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Molecular characterization of MECP2 in Indian Patients with classical Rett Syndrome

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Rett syndrome (RTT) is a neurodevelopmental disorder primarily affecting females. It is caused by mutations in the Methyl-CpG-Binding Protein 2 gene (MECP2). Variants in MECP2 can be identified in 95 to 97% of individuals with Classical RTT using a combination of molecular techniques. Identification of variants is important for confirmation of diagnosis, counseling and prenatal diagnosis. As there is limited Indian data on mutation spectrum of RTT, this study was on a large cohort of classical RTT females. This study is aimed at molecular characterization of MECP2 gene variants in Indian patients with classical RTT.Seventy-two patients fulfilling the revised diagnostic criteria of classical RTT were enrolled and exons 2-4 of MECP2 gene were analyzed by sequencing followed by quantitative analysis. Bioinformatic analysis performed to predict the effect of sequence variations on the function of the MeCP2 protein. A heterogeneous spectrum of MECP2 variants including 13 novel variants were identified with a detection rate of 98.6%. Majority of the variants were distributed in the functional domain of MECP2 with most missense variants clustered in methyl binding domain and truncating variants in inter domain and transcription repression domain of MECP2. Genotype-phenotype correlations revealed that patients carrying early truncating variants presented with a more severe phenotype. This study showed highest detection rate of MECP2 variants in classical RTT patients, emphasizing the importance of using diagnostic criteria, which can further help in providing the diagnostic testing, genetic counseling and prenatal testing.

Biography

Rajni Khajuria has completed her Ph.D in year 2011 and postdoctoral fellowship in year 2013 from All India Institute of Medical Sciences, New Delhi, India. She is senior consultant-Human genetics and Scienfic affairs. She has 16+ years expertise in laboratory Research and diagnostics needs focused in Clinical Molecular genetics and 14+ years of expertise in Genetic counselling. Her special interest includes Rare genetic diseases, Preimplantation Genetic Testing, Prenatal Diagnosis, Prenatal screening, Pre-conceptional screening, Genetic counseling, Reproductive Genetics, Human Genetics, Patient support groups and patient care. She has more than 40 publications in reputed journals, books and conference proceedings.

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