Literature Review of Approaches to Analyzing the Genetic Predisposition to Autism

Gergana Stanilova

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Abstract

Autism is a neurodevelopmental disorder characterized by communication difficulties and repetitive behaviors. Although there are many environmental influences, the intricate genetic component of this disorder must be taken into account and thus, different approaches to its analysis must be considered. Generally, gene annotation, although invaluable, also might prove to be insufficient when it comes to building a full and accurate picture of the genetic factors involved in ASD (Autistic Spectrum Disorder).

1 Introduction

Neurodevelopmental disorders might not affect the vast majority of the population but their importance should not be neglected nevertheless. However, the analysis of the genetic factors contributing to these conditions strongly relies on computational methods due to the size of the human genome. Therefore, exploring different approaches could be crucial to achieving a proper understanding of these genetic predispositions. To that end, I have reviewed three papers describing different techniques of analyzing the anomalies in the genome of ASD patients.

2 Approaches

2.1 Genome Annotation

The authors of all three papers acknowledge the relevance of reliable and abundant genetic information. Some new contributions were made to the Gene Ontology (GO) database [3] regarding a few protein families. Additionally, other researched was based on DNA-annotation resources like the Human Phenotype Ontology (HPO), the Mammalian Phenotype Ontology (MPO) projects [2] and the Autism Database (AutDB) [1].

2.2 Classification

The classification of ASD subjects also has led to some useful insights to the influence of rare copy number variations (CNVs) [2]

2.3 Gene Ranking

An issue which might arise is that the importance of genes which are researched more commonly could be biased. Therefore, a scoring schema is proposed [1] which aims to adjust for such misinterpretations.

References

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