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A genetic tool to analyze de novo mutations in autism spectrum disorder using genetic data in vcf format

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Abstract

Autism Spectrum disorder affects nearly 1-2% of the worldwide population. It is a Neurological Disorder characterized by impaired social communication and interaction, showing signs of repetitive behavior and restricted interests. Even though GWAS studies have identified a large number of genetic markers that could cause Autism, DeNovo mutations also significantly contribute to the cause of Autism Spectrum disorder. Tools for identifying DeNovo mutations such as SNPs, insertions and deletions using analysis such as Identity By State, Inheritance patterns in trios, Copy Number Variation, Parent of Origin duplication all work on SNP Array file format genome data. New generation sequencing file format such as Variant Call Format (VCF) lack tools that performs such analysis. The VCF is a popular standardized text file format for storing genetic data such as SNPs, insertions, deletions and structural variants. The format was developed for the 1000 Genomes Project. Presenting VCFDataPy, a genetic analysis tool written in python that can perform various analysis such as Identity By State (IBS), Inheritance patterns in trios ((mother, father, and child), Parent of Origin of a chromosomal duplication, Copy Number Variation and complete visualization of chromosomes in VCF Files. The tool was applied to identify the DeNovo mutations that caused Autism Spectrum disorder by performing various analysis on genome data provided in VCF file format.

Biography

Siddharth Krishnakumar is a high school Junior in Thomas Jefferson High School for Science and Technology. Siddharth is very interested in bioinformatics research and has presented his research in many conferences as both oral presentation and poster presentation.

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