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MECP2 GENE DYSFUNCTION AND EMERGING MOLECULAR THERAPEUTICS STRATEGIES IN RETT SYNDROME

Ashifa.T.A[a], Beevi Ayisha Banu.J[a], V.Jayashree[b*]

[a] *B.Pharm Student, Department of Pharmacology, School of Pharmaceutical Sciences, Vels Institutes of Science, Technology & Advanced Studies, Chennai*

[b] *Associate Professor, Department of Pharmacology, School of Pharmaceutical Sciences, Vels Institutes of Science, Technology & Advanced Studies, Chennai*

** Corresponding Author: jeya.sps@vistas.ac.in*

Abstract:

Rett Syndrome is a rare X-linked neurodevelopmental disorder that primarily affects females and is mainly caused by mutations in the MECP2 gene. The disorder results in neurological regression, loss of speech and motor skills, seizures, and cognitive impairment. The MECP2 gene plays a crucial role in regulating gene expression and maintaining normal neuronal function. However, recent advances in molecular biology have introduced new therapeutic approaches such as AAV-mediated gene therapy, CRISPR-based gene editing, RNA editing, and MECP2 reactivation strategies. These emerging therapies aim to restore normal gene function and may provide promising disease-modifying treatments for Rett syndrome in the future. This chapter explores the molecular mechanisms of MECP2 dysfunction and highlights current and emerging therapeutic strategies with potential to transform the management of Rett syndrome.

Keywords: Rett Syndrome; MECP2 Gene Mutation; Neurodevelopmental Disorder; Neuronal Dysfunction; Gene Therapy;

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