Dissertation Defense - Leena Sait, PhD Bioinformatics and Computational Biology

April 281, 2022 10:00 AM- 12:00 PM

All are invited to attend the defense. For more information please contact Graduate Coordinator at dstgerma@gmu.edu

Candidate: Leena Sait

Program: PhD, Bioinformatics and Computational Biology

Date: Thursday, April 28, 2022
Time: 10:00 AM

Zoom Link: https://gmu.zoom.us/j/93361201154?pwd=V3lPUjhVWGZrM3RhSXVrdTFPTzVYdz09

Title: Computational Analysis of Autism Spectrum Disorder Biomarkers

Committee Chair: Dr. Iosif Vaisman

Committee Members: Dr. Saleet Jafri, Dr. Alessandra Luchini

ABSTRACT:

Autism spectrum disorder (ASD) is one of the most common neurodevelopmental disorders. Worldwide, ASD tends to have a prevalence of roughly one in 100 children, according to the World Health Organization (WHO). To date, no effective medical treatments for the core symptoms of ASD exists. However, biomarkers capable of detecting and diagnosing ASD can help to translate experimental research results to bench side clinical practices. Biomarker discovery in ASD is complicated by the diversity of core symptoms which comprise deficits in social communication, presence of rigid, repetitive, and stereotypical behaviors, and comorbid medical (e.g., epilepsy) or psychiatric symptoms. This work is aimed at the identification of single nucleotide polymorphisms (SNPs) based on computational approach using SNP genotyping in genomic DNA in a large cohort of ASD patients and unaffected related individuals. We hypothesized that by calculating the distance of alleles between affected and unaffected populations using the Cartesian distance in the space of alleles frequencies, we can identify new putative biomarkers. The dataset retrieved from the Gene Expression Omnibus database (GSE6754) contains more than 6000 samples from 1,400 families. Our studies propose that the SNPs with the highest-ranking distances in three-dimensional genotype count space between all the affected and unaffected subjects in the cohort are likely to be linked to ASD. These results will open new doors for further investigation and future work is expected to help identify the exact genetic mechanisms of ASD that could be used in future research to improve diagnostic and therapeutic interventions. Based on the distance between patients and healthy relatives in the space of mutated alleles, we decided to focus on Fragile X syndrome which is known to be the single leading cause of inherited intellectual disability and autism spectrum disorder. It is found to be on the 1010 top-ranking SNPs targeted in the unbalanced cohort. By performing in silico analysis using computational and other bioinformatics tool which provides a powerful approach to gain a better understanding of the biological systems at the gene/protein level.