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From the Editor's Desk

GWAS, Pathway Enrichment & Regulatory Network Analyses-a robust interface between Genomics and Bioinformatics

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Publishing a Special Issue on advancing research is not only inspiring, but also an enriching experience. This Editorial column gives a bird's eye view of the topic. It's a great delight to introduce this Special Issue, which highlights cutting-edge research on two areas of Advanced Biology-Genomics and Bioinformatics. Genomics on one hand is the study of complete set of genetic material within an organism while Population Genomics is the large-scale analysis of genetic variation across entire genomes of large populations. Bioinformatics on the other hand, serves as a data translator-using computational, algorithm and statistical tools to analyze and decode complex biological data. The synergy between these two areas has revolutionized modern biology and medicine. Both these fields are data driven engine of Life Science, and has given rise to an advanced triad analytical research-GWAS, Pathway enrichment and Regulatory Network Analyses. These three computational methodologies form a link between Genomics and Bioinformatics.

GWAS-Genome-wide association studies is a potent tool kit that identifies genetic variants which are statistically linked with a risk for a disease or specific trait. The process involves scanning complete genome of a vast population in search of minute variations, which is primarily a single nucleotide polymorphism. Ozaki *et al*. in 2002 published the first GWAS on single nucleotide polymorphism linked with myocardial infarction, however, a landmark achievement in GWAS was published in 2005 by Klein *et al.* who identified two major SNPs associated with age-related macular degeneration. In India, the first GWAS for complex conditions- Type 2 diabetes (in two major ethnic groups of India-Indo-European and Dravadian) and Quantitative lipids (within Indian cohorts) was reported by Dr. Dwaiypayan Bharadwaj and his team (2013; 2019). A two-stage pioneer research on GWAS was conducted at the CSIR-Institute of Genomic and Integrative Biology (CSIR-IGIB) and Jawahar Lal Nehru University, New Delhi in collaboration with Indian Diabetes Consortium (INDICO).

Pathway Analysis identifies biological processes, functions or pathways that are significantly over presented in a large set of genes/proteins. **Regulatory Network Analysis** maps how genes, proteins and regulatory elements interact to govern cellular behaviour.

Now the question arises- Why Genome-wide associated studies is crucial in India?

The answer lies in the fact that the Indians have a unique and profound genetic variation as compared to Western population due to complex history of migration, deep archaic ancestry and staunch endogamous practices such as marriages within the caste and consanguineous marriages. During the last 20 years, GWAS has produced spectacular genomic insights on complex disease such as Type 2 diabetes, Parkinson's disease, Arthritis, Crohn's disease, various types of cardiovascular disease, cancer and psychiatric disorders. It compares the genomes of large group of affected individuals and healthy controls to find disease-associated DNA variation.

The Government of India has initiated a massive scientific project-GenomeIndia to create a 'Indian reference genome'. This project aims to prepare a comprehensive catalogue of genetic variations found in the Indian population. It is funded by the Department of Biotechnology, Ministry of Science & Technology. GenomeIndia project is spearheaded by the Centre for Brain Research at the Indian Institute of Science, Bangalore in collaboration with 20 national institutes across India. Indian

Biological Data Centre at Faridabad, Haryana is India's first national life science data repository where all the sequenced data have been archived and stored.

In this backdrop, this Special Issue entitled: ***"Multi-Omics Perspectives on Disease Pathogenesis: Advancing Understanding Through GWAS, Pathway Enrichment, and Regulatory Network Analyses"*** consists of twenty research articles contributed by the innovative researches of one of the premier institutes of India- Apollo Institute of Medical Sciences and Research, Chitoor, India. These research articles basically deal with the integrative triad approach of GWAS, Pathway enrichment and Regulatory Network Analyses to uncover the biological pathways behind various complex disorders in humans. The highlights of the research articles have been summarized:

In the first article ***Timmapuram et al.*** studied the integration of GWAS, functional enrichment, and regulatory analyses to unravel key genetic and molecular mechanisms underlying bipolar disorder. The multi-dimensional approach enhanced the understanding of BD pathophysiology and identified potential biomarkers and therapeutic targets. In the second article, ***Lalitha Sree et al.*** elucidated a robust integrative analysis linking key developmental genes and pathways to VSD pathogenesis, strengthening the molecular understanding of this congenital defect. Its multi-layered bioinformatics approach highlights clinically relevant regulatory networks and potential therapeutic leads. Furthermore, ***Amulya et al.*** in their study reported advance understanding of CHD by integrating multi-omics datasets to uncover novel genetic contributors and regulatory mechanisms. Its comprehensive analytical approach strengthens biological interpretation and identifies potential biomarkers and therapeutic targets. The study of ***Sindhu et al.*** provides valuable genetic insights by integrating GWAS findings with functional and regulatory analyses to identify novel psoriasis-associated loci. Its comprehensive approach strengthens understanding of disease mechanisms and highlights potential therapeutic targets. ***Puthalapattu et al.*** systematically re-evaluated GWAS-identified intelligence genes using complementary bioinformatic approaches, strengthening the biological understanding of cognitive development. Its integration of functional enrichment and gene-specific insights provides meaningful evidence linking neurodevelopmental pathways to childhood intelligence. ***Anil Kishore et al.*** reported significant scientific merit by integrating multi-level bioinformatic analyses to clarify the molecular, metabolic, and regulatory disruptions underlying Tay-Sachs disease. The results provide a strong foundation for future laboratory validation and the development of targeted therapeutic strategies. ***Rajesh Kumar et al.*** in their study integrated multiple databases and analytical approaches to unravel the genetic, metabolic, and therapeutic landscape of polycythemia. Its comprehensive systems-level insights offer a valuable foundation for advancing personalised treatment strategies in the Indian clinical and research context. ***Kukkapalli et al.*** in their study integrated GWAS-derived genetic signals with functional, pathway, and interaction analyses to clarify the molecular basis of iron homeostasis. Its comprehensive approach provides valuable insights that can guide improved diagnostic assessment and personalised therapeutic strategies in populations where iron imbalance is highly prevalent. ***Ravikanth et al.*** in their study integrated GWAS findings with multi-layered functional, regulatory, and metabolomic analyses to unravel the complex genetic drivers of atopic dermatitis. Its comprehensive systems-level approach provides valuable insights that can guide future research and support the development of personalised therapeutic strategies. ***Adiga et al.*** integrated GWAS variants with functional, cellular, and metabolic analyses to unravel the complex genetic and molecular mechanisms driving Amyotropic Lateral Sclerosis. Its multidimensional approach offers valuable insights into disease heterogeneity and highlights potential molecular targets for future therapeutic development. ***Augustine et al.*** reported a comprehensive systems-level analysis linking purine metabolism defects to the neurological and metabolic features of Lesch–Nyhan syndrome. Its integrated bioinformatic approach uncovers key regulatory networks and potential molecular targets, advancing understanding of this rare disorder. ***Vasishta et al.*** reported a comprehensive multi-layered analysis of SCID-linked genes, integrating functional, pathway, regulatory, and interaction data to clarify underlying molecular mechanisms. Its systematic bioinformatic approach highlights key biomarkers and potential therapeutic targets, advancing the current understanding of SCID pathophysiology. ***Brahmaiah et al.*** integrated GWAS, pathway enrichment, regulatory analyses, and metabolomic data to uncover genetic and molecular mechanisms potentially driving prostate cancer. Its multi-dataset bioinformatic approach identifies promising biomarkers and therapeutic targets, strengthening the molecular understanding of disease progression. The work of ***Govardhan et al.*** provides a comprehensive systems-level analysis of childhood obesity, integrating genetic, pathway, and regulatory data to clarify its complex molecular architecture. Its multi-database bioinformatic approach identifies key genes and regulatory networks that may serve as valuable biomarkers and therapeutic targets. ***Padmavathi et al.*** in their study integrated genetic, metabolic, and epigenetic datasets to uncover multifactorial mechanisms driving oropharyngeal carcinoma. Its comprehensive bioinformatic approach identifies key pathways, regulators, and cellular markers with potential diagnostic and therapeutic relevance. ***Kavya et al.*** demonstrated multi-platform computational analyses to unravel the complex genetic, molecular, and metabolic mechanisms underlying hypothyroidism. Its systematic approach offers valuable insights that may guide future diagnostics and therapeutic development for thyroid disorders in the Indian population. ***Anusha et al.*** systematically integrated GWAS, functional enrichment, regulatory analyses, and machine learning to uncover genetic and molecular determinants of Age-related macular degeneration. Its comprehensive multi-omics approach provides valuable insights that may support

biomarker discovery and precision-based therapeutic strategies for AMD. ***Deepthi et al.*** studied multi-database genomic, miRNA, pathway, and metabolomic analyses to elucidate molecular basis of metabolic syndrome. Its comprehensive findings highlight lipid-metabolic dysregulation and miRNA-mediated regulation, offering valuable direction for future therapeutic research. ***Adiga et al.*** performed a comprehensive bioinformatic analyses to decode the molecular and immunological mechanisms underlying measles virus infection. Its findings highlight critical cytokine, STAT-mediated, and inflammatory pathways, offering meaningful direction for future therapeutic and preventive research. ***Dharaneedhar et al.*** in their study employed comprehensive bioinformatic analyses to delineate the immune, molecular, and metabolic mechanisms central to Sjögren's syndrome. This integrative approach highlights key cytokine targets that could inform future precision medicine and therapeutic development.

These research articles shed light on the ongoing advanced studies in India which would further help in using genetic information for clinical research, prediction of diseases via polygenic risk scores, prevention of diseases, empower precision medicine and drug treatment discovery.

In 2025, two emerging research areas-Biomedical Engineering and Computational Biology was added in *Journal of Environmental Biology* to expand its scope. It was a great opportunity to receive an invitation from Dr. Usha Adiga to publish a Special Issue. Her role as the Guest Editor in meticulous curation of this Special issue and ensuring high standards of publication is commendable. I want to take a moment to thank her for this valuable collaboration. We sincerely acknowledge the contribution of Dr. Edulla Venkataravikanth, Associate Editor of this Special issue for his Editorial Coordination. We thank all the experts for contributing their scholarly research articles for this Special issue. Their dedicated research and new perspectives would be instrumental in elevating our field. The peer reviewers of this issue are duly acknowledged for ensuring the scientific rigour of the articles. We look forward for more avenues of collaboration on future publication projects.

I express my sincere gratitude to Professor Divakar Dalela, Executive Editor and Mrs. Kiran Dalela, Managing Editor for continuous support, guidance and trust in successful completion of this scientific endeavour. I extend my sincere thanks to the Editorial Board Members for their contributions in maintaining high scientific standards of the journal. At last, but not the least, I would like to acknowledge and express my deep appreciation to all the team members for their unwavering commitment, collaborative effort, ensuring the highest quality of work in successful completion of this Special Issue.

Before concluding, I would like to share a Chinese proverb with the readers of JEB that says,

"Learning is a treasure, as it follows its owner everywhere"

So, Keep reading and continue learning!'

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