without ASD (p=0.0017). Encoding a member of the SOX family of transcription factors, SOX7 contributes to determining the fate of a cell and the identity in lineages. The encoded protein acts as a transcriptional regulator after forming a protein complex, and leads to the trait of ASD indicating SOX7 as a causal factor for autism. We are currently performing human brain organoid analysis to investigate the biological functions of this finding, which may provide new diagnostic and therapeutic treatments for autism spectrum disorder.

Awards Won:

University of Texas at Dallas: Scholarship of $5,000 per year, renewable for up to four years