Zoologie Innovante

Series 2

Chief Editor
Dr Sr. Beena Jose

Editors
Dr. Meera K M | Sr. Freny Jacob

An initiative of PG Department of Zoology,
Vimala College (Autonomous), Thrissur-9

ZOOLOGIE INNOVANTE SERIES 2

PG Department of Zoology
Vimala College (Autonomous), Thrissur – 680009

Month and Year of Publication	:	September 2021
ISBN	:	978-81-7255-147-6
Printed at	:	The Union Press, Ph : 0487 2973213
Published and Distributed by	:	Sooryagatha (Publishers), Kochi - 682035
Copy Right	:	PG Department of Zoology Vimala College (Autonomous) Thrissur - 9

CONTENTS

MECP2 MUTATIONS IN RETT SYNDROME PATIENTS FROM SOUTH INDIA

ANAGHA.K U,[1] ANITHA AYAPPAN PILLAI,[2] RAHNA PARAKKAL[2], INDU M.S[1], PETRISIA JOSEPH[1]

[1] Vimala College (Autonomous), Thrissur,

[2] Institute for Communicative and Cognitive NeuroSciences, Shornur

* Corresponding author, E-mail: anaghaku246@gmail.com

Abstract

Rett syndrome (RTT) is a progressive neurodevelopmental disorder caused by mutations in the X-linked MECP2 gene and one of the most common causes of mental retardation in females, with an incidence of 1 in 10,000-15,000. For this study, DNA was isolated from peripheral venous blood samples of patients exhibiting features of Rett Syndrome (2-18 years of age) using standard salting out method. DNA quantification was done using qubit fluorometer. Then, the primers for MECP2 gene were designed using Primer 3.0 online tool. This was followed by polymerase chain reaction (PCR). The purified PCR product was used for Sanger sequencing. In our study, out of 20 samples, 11 individuals had MECP2 mutations. The remaining nine samples could be atypical RTT. The mutations were observed in exon 4 for most of the samples. Mutation in exon 3 was found in 2 samples. Of the 11 mutations observed in this study, three were novel mutations. Three benign mutations were also found. The novel mutations include deletion in two samples and insertion in another, causing frameshift mutations. All these three cases were pathogenic variants. Our study mainly aimed to characterize MECP2 gene mutations in South Indian patients with Rett syndrome (RTT)

Keywords: Rett syndrome, RTT, MECP2, DNA Sequencing.

Introduction

Rett Syndrome (RTT) is an X-linked dominant neurodevelopmental disorder that causes progressive loss of motor skills and speech. This disorder was originally described in the 1960s by Andreas. It primarily affects girls, and has an estimated prevalence of 1 in 10,000 to 15,000 females. Rett Syndrome (RTT) is an autistic spectrum disorder with a known genetic basis, Weaving *et al* (2003). It is the 2nd most common cause of intellectual disability in females after Down's syndrome, Zoghbi *et al* (2004). Most babies with Rett Syndrome seem to develop normally for the first 6 to 18 months of age, and then lose skills they previously had, such as the ability to crawl, walk, communicate, or use their hands. There are two types of Rett syndromes; classical Rett syndrome and atypical Rett syndrome. Classical RTT is a neurodevelopmental disorder where in most of the cases carry methyl-CpG binding protein 2 (MECP2) gene mutations on chromosome Xq28, Bebbington (2008).