University of Zabol
Graduate School
Faculty of Sciences
Department of Biology

The Thesis Submitted for the Degree of M.Sc
(in the field of Genetic)

Title

Exom Sequencing in a large family with multiple cases of stuttering

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Autumn 2021

Abstract:

Stuttering is a childhood-speech disorder, intertwined with physiological, emotional, and anxiety factors and characterized by disruptions in normal flow of speech in the form of repetition, prolongation and involuntary hats. The present study, in order to investigate the genetic cause of this disorder in a family with consanguineous marriages that have shown stuttering in different generations, whole exome sequencing and sanger sequencing were used. Data analysis was performed using BWA software for alignment, GATK for variant calling and Annovar for annotation. The obtained variants were examined in three samples and bioinformatic evaluation and segregation analysis of the found variants helped us define probable consequences. Five variants were found in effective stuttering genes, all of which were reported to be benign according to the ACMG guideline. 43 possible variants of other speech and language disorders were reported, among which three variants were found to be homozygous in the coding region, and the results of the analysis predicted them to be relatively indefinitely relatively pathogenic.

The variation TTN gene was selected for sanger sequencing. According to the results obtained from sanger sequencing and variants obtained in genes that cause other speech disorders, it is possible that the proband does not have stuttering speech disorder and the report of the speech therapist is wrong.

KEYWORDS: Stuttering, Whole Exome Sequencing, Bioinformatics, Sanger Sequencing, ACMG guideline, Speech Disorder