OPEN ACCESS Freely available online

ISSN: 2153-0602
Journal of Data Mining in Genomics & Proteomics

Opinion Article

Identification of Rare Genomic Variants in Whole-Genome Sequencing

Zhong Ren*

Department of Bioinformatics, Ohio State University, Columbus, OH, USA

ABOUT THE STUDY

Primary Torsion Dystonia (PTD) is an external vertebral body disease characterized by abnormal posture and movement caused by an uncoordinated or excessive contraction of the active and antagonist muscles. The pathogenesis of PTD is still not fully understood. Several novel disease-associated genes (DYT1-27) have been identified in dystonic syndromes over the last few decades, but the underlying genetic diagnosis remains elusive in the majority of patients. However, almost all cases of primary dystonia have a genetic basis.

At the moment, the focus of genetic disease research is primarily on genes, and gene sequencing is the method of choice. The development of whole genome sequencing, particularly the cost of single human sequencing, has resulted in new opportunities for the study of genetic diseases. Traditional cloning technology has solved many problems, such as too few members in the family, sporadic cases, heterogeneity of gene loci, penetrance, and too many candidate clones in the targeting region. The lack of disease-related structural variations and non-coding region variation in whole genome exon sequencing has been compensated for by genome wide sequencing.

Noncoding mutations, such as promoters, enhancers, introns, and noncoding RNA, can be detected using genome wide sequencing, which is not possible with other methods (including tiny RNA). Chromosomal rearrangements such as inversions, tandem repeats, and deletions can be detected. To achieve genetic evolution analysis and the importance of candidate gene prediction, a large number of genetic differences can be discovered. It encompasses a wide range of disciplines, including clinical medicine research, population genetics research, association analysis, and evolutionary analysis. Whole genome sequencing technology has been tested more widely than exome sequencing technology, and the result analysis is more thorough in the study of genetic disease. Identifying PTD and genetic risk factors has proven difficult thus far, and the introduction of the latest genome-wide sequencing technologies could accelerate progress in these areas. ANO_3 encodes a structurally related homodimeric protein that encodes a Ca+ 2-activated chloride ion channel and a protein of a membrane phospholipid antibody with a different expression pattern. ANO_3 is made up of eight hydrophobic trans membrane helices that function as calcium sensors to regulate calcium homeostasis. The precise function of ANO_3 is unknown, but recent research indicates that it does not function as a Ca+ 2-activated chloride ion channel and may instead function as a Ca+ 2-dependent phospholipid fragment. ANO_3 appears to be involved in the regulation of neuronal excitability and is found in high concentrations in the striatum, hippocampus, and cortex. Mechanisms and pathogens in ANO_3 may cause striatal-neuronal excitability abnormalities, resulting in uncontrollable dystonia movement. ANO_3 mRNA expression is highest in the striatum, 5.30 times that of the frontal cortex, and 70 times that of the cerebellum, and its abnormality can affect the endoplasmic reticulum-related calcium ion gated chloride channel, leading to disease.

At the moment, the disease is primarily treated with drugs and stereotactic surgical procedures, but the treatment is only symptomatic, has many limitations, and the pathogenesis of dystonia is not completely understood; thus, it is necessary to screen new loci of the *DYT* gene, discover new related genes, and study mutant genes and related proteins. The study uses whole genome sequencing technology to examine the pathogenic genes and mutation sites in patients with primary dystonia, as well as the relationship between genotype, clinical phenotype, and prognosis. Detecting genetic mutations in genetic diseases and discovering new genes or mutations can assist us in making the correct molecular diagnosis and providing better genetic information.

Our findings reveal a new mutation that may be pathogenic in known genes with similar phenotypes, laying the groundwork for future research in which more families will be sequenced to uncover more information. Detecting genetic mutations in genetic diseases and discovering new genes or mutations can assist us in making the correct molecular diagnosis and providing better genetic information.

Correspondence to: Zhong Ren, Department of Bioinformatics, Ohio State University, Columbus, OH, USA, E-mail: zhang.ren@imh.edu

Received: 02-Sep-2022, Manuscript No. JDMGP-22-18530; **Editor assigned:** 06-Sep-2022, PreQC No. JDMGP -22-18530 (PQ); **Reviewed:** 20-Sep-2022, QC No JDMGP-22-18530; **Revised:** 27-Sep-2022, Manuscript No. JDMGP-22-18530 (R); **Published:** 04-Oct-2022. DOI: 10.4172/ 2153-0602.22.13. 262.

Citation: Ren Z (2022) Identification of Rare Genomic Variants in Whole-Genome Sequencing. J Data Mining Genomics Proteomics. 13:262.