National Institute of Technology Rourkela

Departmental Seminar

Seminar Title : Predicting the role of RNA binding proteins interaction with the mRNAs transcribed from 15q11.2 BP1-BP2

microdeletion region

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Supervisor : Santosh Kumar #2787 Venue : LS Seminar Hall Date and Time : 17 Mar 2025 (16:30 hrs)

Abstract : The Burnside-Butler susceptibility area, a microdeletion region in 15q11.2 BP1-BP2, is linked to neurodegenerative

disorders. The four conserved, non-imprinted protein-coding genes in this region are TUBGCP5, CYFIP1, NIPA1, and NIPA2. This study aims to investigate the interaction of RNA-binding proteins (RBPs) with the mRNAs of four genes in the microdeletion area and their probable function to better understand the pathogenic effects when deleted. In-silico study was done by intersecting the eCLIP data from ENCODE with the genomic position of the four genes located in 15q11.2 BP1-BP2 susceptibility region to find out the RBPs binding to the region. Then the significant RBPs were filtered for each gene and the binding of RBPs FASTKD2 and EFTUD2 with the mRNAs encoded from CYFIP1 and TUBGCP5 genes were done using in-vitro WEMSA experiments. Our result indicates that most of the RBPs interacting with the susceptibility region are involved in post-transcriptional regulation of the concerned genes. RBPs binding to UTRs, coding region, and junction region were found. The fact that these proteins bind to exon-intron junctions implies that they may be involved in the splicing process. In addition to their functional relevance in normal development, or lack thereof in neurodevelopmental disorders, this work may contribute to a better understanding of the complex connection between RBPs and mRNAs within this area. This insight will aid in the development of improved treatment techniques.

Keywords: 15q11.2 microdeletion, WEMSA, BP1-BP2, eCLIP, RBPs