Genomics India 2019

Genomics India 2019 Conference
Transforming lives with Genomics
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Programme

Day 1 I 24th January, 2019					
Time	Event code	Event			
8.30 AM		Registration			
09:30	1.1	Inauguration - Lighting of lamp by dignitaries			
9.35AM		Welcome address by Dr. Sudha N Rao			
9.40 AM		Inaugural address by Dr. Raja Mugasimangalam			
09:55 AM		Remarks by Dr. Samir K Brahmachari [AcSIR]			
10:00 AM		Key note address by Dr. Murali Panchapagesa Muthuswamy [Jananom Pvt Ltd]			
10:10 AM		Key note address by Dr. K. VijayaRaghavan [NCBS] and Dr. Krishnamoorthy Kannan [GGSIPU]			
10:25 AM		Vote of thanks by Dr. Venkatesh Krishnamurthy			
10.30 - 11:00 AM		Tea and Poster session			
11:00 - 12:30 PM	1.2	Session 1.2A- Genomics Research and Translation	1.2	Session 1.2B - Genomics in Crop Research	
		Dr. B.S. Ajaikumar [HCG]		Dr H S Subramanya [IBAB]	
11:00 AM	1.2A.1	Dr. Kumarasamy Thangaraj [CCMB]	1.2B.1	Dr. Ramanathan Vairamani [Metahelix]	
		Burden of recessive diseases in South Asia: lesson from genomics and future perspectives		Relevance of Genomics in Industry Crop Improvement Programs – From MAS to GS – A sea change	
11.25 AM	1.2A.2	Dr. Kumar Somasundaram [MCBL, IISc]	1.2B.2	Dr Sheshshayee M.S [UAS, GKVK]	
		Whole exome sequencing of glioblastoma: mutations in CALCR identify highly aggressive patients with poor prognosis		Trait introgression to improve water productivity in Rice - A genomic and Phenomic approach	
11.45 AM	1.2A.3	Dr. B.S. Ajaikumar [HCG]	1.2B.3	Dr Kshitish Acharya [Shodhaka Life Sciences Pvt. Ltd.]	
		Clinical application of Gene sequencing and other Genomic related testing in Oncology practice			
12:05 PM	1.2A.4	Dr. Mitali Mukerji [IGIB]	1.2B.4	Dr. R. Yasodha [IFGTB]	
		From Biological intelligence to Artificial Intelligence: Traversing the path in genomics for affordable and stratified P4 medicine solutions		Exploring the Teak Genome: Insights and Challenges	
12:30 to 1:30 PM		Lunch and Poster session			
1:30 - 3:00 PM	1.3	Session 1.3A- Genomics Research and Translation	1.3	Session 1.3B - Genomics in Plant Protection	
		Dr. Sanjeev Jain [NIMHANS]		Dr. Jagadish Mittur (KITS)	
1.30 PM	1.3A.1	Dr. Gundu H R Rao [University of Minnesota, USA]	1.3B.1	Dr. Malali Gowda [TDU – FRLHT: Bangalore]	
		Cardiometabolic Diseases: Micronutrients to Micro-RNAs			
1.55 PM	1.3A.2	Dr. Sunil Raghav [ILS]	1.3B.2	Dr. Hitendra Kumar [CCMB]	
		Immunogenomics identifies direct control of NCoR1 on dendritic cell immune tolerance		Marker assisted breeding for disease/pest resistance	
2.15 PM	1.3A.3	Dr. Radha Venkatesan [MDRF]	1.3B.3	Dr Bhuban Mohan Panda [Ajeet Seeds]	
		Genomics of monogenic diabetes-From bench to bedside		Improvement in Crop Production and Protection through Genomic Approach	
2.35 PM	1.3A.4	Dr. Sanjeev Jain [NIMHANS]	1.3B.4	Dr. Naghabushan Ithal [Monsanto- Bayer]	
				Gene discovery and optimization for pest control	
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3:30 - 5:00 PM	1.4	Session 1.4A - Genome Sequence to Genomic medicine	1.4	Session 1.4B- Genomics and Microbial world	
		Dr. Vani Brahmachari [University of Delhi]		Dr. Niyaz Ahmed [ICDDR, Bangladesh]	
03:30 PM	1.4A.1	Dr. Rajan Dewar [University of Michigan]	1.4B.1	Dr Rahul Roy [IISc]	
		NGS applications in preventable and curable cancers - lowering the costs for global health delivery.		Understanding Flavivirus biology and evolution using sequencing	
03:55 PM	1.4A.2	Dr. Amit Dutt [ACTREC]	1.4B.2	Dr. Niyaz Ahmed [ICDDR, Bangladesh]	
		Translating Cancer Genomics to Medicine		Genomics Landscapes of Chromosomal and Plasmid Diversity in Multiple Drug Resistant Bacteria: Implications for Global Infection Control Priorities	
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		Vitamin B12 deficiency alters DNA Methylation leading to atherogenic risk			
5- 6:30 pm	1.5	Plenary Session			
	1.5	Dr. Rakesh Mishra [CCMB]			
	1.5A	Dr. Samir K Brahmachari [AcSIR]			
		Genomics Journey from Bangalore to Delhi and Back			
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		Genomics: Enabling Precision Medicine For Cancer			
6:45 - 7:30 PM	1.6	Awards night Chaired by Dr. Rakesh Mishra + Cultural session by Datar Institute Of Fine Arts			

07:30 PM	1.7	Dinner

Day 2 I 25th January, 2019				
Time	Event code	Event		
9:30- 11:00 AM	2.1	Session 2.1A – Big Data, AI, P4medicine	2.1	Session 2.1B - Genomics Biomarker and Vaccine
		Dr. Ramesh Hariharan [Strand Life Sciences]		Dr. Ashish Kumar Das [BITS Pilani]
9.30 AM	2.1A.1	Dr. Anurag Agarwal [Director CSIR-IGIB]	2.1B.1	Dr. Nilesh Mehta [Premier Medical Corporation]
		Artificial Intelligence (AI), Intelligence Augmentation (IA) and Intelligent Infrastructure (II) in Healthcare		
9.55 AM	2.1A.2	Dr. Ramesh Hariharan [Strand Life Sciences]	2.1B.2	Dr. Ashish Kumar Das [BITS Pilani]
		Data, AI and How we Should be Thinking About It in India		Network HUB genes and Ribosomal RNA:Possible targets for diagnostics?
10.15 AM	2.1A.3	Dr. Srinivasan Ramachandran [IGIB]	2.1B.3	Dr. Kar Muthumani [The Wistar Institute]
				Utilization of Genomic Approaches for Developing Immune and Alternative Therapeutic Interventions for Emerging and Re-emerging Infectious Diseases
10.35 AM	2.1A.4	Dr. Michael Sagner [Sports medicine]	2.1B.4	Dr. Yogesh Shouche [NCCS]
		From Systems Biology to 21st Century P4 Medicine		
11.00 - 11:30AM		Tea Break and Poster session		
11:30 - 1:00PM	2.2	Session 2.2A - Genome Informatics	2.2	Session 2.2B - Genomics in Bioproducts
		Prof. Samir K.Brahmachari		Dr. Gundu H R Rao [University of Minnesota, USA]
11.30 AM	2.2A.1	Dr. Shibichakravarthy Kannan [Theranosis Life Sciences]	2.2B.1	Dr. Prashanth D'Souza [Natural Remedies]
		Demystifying Liquid Biopsies - finding the needle in the haystack		Modulation of Chicken Caecal Microbiota by a Phytogenic Feed Additive,
				Stodi®: A Metagenomic Analysis
11.50 AM	2.2A.2	Dr. Debasis Dash [IGIB]	2.2B.2	Dr. Pradip Nair [Biocon]
		Challenges and Solutions in Genomics and Proteomics		
12.10 AM	2.2A.3	Dr. G.P.S Raghava [IIT-Delhi]	2.2B.3	Dr. Panduranga Rao [Biovet]
12.30 PM	2.2A.4	Dr. Vidya Niranjan [RVCE]	2.2B.4	BBS-3
12:50 - 2:30 PM		Lunch and Poster session		
2:30 - 3:45 PM	Moderator	Panel Discussion on Genome Science: Education, Enlightenment and Employability		
		Dr. Krishnamoorthy Kannan [GGSIPU]		
		Dr. P. Hari		
		Dr S. Krishnaswamy		
		Dr. Jitendra Kumar [Bioinnovation Centre]		
		Dr. Taslimarif Saiyed [CCAMP]		
		Dr Upendra Nongthomba [IISc]		
		Dr Maulishree [KBITS]		
		Dr S Karthikeyan [VIT]		
3:50 - 5:00 PM	2.3	Session 2.3A - Genomics for Startups & incubators	2.3	Session 2.3B - New Products for the Genomics World
		Dr. Taslimarif Saiyed [CCAMP]		Dr. Jitendra Kumar [Bioinnovation Centre]
3:50-4:05 PM	2.3A.1	Mr C Sivasankaran [Aiwo Ltd]	2.3B.1	Dr Bhupinder Hundle (Oxford Nanopore)
		Prebiotics, Probiotics & Metagenomics: The answer to lifestyle disorders		Real-time, high-throughput DNA/RNA sequencing by Oxford Nanopore
4.05- 4.20 PM	2.3A.2	Dr Shrilakshmi Desiraju [Triphase]	2.3B.2	Dr. Cynthia Gong [Novogen Ltd]
		Life outside cold storage probiotic innovation		
4.20- 4.35 PM	2.3A.3	Dr. Anand Lakshmanan [SIRPI]	2.3B.3	Dr Chandrashekaran Siddamadappa [Genei Laboratories Pvt Ltd.]
				Genomics Reagents and Kits from India
4:35- 4.50 PM	2.3A.4	Dr Naveen Kulkarni [Quantazyme]	2.3B.4	Dr. Raja Mugasimangalam
5:00 - 5:30 PM		Concluding session followed by Tea		

Abstracts
from
Speakers

Session 1.2A- Genomics Research and Translation

Dr. Kumarasamy Thangaraj
Chief Scientist,Basic Sciences- Genetics, CCMB, Hyderabad

Title: Burden of recessive diseases in South Asia: lesson from genomics and future perspectives

Abstract: South Asia is inhabited by about 5,000 anthropologically well-defined populations, many of which are endogamous communities with significant barriers to gene flow due to sociological, linguistic and cultural factors that restrict inter-population marriage. To assess the impact of endogamy, we have analysed samples from more than 2,800 individuals from over 275 distinct South Asian groups from India, Pakistan, Nepal, Sri Lanka, and Bangladesh using about 600,000 genome-wide markers. We found that 81 out of 263 unique South Asian groups, including 14 groups with estimated census sizes of over a million, have a strong founder event than the one that occurred in both Finns and Ashkenazi Jews in the West – these are founder groups known to have large numbers of recessive diseases. We identified multiple examples of recessive diseases in South Asia that are the result of such founder events. Our study provides opportunity for discovering population-specific disease causing genes in communities known to have strong founder events. Mapping of mutations that are responsible for population-specific disease would help in developing strategies for diagnosis, counseling, management and modifying the clinical course of these disorders and to reduce the disease burden among South Asians.

Dr. Kumar Somasundaram
Professor, Department of Microbiology and Cell Biology

Title: Whole exome sequencing of glioblastoma: mutations in CALCR identify highly aggressive patients with poor prognosis

Abstract: Despite significant advances in the understanding of the biology, the prognosis of glioblastoma (GBM) remains dismal. The objective was to carry out whole exome sequencing (WES) of Indian glioma and integrate with that of The Cancer Genome Atlas (TCGA) to find clinically relevant mutated pathways. We carried out WES of different glioma samples and compared to that of TCGA cohort. Further, an integrated analysis of mutated genes from Indian and TCGA cohorts was carried out to identify survival association of pathways with genetic alterations. Patient-derived glioma stem-like cells, glioma cell lines and mouse xenograft models were used for functional characterization of mutational inactivation of Calcitonin Receptor (CALCR). The results of this investigation will be presented.

Dr. Mitali Mukerji [IGIB]

Senior Principal Scientist, Genomics and Molecular Medicine, Ayurgenomics, CSIR-IGIB, DelhiTitle: From Biological intelligence to Artificial Intelligence: Traversing the path in
genomics for affordable and stratified P4 medicine solutions

Abstract: A new paradigm of network medicine for developing more effective and affordable health care solutions is felt. This is because human individuality is an outcome of evolution and adaptations over time, variability in which can also lead to emergence and evolution of new couplings in spatio-temporal scales. P4 medicine aims to integrate information along organizational, spatial and **temporal dimensions to tailor an individual's** treatment more effectively. We need innovative solutions to implement P4 medicine in ethnically and culturally diverse Indian populations not only for management of health but also for addressing the precision intervention needs in rare and complex diseases. Integration of the holistic concepts of P4 medicine of Ayurveda that is in practice over 5000 years in modern efforts through Ayurgenomics can augment the developmental needs in this area. Research highlights of an innovative integrative genomics framework that relies on understanding biology through natural variations in genomes, individuals and **populations and its large scale implementation through 'Big Data" analytics** framework would be presented.

Dr. B.S. Ajaikumar [HCG]

Chairman and Chief Executive Officer, HealthCare Global Enterprises Ltd, Bangalore
Title: Clinical application of Gene sequencing and other Genomic related testing in Oncology practice

Abstract: Despite recent advances in the treatment of cancer with the advent of better surgical expertise, radiotherapy techniques and chemotherapeutic regimes over the past 20 years, the outcome and 5 year survival of most advanced malignancies has improved only marginally. Many major drugs have initial response but the non-responder rates vary between 30% and 70%. Precision medicine in Cancer, primarily driven by developing critical cohort/population specific data has become an area of great interest to redefine our understanding of cancer progression, treatment response and prognosis. The integration of "Digital Pathology" **for an expert opinion on the biology of the tumor,** "Next Generation Sequencing (NGS)" **of the cancer genome from "tissue biopsy" and from the cir**culating **tumor DNA(ctDNA) in blood sample or** "liquid biopsy' enables us to identify specific molecu-lar defects, new biomarkers and make accurate diagnosis, rationalize treatment decision, prognosticate the disease and also monitor response/ resistance in Cancer **patients.** "Radiogenomics" is playing a significant role in understanding genetic variation associated with response to radiation through analysing radio-sensitivity index (RSI) and also correlate between cancer imaging features and gene expression.

Thus, the success of "Genomic Medicine" can already be seen in clinical practice. Since the publication of Human genome project in 2000, integration of many cost effective diagnostic platforms, there has been a significant increase in our understanding of the biology of each cancer making the disease highly treatable. Cancer is no more a deadly disease but considered as chronic lifestyle disease, if the cure is done in proper way i.e. the right way the first time"

Session 1.2B - Genomics in Crop Research

Dr. Ramanathan Vairamani [Metahelix]
Chief Technology Officer, Metahelix Life Sciences Ltd, Bangalore
Title: Relevance of Genomics in Industry Crop Improvement Programs – From MAS to GS – A sea change

Abstract: Plant Breeding has progressed by leaps and bounds with the availability of technologies that have made genotypic characterization on a large scale affordable. Molecular Breeding has become an integral part of all crop improvement programs. From its use for specific major QTLs to Genomic Selection which takes a wholesome approach for selection. The power of genomic tools can be realized very well only if it is combined with high quality phenotypic/agronomic data. Progress is also being made to enable high throughput precision phenotyping at the field level. This combination will enable breeders to sample a large variety of genetic combinations to choose from even before going to the field.

Dr Sheshshayee M.S

Chief Scientist,Professor, Department of Crop Physiology, UAS, GKVK, Bangalore

Title: Trait introgression to improve water productivity in Rice – A genomic and Phenomic approach

Dr. Kshitish Acharya [Shodhaka Life Sciences Pvt. Ltd.

Founder Director, Shodhaka Life Sciences Pvt. Ltd.

Title: Metagenomics and functional genomics approaches can enhance the discovery rates: examples from plant research

Abstract: The utility of the functional genomics (mainly the transcriptomics and proteomics) and metagenomics in the context of crop research will be discussed. It is exciting to see that well-structured conferences and workshops are catalyzing the interest of many traditional research organizations/researchers in the modern, holistic methods of molecular research. For example, during a discussion session in one of the workshops conducted by KBITS, GoK, scientists from pure agriculture and horticulture background seem to be convinced that the 'omics' approaches can be more efficient in addressing a variety of crop research goals. But many scientists who are new to such approaches seem to rush and engage with NGS methods, but often do not realize the significance of well-planned experiments and careful data analysis. I will present, as examples, research projects where my research-group was involved in NGS-based approaches: a) generating and comparing gene expression profiles (transcriptomics) in plants receiving differential treatments; and b) metagenomic analysis of microbiomes related to plant species.

Dr. R. Yasodha

Scientist, Division of Plant Biotechnology, IFGTB, Coimbatore

Title: Exploring the Teak Genome: Insights and Challenges

Abstract: Teak (*Tectona grandis L.*f) is one of the world's most valuable timber species with popular properties like extreme durability, dimensional stability, strength, excellent carvability, and distinctive appearance. Teak timber has numerous uses including ship building, indoor and outdoor furniture, flooring, paneling, plywood and decorative veneers. Although teak is native to India, Myanmar and Thailand, currently teak is grown in more than 100 countries because of its demand in the international market. India is the major importer of teak from other countries as Indians consider teak as cultural premises. However, the natural populations including old plantations of teak in India are subjected to climate change effects. Hence, infusion of genetically divergent material for large scale cultivation and conservation of teak genetic resources have become priority. Several studies have indicated the possibilities of genetic gain from tree improvement of teak for timber production. Fast advancing genomic technologies enable the acceleration of conservation and tree breeding programs. They have potential to reveal the dynamics of neutral and adaptive variation in teak populations and the processes underlie spatially explicit patterns of genetic and genomic variation. At this juncture, the efforts made in exploring the teak genome will be discussed.

Session 1.3A- Genomics Research and Translation

Dr. Gundu H R Rao [University of Minnesota, USA]

Emeritus Professor, University of Minnesota Twin Cities , Chief Technology Officer, Stellixir Biotechnologies, Bangalore

Title: Cardiometabolic Diseases: Micronutrients to Micro-RNAs

Abstract: Metabolic diseases such as hypertension, excess weight, obesity, type-2 diabetes and vascular diseases have reached epidemic proportions worldwide. Globally, obesity has increased two-fold and diabetes four-fold in the last three decades. In China during the same period, increase in diabetes and its prevalence has increased by 17-fold. Since the time I visited India, under the United Nations Development Program (TOKTEN) in 1990, I have been working on issues related to the diagnosis, management, and prevention of cardiometabolic diseases (CMDs). Major focus was to develop bilateral programs, between the US scientists and the Indian Research Collaborators. One of the earliest collaborations we initiated, was with the CSI-Holdsworth Memorial Mission Hospital (MH), Mysore in 1990. In this Hospital, research work on low birth weight children is going on since 1936. Based on the studies done at this center and the Epidemiology group at South Hampton UK, David Barker proposed a hypothesis, (Barker Hypothesis) in 1990, that intrauterine growth retardation, low birth weight and premature birth, have a causal relationship to the origins of hypertension, coronary artery disease, and non-insulin dependent diabetes, in middle age. Studies done at the MH and at South Hampton, UK, on Mysore Cohort" since the 30s, have supported Barker's hypothesis to a great extent. Indeed, Harvard researchers have successfully developed micronutrient supplements for intervention of this condition. Contrary to this observation, researchers at the Children's National Hospital (CNH) at Washington DC, USA, have proposed an alternate hypothesis, (Robert's Hypothesis), which relates to the maternal exosomal RNAs and their role in programming the lipid metabolism of the growing fetus. Bilateral studies between the Robert's Laboratory and that of Professor C.S Yajnik at Pune, is funded by the prestigious National Institutes of Health (USA), USA, under the well-known title, "Maternal Adipocyte-Derived Exosomes in the Thin-Fat Indian Baby Paradox." Professor Yajnik in an earlier study had demonstrated this paradox in fat distribution between the Caucasians and Asian phenotypes (Lancet 2004). central hypothesis is that reduced levels of maternal and cord blood adipocyte-derived exosomal microRNAs that target adipogenesis are associated with high infant adiposity. Indeed, our preliminary data support that maternal adiposity/obesity suppresses microRNAs that target adipogenesis pathway members and therefore are predicted to result in increased foetal adipogenesis. Genotypic Technology (www.genotypic.co.in), Bengaluru, is the industrial collaborator in this US-India Bilateral research project. In this brief overview, we will discuss some of the salient findings on this topic of great public health importance.

Dr. Sunil Raghav [ILS]

Scientist D & Ramalingaswami Fellow, Institute of Life Sciences

Title: Immunogenomics identifies direct control of NCoR1 on dendritic cell immune tolerance

Abstract: Dendritic cells (DCs) link innate to adaptive immunity and regulate a fine balance of inflammatory and tolerogenic responses to prevent immune pathology. Conventional Type I DCs (cDC1) cross-present antigens to T cells upon encounter with intracellular pathogens to educate naïve T cells to differentiate into effecter subtypes Th1, Th2 or Tregs. Our overall goal in lab is to understand the transcriptional control of DC responses. It has been reported that strong nuclear receptors (NR) ligands perturb DC responses by changing their activation and cytokine secretion patterns. NRs regulate their target genes by forming complexes with co-repressors like NCoR1. NCoR1 also form complex with other TFs like NFkB and AP-1. We identified that NCoR1 mediated direct repression of immune tolerance in cDCs is essential for development of an optimal immunogenic response. To explore the underlying mechanism we performed integrative genomic analysis of NCoR1 depleted DCs. NCoR1 depletion upregulated a wide-variety of tolerogenic genes in activated DCs, which consequently increased frequency of $CD25^+FoxP3^+$ regulatory T cells (Tregs) *ex vivo* and *in vivo*. Moreover, NCoR1 strongly represses the PU.1 bound super-enhancers on major tolerogenic genes upon activation. NCoR1 depletion reduced RelA activity after activation whereas RelB activity was unaffected providing DCs a tolerogenic advantage. Interestingly, our genomic data showed an enriched anti-viral network as well in activated NCoR1 KD DCs. These DCs secrete high IFNβ1 and inhibit viral infection by generating a cascade of anti-viral responses against negative strand RNA viruses like Sendai, VSV and NDV. We validated the findings *ex vivo* and *in vivo* using control and $NCoR1^{DC-/-}$ animals. NCoR1 has been reported to form complexes with diverse HDACs to repress their target genes. Therefore we speculated that differential HDAC complex with NCoR1 might be controlling these dichotomous antiviral *vs* tolerogenic responses. Here we will discuss further how we identified the mechanisms underlying the transcriptional control of these diverse responses by NCoR1 in DCs.

Dr. Radha Venkatesan

Executive Scientific Officer & Head, Dept. of Molecular Genetics, MDRF

Title: Genomics of monogenic diabetes – from bench to bedside"

Abstract: Type 2 diabetes (T2D), is a common complex disorder and polygenic in nature. In contrast the rarer forms of diabetes like Maturity Onset Diabetes in the Young and Neonatal diabetes are more penetrant and monogenic in nature.

In the case of monogenic diseases such as (MODY) and Neonatal Diabetes, the genetic testing has now come to the realm of clinical practice as these are single gene defects which can be identified by genetic testing. Maturity-onset diabetes of the young (MODY) is an autosomal dominantly inherited form of diabetes. It is usually diagnosed before 25 years of age, MODY is a group of clinically heterogeneous forms of beta cell dysfunction that are defined at the molecular genetic level by mutations in different genes (eg., *HNF4A, GCK, HNF1A, NEUROD1, HNF1B, PDX1, INS*). In Asian Indians, type 2 diabetes occurs earlier and often overlaps with MODY and hence understanding and recognition of MODY subtypes in this population gains importance. Neonatal diabetes occurs much earlier in life, that is in the first year of life. Potassium channel gene defects are major cause of this condition. We have Our work has revealed important insights into the molecular genetics of these monogenic diabetes. Genetic diagnosis, which has become widely available, is of great clinical importance for patients with monogenic diabetes. It helps to understand the pathophysiology of the disease, tailor the optimal antidiabetic treatment, and define the prognosis for the entire family.

One of the most gratifying clinical applications is in the diagnosis of Neonatal Diabetes5which is defined as diabetes occurring in the first 6 months of life. Children with neonatal diabetes with certain specific mutations have been successfully switched over from insulin therapy to oral sulfonylurea which is a great boon to the patient and the family. The time has now come to take genetic testing to the diabetic clinic in the case of monogenic forms of diabetes.

Dr. Sanjeev Jain
Professor,Molecular Genetics Laboratory, NIMHANS, Bangalore

Session 1.3B - Genomics in Plant Protection

Dr. Malali Gowda
Professor and Head, Functional Genomics & Bioinformatics, Trans Disciplinary University (TDU), Bengaluru
Title: Decoding of Medicinal Plants Genomes using Multi-omic approaches

Abstract: Medicinal plants have been used in traditional medicine since ages in India. Plants have used as the raw material for production of several modern drugs. TDU has documented over 5000 medicinal plants and established *in-situ* gene pool conservation sites for over 100 species in various parts of India. Unlike crop plants (e.g., rice), medicinal plants genomes have not been studied. Since past 5 years, our team has attempted to decode and understand the functional genomic elements of Indian medicinal plants including Neem tree (*Azadirachta indica)*, Tulsi or Holy basil *(Ocimum album)*, Sandalwood tree *(Santalum album)*, Seetha tree *(Saraca asoca)* and Bhodhi/Peeple tree (*Ficus religiosa).* Our medicinal plants research platform is the first of its kind in the world, which connect plant science, conservation science and modern medical science. This unique platform will ignite research on medicinal plants and explore the scientific facts beyond the hidden treasure of traditional Indian systems of medicine and agriculture.

Dr. Hitendra Kumar

Senior Scientist, CSIR-CCMB

Title:Marker assisted breeding for disease/pest resistance

Abstract: Marker Assisted Selection (MAS) is a very effective tool for crop improvement that involves use of variations in DNA sequences as flag posts to make selections of varieties that have new and advantageous combinations of genes. MAS can be applied for crop improvement when the trait of interest is present within the gene pool/extended gene pool of the crop of interest. I will present our collaborative work with the Indian Institute of Rice Research, Hyderabad, on development of a rice variety that is resistant to bacterial blight as a case study on how MAS can be gainfully used for crop improvement. This variety, called Improved Samba Mahsuri (ISM), is in commercial cultivation and is becoming popular in rice growing regions where fine grained varieties are cultivated and bacterial blight is a production constraint. Furthermore, we have developed an EMS mutagenized population of the elite rice variety Samba Mahsuri and used it to isolate mutants that exhibit enhanced tolerance to yellow stem borer, a serious insect pest of rice and sheath blight, a serious fungal disease of rice. Mutants have also been isolated that have any one or more of the following characteristics: higher yield, early duration and strong culm. The strong culm characteristic will be particularly useful in coastal areas of our country where it can help in imparting enhanced tolerance to lodging that is caused by cyclonic conditions. Our results to date on characterization of these mutants and the manner in which they can possibly be used to extend the scope of MAS will be discussed.

Dr. Bhuban Mohan Panda

Scientist, Ajeet Seeds
Title: Improvements In Crop Production And Protection Through Genomic Approach

Abstract: To feed the growing population of 9 billion by 20150, and the 70% increase in the demand for agricultural production that is expected to accompany this increase. A broad range of improvements in the global food supply chain is needed through plants science research. Most of the Earth's arable land is already in production and remaining occupied by urbanization, salinization, desertification and environmental degradation, cropland expansion is not a viable approach to food security. Development and deployment of high-yielding crop varieties will make a vital future contribution to sustainable agriculture. Environmental changes impacts food, feed, and fiber crop production. In addition to these environmental stresses, losses to pests and diseases are also expected to increase. Loss caused by these abiotic and biotic stresses, which already result in 30%–60% yield reductions globally. A reduction in losses to pests, pathogens, and environmental stresses is equivalent to producing more, to creating more land and more water. Genomics has played, and will increasingly play, an important role in contributing to the advancement of the global agriculture and agri-food (AAF) sector. Genomic techniques starting from Marker assisted breeding (MAB), Transgenics, Gene Editing, Genotyping by Sequencing (GBS), siRNA and TILLING has supplemented substantially to the growth of agriculture. There is need for data platforms to collect, compare, annotate, combine, analyze, store, interrogate, and share large data sets that combine different types of information, including genotypes, phenotypes, and environmental conditions.

Dr. Naghabushan Ithal

Lead Prospecting and Competitive Intelligence team, Monsanto Research Center, a Subsidiary of Bayer AG at Bangalore
Title: Gene discovery and optimization for pest control.

Abstract: Several global trends are influencing the agriculture today. While raising population and changing economies and diet are increasing the demand for food, limited farmland, climate change and evolution of resistance to current crop protection practices are putting pressure on food supply. So, an integrated approach is needed to meet the future food demand. Genomics play a key role in discovery of resilient crops and crop management systems. In the crop protection space, specifically for pest management, insecticidal microbes and their toxins are being used for decades. The Bt toxins are popular insecticidal proteins for pest control via transgenic approach. Development of insect resistance to Bt toxins necessitates discovery of new sources of proteins with insecticidal activity. However, the difficulty with culturing most of the soil bacteria is a major limitation in finding new sources of insect toxins. Metagenomics is the culture independent technique that helps in overcoming this issue and allow us to discover novel insect toxins from uncultured species. Continuous evolution technique can be used to further improve the activity of the newly discovered toxins. In this presentation, I will discuss some of our approaches towards gene discovery and optimization.

Session 1.4A - Genome Sequence to Genomic medicine

Dr. Rajan Dewar
Associate Professor, Director, Hematology Laboratory, Department of Pathology, University of Michigan
Title: NGS applications in preventable and curable cancers - lowering the costs for global health delivery.

Abstract: Next generation sequencing techniques have been useful for diagnostics, but have not been adopted extensively because of concerns with costs and awareness of novel utilities among clinical communities. Project Samyuktha is an IIT Madras incubated company that has been implementing low cost cervical cancer screening based on their work on rural India in the last 10 years. This lecture will focus on a novel approach to reduce the costs by high volume multiplexing and HPV genotyping. The second cancer that is curable is pediatric leukemia. The speaker will highlight the importance of NGs to identify distinct subtypes of pediatric ALL. We are very keen in working with partners to implement NGS based assays for the above two cancers.

Dr. Amit Dutt
Principal Investigator, Scientist, Wellcome Trust/DBT India Alliance Int Fellow, ACTREC, Mumbai
Title: Translating Cancer Genomics to Medicine

Abstract: Massively parallel Next Generation DNA sequencing technologies has made technically feasible to interrogate the complete set of genomic alterations in a tumor in a systematic, comprehensive manner in a single run. These methodologies are beginning to transform diagnostics by allowing cancers to be classified based on molecular mechanism and allowing clinical trials to be undertaken on more homogeneous groups of patients; and, therapeutics by sparking a new generation of drugs targeted at the molecular alterations that cause cancer. Advances made by Dutt Laboratory leading to the establishment of TMC-SNPdb, first Indian SNP db, to facilitate cancer research using genomics data from Indian origin samples, along with discovery of novel molecular subclasses, new therapeutic targets and biomarkers for clinical development—with detailed mechanistic insights-- in head and neck; lung; and, gallbladder carcinoma will be presented.

Dr. Gautam Arunachal
Department of Human Genetics,NIMHANS

Dr Shantanu Sengupta
Principal Scientist, Cardiovascular Disease Biology Unit, CSIR-IGIB, New Delhi
Title: Vitamin B12 deficiency alters DNA methylation leading to atherogenic risk

Abstract: Maternal nutritional deficiency *in-utero* is known to predict risk of complex disorders like cardiovascular disease, diabetes and many neurological disorders in the offspring and vitamin B12 is one such critical micronutrient. Vitamin B12 deficiency has long been associated with many complex disorders. It is a critical micronutrient that acts as cofactors for the enzymes methionine synthase (methyl Cbl) and methyl malonyl-CoA mutase (Ado-Cbl). A major proportion of Indians adhere to a strict vegetarian diet and hence are deficient in vitamin B12. We have shown the association of low B12 levels with **coronary artery disease (CAD) in Indian population. According to Barker's** *Fetal Origins Hypothesis: Fetal environmental exposures, especially nutrition, can directly affect embryonic development and "program" the child for adult health outcomes'* probably by altering the global epigenetic changes. To understand the effect of maternal micronutrient deficiency we have used rat as a model animal to delineate the genome wide epigenetic changes (DNA methylation) that occur due to the in-utero maternal micronutrient deficiency. The significantly differentially methylated regions have been identified in the B12 deficient rat pups. Simultaneously we performed proteomics experiments in the liver of offsprings. Both DNA methylation and proteomics data hints at altered lipid metabolism via PPAR, which acts as a master regulator for alteration of lipid metabolism. An important facet of mechanistic insight was shown to be the disruption of beta oxidation pathway in the B12 deficient pups. Interestingly the supplementation of Vitamin B12 during parturition and conception reverted back several epigenetic (DNA methylation) marks and protein expression up to control level.

Session 1.4B- Genomics and Microbial world

Dr. Rahul Roy
Assistant Professor, Molecular Biophysics Unit, IISc
Title: Understanding Flavivirus biology and evolution using sequencing

Abstract: RNA viruses mutate and adapt to their host and environment constantly. This makes it difficult to study these pathogens and develop vaccines and antivirals as interventions. We leverage next generation sequencing along with microfluidics to develop tools and methods that allow us to generate insight regarding lifecycle, host-pathogen interaction and evolution of Flaviviruses like Dengue and Japanese Encephalitis viruses. Using direct virus genome sequencing from clinical samples of Dengue positive serum samples from across India, we have mapped the prevalent Dengue strains in the country. We find convergent evolution and emergence of intra-host species in the patients that highlights the importance of understanding the dynamics of viral quasispecies in the host. To this end, we have develop a single virus (RNA) genome sequencing pipeline. Our method can generate the full-length sequence of individual viral RNA molecules along with their copy numbers and thereby providing the tools to track the sequence variations in a viral population.

Dr. Niyaz Ahmed
Senior Director, ICDDR, Bangladesh
Title: Genomics Landscapes of Chromosomal and Plasmid Diversity in Multiple Drug Resistant Bacteria: Implications for Global Infection Control Priorities

Abstract: Molecular epidemiology of pathogenic bacteria has largely been carried out throughfunctionally neutral or inert sequences mostly entailing polymorphic gene loci or repetitive tracts spread throughout bacterialgenomes. However, it is veryimportant toengage functionally relevant markers in order toassigna valid epidemiological context to tracking of pathogens suchastheir acumento acquire multiple drug resistance (MDR), their phenotypic diversity with reference to clinical or community level dynamics of incidence/transmissionas well astheirresponse or refractoriness to treatment. This presentation aims at thediscussion of the aboveideas in light of our works entailinghigh throughput genomics andfunctional epidemiologyof multiple drug resistant bacteriastudied from different settingsin South Asian neighbors, India and Bangladesh. Further, it will also be possible to present and discuss our recent analyses based on an extensive trajectory of plasmidsfrom different compatibility groups entailingenteric and non-enteric pathogens that forma deep landscape of their diversity and plasticity relevant to their propensity to acquire, confer, disseminate,and/orshuffle/shuttle fitness traits across different species and lineagesof pathogenic and environmentally dwelling bacteria. Discussion ofthese findings would enlighten us of the sheer prowessof bacteriato evolve and spread with new fitness advantages such as MDR phenotypes. It is believed that approaches based on such multi-dimensional and multicentric strategies as mentioned above wouldlikely be successful in targeting the spread of MDR pathogens and guiding the global infection control priorities and policies.

Session 2.1A - Artificial Intelligence (AI), Intelligence Augnentation (IA) and Intelligent Infrastructure (II) in Healthcare

Dr. Ramesh Hariharan
CTO, Strand Life Sciences

Dr. Anurag Agarwal
Director, Institute of Genomics & Integrative Biology (IGIB), Delhi

Dr. Vidya Niranjan

Professor & HOD, Computational Biology, R V College of Engineering, Bengaluru

Dr. Michael Sagner

Head of Department Preventive and Lifestyle Medicine, University Hamburg Medical Center, USA

Session 2.1B - Genomics Biomarker and Vaccine

Dr. Ashish Kumar Das
Professor, Department of Biological Sciences, BITS Pilani
Title: Network HUB genes and Ribosomal RNA:Possible targets for diagnostics?

Abstract: Malaria is caused by protozoan parasites belonging to the genus *Plasmodium*. The four most prevalent of the human malaria parasites are *P .falciparum*, *P .vivax*, *P. malariae* & *P.ovale*. These infect humans in tropical and subtropical regions of the world and are thought to cause upwards of 2 million deaths annually. Malaria remains a very important vector borne disease in India, which is estimated as possessing the most disease burden amongst all the countries in the S.E Asia region.

Plasmodium falciparum has long been known to cause complicated malaria including cerebral malaria. It is only recently that *P. vivax* has also been accepted as causing the same. The talk will focus on WGCNA (Whole Genome Co-Expression Network Analysis) to identify HUB genes from microarray analysis of parasite material, from patients exhibiting uncomplicated or complicated malaria, caused by *P. falciparum*. This has provided information about various genes encoding hypothetical proteins which appear to be key players in disease pathogenesis on the parasite side. Some of these could provide possible diagnostic/ therapeutic targets of the future. IFA data of localization patterns of one of the HUB genes will also be presented.

Parasite ribosomal RNA has long been thought of as a diagnostic target in malaria. We will report, in the second part of the talk, a ribosomal capture assay using genus specific probes against the 28S rRNA of the parasite which has shown high sensitivity and will pave the way for capture assays using combinations of genus specific and species specific probes, designed and validated by us.

Dr. Nilesh Mehta
President at Premier Medical Corporation, Watchung, New Jersey

Dr. Kar Muthumani

Director, Laboratory of Emerging Infectious Diseases, Assistant Professor of Vaccine & Immunotherapy Center, The Wistar Institute

Title: Utilization of Genomic Approaches for Developing Immune and Alternative Therapeutic Interventions for Emerging and Re-emerging Infectious Diseases

Abstract: There is an urgent need to develop improved vaccination techniques that provide effective and lasting protection against viral infection. It is thought that the induction of strong T cell responses and cross-neutralizing monoclonal antibodies (mAbs), as well as antibodies that drive ADCC, plays a key role in vaccine-induced protection. The development of vaccines against highly infectious pathogens such as Human Immunodeficiency Virus (HIV-1), Influence A virus (Flu), Dengue virus (DV), Chikungunya virus (CHIKV), Respiratory Syncytial Virus (RSV), Middle Eastern Respiratory Syndrome (MERS) and Zika virus (ZIKV) has been wrought with difficulties. Recent advances in human antibody isolation have uncovered broadly neutralizing antibodies (bNAbs) that are capable of preventing infection against a wide array of viral pathogens. Yet generating and delivering biologically-relevant levels of such antibodies using conventional methods is impractical, often requiring excessive expenses and repeated administrations. Creating new methods of delivering neutralizing mAbs that overcome these constraints could drastically tip the scales in the fight against a number of devastating viral pathogens.

In the current approach, we have constructed an optimized, enhanced DNA plasmid formulation capable of expressing a neutralizing antiviral IgG. A single administration of the IgG plasmid resulted in the generation of IgG molecules in mouse and capable of binding and neutralizing activity against viral target. Importantly, this delivery method resulted in a more immediate increase in antibody levels, lasted for months, and protected against viral challenge. This approach establishes a new platform for delivering protective mAbs safely and effectively. The study has implications for prophylactic and therapeutic strategies against viral infections and other important diseases, especially in resource-limited settings where antibody therapy is cost-prohibitive.

Dr. Yogesh Shouche

Principal Investigator, National Centre for Microbial Resource, NCCS, Pune

Yogesh Shouche NCMR-NCCS Pune 411007

Title: Microbiome initiatives at National Center for Microbiome Research

Abstract: National Center for Microbial Research was established by Department of Biotechnology Government of India in 2008 as Microbial Culture Collection. Today it is the largest collection of microbes under the single roof. It is recognized as Designated Repository by Ministry of Environment, Forests & Climate Change under the Biodiversity Act 2002 and also as International Depositary Authority under the Budapest Treaty by World Intellectual Property Organization.

The researchers are actively involved in Microbial Taxonomy, diversity and microbiome research and have worked on several interesting projects on defining core microbiome of Indian population and the changes within it during dysbiosis, microbiome changes in river ecosystem during Mass Bathing Event, Dep bioshphere microbiome etc.

The talk will summarize salient features of some of these studies.

Session 2.2A - Genome Informatics

Dr. Shibichakravarthy Kannan
Founder & CEO at Theranosis Life Sciences Pvt Ltd, Hyderabad

Title: Demystifying Liquid Biopsies - finding the needle in the haystack

Abstract: Current trends in cancer diagnostics include the concept of liquid biopsy which is now a tested and proven technology that is rapidly impacting the lives of cancer patients. The main purpose of performing a liquid biopsy is to assess the tumor mutational status non-invasively by sequencing the cell free DNA of tumor origin (ctDNA). But there are several clniically important mutations that are found less 0.1% frequency in the patient population. Classic NGS approaches do not have the sensitivity to detect such variants. In this session we will discuss the various wet lab and bioinformatics data analysis approaches to detect ctDNA with confidence.

Dr. Debasis Dash

Senior Principal Scientist, Informatics and Big Data unit, CSIR-IGIB, New Delhi
Title: Challenges and Solutions in Genomics and Proteomics

Abstract: Advances in mass spectrometry-based proteomics have enabled generating large scale tissue wise human proteome data. Despite the ongoing refinement of the human proteome, there are still a lot of missing or unexplored variations in the proteome which has sufficient evidence at the transcript level. This gap in the knowledge of proteome can be filled by employing proteogenomics strategies to unravel proteoforms that majorly arise from sequence polymorphism, alternative splicing and proteolysis. Tissue wise catalogue of proteoforms will aid in understanding fundamental working of biological systems as well as could explain disease inception. Identifying this pool of unexplored proteoforms has always remained challenging due high similarities of identified peptides that necessitates an efficient protein inference solution. A strategy is being devised to explore the human tissuescape of proteoforms which will help to detect diversity among different tissues. Publicly available mass spectrometry datasets like PRIDE contains data from various human tissues which will be searched against the in-house-built customized database containing different transcripts and amino acids variant information from GENCODE and neXtProt using a multi-algorithmic proteogenomic approach [3]. Currently, we have analyzed brain tissue-specific datasets from different regions of the brain to identify several known and novel brain proteoforms and developed a repository for the human brain-specific proteoforms.

Dr. G.P.S Raghava
Head & Professor (CB), PhD, Institute of Microbial Technology, Chandigarh

Dr. Srinivasan Ramachandran
Senior Principal Scientist, Informatics and Big Data Unit, IGIB, Delhi

Title: Genome data analytics in Infectious diseases and in complex diseases

Abstract: Genome data are encased in literature, gene expression datasets and various repositories. I shall present the analysis of these datasets in case of infectious diseases and in complex diseases using the published softwares developed in our group. The analysis reveals the innovative genome strategies used by pathogens. In case of complex diseases revelation includes the common genes in co-morbidity cases, examples of inclusive index, most notably in diabetes associated complications. Significant Challenges remain in genome informatics, which may need disruptive ideas.

Session 2.2B - Genomics in Bioproducts

Dr. Gundu H R Rao
Emeritus Professor, University of Minnesota Twin Cities
Chief Technology Officer, Stellixir Biotechnologies, Bangalore

Bangalore Biotech Start-ups (BBS) company

Dr. Pradip Nair
Principal Scientific Manager, Biocon Limited., Bangalore

Dr. Panduranga Rao
Associate Vice President, Biovet Private Limited

Dr. Prashanth D'Souza
Assistant General Manager at Natural Remedies Private Limited., Bangalore

Title: Modulation of Chicken Caecal Microbiota by a Phytogenic Feed Additive Stodi®: A Metagenomic Analysis

Abstract: Background: The cecal microbiota plays a critical role in gut health and utilisation of nutrients left undigested in the small intestine. Objective: The impact of Stodi® on the composition of caecal microbiota was evaluated in broilers using high-throughput sequencing of 16S rRNA gene amplicons. Materials and Methods: A total of 960 one-day-old Ross 308 chicks were allocated to four groups viz. normal diet (ND), negative control (NTC; challenged with 1.7% magnesium chloride (MgCl2), and Stodi® treatment groups which comprises negative control plus Stodi® (500 and 750 g/ton of feed). MgCl2 was used to increase the cecal moisture content and in turn to disturb the cecal microbiota. Results: Birds challenged with MgCl2 exhibited poor performance traits as compared to ND group, whereas the supplementation of Stodi® (500 and 750 g/ton) improved the performance of broilers. MgCl2 did not produce a notable change in the microbiota, but supplementation of Stodi® (500 and 750 g/ton) produced a statistically significant shift in the microflora in comparison with ND. The Firmicutes to Bacteroidetes ratio was significantly elevated in comparison to ND. The abundance of energy harvesting bacteria belonging to specific families of Lachnospiraceae and Ruminococcaceae were increased by Stodi® supplementation, especially at 500 g/ton of feed. Conclusion: The supplementation of Stodi® was effective in modulating the caecal microbial population in a manner conducive for gut health and performance of broilers, as revealed by the increase in abundance of favourable microflora.

Mr C Sivasankaran
Aiwo Ltd
Title: Prebiotics, Probiotics & Metagenomics: The answer to lifestyle disorders

Abstract: The human gut microbiota contains trillions of microorganisms, including at least 1000 different species of known bacteria with more than 3 million genes (150 times more than human genes). Every individual has a unique footprint of the gut microbiota. The composition of an individual's gut microbiota changes with age, feeding (breast milk) type, birth mode (c-section vs normal) and other factors. The human gut microbiota functions as an organ and is critical for modulation of immune response, colonization resistance, and nutritional needs. The composition of the gut microbiota can be altered with diet, prebiotics and probiotics. For example, the probiotic Lactobacillus Acidophilus has been shown to break down enzymes like casein and gluten, reduce GI concentrations of carcinogenic enzymes, and enhance innate & acquired immunity. Similarly, Inulin a prebiotic has been shown to decrease tissue inflammation, reduce pH in patients with IBD, Crohn's disease and Ulcerative colitis across multiple randomised clinical trials. Prebiotics and Probiotics are easy to consume and when coupled with other treatment approaches can lead to significant clinical improvements across multiple disease modalities. Pre- and pro-biotics will play a key role in changing the current paradigm of curative healthcare to the pre-emptive, preventive healthcare model.

Dr Shrilakshmi Desiraju

Director Business Development, Triphase Pharmaceuticals Pvt.Ltd.

Title:TEMPERATURE STABLE PROBIOTICS (TSP)

The market pain of probiotic industry is non availability of high tempt stable , pH stable probiotic strains. This also results in lower shelf life there by increasing stability issue to the ingredient and the product formed out of the use of the ingredient.

Triphase introduces temperature-stable natural lactobacilli probiotic strain; *Lactobacillus plantarum* (TSP-Lp1) & *Lactobacillus acidophilus* (TSP-La1) have the ability to withstand harsh manufacturing process especially required during making Food /Beverage, confectionaries and Pharma industries.

Most of the probiotics currently available in the market can't survive harsh environment like during manufacturing & acidic environment of the stomach. These currently available Probiotics are enteric coated to withstand harsh conditions. Probiotics that fail to reach intestinal tract alive are less likely to provide digestive & immune support.

Dr. Anand Lakshmanan

CEO, SIRPI.io

Title : Tidyverse Programming Methods for Data Visualization : Get data-ready and be prepared for the AI revolution in healthcare.

Abstract: The open source R Programming language has evolved to become more "human-friendly". Dr. Anand Lakshmanan will discuss practical aspects of the tidyverse programming methods using R that you can implement at your organisation to get organised and be prepared for the AI revolution in healthcare. You'll get a peek into data importing, data cleaning, data processing and data visualizations using programming geared towards non-programmers (typically bio researchers and decision makers). And it's easier than you think !

Naveen Kulkarni
CEO Founder Quantumzyme

Quantumzyme is a biotransformation company, focused on Clean and Green Chemistry. Customer centric research to enhance enzyme activity, selectivity and specificity by applying novel Quantum Mechanics, Molecular Modelling and engineering approaches.

Quantumzyme's uniqueness lies in its advanced framework "QZyme Workbench®" that offers advanced solutions for specific biocatalysis requirements.

QZyme Workbench® has already been acknowledged by customers worldwide in successful design of the enzymes that has achieved remarkable results in industrial scale.

Session 2.3B - New Products for the Genomics World

Dr Bhupinder Hundle (Oxford Nanopore)

Director, Distribution Sales, Oxford Nanopore Technologies

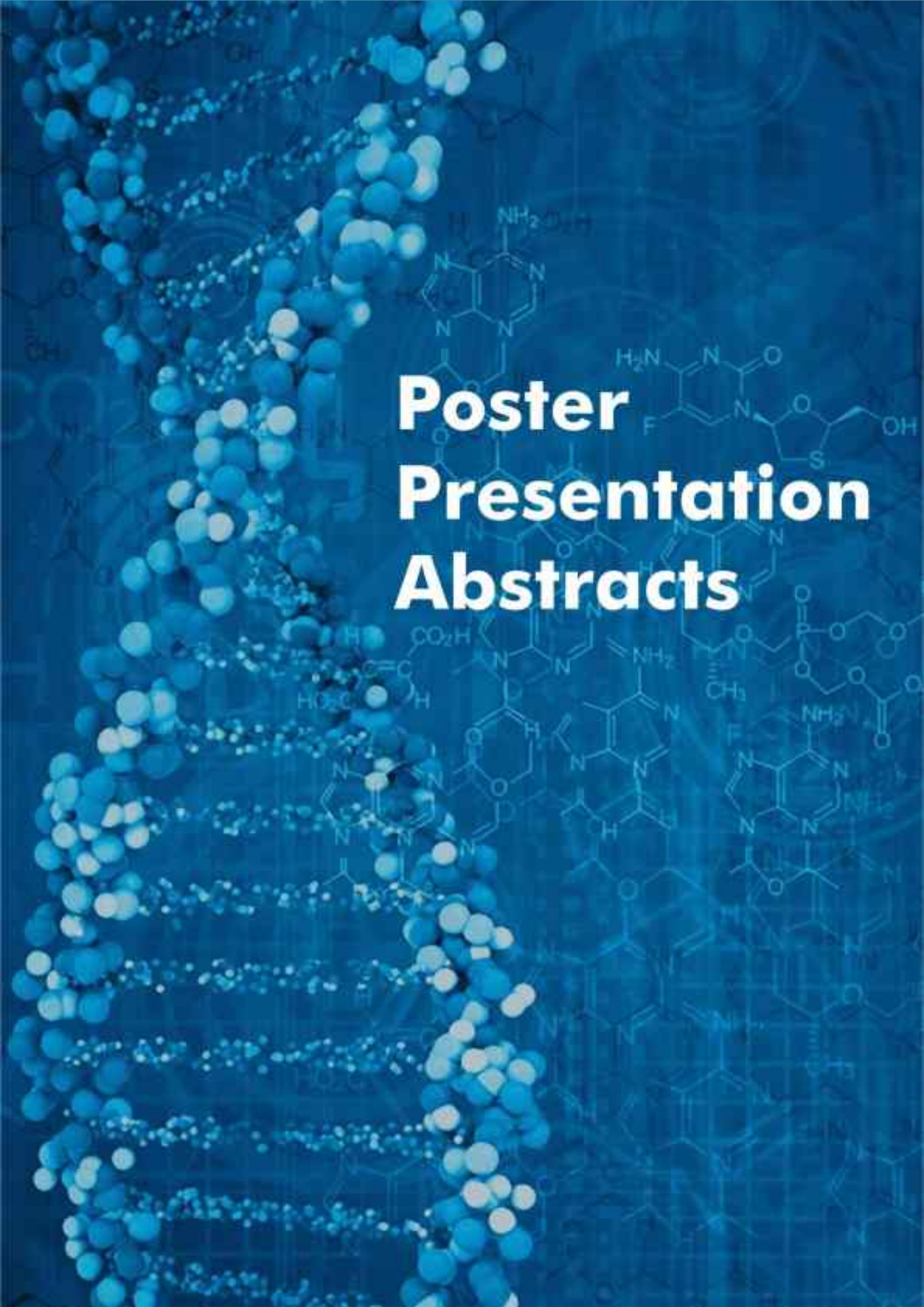Title: Real-time, high-throughput DNA/RNA sequencing by Oxford Nanopore

Abstract: Oxford Nanopore Technologies has developed the world's first and only nanopore DNA sequencer, the MinION. The MinION is a portable, real time, long-read, low cost device that has been designed to bring easy biological analyses to anyone, whether in scientific research, education or a range of real world applications such as disease/pathogen surveillance, environmental monitoring and food chain. The pocket-sized MinION is a powerful and portable sequencing device that can deliver high volumes of long read sequence data. Our novel, electronics-based DNA/RNA sequencing technology is being used for a range of biological research applications. These include large scale human genomics, cancer research, microbiology, plant science and environmental research. Nanopore sequencing is also being explored beyond research, where it has the potential to provide rapid, meaningful information in the fields of healthcare, agriculture, food and **water surveillance and education. Oxford Nanopore's proprietary technology is fully** scalable for any requirement. Small formats such as Flongle address the need for on-demand, rapid, smaller tests or experiments, and can be used in labs or in the field. The benchtop GridION X5 can run up to five MinION Flow Cells at a time, on-demand, for larger genomics projects. The recently launched PromethION is the largest format for nanopore sequencing, designed to offer on-demand use of up to 48 Flow Cells – each of which can offer more than 100Gb of sequencing data.

Cynthia Gong

Sales Director, Novogen Ltd.

Dr. Chandrashekaran Siddamadappa
Chairman & Managing Director, Genei Laboratories Pvt Ltd.

Title: Genomics Reagents and Kits from India

Dr. Chandrashekaran Siddamadappa
Chairman & Managing Director, Genei Laboratories Pvt Ltd.

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Poster
Presentation
Abstracts

MUTATION ANALYSIS IN MLPA NEGATIVE DUCHENNE MUSCULAR DYSTROPHY: NEXT GENERATION SEQUENCING AS A DIAGNOSTIC TOOL PRIOR TO MUSCLE BIOPSY.

Polavarapu K [1] , Saroja M [2] , Gunasekaran S[1], Preethish-Kumar V [1] , Sekar D [1] , Nashi S [1] , Vengalil S [1] , Thomas P [1] , Krishnamurthy V[2], Rao S [2] ,Nalini A [1]

[1] National Institute of Mental health and Neurosciences (NIMHANS), Bengaluru Karnataka, India

[2] Genotypic Lab, R&D Division, Bengaluru Karnataka, India

Background: Duchenne muscular dystrophy (DMD) is the most severe and common form of childhood muscular dystrophies resulting from mutations in Dystrophin gene (Xp21), affecting about 1 in 3500 boys worldwide. While 75% patients have deletions/duplications of one or more exons, smaller mutations - point mutations, insertion/deletions (INDELs) etc., occur in 20-25% who require direct sequencing of entire DMD gene or invasive muscle biopsy for diagnostic confirmation. Next generation sequencing (NGS) offers a cheaper and higher throughput alternative to traditional Sanger sequencing and can be developed as a single diagnostic platform to identify both copy number variations (CNVs) and small mutations in DMD patients.

Methodology: Clinically suspected and/or biopsy confirmed DMD children in whom MLPA (Multiplex ligation-dependent probe amplification) did not identify deletions/duplications of exons were recruited from our Neuromuscular clinic. Custom probe design for DMD gene was created using Agilent's SureDesign™ tool with a total capture size of 2.077Mbp to cover entire gene (exons, introns and promoter regions) at least twice. After obtaining informed consent/assent, DNA extracted from blood was used to prepare libraries and sequenced on NextSeq™ (Illumina).

Results: Mutational analysis was performed in 64 MLPA negative DMD children with mean age of 7.87±2.3 (range:3-13 years). Majority were sporadic cases and family history (X-linked recessive) is present in only six cases (9.37%). Biopsy was performed in 51 boys (79.7%) and showed loss of Dystrophin on immunohistochemistry. Target NGS identified hemizygous mutations of DMD gene in 58/64 children (90.6%). Nonsense mutations resulting in stop codon were most common at 54.7% (35/64) followed by frameshift mutations due to small INDELs 21.9% (14/64). Small INDELs of up to 10 bases were identified accurately. Variants affecting splicing occurred in 8/63 (12.5%) out of which six involved invariant GT and AG dinucleotides at the 5' end (splice donor) and 3' end (splice acceptor) of introns. Missense mutation was identified in only one patient. Except for one splice mutation where variation occurred nine bases upstream of exon 12, all other variants were classified as pathogenic (55) or likely pathogenic (2) as per ACMG guidelines. In total there were 57 unique variants among which 60% (34) were novel and only one mutation (p.Arg539*) recurred in two unrelated patients. Unlike larger CNVs, which occur predominantly in exons 45–55 and 2–10 for deletions and duplications respectively, small mutations lacked any hot spot regions and more uniformly spread across coding region with exons 30 and 44 having most number of mutations (4 each). In thirteen patients who did not undergo biopsy and clinical suspicion was high, we were able to accurately identify pathogenic mutations. Mutations were not identified in only six cases (9.5%), where possibility of deep intronic variants/complex rearrangements should be considered.

Conclusion: In this study we describe spectrum of smaller mutations in a large cohort of DMD children from India. Accurate diagnosis of these mutations is important to identify potential cases who can benefit from mutation specific therapies like nonsense read-through and NGS offers a valuable diagnostic screening tool before contemplating invasive muscle biopsy in clinically suspected DMD patients.

IDENTIFICATION OF MUTATIONS IN A COHORT OF UNCLASSIFIED INHERITED MUSCLE DISORDERS BY TARGETED NEXT GENERATION SEQUENCING.

Polavarapu K [1], Joshi A [2], Preethish-Kumar V [1], Santhosh R [1], Mathur A [2], Nayak S [2], Ambawat S [2], Nashi S [1], Vengalil S [1], Narayanappa G [1], Faruq M [2], Nalini A [1]

[1] National Institute of Mental health and Neurosciences (NIMHANS), Bengaluru Karnataka, India

[2] CSIR-Institute of Genomics and Integrative Biology, New Delhi, India

Background: Inherited myopathies and muscular dystrophies are a genetically heterogenous group of neuromuscular disorders which often present with overlapping clinical and pathological phenotypes. Although muscle histochemistry, immunostaining, western-blot and electron microscopy techniques offer important clues, these are often found insufficient to provide a conclusive diagnosis. With rapid evolution of gene discovery, accurate molecular diagnosis is essential for these disorders to provide appropriate genetic counselling, prognostication and identification of disease burden essential for future therapeutic research. Targeted Next generation sequencing (NGS) with gene panels are emerging as quick low-cost methods to diagnose mendelian disorders.

Methodology: Randomly selected patients with clinical features suggestive of Limb girdle muscular dystrophies (LGMD), distal myopathies (DM) and congenital myopathies (CM) evaluated over last 10 years in our Neuromuscular clinic were taken up for genetic analysis by Targeted NGS with a gene panel constituting 53 known muscular dystrophy and myopathy genes. DNA extracted from blood was utilized for library preparation as per Agilent SureselectQXT protocol followed by sequencing on Hlseq Illumina platform.

Results: We performed Target NGS in 45 patients with mean age of 21±11.7 (3-49 years) and male/female ratio of 3:2. Broad clinical phenotypes included LGMD: 31 (68.9%), CM: 9 (20%), DM: 5 (11.1%). Majority of cases were sporadic: 33 (73.3%), while family history was present in 12 (26.7%): Autosomal dominant inheritance in 7, recessive history in 3 and suspected X-linked inheritance in 2 families. Serum Creatine kinase was elevated in 82.2% (>5 times: 22, <5 times: 15). Muscle biopsy reports available in 42 patients showed muscular dystrophy in 25 (60.9%). Myopathic features were reported in 17 patients showing varying degree of cytoplasmic inclusions in 9 cases and rimmed vacuoles in 4. Putative disease-causing variants in 17 genes were identified in 31 patients (68.8%) and diagnosis was possible in 21 (46.66%) where clinical suspicion was supportive, and variants were either reported pathogenic or novel rare variants (MAF=0) recognized as damaging/probably damaging by in-silico predictions (Table 1). In another 10 cases, variants were identified with uncertain significance/not exactly correlating with phenotype and need to be further characterized. Most common genes affected were CAPN3 (Calpain-LGMD2A) in 5 patients followed by DYSF (Dysferlin-LGMD2B), EMD (Emerin-EDMD) in 4 cases each, RYR1 (Ryanodine receptor1-CM) in 3 and GMPPB (LGMD 2T), LMNA (LGMD 1B/EDMD) in 2 each.

Conclusion: Targeted gene panel NGS offers a quick cost-effective strategy with good diagnostic yield in inherited muscle disorders, when complimented with good clinical phenotyping.

Title: INFLUENCE OF CYP2C9 GENETIC POLYMORPHISM ON VALPROIC ACID-INDUCED ADVERSE EFFECTS IN EPILEPTIC PATIENTS OF SOUTH INDIA ORGIN.

Kesavan R, Kirubakaran R and Sunil K Narayan*

Department of Pharmacology and Neurology*, JIPMER Pondicherry 605006

Introduction: Valproate is one of the commonly used anti-epileptic drugs. It is a drug with broad spectrum antiepileptic activity and it is being used as first-line agent in most of the seizure disorder. Though valproic acid is well tolerated by majority of the patients, but when adverse drug reactions emerge it demands either dose reduction or withdrawal. Valproate also shows large inter-individual variation in its pharmacokinetics which could explain the differences in response or ADRs among patients. The variation in pharmacokinetics can be due to an influence by genetic or non-genetic factors. The most commonly observed polymorphisms are CYP2C9*2 (430 C > T) and CYP2C9*3 (1075 A>C). The genetic composition of South Indian population is different from other major ethnic populations. Hence this study was planned to find out the strength of association between CYP2C*2, CYP2C9*3 polymorphisms and valproate-induced adverse effects.

Material and Methods Cases and controls were recruited from epilepsy clinic in a tertiary care hospital. Individuals with suspected or confirmed ADR to valproate monotherapy are considered as cases. Controls were the subjects on valproate monotherapy for more than 3 months without any evidence of ADR. 79 cases and 79 controls were recruited. 3-5 ml of venous blood was collected for plasma total valproic acid level estimation and DNA extraction for genotyping. Genotyping of CYP2C9 *2 (C > T, rs1799853; assay ID: C 25625805 10), and CYP2C9 *3 (A > C, rs1057910; assay ID: C 27104892 10) was done by Real Time Thermocycler (ABI Prism 7300, Foster City).

Results There was a significant difference in plasma valproic acid and dose of valproate (mg/kg//day) between cases and controls. When adjusted for dose, BMI, valproate levels and other genotypes, CYP2C9*3 allele was associated with 3 times the risk of developing ADR to VPA. There is no significant influence of CYP2C9 polymorphisms on plasma valproic acid levels.

Conclusion The genotyping of CYP2C9*3 allele in epileptic patients of south India can be used to reduce VP-induced ADR and optimize the therapy for maximum benefit.

CONTACT INFORMATION: Dr Kesavan R Associate Professor Dept of Pharmacology JIPMER Pondicherry 605006 kes7ramasamy@gmail.com Mobile : 9360532592

DE NOVO SEQUENCING AND HYBRID ASSEMBLY OF BIOFUEL CROP JATROPHA CURCAS L: INSIGHTS ON GENE ANNOTATION, COMPARATIVE GENOMICS AND TRANSCRIPTOME ANALYSIS WITH IDENTIFICATION OF QUANTITATIVE TRAIT LOCI FOR GEMINIVIRUS RESISTANCE

Nagesh Kancharla[1], Saakshi Jalali[1], J. V. Narasimham[1], Vinod Nair[1], Vijay Yepuri[1], Bijal Thakkar[1], Neeta Madan[1], Arockiasamy S[1*]

[1] Reliance Technology Group, Reliance Industries Limited, Navi Mumbai, Maharashtra, India;

Jatropha curcas is an important perennial, drought tolerant plant identified as a potential biodiesel crop. We report here the hybrid de novo genome assembly of *Jatropha curcas* generated using Illumina and PacBio sequencing technologies and identification of quantitative loci responsible for geminivirus resistance.

In this study, we generated scaffolds of 265.7 Mbp in length which is corresponding to 84.8% of the gene space using BUSCO analysis. Additionally, 96.4% of predicted protein-coding genes were captured in corresponding tissues transcriptome data, which reconfirms the accuracy of the assembled genome. The genome was utilized to identify 12,103 dinucleotide repeat SSR markers, which were exploited in genetic diversity analysis to identify genetically distinct lines. A total of 207, polymorphic SSR markers were employed to construct a genetic linkage map for *Jatropha* Mosaic Virus (JMV) resistance using interspecific F_2 mapping population involving *Jatropha curcas* and *Jatropha integerrima* as parents. QTL analysis showed the identification of three minor quantitative trait locus (QTL) for JMV resistance and validated in an alternate recipient genetic background F_2 mapping population. These validated QTLs were utilized in Marker Assisted Breeding (MAS) for developing *Jatropha* Mosaic Virus resistance hybrids in Jatropha. Comparative genomics of oil-producing genes across selected oil producing species revealed 27 conserved genes and 2,986 orthologous protein clusters in *Jatropha*. This reference genome assembly gives an insight into the understanding of the complex genetic structure of *Jatropha* and serves as source for the development of agronomically improved virus resistant and oil producing species.

THE GENOMIC LANDSCAPE OF CYP2D6 ALLELIC VARIANTS IN INDIAN POPULATION

Ambily Sivadas, Surabhi Rathore, Lipi Thukral, Vinod Scaria

* CSIR-IGIB, Delhi

Background: Genetic variation plays a prominent role in altering the therapeutic response of individuals and thereby represents the fundamental principle of precision medicine. Cytochrome P450 (CYP) is an important family of heme-thiolate-containing enzymes which metabolizes a large number of drugs in use today. Fifteen percent of the total drug metabolism is performed by CYP2D6 enzyme which is characterized by high inter-individual variation in its efficiency and quantity.

Objective: The aim of the study was to investigate the allelic diversity and differential distribution of CYP2D6 alleles in the Indian population. We also explored the functional consequence of alleles unique to Indian population using in silico approaches.

Materials & Methods: The phased genetic variation data for South Asian populations including structural variations was obtained from 1000 Genomes Project in VCF format and processed using in-house haplotyping scripts to determine the CYP2D6 allelic diplotype of individuals. The Indian haplotype frequencies were estimated and compared with the frequencies of other global populations. The functional prediction of Indian-specific haplotypes were performed using pathogenicity scores from SIFT, CADD and also using molecular dynamics (MD) simulations.

Results: The most predominant CYP2D6 alleles were identified as *1 (40.2%), *2 (21.4%), *41 (12%), *4 (8.6%), *10 (3.8%) and *5 (2.6%). We report six low frequency alleles (*86, *7, *111, *112, *113, *99) that are specific to Indian population with a cumulative frequency of 5%. The phenotype frequencies are 87.8%, 1.8%, 4.9%, 2.9%, 2.4% for Extensive, Ultrarapid, Intermediate, Poor and Unknown metabolizer status. We observed significant differences in the CYP2D6 allele frequencies between Indian subpopulations. Four out of the six Indian-specific haplotypes had unknown functional status. Pathogenicity prediction scores from SIFT and CADD identified three of them to have lower function or no function. Apo MD Simulations (500ns) of the wildtype and two mutants (*86 and *113) revealed large conformational changes and longer time to convergence in case of the mutant structures compared to the wildtype thereby supporting our predicted functions.

Conclusion: Large-scale comprehensive analysis of publicly available Indian whole genomes revealed unique CYP2D6 alleles in the population with unknown functional consequences. Functional analysis using in silico approaches predicts lower or no function for these alleles which affect about 5% of the population. Comprehensive characterization of CYP2D6 alleles in the population can aid precise phenotyping of the metabolizer status of individuals and help minimize undesirable therapeutic response.

STUDY OF EARLY STAGES OF HNSCC : THE ROLE OF CIRCULAR RNA AND MICRO RNA.

Ms. Shaheerah Khan
Institute Of Life Sciences, Bhubaneshwar, Odisha

Abstract: India is amongst the top three countries having the highest rate of Oral cancer. The detection of it in early stage and also with a non invasive technique like 'Liquid Biopsy' is the requirement. Circular RNAs are a novel member of the noncoding RNAs. With technologies like next-generation sequencing (NGS), especially RNA-seq technology, over 30,000 circRNAs have already been discovered. circRNAs are exceptionally stable molecules and some have been shown to function as efficient microRNA sponges. CircRNAs play important roles in the carcinogenesis of cancer, in many aspects of malignant phenotypes, including cell cycle, apoptosis, vascularization, and invasion,RNA sponge, and binding to RBP. The main objective is to see if these circularRNAs can act as biomarkers for HNSCC, in the primary stage of cancer.

A complete picture of how circularRNA-microRNA interaction effects the microRNA expression in various cancers would be of immense value for researchers. The current work explores the interaction between circularRNAs, microRNAs and its gene target, studying the correlation based upon their expression, by utilising the concepts of different branches of Bioinformatics such as Data Mining, NGS, Network Biology and Systems Biology.

The study aims at first integrating this information in a database. Analysis of these circularRNAs and integrating TCGA to find the Driver genes and mutations, and potential microRNA helps predict circularRNAs as microRNA sponge. The Study further aims to find signatures in Oral Lichen Planus.

These regulatory molecules have been found to be involved in various cancers and due to their stability can be detected in blood thereby providing a new opportunity for design of new diagnostic tools. The future work involves modelling of these circularRNAs, so as to design small molecule modulators that has the potential to open a new avenue in Drug Design and Discovery.

SNPRIORITY: A SYSTEMS GENOMICS PARADIGM TO FILTER DISEASE CAUSATIVE SNP

*Praharshit Sharma (Member- ISCB and ACS, usa)
Former SRF: TRI, Thrombosis Research Institute- Bengaluru.
Bioinformatics Research Fellow, Birla Institute of Scientific Research Jaipur.

Background: Based on the first-of-its kind "Systems Genomics approaches" workshop in India, having attended by the presenting author (Courtesy of [1]) that concluded recently, this work is an endeavor toward the same in terms of programmatic pipeline after variant annotation stage is past.

Objective: To abridge the computational pipeline of "Finding needle-in-a-Haystack", so to say pin-point the "One (or Couple of) "Exonic" single nucleotide polymorphisms (SNP) including synonymous mutations thereby validate the association of genotype to disease-phenotype diagnosed in clinic.

Materials & Method: After variants are duly annotated using ANNOVAR web-based interface [2] by supplying the respective variant call format (VCF) files for three typical phenotypes, namely "bleeding disorder", "short stature" and "epilepsy" respectively and correspondingly, the resultant variant annotation metadata (CSV, or XLS formats) is subject to a simple three-line R-script using various parametrized constraints to filter the unique SNPs for each of the clinical phenotypes, duly supplemented by exhaustive gene panels manual curation in order to obtain consensus.

Results and Conclusion: Upon filtering the SNPs as per above-mentioned procedure, the four-cases of Autosomal Dominant or Recessive and Homozygosity or Heterozygosity thereof are addressed carefully using a systematic lookup of the OMIM db, Online Mendelian Inheritance in Man database [3]. As the compendium of variant filtering and annotation is gathered over time, the far-fetched dream of genome-to-phenome can be realized. Further, an "R-package" can be deployed for above purpose.

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INDENTIFICATION OF STAGE-WISE miRNA SIGNATURE AS POTENTIAL BIOMARKER IN HEAD AND NECK SQUAMOUS CELL CARCINOMA

Sugandh Kumar

Institute Of Life Sciences, Bhubaneshwar, Odisha

The head and neck squamous cell carcinoma (HNSCC) is one of the predominant causes of cancer-related casualties globally. The high mortality rate of HNSCC is due to late diagnosis in the majority of cases. The early detection of primary tumour and prevention of relapse are the major research challenges associated with HNSCC. The early stage markers may prove to be essential tools in the timely diagnosis of the disease and improved clinical outcome. However, currently, not one molecular marker has been widely accepted for routine use in managing patients with HNSCC. Therefore, a study relating the association between clinical stages and gene expression profiles is very much required. The comparative analysis of expression profiles between early and late stages has uncovered miRNAs with stage-dependent alterations in expression in various cancers. These miRNAs play important roles in oncogenesis and chemo-resistance. However, no study has reported the relationship between clinical stages and miRNA expression profiles at the genome level in HNSCC. We have used the different tools (limma-voom DESeq2 and edgeR) to rank differentially expressed miRNAs in a stage-wise (TNM) manner. The Cox regression was used to identify stage-specific miRNA signatures. Further, the Kaplan-Meier survival analysis revealed the prognostic value of identified miRNAs. The functional enrichment (Pathway and Gene Ontology) analysis suggested that the target genes of identified miRNAs may be involved in the ECM degradation, EMT and cellular metabolic processes, Integrine, IL-10 in early stages. We hope the current studies will be useful for the development of diagnostic and therapeutic approaches for HNSCC. Key words: Biomarker, DEG, miRNA-mRNAs, survival analysis, Pathways.

BAR-HRM TECHNOLOGY FOR AUTHENTICATION OF SPICES AND DETECTION OF ITS ADULTERANTS

Nayana G N, Rajashekhar V Ballari, Ramya Shanivarsanthe Leelesh, and Abhijeet Tibile, Krishna Prasad and Raja Mugasimangalam*

QTLomics Technologies Pvt Ltd, Bengaluru. Email: raja@qtlomics.com, Website:www.qtlomics.com.

Background: Spices are low volume high valued commodity of agriculture produce and they significantly contribute to the GDP of the country. Common spices namely black pepper, turmeric, ginger, cardamom, cinnamon etc., are heavily adulterated in the market which is of a significant concern to end users in term of value and health risk. Identification and authentication of the spices are important for consumers, importers and exporters. Lack of robust, cost effective, definitive and analytical tools/methods are impeding the process of identification and authentication. DNA based molecular tools are ideal to detect and authenticate the spices.

Objective: To develop BAR- HRM kit for authentication and detection of adulteration of spices.

Materials and methods: DNA was isolated from different spices from both authentic spices and commonly known adulterants using modified CTAB extraction method. DNA was quantified using Nanodrop spectrophotometer. Primers were designed targeting the chloroplast genes (rbcL and matK) and nuclear genes (ITS1 and ITS2). Bar-HRM PCR was carried out using the primers for different spices along with their adulterants.

Results and discussion: Bar-HRM method showed different melting curve, differential plot and clear temperature shift for different spices and the adulterants which directly correlates to robust identification of spices. Sensitivity of method to detect adulteration was carried out by using different proportion of the DNA samples of different spices. The method is sensitive to detect 1% adulteration. Bar HRM method developed is an accurate, convenient and rapid tool for market supervision, and accurate, fast/rapid, cost effective and robust tool and method for spices authentication and detection of adulteration.

GENOMICS APPROACHES TO DECIPHER DIVERSITY AND FUNCTIONAL ATTRIBUTES OF CELLULASES FROM PSYCHROPHILIC ENVIRONMENT OF SELECTED SITES OF NE INDIA

Santosh K Gupta1,2#, Rajesh K Prasad1, Soumya Chatterjee1*, Sonika Sharma1, Sibnarayan Datta1, Mohan G Vairale1, Rajeev Gupta1, Sanjai K Dwivedi1
1. Defence Research Laboratory, DRDO, Tezpur 784001, Assam
2. Dept of Biotechnology, Bharathiar University, Coimbatore 641046, Tamil Nadu
 # presenting author; *corresponding author

Abstract: Diversity of microbial enzyme is key to know various ecosystem processes. In environmental conditions, the diversity of bacteria cannot be ascertained only through laboratory cultures. Culture-independent sequencing of environmental samples are important to acquire information on qualitative and quantitative diversity of the organisms and their genome to determine functional potential. NGS based metagenomics is an evolving tool that can decipher several information using multi-features bioinformatics approaches like, usage of mono-codon, single amino acid, ORF length coverage, Z-curve features, bioinformatic and structural modelling etc. Using deep stacking networks learning model for prediction of multiple genes and gene products, metagenomics can be helpful even for working on hydrolytic enzymes, like, novel cellulases and its glycoside hydrolases gene. In nature, especially in the high altitude regions, cellulases can be exploited to convert lignocellulosic biomass to value-added products related to waste biodegradation and other industrial applications like generation of bioethanol. These industrial applications require cellulolytic activity under psychrophilic conditions. The present work aims at the diversity profiling of the selected high altitude regions (above 10000 ft) around Tawang, Arunachal Pradesh of NE of india. Naturally aggegated waste samples were collected from six selected sites for assessing both bacterial diversity (through NGS platform) and lab cultures (for culturable bacteria using PCR studies). Cellulase activities were assessed at different set of temperature using carboxy methyl cellulose (CMC at 1%). Out of 40 different isolates from six sites, above 27% of bacteria isolates displayed the ability to degrade carboxymethyl-cellulose, indicating the presence of cellulolytic activity. This study is important to determine natural degradation of cellulose at psychrophilic conditions. Keywords: metagenomes; biotechnology; cellulase; glycoside hydrolase.

IMMUNO-INFORMATICS ANALYSIS OF THEILERIA GENOME FOR IDENTIFICATION OF NOVEL VACCINE CANDIDATES AND DESIGN OF A MULTI-EPITOPE BASED VACCINE CANDIDATE

Ms. Prajna Parimita Kar, Anand Srivastava

National Institute of Animal Biotechnology

Theileriosis poses a serious threat to ruminants and is prevalent in tropical and subtropical countries causing huge economic losses to farmers. It is a tick-borne disease, caused by Theileria, an apicomplexan parasite. Further, with increasing cases of resistance to commonly used drugs, it is highly desirable to develop better and cost-effective vaccines against theileriosis. The only available vaccine, live attenuated parasite vaccine, has many drawbacks and hence is unsuitable for controlling this disease. With the availability of whole genome sequence of this pathogen, it is possible to predict the antigen which may play a great role in the production of immune response. In this study, we have used an immuno-informatics driven genome-wide screening strategy to identify potential vaccine targets containing important and effective dominant immunogens against Theileria. The proteins with a probability of plasma membrane localization or GPI anchor were screened from Theileria annulata proteome. The non-homologous proteins to the host (bovine) were selected and their antigenicity was analysed. Further the antigenic proteins were analysed for the B cell and T cell epitopes. The identified B cell epitope mapped in the modelled structure of the proteins. A total of 19 linear epitopes in 12 proteins, exposed in the extracellular space and having the potential to induce protective antibodies, were obtained. Additionally, CTL epitopes, which are peptides with 9-mer core sequence, were also identified, modeled and docked with bovine MHC-I structures. The CTL epitopes showing high binding energy with the bovine MHC-I were further engineered in-silico to design a putative multi-epitope vaccine candidate against Theileria parasites. The docking studies and molecular dynamics studies with the predicted multi-epitope vaccine candidate and modelled bovine TLR4 exhibited strong binding energy, suggesting that the complex is stable and the putative multi-epitope vaccine candidate can be a potentially good candidate for vaccine development.

ZAP INSERTS™: A NOVEL METHOD FOR MOLECULAR CHARACTERIZATION OF GMOS AND CRISPR GENE EDITED PLANTS BY NGS

Rajashekhar V Ballari, Nayana G N, Keya Mukherjee, Abhijeet Tibile, and Raja Mugasimangalam*

Qtlomics Technologies Pvt Ltd, Bengaluru.
Contact number: +91-6361531573.
Email: contact@qtlomics.com, www.qtlomics.com

Background: In Agriculture GMOs (Genetically modified Organisms) technology is applied for pest resistance, herbicide resistance, abiotic stress tolerance and also for vitamins and protein enhancement leading to higher nutritive values. Regulatory approval of genetically engineered crops requires defining the insertion events, characterization, and description of the inserted genetic material, sequence data and of the flanking region bordering the site of insertion. Southern blot and polymerase chain reaction (PCR) and molecular biology techniques combined with Sanger sequencing are commonly applied approaches. These techniques are both time- and resource-consuming and never can comprehensively characterize all insertion events. Next-generation sequencing (NGS) is the preferred alternative to southern blotting and Sanger sequencing.

Objective: To develop NGS based, comprehensive, robust and cost-effective method for the characterization of GMOs and CRISPR edited plants.

QTLomics team developed ZAP inserts™ that applies NGS in a novel workflow to elucidate and characterize all the insertion events. Insert target-specific PCR was carried out. Nanopore DNA library was prepared using pooled PCR products and Sequencing was carried out on the MinION sequencer (Oxford Nanopore Technology). Novel NGS analysis work flow was developed to elucidate the inserts quickly.

Result and Discussion: We have developed a novel method to characterize the transformation event by a unique PCR and NGS based strategy using Nanopore MinION sequencing called as ZAP inserts™. Further bioinformatics analysis showed insert and flanking sequences of transgenic DNA. The developed method is robust, less time consuming and cost effective and applied successfully to commercial samples. QTLomics has applied ZAP inserts™ method for successfully characterizing recombinant inserts in Tobacco, Rice, cotton, eggplant (brinjal), tomato and eucalyptus.

PREDICTION OF TUBERCULOSIS DRUG-RESISTANCE USING WHOLE GENOME SEQUENCING DATA

Arup Ghosh[1] , Viplov K. Biswas[1], and Sunil K. Raghav[1]

Infectious Disease Biology, Institute of Life Sciences, Bhubaneswar, Odisha, INDIA

According to recent WHO Global tuberculosis report 2017, India accounts for more than 25% of global tuberculosis cases. Also, India has the second highest rate of multidrug resistance(MDR) cases and the highest mortality rate. Although the drug-sensitive cases had a higher cure rate the MDR and XDR TB cases had only 54% and 30% were only successfully treated. Prevalence four major lineages *Mycobacterium tuberculosis*, East African Indian (EAI), Beijing, Central Asian Strain(CAS), Euro-American (EA) are observed across India. The major challenges prevail with the current TB diagnostic strategies are the time required for culture-based tests(2-4 weeks) resulting less number of patients follow-up with the treatment and commercially available diagnostics fail to account for novel compensatory mutations leading to drug resistance development. This suggests the need for more specific molecular diagnostics to be developed using whole genome sequencing. In this study, we have tried to aggregate the single nucleotide mutations from publicly available tuberculosis whole genome sequencing data from India and use them to predict drug-resistant phenotype using machine learning algorithms.

Currently, there are only a handful whole genome sequencing studies of tuberculosis (clinical strains) has been conducted in southern parts of India. Till date, we have collected 297 clinical isolate sequencing data from NCBI Sequence Read Archive (SRA) of which 37 are multidrug resistance (MDR) and 62 monoresistant (to either isoniazid, streptomycin or quinolones).

Using minimum base quality filter of 30, minimum map quality filter of 20 minimum depth quality filter of 20 we identified more than ten thousand unique mutations combining all the isolates. The prediction accuracy using different machine learning models for different antibiotics were between 60-80%. The lower prediction accuracy is due to a lower number (20% of total dataset) of isolates belonging to the category and demonstrates the requirement of more whole genome sequences form more drug-resistant isolates. Application of region wise drug-resistant tuberculosis whole genome sequencing will help us to develop more precise drug resistance diagnostics tests.

RGS PROTEINS AND THEIR ROLE IN CANCER ETIOLOGY

Dikshaya P.[1], Varun PC. [1], Karyala Prashanthi*[2], Amrita Roy*[1]

Department of Biochemistry, Indian Academy Degree College
Department of Biotechnology,Indian Academy Degree College

ABSTRACT: Cancer with its unregulated cell division and resistance to apoptosis are frequently associated with dysfunctional activation/ suppression of GPCRs and associated proteins. And considering the great diversity in GPCRs and their associated proteins, their involvement do suggest myriad possibilities. Consequently, RGS (Regulators of G Protein Signalling) proteins—as one of the major regulators of GPCR signalling—have emerged as a crucial factor governing cancer etiology and had already generated considerable research interest.

In the last decade or so, several independent research groups have indicated the altered expression level of several RGS proteins in various kinds of tumors and the implications of such altered expression have also been sporadically investigated. But a clearer picture regarding the role of the RGS proteins in tumor biology is yet to emerge. In this respect, a bioinformatics based approach was employed to screen the expression level of some RGS proteins between normal and tumor tissues (Breast and Lungs) by using several microarray datasets available in public domain. The expression level of RGS17 and RGS4 was found to be greatly upregulated in lung and breast cancers respectively compared to their normal counterparts. The datasets were further analyzed to determine the correlation of target RGS proteins—RGS17 in lung cancers and RGS4 in breast cancers—with their protein interactors. The protein network that emerged shed a light of the dynamics between the RGS proteins and their interacting partners and might be further exploited to understand the overall effect of RGS proteins on tumor etiology.

GENOMIC INSIGHTS OF VIBRIO HARVEYI RT-6 STRAIN, FROM INFECTED "WHITELEG SHRIMP" (LITOPENAEUS VANNAMEI) USING ILLUMINA PLATFORM

*[1]Thirugnansambandam, R., [1]Inbakandan, D., and [2]Kirubagaran R.

[1]Centre for Ocean Research, Sathyabama Institute of Science and Technology, Chennai - 600 119, Tamil Nadu, India.

[2]Marine Biotechnology Division, National Institute of Ocean Technology, Ministry of Earth Sciences, Govt. of India, Chennai - 600 100, Tamil Nadu, India.

Abstract:

Background: *Vibrios* are the phenomenal test models for genomic classification because they are abundant in the aquatic environment, related with a wide range of marine life. The pathogenicity of "Vibriosis" in Shrimps impose prominent menace to the sustainable growth of maricultural economy. Often the disease outbreak is associated erroneously with *Vibrio harveyi* and closely related species. The present study was investigated to explore the molecular insight of complete genome of the strain *V. harveyi* RT-6.

Methods:*Vibrio harveyi* strain was isolated from an infected shrimp, *Litopenaeus vannamei* collected from aquaculture ponds, India (12.1899° N, 79.9249° E). The whole genome sequencing (WGS) was performed using the Illumina Hiseq 2500 platform and assembled *de novo* using SPAdes and Velvet optimiser. Furthermore, the gene prediction and annotation were performed by a rapid prokaryotic genome tool-Prokka.

Results:The genome of *V. harveyi* RT-6 strain has one circular chromosome with 6,374,398 bp long. *V. harveyi* RT-6 strain contains 5912 predicted genes with an average of 45.7% GC content. A total of 94 known genes associated with pathogenicity were identified and 36 genes were found to be responsible for virulence factors. Furthermore, 1088 unigenes were subjected to Gene Ontology (GO) terms, and 5730 predicted proteins were annotated with Clusters of orthologous (COGs), 3401 unigenes were Kyoto Encyclopedia of Genes and Genomes (KEGG) functional groups, 13 insertion sequences-(IS) and one incomplete prophage region were also identified.

Conclusion:The whole genomic sequencing of *V. harveyi* RT-6 contains clues to pathogenicity and spurs the development of more powerful approaches to further study on genomic basis of complex hereditary traits.

NCOR1 CONTROLS THE TOLEROGENIC PROGRAM IN DENDRITIC CELLS

Abdul Ahad1,2, Shuchi Smita1,2, Mathias Stevanin3, Gyan Prakash Mishra1,4, Bhawna Gupta4, Hans Acha-Orbea3, Sunil K. Raghav1,2,*

1Immuno-genomics & Systems Biology Laboratory, Institute of Life Sciences (ILS), Bhubaneswar, India
2Manipal Academy of Higher Education, Manipal, India
3Department of Biochemistry CIIL, University of Lausanne, Epalinges, Switzerland
4Department of Biotechnology, Kalinga Institute of Industrial Technology (KIIT), Bhubaneswar, India

Nuclear receptor co-repressor 1 (NCoR1) has been studied as an important regulator of gene expression in a variety of contexts. We show here that NCoR1 strongly represses the tolerogenic program in DCs that allows them to develop pathogen-specific immunogenic responses. In addition, NCoR1 depletion in DCs upregulates a wide variety of tolerogenic genes irrespective of activation and as a consequence, promotes regulatory T cell (Treg) differentiation in vitro and in vivo. Mechanistically, NCoR1 represses the aryl hydrocarbon receptor (AhR) gene expression which known to form a dynamic interaction with NFkB subunits especially with RelB to induce variety of anti-inflammatory genes. We identified that NCoR1 masks the PU.1 bound super-enhancers present on tolerogenic genes after DC activation that are subsequently bound by activating transcription factors like NFkB (RelB). We found that AhR expression is enhanced in NCoR1 depleted DCs and it interacts with RelB in NCoR1 KD DCs. Which may be leading to enhanced RelB binding on tolerogenic genes leading to their enhanced expression.. Furthermore, the bacterial and parasite infection in NCoR1 DC-/- animals enhanced Treg development with a concomitant increase in disease burden. Likewise, adoptive transfer of activated NCoR1 KD DCs in helminth- infected mice increased both Tregs and intestinal worm load. Altogether, our results highlight NCoR1 as a promising target to generate tolerogenic DCs.

DESIGNING OF CLAUDIN-4 INHIBITORS BY STRUCTURE BASED VIRTUAL SCREENING, MOLECULAR DYNAMICS AND IN VITRO STUDIES

Majji Rambabu and Sivaraman Jayanthi*

*Corresponding Author:
School of Bio Sciences and Technology, Vellore Institute of Technology, Vellore-632014

ABSTRACT

Background: Claudin-4 (CLDN4) is a vital member of tight junction (TJ) proteins that is often overexpressed in cancer and other malignancies.

Objective: To identify inhibitors of Claudin-4 for cancer.

Methods: The three-dimensional (3D) structure of human CLDN4 was constructed based on homology modelling approach. A total 2,65,242 molecules from the National Cancer Institute (NCI) database has been utilized as a dataset for this study. In the present work, structure-based virtual screening is performed with the NCI database using Glide.

Results: By molecular docking, ten candidate molecules with high scoring functions which binds to the active site of CLDN4 were identified. Subsequently, molecular dynamic simulations of membrane protein were used for optimization of the top three lead compounds (NCI110039, NCI344682 and NCI661251) with CLDN4 in a dynamic system. The lead molecule from NCI database NCI11039 (Purpurogallin Carboxylic Acid) was synthesized and cytotoxic properties were evaluated with A549, MCF7 cell lines. Our docking and dynamics simulations predicted that ARG31, ASN142, ASP146 and ARG158 as critically important residues involved in the CLDN4 activity.

Conclusion: Finally, three lead candidates from the NCI database were identified as potent CLDN4 inhibitors. Cytotoxicity assays had proved that purpurogallin carboxylic acid had an inhibitory effect towards breast (MCF7) and lung (A549) cancer cell lines. Computational insights and *in vitro* (cytotoxicity) studies reported in this study are expected to be helpful for the development of novel anti-cancer agents.

Keywords: claudin-4, homology modeling, molecular dynamics, virtual screening

STUDY ON COLD STRESS RESPONSIVE DIFFERENTIALLY EXPRESSED GENES IN RICE CULTIVARS

Rajani Kumari, Bobby Bharti, Dev Mani Pandey*

Department of Bio-Engineering, Birla institute of Technology, Mesra, Ranchi, Jharkhand, India.

*Presenting & Corresponding E-mail Id: dmpandey@bitmesra.ac.in, 09431900315

Abstract

Rice (*Oryza sativa* L.), one of the most important crops, has now emerged as an ideal model species for the study of crop genomics due to its commercial value and relatively small genome size. Cold is one of the abiotic stress that significantly affect the agriculture worldwide, resulting in substantial losses in crop yield. To address this problem, we must first understand how plants respond to adverse conditions by Genomic studies with the goal to identify the genes involved and their expression patterns during stress perception and response. The present study considered Transcription factors/regulators and their differential expression analyses and co-expression networking under cold stress condition in four rice cultivars namely Jinbubyeo, BR29, IR114 and IR20. The candidate genes chosen for expression studies were GLABROUS1 enhancer-binding protein genes, vascular plant one zinc finger protein gene, Arabidopsis RESPONSE REGULATORS Type-B gene, *ULTRAPETALA* gene and LEAFY gene. Cold stress treatment was given to 3 weeks old plants followed by subsequent RNA isolation, gene expression analysis by Real Time PCR and further *in-silco* analysis of the obtained data. Real-Time PCR based differential expression analysis of above mentioned genes in different tissues under cold stress condition was observed. Differential expression pattern was further analyzed by Weighted gene co-expression analysis based on Real Time PCR data indicating that some genes are highly correlated and of biologically significant. Our study uncovers gene-network involved in cold stress responses which are differentially expressed in different tissues of rice plant and the gene conferring tolerance, belonging to different functional classes, thus providing important insight into the functional basis of cold stress tolerance in rice cultivars.

LONG READ SEQUENCING TECHNOLOGY TO IDENTIFY VARIANTS IN COMPLEX GENOMIC REGIONS ASSOCIATED WITH CLINICAL CONDITIONS

Arvind Ramesh[1], Kumar Gautam Singh[1], Sudha Rao[1*]

[1*]Dhiti Omics Technologies Private Ltd, Bengaluru-560064, India.
Email: contact@dhitiomics.com

Abstract

Massive parallel sequencing technologies have rapidly transformed clinical genomics by enabling detection of variants on a genomic scale in patients with genetic disorders. However, due to short read length, CNV and SNV detection in genes with highly homologous sequences remain a challenge. Molecular techniques like microarray and exome sequencing cannot detect balanced reciprocal translocations the characterization of which remains critical to identify the gene associated with the clinical condition. Here we discuss case studies where long read Nanopore sequencing has enabled sequencing of genes with homologous sequences, complex regions with high GC content and helped mapping of translocation to basepair resolution.

METAGENOMIC ANALYSIS OF MICROFLORA OF DENTAL PLAQUE FROM CHILDREN WITH SEVERE EARLY CHILDHOOD CARIES USING OXFORD NANOPORE TECHNOLOGY

Dr Shruthi Igoor, Dr Latha Anandakrishna

Dept of Pedodontics and Preventive Dentistry, FDS, RUAS

Introduction: Severe Early childhood caries is a prevalent public health problem amongyoung children and little is known about the microbiota associated with it. Therefore, there is a need to determine the oral microbiome using sensitive methods such as Oxford Nanopore technology.

Aims: To conduct a metagenomic analysis to identify microflora of dental plaque fromchildren with Severe Early Childhood caries using Nanopore technology

Methods: The metagenomic analysis included 13 plaque samples which were collected from children with S-ECC and the samples were placed in Eppendorf tubes and transferred to the laboratory and subjected to DNA extraction and Nanopore sequencing as per protocol.

Results: At the species level, Actinomyces oris, Veillonella parvula, Prevotella intermedia, Streptococcus oralis, Pseudopropionibacterium propionicum, Fusobacterium nucleatum, capnocytophaga sputigena etc were found to be among the most dominating bacteria in dental plaque.

Conclusion: ECC is a polymicrobial disease and the oral microbiota associated with it includes species such as Actinomyces, Veillonella, Prevotella, Pseudopropionibacterium, Fusobacterium etc. Oxford nanopore technology has the potential to analyse metagenomically bacteria which were previously undetected and provide avenues for further research.

PEERING INTO CLINICAL GENETICS: THE MARKET AND ITS CHALLENGES

Dr. Anindita Saha Deb, Dr. Monica Supriya Rao*, Dr. K.S. Murthy
Agri-Business Division, ITC Ltd.
*Corresponding author – monica.supriyarao@itc.in

Clinical genetics is the medical speciality which provides diagnostic service, care, genetic counselling and treatment for individuals or families with or at risk of conditions which may have a genetic basis. According to the report the global genetic testing market was valued at US$5.060 billion in 2018 and is projected to expand at CAGR of 12.94 % over the forecast period to reach US $ 9.296 billion by 2023[1]. There are three major genetic testing methodologies i.e, cytogenetic (chromosome structure), biochemical (protein function), and molecular (DNA sequence). Prenatal, cancer, haematology and neurology are also included in a substantial proportion of the new tests. Rise in the adoption of health care, IT solutions for genetic testing, increase in the prevalence of cancer and cardiovascular diseases, and benefits offered by genetic testing (such as specificity and early identification) are expected to have a positive impact on the global genetic testing services market during the forecast period. Genetic and congenital abnormality is the second most common cause of infant and childhood mortality i.e, 25-60 per 1000 births in India[2]. Currently traditional prenatal screening method is having a market size of over 60%, bound to be a thriving market in India by 2024[3]. Along with birth defects, aneuploidies and familial cancer have also become an emerging issue. Technologies associated with clinical genetics are NGS (next generation sequencing), FISH (Fluorescence in situ hybridization), RTPCR (Real Time Polymerase Chain Reaction), Sanger sequencing, Microarray, MLPA (Multiplex ligation dependent probe amplification). In India regulation of genetic diagnostics services is presently governed by the ICMR (Indian Council of Medical Research) guidelines for biomedical research as no separate standards are available for the same. Prenatal diagnostic services are more stringently regulated by preconception and prenatal diagnostic techniques (Prohibition of Sex section) act 1994, which is primarily intended to curb the practice of prenatal sex determination. However, separate laboratory standards are required for genetic diagnostics laboratory as presently most laboratories are following international guidelines or appropriate modification by individual quality control standards. Despite a thriving market for genetic testing in India only 2% of population are covered by insurance. Affordability is another major constraint due to high cost of the instruments, skills and techniques. As per WHO report, Public sector accounts only for 30% of the total healthcare expenditure in the country, whereas it is above 50% in other developing countries and 83 % in UK. Despite numerous reports of new syndromes from India, there has been scarce development in the area of gene mapping. The growing prevalence of genetic disorder has been instrumental in driving the market. Moreover, the reduction in genetic testing prices globally has increased the number of individuals under growing the genetic tests since it has a scope beyond the medical field because of its ability to provide the ancestral history about an individual. However, concerns regarding the confidentiality and misleading advice along with the difficulty in interpreting results has limited the market growth.

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*Corresponding author – monica.supriyarao@itc.in

Food Authenticity Testing – Market Drivers and Discovering Technologies

Geetanjali Chakraborty, Dr. Monica Supriya Rao*, Dr. K.S. Murthy

Agri-Business Division, ITC Ltd.
*Corresponding author – monica.supriyarao@itc.in

Changing demographics and the need to know what we eat have fuelled a drastic rise in consumer awareness and hence the demand for certification by government and regulatory bodies. Based on test target, the food authenticity testing market can be segmented into testing for economically motivated adulteration, testing for false labeling of geographical and botanical origin, speciation of meat and fish, and testing for ageing. In the 5.3 million USD global food authenticity testing market in 2017, meat speciation accounted for one-third share and is expected to witness the highest growth rate, registering a CAGR of 8.7% during the forecast period 2018-2022[1]. DNA based testing is projected to be the largest and fastest growing technology with CAGR 8.1% and a projected market value of USD 2325.9 million[2] by 2022, due to reliability, quick results and trace quantification limits. Regulatory giants like the USFDA and the European Union, lay down the legislative requirements for the food sent to markets. The inception of regulations in emerging economies like India and China that have entered the global food trade are expected to increase the demand for testing services. Due to the sheer volume of the global meat market, there has been focus on technologies for meat speciation, like immunoassay-based techniques and electrophoretic techniques. However, by virtue of their intrinsic constraints, these technologies have been superseded by the recent molecular DNA-based methods. DNA-based technologies – mainly PCR and RTPCR, are in the spotlight because of their quantitative capabilities, better sensitivity, and rapidity. DNA is a molecule of choice for species identification due to its stability and high copy numbers even in heat treated products. Next-Generation Sequencing (NGS), a high-throughput sequencing method, has revolutionized meat speciation studies in terms of speed, read length and throughput, along with cost-reduction. Variants of chromatographic and spectroscopic techniques are an attractive option due to the speed of analysis and minimal sample preparation but they fail to quantify adulteration in some cases - such as cooked meat, which has more complex chromatographic patterns. Even with the advent of these methods, consumers need to be vigorously assured that players across the supply **network are operating in the consumer's interest and not in their own financial gain.** Traceability systems need to go beyond documented requirements to meet consumer demands for a transparent system. Block-chain technology is a software product that allows online storage and conversion of data in a decentralized, secure, and transparent way. Under this system, the end product is traceable at every point of its processing or packaging and all the data can be retrieved in less than two minutes. Despite all these advancements, the food authenticity testing market still has to overcome challenges like the affordability of testing, inappropriate sample collection and standardization, complex matrices, and lack of harmonization of regulations in developing countries. But with the implementation of increasingly strict regulations and highly publicized instances of food fraud serving as market drivers, the industry is set to grow at a momentous pace in the next few years.

1,2: Markets and Markets - Food Authenticity Testing Market: Global Forecast to 2022

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eurofins

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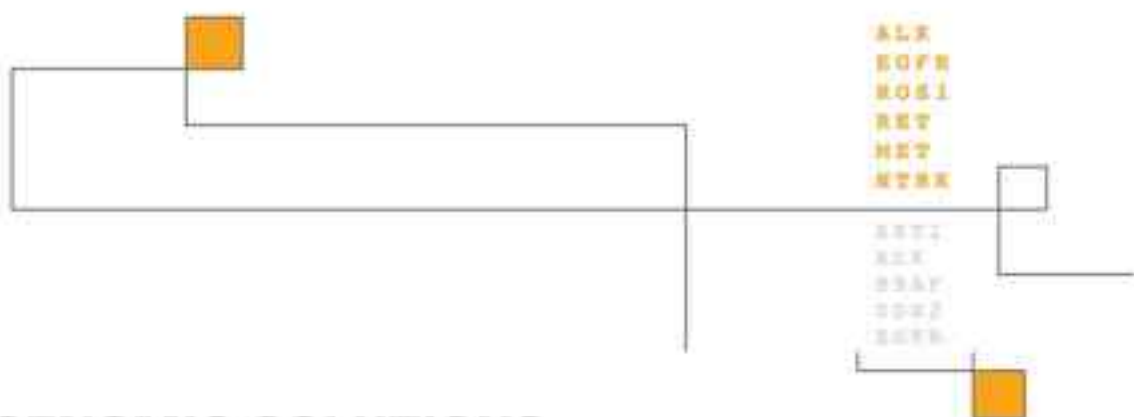Washington, D.C.
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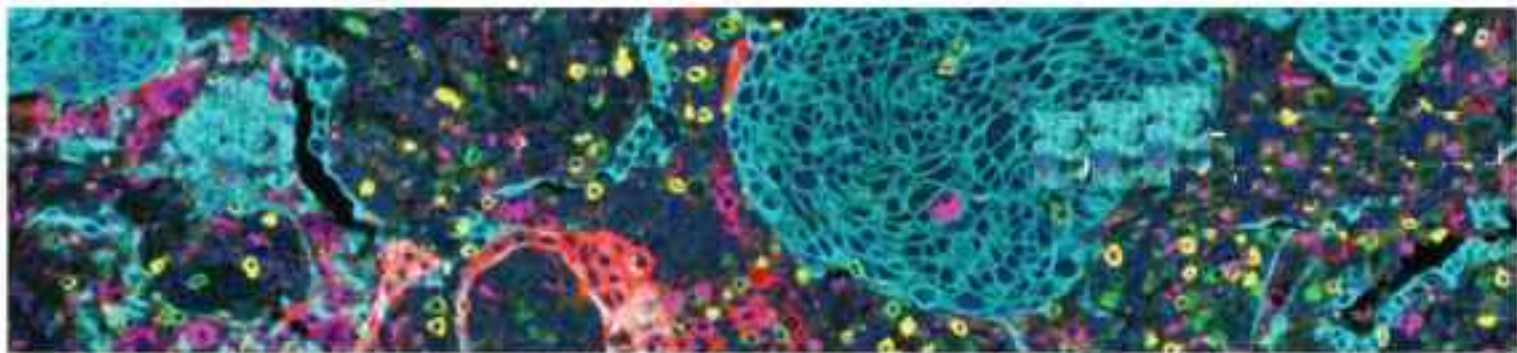
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A comprehensive liquid biopsy solution (IVD) for early detection of cancer metastasis (secondary tumor) and monitoring cancer treatment outcomes

Opportunity:

Currently Cancer diagnostics has many problems - Tissue biopsies are invasive and difficult to perform in certain cases, PET CT scans expose the patients to unnecessary radiation toxicity, inability to detect cancer spreading or late diagnosis of metastasis
Tumor Tissue Biopsy: Most common surgical procedure to diagnose cancer, cannot be repeated once primary tumor is removed. There is no way to assess the disease status six months later. Surgery is known to cause cancer spreading to distant organs
Diagnostic Imaging: PET (positron emission tomography) CT scans are used to monitor disease progression. It is recommended once every three months only to avoid toxicity. But cancer does not stop for anyone and continues to grow undetected during this interval.

Only 40% of the patients respond to standard chemotherapy regimens. Personalized medicine is the need of the hour

Solution: Liquid Biopsy

Non Invasive approach to detect circulating tumor cells - rogue cancer cells running around in the bloodstream and likely cause of secondary metastasis
More thorough early detection system with a statistical confidence of 99%. Identifies the cancer subtype for each individual (personalized medicine) based on tumor mutational profiling (DNA sequencing) to recommend targeted therapies - "right drug for the right patient"

Three types of liquid biopsy:
(1) CTCs - circulating tumor cells found in the peripheral blood - these are live cancer cells - usually decreases post treatment
(2) ctDNA - circulating tumor DNA is a subset of cell free DNA - representative fraction of dead apoptotic cancer cells - usually increases post treatment
(3) Exosomes - extracellular vesicles shed by primary tumors - cargo is made of protein biomarkers and micro RNA species that are suspected to prime the target organs for secondary metastasis

TheraSense™ Platform to detect circulating tumor cells (CTCs) using patent pending innovative microfluidics lab-on-a-chip technology and computer vision (machine learning) to detect the rare cancer cells among billions of normal blood cells - finding the needle in a haystack

Why is this a business of tomorrow?

1. Ability to detect cancer metastasis at least 4 to 6 weeks ahead of imaging
2. Ability to detect both CTC clusters and singlets without use of antibodies (Competitors can do only single cells; there is no commercial product yet for CTC clusters)
3. Ability to detect cancer cells undergoing epithelial mesenchymal transition (EMT) - other platforms fail to detect such transformed CTCs
4. Ability to enrich viable CTCs for downstream applications
5. Digital Pathology compatible for rapid diagnosis and reporting

Market Size:

Oncology in vitro diagnostics segment - liquid biopsy market expected to reach $28.6 billion, with CTC market at $11.6 billion by 2022
Growing interest in liquid biopsy fueled by targeted therapies
Total Addressable Market (TAM) is 14 Million, and SAM at 1.7 Million
Cancer is the second leading cause of death globally, and is responsible for an estimated 9.6 million deaths in 2016 - about 1 in 6 deaths is due to cancer
Economic loss due to cancer - US $ 1.16 trillion

Key Founders & Advisors:

Dr. Shibichakravarthy Kannan, MBBS, PhD
Founder & CEO - Trained at MD Anderson Cancer Center, Houston, TX.
Previously involved with Novartis Oncology, Mapmygenome (23andme), Datar Genetics, Apollo Hospitals

Dr. Vennila Dharman, MBBS, MPH
Co-Founder & COO - ECFMG certified family physician. Previously involved with Novartis Communications, Cancer Epidemiology & Prevention, US FDA regulations.

Supported by an execution team of 7 research scientists

Key Milestones & Traction:

Liquid biopsy is US FDA and CE IVD approved for lung, breast, colorectal and prostate cancers - recently melanoma and renal cell cancers were added
Gastrointestinal cancers such as liver and pancreas are recognized
Every year new indications (clinical use cases) are being added to growing list of liquid biopsies and companion diagnostics
NCCN and ESMO guidelines already included ctDNA (EGFR) for lung cancer

"Theranosis team has completed the working prototype and started the clinical validation phase of their product development - focus on breast, ovarian and cervical cancer"
Strategic partnerships with key B2B service providers
Strategic Alliances with top cancer hospitals in South India in progress

Awards and Recognition:

USD 5000 Rare Genomics Grant winner
ET Power of Ideas winner 2016

Key Reasons to Engage

Challenging the status quo of conventional cancer treatment landscape
Promoting personalized medicine with precision oncology
Strong founding team with required domain expertise
Idea and business plan validation by Philips Healthworks program

•Bangalore Bioinnovation Centre (BBC) is envisioned to be a state of the art incubation centre catering to the needs of start ups in the broad areas of Life Sciences

•The Bangalore Bioinnovation Centre (BBC) is an initiative of Karnataka Biotechnology and Information Technology Services (KBITS), Dept of IT, BT and S&T, Government of Karnataka with a liberal funding support from Department of Biotechnology (DBT), Government of India. It is located within Bangalore Helix Biotechnology Park at Electronic City. The Centre is a world class Incubation Centre with Central Instrumentation Facility in a 10 Acre campus with total built up area of above 50,000 sq ft.

•The Centre caters to the broad areas of Life Sciences i.e, Healthcare (MedTech/ Pharma/Bio-Pharma), Agriculture, Food/ Nutrition, Industrial Biotechnology Environmental Biotechnology.

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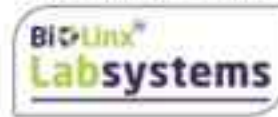
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