Abstract

Intellectual disability (ID) is a neurodevelopmental disorder, which cause great socioeconomic burden. The advent of high-throughput sequencing technologies has led to an exponential increase the in deciphering of novel disease causing genes in highly heterogeneous disease. A novel missense mutation in *RNFT2* gene was detected by Next-Generation Sequencing (NGS) in two Iranian families with syndromic autosomal recessive intellectual disability (ARID). *RNFT2* encodes a ring finger transmembrane protein that involve in ubiquitination and protein degradation which is electron carrier activity and ion-self cluster binding. In silico simulation show that this point mutation led to change in protein structure and low affinity of the Zink. Furthermore, RNAi-mediated knockdown of *CG13605*, the *RNFT2* ortolog in *Drosophila melanogaster* brain led to behavioral defect in learning and memory. In conclusion, our funding is supported the possible role of *RNFT2* in brain development, learning and form of memory which can led to cognitive impairment.

key words: Autosomal recessive intellectual disability, *RNFT2*, *Drosophila melanogaster*, *Kenyon cells.*

University of zabol

Graduate school

Faculty of science

Department of Biology

The Thesis submitted for the Degree of M.Sc

(in the field of Genetic)

Title:

Functional study of *RNFT2* gene in causing of Autosomal Recessive Intellectual Disability

Supervisore:

Dr. M. Haddadi

Dr. H. Najmabadi

By:

Reza Ataei Jaliseh

Jun 2018