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Notice for the PhD Viva Voce Examination

Mr Arun Meyyazhagan (Registration Number: 2071601), PhD scholar at the School of Sciences, CHRIST (Deemed to be University), Bangalore will defend his PhD thesis at the public viva-voce examination on Wednesday, 31 July 2024 at 10.00 am in Room No. 044, Ground Floor, R & D Block, CHRIST (Deemed to be University), Bengaluru - 560029.

Title of the Thesis	:	**Biomarkers of Autistic Study: Biochemical, Genomics, Epigenetics and Cytogenetic Signatures**
Discipline	:	**Biotechnology**
External Examiner (Outside Karnataka)	:	**Dr Anand Prem Rajan** Associate Professor School of Bio Sciences and Technology Vellore Institute of Technology Vellore - 632014 Tamil Nadu
External Examiner (Within Karnataka)	:	**Dr Mathivanan Jothi** Associate Professor Department of Human Genetics National Institute of Mental Health and Neuroscience Bengaluru – 560029 Karnataka
Supervisor	:	**Dr P Manikantan** Associate Professor Department of Life Sciences School of Sciences CHRIST (Deemed to be University) Bengaluru – 560029 Karnataka

The members of the Research Advisory Committee of the Scholar, the faculty members of the Department and the School, interested experts and research scholars of all the branches of research are cordially invited to attend this open viva-voce examination.

Registrar

Place: Bengaluru
Date: 23 July 2024

ABSTRACT

Autism is a multifaceted neurodevelopmental condition marked by significant challenges in social interaction, communication, and repetitive behaviors. Over the past two decades remarkable increase in the prevalence of autism has been witnessed, with incidence rates soaring to approximately 1 in 150 children during the 2000s. Given the pronounced phenotypic variability and genetic heterogeneity for autism, cytogenetic investigations are indispensable for accurate clinical diagnosis. This study aims to elucidate the behavioral phenotypes of autistic individuals in South India, utilizing standardized diagnostic criteria from the DSM-IV and ATEC open questionnaires. These tools are critical in establishing consistent and reliable behavioral profiles among the study's participants. In addition, the investigation reveals that metabolic factors, encompassing a range of hormones, neurotransmitters, and oxidative ions, are integral to the progression and manifestation of autistic symptoms.

The interplay of such metabolic elements suggests a complex biochemical underpinning of autism, highlighting potential pathways for targeted therapeutic interventions. Another pivotal aspect of this study is the identification and analysis of two major causative genes, NRXN1 and CNTNAP2. These genes are shown to play significant roles in the spectrum of genotypes that contribute to the severity and heterogeneity of autism. NRXN1 and CNTNAP2 are implicated in neural connectivity and synaptic functioning, which are crucial for proper neurological development and function. Mutations or alterations in these genes can disrupt these processes, leading to the diverse and complex symptomatology observed in ASD. The findings of this research emphasize the importance of integrating biochemical, genomic, epigenetic, and cytogenetic signatures to advance our understanding of autism. By delineating the roles of specific metabolic and genetic factors, this study provides a more nuanced perspective on the etiology and progression of autism. Such insights are vital for the development of more precise diagnostic tools and personalized therapeutic strategies, ultimately aiming to improve the quality of life for individuals with autism and their families.

Keywords: Autism, chromosomal alterations, single nucleotide polymorphisms, metabolic signatures, genetic heterogeneity.

Publications:

1. **M. Arun**, B. Balasubramanian, H. Kuchi Bhotla, M. Easwaran, S. Kumar, A. Karthick Kumar, V. Anand, S. Keshavarao, and M. Pappusamy - Genetic and cytogenetic screening of autistic spectrum disorder: Genotype-phenotype profiles," *Meta Gene*, vol. 29, 2021, doi: 10.1016/j.mgene.2021.100924.
2. **M. Arun**, B. Balasubramanian, M. Easwaran, A. Karthick Kumar, S. Kumar, H. Kuchi Bhotla, M. Pappusamy, V. Anand, A. Thangaraj, T. Kaul, S. Keshavarao, and R. Cacabelos - Biomarker study of the biological parameter and neurotransmitter levels in autistics," *Molecular and Cellular Biochemistry*, vol. 474, 2020, doi: 10.1007/s11010-020-03851-2.
3. **M. Arun**, B. Balasubramanian, S. Kathannan, K. K. Alagamuthu, M. Easwaran, S. Shanmugam, M. Pappusamy, H. K. Bhotla, S. Mustaqahamed, V. A. Arumugam, T. Kaul, and S. Keshavarao, "Scrutinizing the molecular, biochemical, and cytogenetic attributes in subjects with Rett syndrome (RTT) and their mothers," Epilepsy & Behavior, vol. 111, 2020, doi: 10.1016/j.yebeh.2020.107277.