ABSTRACT BOOK

جامعة الإمارات العربية المتحدة
United Arab Emirates University

FIRST UAEU SYMPOSIUM ON BIOLOGICAL SCIENCES

"GENOMICS & BIOINFORMATICS"

UNITED ARAB EMIRATES UNIVERSITY
USBS
GENOMICS & BIOINFORMATICS
2016
KHALIFA CENTER FOR GENETIC ENGINEERING & BIOTECHNOLOGY
DEPARTMENT OF BIOLOGY

November 13-15, 2016

IT Building
United Arab Emirates University
Al Ain, United Arab Emirates

Organized by

Department of Biology, College of Science &
Khalifa Center for Genetic Engineering and Biotechnology
United Arab Emirates University

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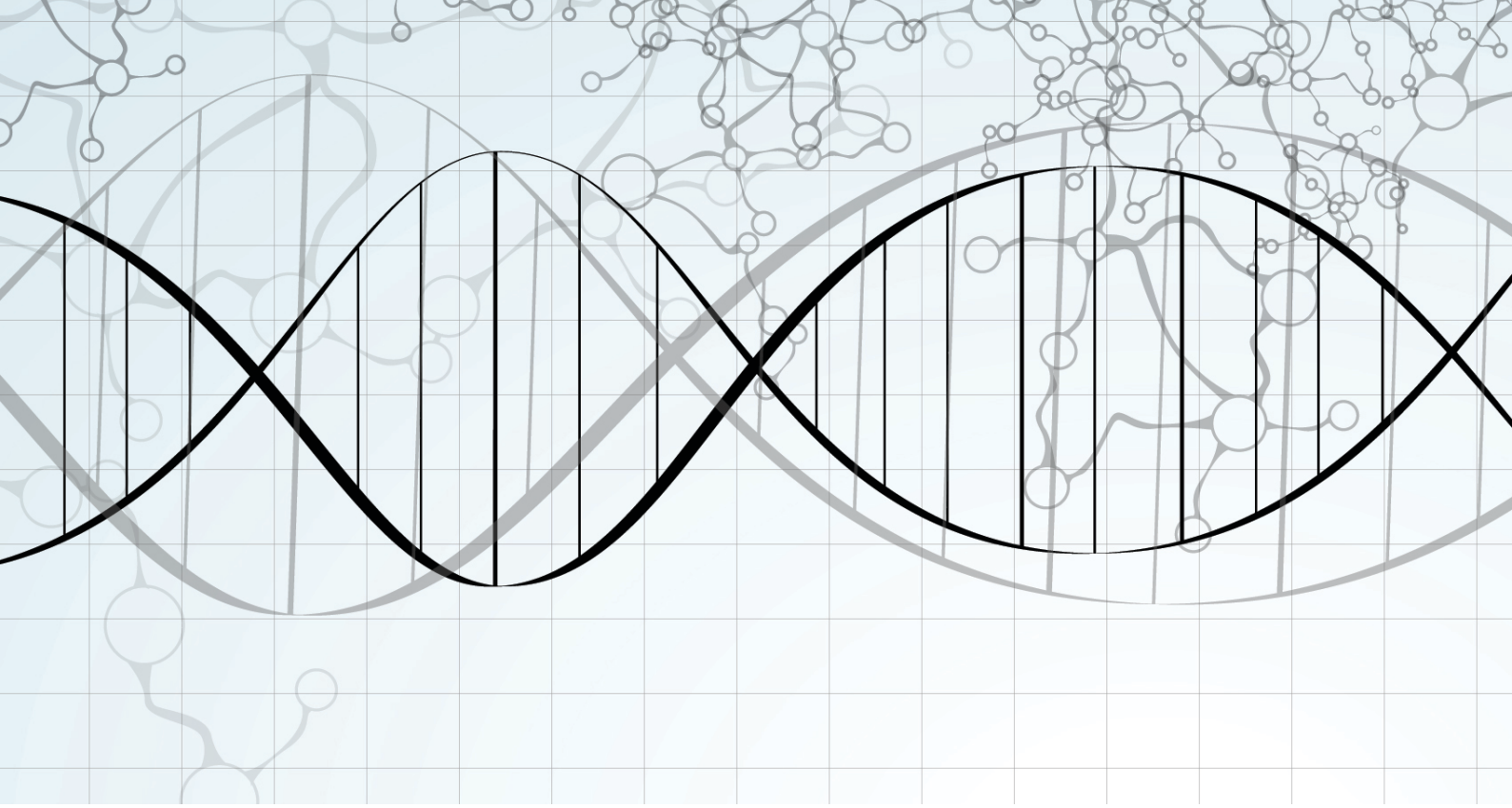


Program

DAY 1: SUNDAY, NOVEMBER 13, 2016

Location: Hilton Al Ain

5:00 pm – 6:00 pm	**REGISTRATION**
6:00 pm – 6:15 pm	**WELCOME ADDRESS** ***Dr Khaled Amiri*** *Chairman, Organizing Committee*
6:15 pm – 6:30 pm	**OPENING ADDRESS** ***Prof Mohamed Albaili*** *Vice Chancellor, UAE University*
6:30 pm – 7:30 pm	**PLENARY LECTURE** ***Prof Eduardo Blumwald*** *University of California, Davis, USA*
7:30 pm – 9:30 pm	**DINNER**

DAY 2: MONDAY, NOVEMBER 14, 2016
Location: IT Building, United Arab Emirates University

	SESSION 1: GENOMICS Chairs: Dr Khaled Hazzouri & Dr Pere Arús
9:00 am – 9:50 am	**KEYNOTE LECTURE** K1. Disruptive technologies for understanding and improving disease resistance in crop plants ***Prof Richard Michelmore*** *University of California, Davis, USA*
9:50 am – 10:20 am	**INVITED TALK** I1. New genomics-based approaches for perennial crop breeding: peach as an example ***Dr Pere Arús*** *Center for Research in Agricultural Genomics, Barcelona, Spain*
10:20 am – 10:40 am	**COFFEE BREAK**
10:40 am – 11:10 am	**INVITED TALK** I2. Building genomic tools for plant breeding: Our experiences improving red clover, ryegrass, and Bracharia ***Dr José de Vega-Bartol*** *The Genome Analysis Center, Norwich, UK*
11:10 am – 11:40 am	**INVITED TALK** I3. Redox strategies for crop improvement ***Dr Pavel Kerchev*** *Ghent University, Ghent, Belgium*
11:40 am – 11:55 am	O1. Whole genome re-sequencing of date palms yields insights into diversification of a fruit tree crop ***Dr Khaled Hazzouri*** *New York University, Abu Dhabi, UAE*
11:55 am – 12:10 pm	O2. Genomics and transcriptomic profiles of imatinib resistance in gastrointestinal stromal tumor ***Dr Asmaa Elzawahry*** *National Cancer Center, Japan*
12:10 pm – 1:40 pm	**LUNCH & POSTER PRESENTATIONS**

SESSION 2: TRANSCRIPTOMICS
Chairs: Dr Synan AbuQamar & Dr R. Manimekalai

1:40 pm –2:30 pm	**KEYNOTE LECTURE** K2. Genetic characterization of salinity tolerance traits to increase salinity tolerance of crops ***Prof Mark Tester*** *King Abdullah University of Science and Technology, Saudi Arabia*
2:30 pm – 3:00 pm	**INVITED TALK** I4. Alternative splicing in FMR1 premutation carrier ***Prof Flora Tassone*** *University of California, Davis, USA*
3:00 pm – 3:30 pm	**INVITED TALK** I5. Transcriptomics to unravel functional markers for crop improvement ***Dr R. Manimekalai*** *Sugarcane Breeding Institute, Indian Council of Agricultural Research, Coimbatore, India*
3:30 pm – 4:00 pm	**COFFEE BREAK**
4:00 pm – 4:15 pm	O3. A genome-wide identification of the miRNAome and transcriptome in response to salinity stress in the date palm tree (Phoenix dactylifera L.) ***Dr Mahmoud Yaish*** *Sultan Qaboos University, Oman*
4:15 pm – 4:30 pm	O4. De novo transcriptome analysis of non-model plant Andrographis paniculata using mRNA-Seq data for gene discovery and marker identification ***Mr Vivek Chadramohan*** *Siddaganga Institute of Technology, India*
4:30 pm – 4:45 pm	O5. Identification of genes involved in responses to environmental stress using reverse genetic approaches ***Dr Synan AbuQamar*** *United Arab Emirates University, UAE*
4:45 pm – 6:30 pm	**WORKSHOP** **(Selected participants only due to space limitations)** W1. From FASTQ to BAM: accurate alignment of short reads and references ***Dr José de Vega-Bartol*** *The Genome Analysis Center, Norwich Research Park, Norwich, UK*

DAY 3: TUESDAY, NOVEMBER 15, 2016
Location: IT Building, United Arab Emirates University

	SESSION 3: SYSTEMS BIOLOGY Chairs: Dr Ghada Al-Kafaji & Dr Roderic Guigó
9:00 am – 9:50am	**KEYNOTE LECTURE** K3. Unraveling metabolic pathways regulating fruit ripening ***Prof Eduardo Blumwald*** *University of California, Davis, USA*
9:50 am – 10:20 am	**INVITED TALK** I6. The human transcriptome across tissues and individuals ***Dr Roderic Guigó*** *Center for Genomic Regulation, Barcelona, Spain*
10:20 am – 10:40 am	**COFFEE BREAK**
10:40 am – 11:10 am	**INVITED TALK** I7. Cross-talk between intragenic epigenetic modifications and exon usage across developmental stages of human cells ***Dr Siba Shanak*** *The Arab American University, Jenin, Palestine*
11:10 am – 11:25 am	O6. Circulating microRNA-126 in peripheral whole blood as a potential biomarker for type 2 diabetes-related vascular complications ***Dr Ghada Al-Kafaji*** *Arabian Gulf University, Bahrain*
11:25 am – 11:40 am	O7. Rice root germin-like protein 2 gene promoter (OsRGLP2) is responsive to different plant signaling molecules in potato ***Dr Tariq Mahmood*** *Quaid-i-Azam University, Islamabad, Pakistan*
11:40 am – 11:55 am	O8. From dissecting complex networks in Arabidopsis to revealing new developmental mechanisms and adaptive strategies in date palm ***Dr Ikram Blilou*** *Wageningen University, Netherlands*
12:00 pm – 1:30 pm	**LUNCH & POSTER PRESENTATIONS**

SESSION 4: BIOINFORMATICS/PROTEOMICS
Chairs: Dr Rabah Iratni & Dr Marek Mutwill

1:30 pm – 2:20 pm	**KEYNOTE LECTURE** K4. Strategies for building and annotating high quality genome sequences ***Dr Ivo Gut*** *Centro Nacional de Análisis Genómico, Barcelona, Spain*
2:20 pm – 2:50 pm	**INVITED TALK** I8. Gene module multiplication drives pathway expansion in plants ***Dr Marek Mutwill*** *Max Planck Institute of Molecular Plant Physiology, Potsdam, Germany*
2:50 pm – 3:20 pm	**INVITED TALK** I9. Dissection of climacteric fruit ripening using genetic and genomic resources in melon ***Dr Jordi Garcia-Mas*** *Center for Research in Agricultural Genomics, Barcelona, Spain*
3:20 pm – 3:40 pm	**COFFEE BREAK**
3:40 pm – 4:10 pm	**INVITED TALK** I10. The 1000 Arab Genome: Characterizing the genome of ethnic groups in UAE. ***Dr Habiba Alsafar*** *Khalifa University Center for Biotechnology, Abu Dhabi, UAE*
4:10 pm – 4:25 pm	O9. Integrated genomic analysis of the human mitochondrial transcriptome ***Dr Youssef Idaghdour*** *New York University, Abu Dhabi, UAE*
4:25 pm – 4:40 pm	O10. Anti-breast cancer activity of carnosol in vivo and in vitro and in silico analysis of its target interactions ***Dr Rabah Iratni*** *United Arab Emirates University, UAE*
4:40 pm – 6:15 pm	**WORKSHOP** W2. Current and upcoming DNA sequencing technologies ***Prof Richard Michelmore*** *University of California, Davis, USA*
6:15 pm – 6:30 pm	**CLOSING ADDRESS** ***Dr Khaled Amiri*** *Chairman, Organizing Committee*

Posters

P1. Using of whole genome bisulfite sequencing (WGBS) to identify gene expression profile regulated by cytosine methylation under salinity stress in date palm
Miss Ibtisam Rashid Said Al Harrasi
Sultan Qaboos University, Oman

P2. Comparative genomics study for Acidithiobacillus ferrooxidans
Dr Rajesh Patel
Hemchandracharya North Gujarat University, India

P3. Analysis of Mendelian randomization studies of biomarkers and type 2 diabetes: observations from analysis
Dr Nadia Hussain
Al Ain University of Science and Technology, UAE

P4. Identification of Arabidopsis candidate genes in response to biotic and abiotic stresses using comparative microarrays.
Mr Arjun Sham
United Arab Emirates University, UAE

P5. Expression profiling of Arabidopsis WRKY33 mutants in response to Botrytis cinerea
Ms Shamma Al-Shamsi
United Arab Emirates University, UAE

P6. CRELD1 gene may predispose the risk of atrioventricular septal defects in Down Syndrome patients of North Indian origin
Ms Ambreen Asim
Sanjay Gandhi Post Graduate Institute of Medical Sciences, India

P7. Identification of premutation and grayzone FMR1 carrier status in reproductive age women by TP-PCR
Mrs Deepika Delsa Dean
Sanjay Gandhi Post Graduate Institute of Medical Sciences, India

P8. Novel NR1I2 mutations as probable cause of intellectual disability
Dr Inusha Panigrahi
PGIMER, India

P9. Genetic characterization of Epinephelus marginatus in the Mediterranean Sea: Contribution of microsatellite markers
Miss Aziza Elgid
INSTM, Tunisia

P10. Novel mutation in family with WNT 1- related osteoporosis
Dr Siyaram Didel
PGIMER, India

P11. Analysis of CGG repeat status at FMR1 gene in premature ovarian insufficiency cases: A study from SGPGIMS, Lucknow
Dr Sarita Agarwal
Sanjay Gandhi Post Graduate Institute of Medical Sciences, India

P12. Metagenomic Analysis of Soil Microbes and Risk Assessment of Transgenic Cotton in Northern Karnataka, India using DGGE and ARDRA techniques
Dr Devarajan Thangadurai
Karnatak University, India

P13. BS-Seq analysis pipeline for targeted next generation sequencing data
Mr P Sandeep Mallya
Manipal University, Manipal, India

P14. Prediction of transcription factor binding sites in differentially expressed genes for water stress in rice mediated by Pseudomonas fluorescens
Mrs Abida P.S.
Kerala Agricultural University, India

P15. Identifying a vovel deletion mutation in exon 3 of phenylalanine hydroxylase (PAH) gene in phenylketonuria (PKU) patients of UAE population.
Dr Muhammad Shahid Nazir
University of Modern Sciences, UAE

P16. Genome-wide analysis revealed that DNA methylation is a dynamic regulator of salt and osmotic stress in rice
Dr Fiaz Ahmad
Bahauddin Zakariya University, Pakistan

P17. Identification of superior buffalo bulls for increasing milk production through genome analyses
Miss Saher Islam
University of Veterinary & Animal Sciences, Lahore, Pakistan

P18. Morphology, physiology and microenvironment determine the morphogenetic potential of cell types of leaf callus of pearl millet cultivars.
Dr T.V.R. Lakshmi
University of Modern Sciences, UAE

P19. Whole genome sequencing and gene annotation of almond (Prunus dulcis)
Ms Sumisha Habeeb
UAE University, UAE

P20. Evaluation of suitable reference genes in date palm (Phoenix dactylifera L.) under drought and salinity stress for quantitative real-time PCR
Mr Himanshu Vishwas Patankar
Sultan Qaboos University, Oman

P21. Overexpression, purification & antimicrobial activity of recombinant human ß defensin 3
Dr Zaigham Abbas
University of Punjab, Pakistan

P22. Construction, expression and purification of thymosin α1-Azu28 fusion protein for targeted cancer therapy
Mr Muhammad Shahbaz Aslam
University of Punjab, Pakistan

P23. Testimony of rbcL and matK loci for eight United Arab Emirates native plant species
Dr Alagappan Kumarappan
University of Modern Sciences, UAE

P24. Using novel remote sensing approaches and carbon isotope discrimination to predict drought tolerance in IPT transgenic creeping bentgrass in the field
Dr Ramzi Belkhodja
Khalifa Center for Genetic Engineering and Biotechnology, UAE

P25. Anti-malarial drug discovery using integrative biology and systems biology
Dr Vishal Mevada
Hemchandracharya North Gujarat University, India

P26. In-silico design of inhibitors for Cyclin Dependent Kinase 2 molecule in Breast Cancer: a computational approach
Mrs Sreejisha P.S.
United Arab Emirates University, UAE

P27. Synthesized novel Fmoc-2-aminothiozole: protective against cadmium induced apoptosis, teratogenic effect on zebrafish (Danio rerio) embryos and molecular docking studies
Ms Vaishnavi M
Siddaganga Institute of Technology, India

P28. Structural insights into the polypharmacological activity of dietary flavonols on serine/threonine kinases
Ms Bincy Baby
UAE University, UAE

P29. Inhibiting non-receptor tyrosine kinases associated with prostate cancer using polypharmacological natural compounds
Ms Priya Antony
UAE University, UAE

P30. Bioinformatic methodologies reveal metagenomic Depiction of Indian hot springs' microbial community
Dr Pravin Dudhagara
Veer Narmad South Gujarat University, India

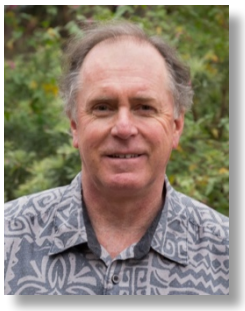
P31. Advanced Statistical Methods in Genetic Association studies
Dr Suresh Kumar Sharma
Panjab University, Chandigarh, India

P32. eHealth applications for clinical genetics
Dr Muhammad Jawad Hashim
United Arab Emirates University, UAE

P33. Artificial intelligence (AI), genomics and personalized medicine
Mr Shaheen N Shah
Genomics Central, India

Keynote Speakers

Prof. Richard W. Michelmore



After studying Natural Sciences at Cambridge, Richard Michelmore joined the faculty of the University of California at Davis in 1982. Richard was the founding Director of the Genome Center at UC Davis in 2003; since then he has overseen the recruitment of over sixteen genomics and bioinformatics faculty and the development of five service cores. He is currently a Distinguished Professor in the Departments of Plant Sciences, Molecular and Cellular Biology, and Medical Microbiology and Immunology. Richard has published over 160 scientific papers. His multidisciplinary research utilizes a synthesis of molecular, genetic and evolutionary approaches (http://michelmorelab.ucdavis.edu). His interests span basic research into the molecular basis of specificity in plant-pathogen interactions to translational plant genetics and crop improvement. His research is focused on comparative and functional genomics with an emphasis on plant disease resistance and pathogen variability. In addition, his program coordinates and hosts the bioinformatics component of the Compositae Genome Project. Richard's interests also include applications of next-generation DNA sequencing approaches to all areas of biology and its imminent impact on society in general. In particular, he aims to exploit such approaches for information-driven deployment of resistance genes in plants to provide durable disease resistance. In addition, he is interested in fostering research to enhance food security internationally.

Prof. Eduardo Blumwald

Dr. Eduardo Blumwald, Ph.D. serves as a Distinguished Professor of cell biology at the University of California, Davis and as a Faculty at the Lawrence Berkeley National Laboratory. Dr. Blumwald's research program is multidisciplinary in nature, combining physiology, biochemistry, molecular biology and omics. The general objectives of his work are: (i) the study of the cellular and molecular mechanisms that regulate ion homeostasis in plants; (ii) to characterize the responses of plants to environmental stress (e.g., salt, drought, and heat) and to engineer transgenic stress-tolerant crops; (iii) the study of the biochemical and molecular bases of sugar and acid accumulation in fruits and (iv) the development of genomic resources for the improvement of fruit quality. Dr. Blumwald serves as a Editorial Board Member and Associate Editor of several scientific journals and as the Editor-in-Chief of Plant Science.

Prof. Mark Tester

Mark Tester is Professor of Bioscience at King Abdullah University of Science and Technology (KAUST), Saudi Arabia. He was previously in Adelaide, where he was a Research Professor in the Australian Centre for Plant Functional Genomics and Director of the Australian Plant Phenomics Facility. Mark led the establishment of this Facility, a $55m organisation that develops and delivers state-of-the-art phenotyping facilities, including The Plant Accelerator, an innovative plant growth and analysis facility. He now leads a research group in which forward and reverse genetic approaches are used to understand and manipulate traits that contribute to salinity tolerance and improve this in crops such as barley and tomatoes. His aspiration is to unlock seawater, by developing a new economically viable agricultural system where salt-tolerant crops are irrigated with partially desalinised seawater or brackish groundwater.

Professor Tester obtained his Bachelor's degree in botany from the University of Adelaide in 1984, and his PhD in biophysics from the University of Cambridge in 1988. He was awarded a Junior Research Fellowship from Churchill College, Cambridge in 1988, a BBSRC (UK) Research Development Fellowship in 2001, and an Australian Research Council Federation Fellowship in 2004. He moved to Saudi Arabia in 2013.

Dr. Ivo Gut

Ivo Gut is a Director of the Centro Nacional de Análisis Genómico (CNAG) in Barcelona, one of the largest European genome sequencing operations, which he established in 2010.
His research interests are genomics, genetics, high-throughput nucleic acid analysis methods, proteomics, implementation of –omics methods, omics technologies, automation bioinformatics, data analysis, disease gene identification, cancer genomics, agrogenomics. He has more than 20 years experience in high-throughput nucleic analysis (genotyping and sequencing DNA, RNA and DNA methylation), technology development in nucleic acid and protein analysis. He was Head of Technology Development and Associate Director at the Centre National de Génotypage (CNG) – CEA (1999-2009), where he established the highest throughput genotyping platform in Europe and executed many genome-wide association studies, he initiated and was the coordinator of the EU-funded Project READNA in which 2nd, 3rd and 4th generation nucleic acid analysis technologies were developed. READNA Consortium was awarded with "Stars of Europe" Prize from the French Ministry of Higher Education and Research in 2013. He received his PhD in Physical Chemistry from the University of Basel in 1990. After his appointments as Research Fellow at Harvard Medical School and Imperial Cancer Research Foundation of London, he led a group in the Department for Vertebrate Genomics at Max-Planck-Institute for Molecular Genetics. He is author of more than 300 research papers, 11 reviews and 12 book chapters, cited over 21 000 times, inventor of 25 patents or patent applications, founder of 4 biotech start-ups, and serves on numerous international advisory boards.

Abstracts:
Oral Presentations

KEYNOTE LECTURES

K1. Disruptive technologies for understanding and improving disease resistance in crop plants

Prof. Richard Michelmore
The Genome Center & Department of Plant Sciences, University of California, Davis.

Although more remains to be learnt, great strides have recently been made in the understanding of the molecular and genetic basis of disease resistance in plants. It is now time to deploy this knowledge to provide more durable disease resistance. Much of these advances have been enabled by improvements in analytical technologies and further advances are anticipated. In particular, high throughput DNA sequencing enables detailed analysis of crops and their pathogens. It is now possible to characterize variable pathogen populations and use this information for the rational deployment of resistance genes so as to maximize the evolutionary hurdle for the pathogen to become virulent.

Much of our work over the past thirty years has focused on resistance to downy mildew in lettuce. Lettuce (*Lactuca sativa*) is one of the most valuable vegetable crops and downy mildew, caused by *Bremia lactucae*, is the most important foliar disease of lettuce worldwide. The use of resistant varieties is the most effective method for controlling this disease; however, pathogen variability has led to the rapid defeat of individual resistance genes. Over 50 resistance genes have been identified and lettuce downy mildew is one of the best genetically characterized plant diseases. Whole genome sequencing of multiple genotypes has allowed the identification of candidate resistance genes in the host and virulence factors in the pathogen. Knowledge-driven deployment of effective resistance genes as gene pyramids provides the opportunity for more durable resistance to *B. lactucae*. Gene stacking using genome editing has the potential making this process more efficient. In addition, host-induced gene silencing of vital pathogen genes presents potentially insurmountable evolutionary hurdles for the pathogen to overcome in order to become virulent.

K2. Genetic characterization of salinity tolerance traits to increase salinity tolerance of crops

Prof. Mark Tester
King Abdullah University of Science & Technology, Saudi Arabia

Forty percent of the world's food is produced under irrigation, and this is directly threatened by over-exploitation of water resources and changes in the global environment. In this talk, the focus will be on the use of forward genetic approaches for discovery of genes related to salinity tolerance in barley, rice and tomatoes. Rather than studying salinity tolerance as a trait in itself, we dissect salinity tolerance into a series of components that are hypothesised to contribute to overall salinity tolerance.

For barley, two consecutive years of field trials were conducted at the International Center for Biosaline Agriculture, a site with sandy soil and very low precipitation. Drip irrigation systems allowed the control of salinity by supplying plots with low (1 dS/m) and high salinity water (17 dS/m). A barley Nested Association Mapping (NAM) population developed by Klaus Pillen has been used to dissect physiologically and genetically complex traits in response to salt stress. Ten traits related to yield and yield components (e.g. days to flowering, harvest index, 100 seed mass) were recorded and five stress-indices were derived from each of these measurements. We have identified two significant loci located on the long arms of chromosomes 1H and 5H, which are both associated with several traits contributing to salinity tolerance, namely days to flowering, days to maturity, harvest index and yield.

For tomatoes, the focus is on genetics of tolerance in wild tomatoes, specifically *Solanum galapagense, Solanum cheesmaniae* and *Solanum pimpinellifolium*. An association genetic approach is being taken. High quality genome sequences have been made, and genotyping-by-sequencing undertaken. Tomatoes have been phenotyped in The Plant Accelerator and in the field, and analyses are currently in progress.

The application of this approach provides opportunities to significantly increase abiotic stress tolerance of crops, and thus contribute to increasing agricultural production in many regions.

K3. Unraveling metabolic pathways regulating fruit ripening

Prof. Eduardo Blumwald
Department of Plant Sciences, University of California, Davis, USA

Ripening is a process regulated by a large number of genes that control the progressive fruit maturation. During ripening, fruits undergo several physiological and biochemical modifications that influence properties associated with fruit quality, such as texture (fruit softening), color (chlorophyll degradation and accumulation of non-photosynthetic pigments), aroma (production of volatile compounds) and taste (increase in sugars and decline in organic acids). Traditionally, fruit ripening has been defined as either climacteric or non-climacteric. Increased levels of autocatalytic ethylene production and high respiration rates during ripening characterize climacteric fruits. Non-climacteric fruits show no increase or autocatalytic ethylene production or respiration rates during ripening.

We characterized and compared two plum cultivars (*Prunus salicina* Lindl. cv Santa Rosa [SM] and its bud-sport mutant cv. Sweet Miriam [SM]). Although SR and SM share the same genetic background, they display contrasting ripening behavior, i.e. climacteric (SR) and non-climacteric (SM). We integrated transcriptomics, proteomics, metabolomics, enzymatics and physiology to assess the biochemical/molecular regulation of ethylene-dependent and ethylene-independent biosynthetic pathways that are associated with fruit ripening in each cultivar. RNAseq analyses identified over 5,000 genes displaying differential expression between the cultivars. Weighted gene co-expression network analysis allowed the identification of several gene modules associated with hormone and sugar homeostasis. Furthermore, several hub genes were identified based on intra-module connections of the co-expression networks, among these were genes encoding a number of transcription factors and key regulators of hormone and carbohydrate metabolism. Targeted analyses of some of these genes facilitated the detection of altered biosynthetic pathways in both plum varieties. For example, in terms of sugar homeostasis, the climacteric variety (SR) displayed a “sucrose-associated” metabolism while the non-climacteric variety (SM) showed a “sorbitol-associated” metabolism. Differences in transcript accumulation of key enzymes regulating the synthesis/degradation of sucrose or sorbitol revealed variety-specific metabolic interactions directly associated with the observed changes in sugar homeostasis.

This experimental system provides a unique tool to study metabolic pathways underlying contrasting fruit ripening behavior. Understanding the steps associated with the regulation of sorbitol/sucrose homeostasis and the increased production of sugar alcohols will enhanced fruit quality and contribute to the development of fruits with a low glycemic index (high sorbitol, less glucose and fructose), adding to the health benefits of fruit consumption.

K4. Strategies for building and annotating high quality genome sequences

Dr. Ivo Gut
Centro Nacional de Análisis Genómico (CNAG-CRG), Barcelona, Spain

At the CNAG we have carried out *de novo* genome assembly and annotation for many different species (*e.g.* Iberian lynx, turbot, cedar aphid, almond, wasp, olive). The objective of each *de novo* assembly is to provide high contiguity and scaffolding with correct order and orientation of contigs so that downstream annotation has the best possible chance to recover the genes and gene structures correctly. Over the years we have refined the sequencing strategies that we apply to optimize contiguity and scaffolding while reducing the overall cost of a *de novo* assemble. Our basic strategies use paired-end whole genome shotgun sequencing together with matepair analysis, fosmid pool shotgun sequencing and fosmid end

sequencing in pools that are all run on Illumina sequencing systems. Computational assembly strategies start with de novo assembly of fosmid pools, followed by error correction with the whole genome shotgun data. Scaffolding is refined with matepair and fosmid end sequences. Throughout we apply a contig breaking and re-assembly strategy. We are constantly exploring datatypes from different types of sequencers to optimize the quality of an assembly and the cost of building a new genome sequence. In particular, these are the inclusion of fosmid pool sequencing using Pacific Biosciences and Oxford Nanopore Technologies MinIon systems and optical mapping strategies with the Iris system from BioNanoGenomics. After assembly annotation is done using a combination of computational tools and RNA sequencing of mRNA from several different tissues of the species we are sequencing. In this presentation we will discuss the strategies and pipelines that we have developed to achieve high quality reference genomes and annotations for the different *de novo* projects carried out at the CNAG.

SESSION 1: GENOMICS

I1. New genomics-based approaches for perennial crop breeding: peach as an exemple

Dr. Pere Arús
IRTA, Center for Research in Agricultural Genomics CSIC-IRTA-UAB-UB, Spain

Perennial crops, including most sweet fruit, palms, grapes, olives, coffee, most ornamentals, etc. are an important source gastronomic or aesthetic pleasure and provide for vitamins, fiber, antioxidants and other health-related products that complement the calories provided by cereals and other staple foods. They often have long intergeneration periods and are clonally propagated. Their cultivation extends for around 1/8 of the world's total food-producing surface and represents more than 60% of world's patents and breeder's rights titles. Conventional breeding methods consist of phenotypic selection from progeny of crosses between two highly heterozygous parents. The selected plants are then clonally propagated and tested for yield and quality. This scheme is fast and simple, but it has major drawbacks: recombination is used to a very limited extent and trait introgression from exotic sources is practically impossible. We propose three approaches based on the selection of the whole genome with molecular markers, and apply them to peach (*Prunus persica*): a) marker-assisted introgression, where new genes of interest from other *Prunus* species compatible with peach are identified, and peach individuals with a single exotic introgression are recovered in only two backcross generations. An example is provided based on almond × peach hybrids; b) "resynthesis", where it is possible identifying in the selfed progeny of a high-value cultivar: a) genotypes with the same genetic composition as the original cultivar, except for one or few chromosomal regions, and b) two complementary homozygous (or nearly-homozygous) individuals that crossed will produce hybrids with a nearly-identical genotype that the original cultivar, and c) VORIS (varieties obtained by resynthesis and introgression), using the two previous concepts to obtain with a low number of generations a cultivar, nearly-identical to a highly performing one, with a gene incorporated from an exotic source.

I2. Building Genomic tools for plant breeding: Our experiences improving red clover, ryegrass, and *Bracharia*

Dr. José de Vega-Bartol
The Genome Analysis Center, Norwich Research Park, Norwich, UK

Advances in NGS and assembly techniques allow to generate high quality genomes of heterozygous and complex plants that were previously inaccessible. These genomes are a useful tool for breeding and plant research. They can be explored by comparative genomics, used to asset the diversity of natural and synthetic populations, or identify the causal loci of complex agronomic traits. We have produced chromosome-scale genomes of red clover, ryegrass and the tropical forage *Bracharia*. We have sequenced plants of red clover from around Europe to characterize its diversity and domestication, and later used this data to identify useful elite genotypes to enrich the breeding program. We are also using genomic approaches to characterize the progeny in the red clover and *Brachiaria* breeding programs, and to identify the key regulators behind complex traits, like tolerance to acid soils and persistency.

I3. Redox strategies for crop improvement

Dr. Pavel Kerchev

Department of Plant Systems Biology, VIB, Belgium

Department of Plant Biotechnology and Bioinformatics, Ghent University, Belgium

The increased awareness that reactive oxygen species (ROS) can act as signaling molecules has opened new avenues to exploit redox biology for crop improvement. Understanding hydrogen peroxide (H_2O_2) signaling is particularly important due to its relatively long half-life and major production during normal physiological process such as photorespiration. To analyze molecular mechanisms that regulate the response to increased H_2O_2 levels, we have been using *Arabidopsis thaliana* mutants lacking peroxisomal catalase activity (*cat2-2*) needed for H_2O_2 decomposition. By screening for second-site mutations that attenuate the photorespiratory phenotype of *cat2-2*, we isolated a number of mutations that rescued the cell death of *cat2-2* plants under photorespiration-promoting conditions. Using a similar approach, we probed the ability of 10 000 chemicals to alleviate the cell death phenotype of *cat2-2* plants and identified 34 hit molecules. I will discuss the functional roles of two confirmed mutations and highlight the outcomes of the chemical screen. This strategy has the potential to deliver stress-protective agrochemicals and candidate lead genes that can form the basis of better performing crops.

O1. Whole genome re-sequencing of date palms yields insights into diversification of a fruit tree crop

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Date palms (*Phoenix dactylifera*) are the most significant perennial crop in arid regions of the Middle East and North Africa. Here, we present a comprehensive catalogue of approximately seven million single nucleotide polymorphisms in date palms based on whole genome re-sequencing of a collection of 62 cultivars. Population structure analysis indicates a major genetic divide between North Africa and the Middle East/South Asian date palms, with evidence of admixture in cultivars from Egypt and Sudan. Genome-wide scans for selection suggest at least 56 genomic regions associated with selective sweeps that may underlie geographic adaptation. We report candidate mutations for trait variation, including nonsense polymorphisms and presence/absence variation in gene content in pathways for key agronomic traits. We also identify a copia-like retrotransposon insertion polymorphism in the R2R3 myb-like orthologue of the oil palm virescens gene associated with fruit colour variation. This analysis documents patterns of post-domestication diversification and provides a genomic resource for this economically important perennial tree crop.

O2. Genomics and transcriptomic profiles of imatinib resistance in gastrointestinal stromal tumor

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Background The gastrointestinal stromal tumor (GIST) is the most common frequent mesenchymal tumor of the digestive tract. GIST proliferation is driven by gain-of-function mutations in KIT. These characteristics have facilitated the targeted therapies development that based on with tyrosine kinase inhibitors, such as imatinib. Although many clinical studies have demonstrated revolutionized effects of imatinib, more than 80% of patients lastly develop disease progression driven by secondary resistance mutations in KIT kinase domains. However, the full spectrum of genomic and transcriptomic changes behind the resistance remains unknown. Results This study analyzed genomic and transcriptomic changes in drug-sensitive and -resistant cell lines against imatinib. We also looked at an “intermediate” cell-line before reaching the full resistance. We identified SNVs and CNAs from the next-generation sequencing and also the transcriptome from microarrays. For clinical insights, we conducted exome sequencing for two clinical samples with the resistance. Notably, the cell line briefly exposed to imatinib exhibited drastic transcriptional changes, but few genomic changes. Conclusion We suggest that pre-existing cell death-resistant subpopulations are the main cause for full resistance via secondary KIT mutations. The combination of chemotherapy with imatinib and apoptosis pathway-targeting drugs, could limit the emergence of drug-resistant cancer.

SESSION 2: TRANSCRIPTOMICS

I4. Alternative splicing in FMR1 premutation carriers

Prof. Flora Tassone
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Over 40% of males and ~16% of female carriers of a FMR1 premutation allele (55-200 CGG repeats) are at risk for developing Fragile X-associated Tremor/Ataxia Syndrome (FXTAS), an adult onset neurodegenerative disorder while, about 20% of female carriers will develop Fragile X-associated Primary Ovarian Insufficiency (FXPOI), in addition to a number of neurodevelopmental and adult-onset clinical problems (FMR1 associated disorders). Marked elevation in FMR1 mRNA levels have been observed with premutation alleles and the resulting RNA toxicity is believed to be the leading molecular mechanism proposed for these disorders. The FMR1 gene, as many housekeeping genes, undergoes alternative splicing. Using Single molecule real time sequencing (SMRT) (SMRT) and qRT-PCR we have recently reported that, although the relative abundance of all FMR1 mRNA isoforms is significantly increased in the premutation group compared to controls, there is a disproportionate increase, relative to the overall increase in mRNA, in the abundance of isoforms spliced at both exons 12 and 14. In total, we confirmed the existence of 16 out of 24 predicted isoforms in our samples. However, it is unknown, which isoforms, when overexpressed, may contribute to the premutation pathology. To address this question we have further defined the transcriptional FMR1 isoforms distribution pattern in different tissues, including heart, muscle, brain and testis derived from FXTAS premutation carriers and age-matched controls. Preliminary data indicates the presence of a transcriptional signature of the FMR1 gene, which clusters more by individual than by tissue type. We identified additional isoforms than the 16 reported in our previous study, including a group with particular splice patterns that were observed only in premutations but not in controls. Our findings suggest that the characterization of expression levels of the different FMR1 isoforms is fundamental for understanding the regulation of the FMR1 gene as well as for elucidating the mechanism(s) by which “toxic gain of function” of the FMR1 mRNA may play a role in FXTAS and/or in the other FMR1-associated conditions. In addition to the elevated levels of FMR1 isoforms, the altered abundance/ratio of the corresponding FMRP isomers, or gene methylation, could affect the overall function of FMRP in premutations.

I5. Transcriptomics to unravel functional markers for crop improvement

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Agricultural crops are put into various biotic / abiotic stress factors during their growth phases. The vagaries of climatic conditions and thereby the changes in the scenario of pest / pathogen greatly affect the production and productivity of crops. This abstract perceives the potential of transcriptomics in deciphering the interaction between crops and stress factors to develop functional markers for crop improvement. In particular, whole genome transcriptome sequence information utilized to understand the association between the crop and biotic / abiotic stress factors. Transcriptomics-based analysis of phytoplasma (plant pathogen) associated coconut root wilt diseases highlighted the differentially expressed genes involved in physiological response of the disease palms. The genes implicated in water stress, response to ethylene, cell wall biosynthesis were largely up-regulated and genes related to photosynthesis and plant defence such as chitinase, LRR domain, glucosidase, pathogenesis related (PR) were down-regulated in diseased palms. However, the same pathogen in arecanut palm is associated with yellow leaf disease exhibits a different scenario. The genes involved in plant-pathogen interaction pathways were differentially expressed, specifically, genes such as, LLR family protein and apoptosis inducing factor were up-regulated and RUBPcase and most of the chloroplast related (plastocyanin, thioredxoin) genes were down regulated in diseased condition. The transcriptome of sugarcane under oxidative stress showed the differential expression of zinc ion binding proteins under stress. There is significant difference in miRNA profiles of cultivated variety and wild species of sugarcane under oxidative

stress. The co regulated miRNA and transcripts in particular condition helps to identify the positively regulated genes for further improvement of crops through transformation / MAS. Transcriptome is an alternative to genome sequence especially for complex polyploid crops for developing markers. The whole transcriptome can be used to develop expressed QTL (eQTL) and it is possible to map on to a segregating population for development of functional markers and would aid applying genomic selection strategies in plant breeding.

O3. A genome-wide Identification of the miRNAome and transcriptome in response to salinity stress in the date palm tree (*Phoenix dactylifera* L.)

Dr. Mahmoud Yaish
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Date palm is an important tree in arid and semiarid regions however; in the recent years this plant experiences an enormous challenge due to excessive soil salinity which causes a significant yield loses. Despite the existence of a certain level of salt adaptation capability, the molecular mechanism behind this tolerance is completely unknown in this plant. This mechanism likely encompasses the involvement of epigenetic factors such as small non-coding RNA (sRNA) molecules known as miRNA which usually play a role in the post-translational modifications when plants exposed to abiotic stresses including salinity stress. In this project, four libraries of sRNAs were constructed and a high-throughput Solexa sequencing approach has been employed to generate a genome-wide assessment of miRNAome in response to salinity of date palm seedlings. A total of 251 million raw reads were obtained from NaCl-treated and untreated leaves and roots libraries including a 153 unique conserved, 89 nonconserved and a 180 putative novel unique miRNA/miRNA* sequences all were encoded by appropriate precursors. Sequence analysis showed some predicted miRNA gene targets have a potential function in salt adaptation mechanism. While global differential miRNAome expression analysis showed that most miRNAs repressed in roots upon exposure to salinity, the same analysis showed that the miRNA expression in leaves is slightly down regulated under the same conditions. The expression level of some miRNA was experimentally tested using qRT-PCR suggesting that these miRNAs might play a role in gene expression regulation in date palm. The miRNAome discovered in this study provided insights into molecular aspects of miRNA-mediated gene expression and therefore these results would provide a basis for further unravelling the mechanism on how miRNAs can control salinity adaptation mechanism in date palm.

O4. De novo transcriptome analysis of non-model plant *Andrographis paniculata* using mRNA-Seq data for gene discovery and marker identification

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Co-authors: Vivek Chandramohan, Anubhav Kaphle, Sindhu Gangarudraiah, Mohit Jhunjhunwala

Andrographis paniculata Nees, known as 'Kalamegha' in Sanskrit, is an herbaceous plant in the family Acanthacea and is known to have a broad range of pharmacological potential. However, Andrographis being a non-modal plant, genomic analysis is limited by very small quantity of publicly available annotated sequence data. Our research, thus, focuses on employing transcriptomic analysis based on NGS to generate transcript information about the plant and characterize metabolic pathway related to diterpenoids biosynthesis. Paired-end reads were downloaded from the NCBI SRA database of ID SRR1292497 and were quality-checked and trimmed to produce clean reads. We assembled in total of 197,537,498 reads de novo using Trinity assembler, and CLC Genomics Workbench (CLC) using various k-mer values to optimize the assembly. Trinity outputs a total of 86,215 transcripts and 64,488 trinity 'genes' with average length of 1156.63 and N50 of 2111. %GC content was calculated to be around 42.56. RSEM was used to calculate relative abundance of isoforms in the experiment pool of mRNA. The contigs were extracted and used as queries in BLASTx against the Ref-Seq protein database (plant division). The

majority of contigs produced significant hits with expectation values under 1.0E-5 and showed similarity with *Vitis vinifera* and *Sesamum indicum*. Blast2GO tool was used to functional-annotate the obtained transcript sequence. In addition, we identified simple sequence repeat motifs in transcripts using MISA tool. We used KASS online annotation from KEGG to elucidate the genes involved in di-terpenoid biosynthesis. The transcripts set generated here provide a resource for gene discovery and development of functional molecular markers. In addition, the strategy for de novo assembly of transcriptome data presented here will be helpful in other similar transcriptome studies.

O5. Identification of genes involved in responses to environmental stress using reverse genetic approaches

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Transcriptional reprogramming forms a major part of a plant's response to environmental stress. We investigated the effects of combinations of biotic and abiotic stresses on the transcriptome level of *Arabidopsis* genome using comparative microarrays. We showed a unique program of gene expression was activated in response to each biotic and abiotic stress. In addition, abiotic stress-induced genes were commonly regulated with *Botrytis cinerea* infection. The *Arabidopsis* cell wall e*xpansin-like A2* (*EXLA2*) gene was identified based on its down-regulation in response to infection by the necrotrophic pathogen *B. cinerea*, and on the reduced susceptibility of its mutants to the same pathogen. The *exla2* mutants also enhanced tolerance to the phytoprostane-A_1 (PPA_1). Our results suggest that the absence or down-regulation of *EXLA2* leads to increased resistance to *B. cinerea* in a COI1-dependent manner, and this down-regulation can be achieved by PPA_1 treatment. The *EXLA2* is significantly induced by salinity and cold, and exogenous application of Abscisic acid (ABA). The *exla2* mutant also showed hypersensitivity towards increased salt and cold, and this hypersensitivity required a functional ABA pathway. Overall, *EXLA2* appears to be important in response to environmental stress, particularly in the pathogenesis of necrotrophic pathogens and tolerance to abiotic stress. Future directions to further analyze the functions of commonly expressed genes in response to environmental stress will increase our understanding of the plant stress response.

SESSION 3: SYSTEM BIOLOGY

I6. The human transcriptome across tissues and individuals

Dr. Roderic Guigó

Center for Genomic Regulations, Universitat Pompeu Fabra, Spain

The pilot phase of the Genotype-Tissue Expression (GTEx) project has produced RNASeq from 1,641 samples originated from up to 43 tissues from 175 post-mortem donors, and constitutes a unique resource to investigate the human transcriptome across tissues and individuals. Clustering of samples based on gene expression recapitulates tissue types, separating solid from not solid tissues, while clustering based on splicing separates neural from non-neural tissues, suggesting that post-transcriptional regulation plays a comparatively important role in the definition of neural tissues. About 47 % of the variation in gene expression can be explained by variation of across tissues, while only 4% by variation across individuals. We find that the relative contribution of individual variation is similar for lncRNAs and for protein coding genes. However, we find that genes that vary with ethnicity are enriched in lncRNAs, whereas genes that vary with age are mostly protein coding. Among genes that vary with gender, we find novel candidates both to participate and to escape X-inactivation. In addition, by merging information on GWAS we are able to identify specific candidate genes that may explain differences in susceptibility to cardiovascular diseases between males and females and different ethnic groups. We find that genes that decrease with age are involved in neurodegenerative diseases such as Parkinson and Alzheimer and identify novel candidates that could be involved in these diseases. In contrast to gene expression, splicing varies similarly among tissues and individuals, and exhibits a larger proportion of residual unexplained variance. This may reflect that that stochastic, non-functional fluctuations of the relative abundances of splice isoforms may be more common than random fluctuations of gene expression. By comparing the variation of the abundance of individual isoforms across all GTEx samples, we find that a large fraction of this variation between tissues (84%) can be simply explained by variation in bulk gene expression, with splicing variation contributing comparatively little. This strongly suggests that regulation at the primary transcription level is the main driver of tissue specificity. Although blood is the most transcriptionally distinct of the surveyed tissues, RNA levels monitored in blood may retain clinically relevant information that can be used to help assess medical or biological conditions.

I7. Cross-talk between intragenic epigenetic modifications and exon usage across developmental stages of human cells

Dr. Siba Shanak

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Arab American University- Jenin, Palestine

Differential exon usage has been reported to affect the large majority of genes in mammalian genomes. It has been shown that different splice forms sometimes have distinctly different protein functions. Here, we present an analysis of the Human Epigenome Atlas (version 8) to connect the differential usage of exons in various developmental stages of human cells/tissues to differential epigenetic modifications at the exon level. We found that the differential incidence of protein isoforms across developmental stages is often associated with changes in histone marks as well as changes in DNA methylation in the gene body or the promoter region. Many of the genes that are differentially regulated at the exon level were found to be associated with development and metabolism.

Differential exon usage is a mechanism used by complex organisms to increase the usability of the gene-coding regions, so that several different proteins are expressed from the same chromosomal position. Epigenetics studies inherited modifications to genes that do not belong to the raw DNA sequence, but nevertheless modulate gene expression. Epigenetics is well-associated with alternative splicing in the gene body, but the connection to distinct developmental stages has not been addressed

so far. Here we show that a sizeable number of genes that are essential for development show strong associations between differential exon usage and epigenetic modifications.

O6. Circulating microRNA-126 in peripheral whole blood as a potential biomarker for type 2 diabetes-related vascular complications

Dr. Ghada Al-Kafaji
Arabian Gulf University, Bahrain

Patients with type 2 diabetes mellitus (T2D) develop serious complications that reduce their quality of life and life expectancy. Biomarkers for early detection of the disease and identification of individuals at risk of developing complications would be clini¬cally significant to improve the care of these patients. Small non-coding RNAs called microRNAs (miRNAs) regulate gene expression and participate in diverse cellular processes such as metabolism, inflammation responses, and angiogenesis; and their dysregulation has been associated with various diseases including T2D and its related vascular complications. Recently, miRNAs have been shown to be stalely present in the circulation and can be used as biomarkers for disease diagnosis or prognosis. MiR-126 is highly enriched in endothelial cells and altered its circulating levels has been reported in the blood of T2D patients. The aim of this study was to investigate the expression of circulating miR-126 and to assess its potential as a blood-based biomarker for diabetic nephropathy (DN) and coronary artery disease (CAD) in T2D patients. The expression of miR-126 was evaluated in peripheral whole blood using TaqMan-based quantitative real time PCR from four groups: T2D patients without microvascular or macrovascular complications (n=52), DN patients (n=50; 29 with microalbuminuria and 21 with macroalbuminuria), CAD patients (n=50) and non-diabetic healthy controls (n=50). The expression levels of circulating miR-126 were significantly decreased in T2D patients and further decreased in DN patients and CAD patients compared to non-diabetic healthy controls ($P<0.05$). Multivariate logistic regression analysis confirmed the independent association of lower miR-126 levels with T2D, DN and CAD ($P<0.05$). In the group of patients with DN, circulating miR-126 negatively correlated with albuminuria and positively with glomerular filtration rate, and in addition, negatively correlated with fasting glucose (FG), glycated hemoglobin (HbA1c), triglyceride and low density lipoprotein (LDL) ($P<0.05$). Moreover, stepwise multiple regression analysis identified albuminuria as a significant predictor of miR-126 ($P<0.05$), and receiver operating characteristic (ROC) analysis revealed a significant ability of blood miR-126 to differentiate between T2D patients, DN patients, and non-diabetic healthy controls ($P<0.05$). In the group of patients with CAD, circulating miR-126 negatively correlated with FG, HbA1c and LDL ($P<0.05$). Furthermore, ROC analysis showed that miR-126 in peripheral blood was able discriminate between T2D patients, CAD patients, and non-diabetic healthy controls ($P<0.01$). Our data suggest that decreased expression of miR-126 is association with diabetic renal and cardiac defect, and that circulating miR-126 may be a promising blood-based biomarker for identification of T2D patients at risk of developing DN and CAD.

O7. Rice root germin-like protein 2 gene promoter (OsRGLP2) is responsive to different plant signaling molecules in potato

Dr. Tariq Mahmood
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Co-authors: Tariq Mahmood, Faiza Munir

Germins and germin-like proteins (GLPs) signify a vital plant proteins family taking part in numerous stress linked and developmental activities. In plants under stress situations the signaling cascades are generated, initiating various physiological changes that eventually stimulate the expression of particular gene batteries conferring tolerance in response to stress conditions. In present study, regulation of the Oryza sativa root expressed germin-like protein 2 gene promoter (~1100-bp) ligated upstream to to a reporter gene (GUS) was ascertained in response to different plant signaling molecules in transgenic potato plants. Four major signaling molecules i.e. jasmonic acid (JA), salicylic acid (SA), abscisic acid (ABA) and hydrogen peroxide (H_2O_2) were exogenously applied, in response to which the

OsRGLP2 promoter activity was found to be differently stimulated. As revealed through quantitative real time PCR analysis. The finding suggested involvement of JA, SA, ABA and H2O2 dependent signaling pathways in stimulation of OsRGLP2 promoter. This work is a valuable addition to the information being gathered about the mechanisms through which the GLPs are stimulated and exert their role during exposure of plants to various stresses.

O8. From dissecting complex networks in Arabidopsis to revealing new developmental mechanisms and adaptive strategies in date palm

Dr. Ikram Blilou
Wageningen University Research, Netherlands

Transcription factors function as cell fate determinants. In the Arabidopsis roots, the transcription factors SHORT-ROOT, SCARECROW and the BIRD proteins control asymmetric cell division and endodermal specification through a complex network involving protein complex formation and transcriptional regulation. Here we report the spatial distribution of these complexes using Förster resonance energy transfer measured by fluorescence lifetime microscopy, we show that differential transcription factor conformation and promoter specific distribution in living roots imply a complex spatial regulatory network at the level of cell-specific transcription factor interactions. In Date palm, *Phoenix dactylifera*, we identified components of the described network and mapped their expression at the cellular resolution and we are currently studying their function in both Arabidopsis and date palm. Date palm are among the few plants adapted to drought and to high levels of soil salinity, however, the molecular mechanisms conferring date palm tolerance to these conditions remain largely unknown. Recently, we have shown that selected date palm root bacterial endophytes alleviate drought stress. Using the model plant Arabidopsis, we report that date palm bacteria promote root growth and increase system architecture through modulating the plant hormone auxin. We also show that these bacteria act through the epidermal layer and promote root tolerance to salinity and drought in Arabidopsis and date palm plants. Our experimental data combined with mathematical models strongly suggest that this increased tolerance is mediated by changes in auxin distribution and transport. These data provide a new entry in using date palm growth promoting bacteria as a biotechnological approach to promote tolerance to drought and salinity in crops. In addition, understanding developmental process in date palm and using omics approaches will be very instructive towards understanding strategies for adaptation to desert conditions.

SESSION 4: BIOINFORMATICS/PROTEOMICS

I8. Gene module multiplication drives pathway expansion in plants

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The survival of species depends on their ability to adapt to new environments. Adaptive innovations require genetic material, mainly provided by gene duplications, which can lead to new or alternative pathways. However, the principles behind the emergence of such pathways are not understood. Here, we show that approximately one third of the genes of a plant's genome participate in hundreds of alternative pathways, which were primarily generated by single gene duplications. We determined the copy number of alternative pathways by searching for sets of genes, termed gene modules, that occur multiple times in gene co-function networks of eight plant species. We found that plants employ a genetic copy-and-paste principle to increase the number and diversity of gene modules. Our findings demonstrate that gene module multiplication has provided the capacity for plants to increase their repertoire of cellular functions.

I9. Dissection of climacteric fruit ripening using genetic and genomic resources in melon

Dr. Jordi Garcia-Mas
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Melon (*Cucumis melo* L.) is a cucurbit of high economic value and an interesting model system to study fruit ripening, as both climacteric and non-climacteric varieties exist in this species. The availability of the genome sequence and re-sequencing of several melon accessions provide powerful tools to help characterizing genes involved in climacteric fruit ripening. An introgression line (IL) population in the Piel de Sapo (PS, non-climacteric) background containing introgressions from PI 161375 (SC, non- climacteric) has been used to characterize two QTLs, *eth6.3* and *eth3.5*, which provide climacteric ripening to PS. *eth6.3* has been cloned, and *eth3.5* has been mapped in a short genomic interval. A new RIL population obtained after crossing PS with the Védrantais (Ved) climacteric melon type revealed that climacteric ripening is a complex trait. The combination of these genetic resources with genomic tools will help dissecting the mechanisms that control climacteric ripening in melon.

I10. The 1000 Arab Genome: Characterizing the genome of ethnic groups in UAE.

Dr. Habiba Alsafar
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There is an intolerable gap in the human genome landscape. Human Genome Organisation (HUGO), Haplotype Map (HapMap) and other international consortia have all neglected to include ethnic groups of the Arab-speaking world in their sequencing efforts. The genomic organization of ethnic groups within Arabia is poorly defined despite the region being the crossroad of human migration out of Africa some 50,000 to 85,000 years ago as well as being the hub of bidirectional flow between traders from 3 continents in more contemporary history. There are many examples where lack of information is hindering the provision of adequate clinical services. This project is a bold step to address the deficiency in genomic data on populations of the Middle East. Serendipitously, the recent advancements in DNA sequencing technology have coincided with the need to improve our understanding of the genomic organization in the Arabian population.

O10. Integrated genomic analysis of the human mitochondrial transcriptome

Dr. Youssef Idaghdour
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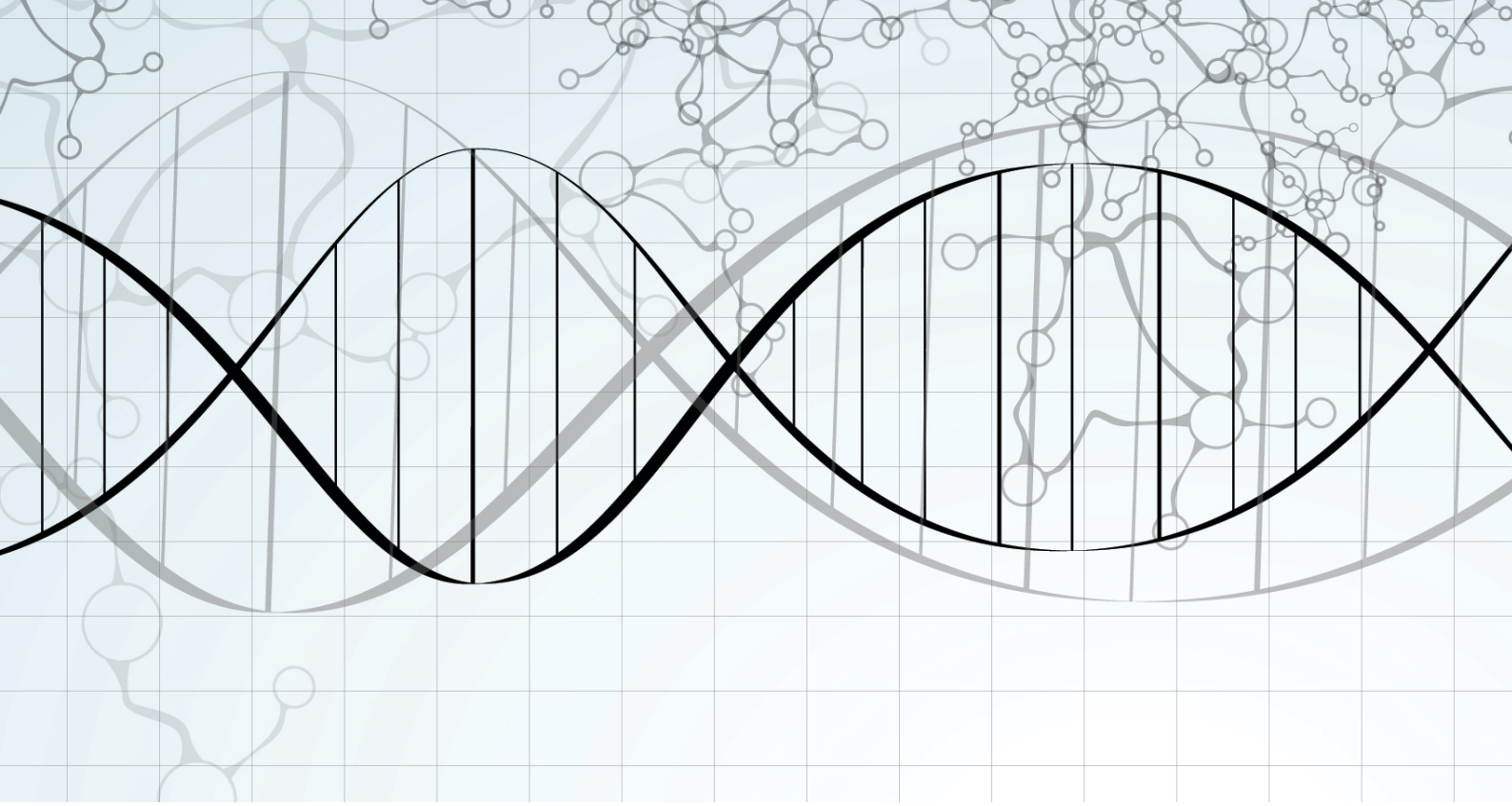
Mitochondria are vitally important for basic cellular function. Sequence variation in mitochondrial DNA has been associated with a large number of diseases, as well as fundamental processes such as aging. However, little is known about the extent and nature of transcriptomic and epigenetic variation in mitochondrial RNA and its downstream effects. Using deep RNA sequencing from large number of human samples and an elegant bioinformatic approach, we document mitochondrial RNA sequence variation and quantify the levels RNA processing and modification in population-scale data. We find remarkable levels of heteroplasmy and variation across individuals, as well as sites that show consistent signatures of epigenetic modification. Most interestingly, we find extensive variability at specific mitochondrial tRNA positions and report nuclear genetic and gene expression variation associated with it. The observed large-scale variation observed at these sites could have a significant effect on mitochondrial function.

O11. Anti-breast cancer activity of carnosol in vivo and in vitro and in silico analysis of its target interactions.

Dr. Rabah Iratni
Department of Biology, College of Science, United Arab Emirates University, UAE

We have previously demonstrated that carnosol, a natural compound, inhibited *in vitro* cell viability and colony growth, and induced cell cycle arrest, autophagy and apoptosis in human breast cancer cells. In the present study, we evaluated the ability of carnosol to inhibit tumor growth and metastasis *in vivo*. Using chick embryo tumor growth assay, we showed that carnosol significantly and markedly suppressed tumor growth and metastasis of breast cancer. Moreover, we found that non-cytotoxic concentrations of carnosol decreased the migration and downregulated the expression and the activity of MMP-9 in the triple negative breast cancer (TNBC) MDA-MB-231 cells. Mechanistically, we demonstrated that carnosol promoted proteasome-dependent degradation of p300 in all the panel of breast cancer cells analyzed. Furthermore, in addition to promoting p300 degradation, carnosol inhibited the acetyltransferase activity of recombinant p300. *In silico* docking analysis indicated that carnosol might inhibit p300 HAT activity by blocking the entry of acetyl-CoA binding pocket of the catalytic domain. This study further confirms carnosol as a promising anti-breast cancer therapeutic compound, and identifies it as a new inhibitor of p300 to be added to the panel of inhibitors identified so far**.**



Abstracts:
Poster Presentations

POSTER ABSTRACTS

P1. Using of Whole Genome Bisulfite Sequencing (WGBS) to identify gene expression profile regulated by cytosine methylation under salinity stress in date palm

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Date palm (*Phoenix dactylifera* L.) is the most important domesticated tree in the arid and semiarid countries of the Middle East and North Africa. In recent decades, soil salinity has become a global problem especially in agricultural lands of arid and semi-arid regions. There is growing evidence that DNA methylation plays an important role in regulating gene expression in response to abiotic stress. Therefore, this study aims to investigate differential DNA methylation status of three sequence contexts (mCG, mCHG, mCHH, where H = A, C or T) that occur in date palm under salinity stress and control conditions using cytosine methylome sequencing (methylC-seq) technology. The study also aims to identify salinity stress-responsive genes regulated by DNA methylation based on metylome and RNA sequencing analysis. Pooled DNA and RNA samples from the control and salinity treatments were analyzed. The analysis covered up to 324,987,795 and 317,056,091 total reads of the control and salinity-treated samples, respectively. Among these, 19,209,086 (5.9%) from the control and 19,185,920 (6.0%) from the salinity-treated sample were aligned to unique positions of mCGs and covered 40% and 41% of the total genomic cytosine(s) with an average read depth 0f 17 fold coverage per DNA strand for both with a bisulfite conversion rates of 99% and 98% for the control and treated DNA pools, respectively. The cluster analysis of top 100 differentially methylated sites (DMSs) of mCG, mCHG and mCHH showed that the majority of the mCHH sites were hypermethylated in the genome of plants grown under the control condition. RNA sequencing revealed the presence of around 6,000 differentially regulated genes in response to salinity however, the relationship between DNA methylation and transcriptome abundance is yet to be determined. This work provides a new insight on date palm epigenetic regulation mechanisms in response to salinity stress.

P2. Comparative genomics study for *Acidithiobacillus ferrooxidans*

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Co-authors: Rajesh Patel, Vishal Mevada, Rajesh Chaudhari and Ghelani Anjana

Acidithiobacillus is the most important genus of chemolithotrophs that metabolize sulfur. They can be isolated from rivers, canals, acidified sulfate soils, mine drainage effluents and other mining areas. Acidithiobacillus are adapted to wide variations of temperature and pH and can be readily isolated and more enriched. The genus has emerged as an economically significant bacterium in the field of leaching of sulfide ores. In present work an attempt has been carried out to compare the genomic information of Acidithiobacillus sp. GGI-221 strain of Indian origin with two well-studied genome Acidithiobacillus ferrooxidans ATCC 53993 and Acidithiobacillus ferrooxidans ATCC 23270. ATCC 23270 strain was having the highest genome size of 2982,397 bp followed by 2,885,038 bp and 2,284,565 bp respectively for ATCC 53993 and GGI 221 strain. Maximum 3086 coding sequences were reported for ATCC 23270 followed by 2953 and 2694 coding sequences respectively reported for ATCC 53993 and GGI 221 strain. Maximum 83 RNAs gene were reported for ATCC 23270 followed by 52 and 41 RNAs for ATCC 53993 and GGI 221 strain respectively. Total 27 subsystem based annotation was resulted from the RAST based annotation process. Only three subsystem Photosynthesis, Motility Chemotaxis and Secondary Metabolism were absent in all the three strain. Subsystem for Dormancy and Sporulation was similar for all the three strain. For

subsystem Potassium Metabolism, Iron acquisition and metabolism, Phosphorus Metabolism and Carbohydrates subsystem category higher subsystem feature count was reported for GGI 221 strain with comparison to other strain. For remaining 19 subsystem category less subsystem feature count was obtained for GGI 221 strain with respect to ATCC 23270 followed by ATCC 53993. In comparison with GGI 221 and ATCC 23270 total 1362 protein were reported out of which 1090 were common while 18 proteins were present. The comparative genomics analysis was based upon Rapid Annotation using Subsystem Technology (RAST) comparison and whole genome alignment approach to find out the similarity and difference in the strain for understanding molecular evolution process and their application regards to effective biotechnological application of strain.

P3. Analysis of Mendelian randomization studies of biomarkers and type 2 diabetes: observations from analysis

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In epidemiology, many biomarkers are noted to be associated with type 2 diabetes (T2D) risk. The aim of this study was to identify and summarize current evidence for causal effects of biomarkers on T2D.

A systematic literature search in PubMed and EMBASE (until October 2015) was done to identify Mendelian randomization studies that examined potential causal effects of biomarkers on T2D. Data from two large-scale genome-wide association studies (GWAS) were used: the Meta-Analyses of Glucose and Insulin-related traits Consortium (MAGIC) for glycemic traits and Diabetes Genetics Replication and Meta-analysis (DIAGRAMv3) for T2D. GWAS summary statistics were extracted for the same genetic variants, which were used in the original Mendelian randomization studies. Twelve out of 21 biomarkers (from 32 studies) have been reported to be causally associated with T2D in Mendelian randomization. Most biomarkers that were investigated were from a single cohort study or population. Of the 12 identified biomarkers, nominally significant associations with T2D or glycemic traits were achieved for those genetic variants related to bilirubin, pro-B-type natriuretic peptide, Delta-6 desaturase and dimethylglycine. Several Mendelian randomization studies have investigated the nature of associations of biomarkers with T2D. However, there were only few biomarkers that may have causal effects on T2D. Further research is required to evaluate the causal effects of multiple biomarkers on T2D and glycemic traits using data from large-scale cohorts.

P4. Identification of Arabidopsis candidate genes in response to biotic and abiotic stresses using comparative microarrays

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Plants have evolved with intricate mechanisms to cope with multiple environmental stresses. To adapt with biotic and abiotic stresses, plant responses involve changes at the cellular and molecular levels. We investigated the role of cyclopentenones in mediating plant responses to environmental stress through TGA (TGACG motif-binding factor) transcription factor, independently from jasmonic acid. Candidate genes were identified by comparing plants inoculated with Botrytis cinerea or treated with heat, salt or osmotic stress with non-inoculated or non-treated tissues. About 2.5% heat-, 19% salinity- and 41% osmotic stress-induced genes were commonly upregulated by B. cinerea-treatment; and 7.6%, 19% and 48% of genes were commonly downregulated by B. cinerea-treatment, respectively.

Our results indicate that plant responses to biotic and abiotic stresses are mediated by several common regulatory genes. Comparisons between transcriptome data from Arabidopsis stressed-plants

support our hypothesis that some molecular and biological processes involved in biotic and abiotic stress response are conserved. Thirteen of the common regulated genes to abiotic and biotic stresses were studied in detail to determine their role in plant resistance to B. cinerea. Moreover, a T-DNA insertion mutant of the Responsive to Dehydration gene (rd20), encoding for a member of the caleosin (lipid surface protein) family, showed an enhanced sensitivity to B. cinerea infection and drought. Future research directions towards a better dissection of the potential crosstalk between B. cinerea, abiotic stress, and oxylipin signaling are of our particular interest.

P5. Expression profiling of Arabidopsis WRKY33 mutants in response to *Botrytis cinerea*

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Botrytis cinerea is a necrotrophic fungus that causes plant diseases on a wide range of crops. The WRKY33 transcription factor was reported for resistance to *B. cinerea*. We compared Arabidopsis WRKY33 overexpressing lines and wrky33 mutant that showed altered susceptibility to *B. cinerea* with their corresponding wild type (WT) plants using the high-throughput microarray gene expression analysis. In WT, about 1660 genes (7% of the transcriptome) were up regulated and 1054 genes (5% of the transcriptome) were down-regulated at least twofold at early stages of inoculation with *B. cinerea*, confirming previous data of the contribution of these genes in *B. cinerea* resistance. The expressions of *B. cinerea*-regulated genes, encoding for proteins and metabolites involved in pathogen defense and non-defense responses, seem to be dependent on a functional WRKY33 gene. The expression profile of OPDA- and PPA1-treated Arabidopsis plants in response to *B. cinerea* revealed that cyclopentenones can also modulate WRKY33 regulation upon inoculation with *B. cinerea*. These results support the role of electrophilic oxylipins in mediating plant responses to *B. cinerea* infection through the TGA transcription factor. Further investigations to elucidate the function and mechanism of cyclopentenone metabolism during *B. cinerea* and other necrotrophic pathogens infections are underway.

P6. CRELD1 gene may predispose the risk of Atrioventricular septal defects in Down Syndrome patients of North Indian Origin

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Down syndrome (DS), also called as trisomy 21, is one of the most leading cause of intellectual disability. DS is associated with number of phenotypes including Congenital Heart Disease (CHD), Leukemia, Alzheimer's disease, Hirschsprung's diseases and others. DS affects about 1 in 700 live births. Objectives: To explore the role of CRELD1variants on congenital heart defects, we sequenced the exon 4,9,10 of CRELD1in the samples from North India. Sixty participants were included in the genetic association study and they were stratified as DS with atrioventricular septal defect (AVSD), DS without AVSD and AVSD without DS cases. A significant association was found between DS having AVSD and seven polymorphisms which includes rs7699000959 (c.368 G>A), rs73118372 (c.1136T>C, present at 10th exon-intron junction), rs3774207 (c.1119C>T, present at 10th exon-intron junction), rs9878047 (c.1049-129T>C), two deletions of A at positions 9891 and 10281 and g.9768T>A, rs377654613 (c.447 G>A, present at exon 4). Mutation Taster software (http://mutationtaster.org/) and NCBI blast (http://blast.ncbi.nlm.nih.gov/Blast.cgi) was used to predict the variant. Since these variants are, present only DS having AVSD group, so we hypothesize that these variants may increase the risk of AVSD in DS patients.

P7. Identification of premutation and grayzone Fmr1 carrier status in reproductive age women by TP-PCR

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Fragile X Syndrome (FXS), the second most common type of X linked mental retardation is caused due to silencing of FMR1 gene due to hyper expansion and hypermethylation of CGG repeat at its 5'UTR. Existing definitions describe the normal range of FMR1 CGG repeats as 6–44, the Gray zone (GZ) range as 45–54 repeats, permutation (PM) range as 55–200 repeats and FXS positive full mutation (FM) cases with >200 repeats. Though asymptomatic, PM and GZ carrier female are at a high risk of transmitting expanded allele in the subsequent generation. Owing to high prevalence of these alleles in the general population (1 PM in 113–259 females and 1 GZ in 66 females), screening of women of reproductive age for identification of PM and GZ carrier is of great clinical utility. As there is no cure for FXS thus timely identification of carriers will open doors for genetic counselling and option of anti-natal diagnosis for at risk women and thereby reduce the incidence of FXS in the society. This Paper focuses on doing the molecular characterization of CGG repeat sequence at FMR1 gene in women of reproductive age by using indigenously developed TP-PCR method for identification of carriers. Genomic DNA was extracted from 150 reproductive age women (17years to 40 years). TP- PCR amplification for FMR1 allele is conducted and the amplicons generated were subjected to fragment analyses and results were documented. TP-PCR analysis of 150 samples of reproductive age women identified 1.3% (2 of 150) subjects with alleles in the GZ region, 49 and 50 CGG repeats, while rest 98.7% (148 of 150) subjects with alleles in the normal range (29 to 42 repeats). No PM carrier was identified among the studied cohort. Knowledge of premutation allele frequency in the general population will establish an estimate of FXS burden and offering prior genetic counselling may improve the quality of life, both mentally and financially .However in spite of clinical significance of conducting screening program for identification of the carrier status of reproductive age women, such objective had greatly suffered due to unavailability of accurate, cost effective and rapid molecular techniques. The indigenously developed TP-PCR in our study is cheaper and in comparison to available commercial kits and thus it has proved to be economically more feasible to be used in screening program.

P8. Novel NR1I2 mutations as probable cause of intellectual disability

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Human NR1I2 or PXR serves as a key transcriptional regulator of the CYP3A4 gene. It is involved in transport through the blood brain barrier, and interacts with P-glycoprotein or Abcb1. These interactions have been explored in developing treatment for Alzheimer's disease. Mental retardation has been linked to mutations in the HSD17B10 gene, which alters the neuronal steroid and isoleucine metabolism. We found homozygous NR1I2 mutations in a child with unexplained intellectual disability. The screening for malformations through radiological and imaging techniques was negative. Karyotyping done ruled out any aneuploidies. Targeted next gen sequencing (NGS) revealed mutations in the NR1I2 gene-c.242G>A in exon 2 in homozygous state. This variant is rare in population databases and predicted to be damaging on in silico analysis (Polyphen). The parents were nonconsanguineous but carriers for the same mutation. We hypothesize that Nr1I2 mutations might be causing the phenotype-microcephaly, and intellectual disability in the child by altering the neuronal steroid metabolism. Keywords: blood brain barrier, pregnane X receptor, mental retardation, microcephaly, steroid metabolism.

P9. Genetic characterization of *Epinephelus marginatus* in the Mediterranean Sea: Contribution of microsatellite markers

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In this study, we used seven microsatellite markers to analyze the diversity and genetic differentiation of populations of grouper *Epinephelus marginatus* from the Mediterranean Central (Tunisia and Libya). The mean number of alleles per locus and the proportion of polymorphic locus showed the existence of high variability. The two populations analyzed have the same rate of heterozygosity (He=0.5 ± 0.35), these values are similar to those observed in other marine fish. An applied test on the two samples showed significant departure from Hardy-Weinberg equilibrium (FIS= 0.177; P< 0.001) and FST value (0.024; p<0.05). The GAG45 and GAG007 loci were responsible of the observed heterozygote deficit.

P10. Novel mutation in family with WNT 1- related osteoporosis

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Osteogenesis imperfecta (OI) is an inherited disorder with osteoporosis and recurrent fractures. Children presenting with recurrent fractures and bowing of limbs have severe form of the disorder.WNT-1 homozygous patient have more frequent fractures while heterozygous carriers of the mutation in WNT-1 gene are also found to have early onset osteoporosis. We identified a family with WNT-1 mutation out of cohort of 30 patients of OI under follow up in the genetic clinic. The index case, a 6-month old showed 41bp deletion in splice region following exon 1 of WNT-1 gene in homozygous state on Sanger sequencing. The mutation was reported as likely pathogenic on bioinformatic analysis. To further characterize the significance of the mutation we studied his mother who is 30year old with blue sclera but no fracture. Her DEXA scan of lumber spine showed osteoporosis and she was heterozygous for the mutation. Next generation sequencing(NGS)done for the child didn't show any significant variation in other OI genes including COL1A1, COL1A2, SERPINH1, CRTAP, LEPRE1, PP1B, 1F1TM5 and BMP1 genes. Father also had history of backache but was not available for evaluation. Thus we conclude that this novel variant identified in the child with OI is likely cause for the disease.

P11. Analysis of CGG repeat status at Fmr1 gene in premature ovarian insufficiency cases: A study from SGPGIMS, Lucknow

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Fragile X Syndrome (FXS), the second most common type of X linked mental retardation is caused due to silencing of FMR1 gene due to hyper expansion and hypermethylation of CGG repeat at its 5'UTR. Current definitions describe the normal range of FMR1 CGG repeats as 6–44, the intermediate range as 45–54 repeats, and the premutation range as 55–200 repeats. Those affected by FXS have >200 repeats (full mutation). PM alleles are highly unstable and get expanded to FM when maternally transmitted in 50% of the offspring. PM is often associated with Primary ovarian insufficiency (POI). In spite of ovarian dysfunction 5-10% of POI women can still conceive and have a viable pregnancy with a 50% chance of giving birth to a FXS positive offspring. This study aims to test FMR1 PM status by using indigenously developed TP-PCR, for all women presenting with POI in order to rule out the chance of being PM positive as a cause of ovarian insufficiency and to avoid the load of this disorder in the society. Genomic DNA was

extracted from 79 POI women with average age of 30.02+5.94 years (17years to 40 years). TP- PCR amplification for FMR1 allele is conducted and the amplicons generated were subjected to fragment analyses and results were documented. Molecular screening of 79 POI subjects resulted in identification of 2 PM positive (2.5%) subjects with 80 and 67 CGG repeat number. The remaining 77 POI subjects had CGG repeats in the range of 29 to 40 repeats, normal range. Extended family screening of PM positive POI subjects revealed maternal transmission in one of the subject while other subject was found to have a family history of FXS with her nephew being FM positive. Conclusions: Screening of POI women for identification of PM alleles is of great clinical utility due to the inherent risk of these alleles to expand FM. As the indigenously developed TP-PCR in our study is cheaper and rapid in comparison to available commercial kit and thus can meet high diagnostic output requirement, it has proved to be economically more feasible to be used in screening program. The identification of PM carrier status among the POI/POF subjects will serve dual purpose of identifying the cause for ovarian dysfunction as well as providing prenatal diagnosis to prevent the birth of FM male child (Fragile X syndrome). We at SGPGI have established this technique indigenously and are providing nationwide service for routine diagnosis and screening for FXS.

P12. Metagenomic analysis of soil microbes and risk assessment of transgenic cotton in Northern Karnataka, India using DGGE and ARDRA techniques

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Metagenomics, the study of genomes from microbial communities than individual species is the novel approach to deal with 99 % of microorganisms that are unculturable and exists in several extreme environments. This approach has the potential to reveal genetic diversity, population structure and functional ecosystems of diverse microorganisms. With the advent of molecular and genomic tools such as polymerase chain reaction (PCR), denaturing gradient gel electrophoresis (DGGE) and amplified ribosomal DNA restriction analysis (ARDRA), it is possible to reveal and understand the microbial diversity patterns from diverse ecological niches. In the current study, the risk assessment of transgenic cotton on soil microbial community was carried out using DGGE and ARDRA techniques. Transgenic and non-transgenic cotton cultivated soil samples were collected from three districts of Northern Karnataka in India (Dharwad, Belgaum and Bagalkot). Metagenomic DNA was isolated from transgenic and non-transgenic cotton cultivated soil samples and the same has been subjected for PCR, DGGE and ARDRA analysis using several microbial species specific universal primers to analyze the possible effect of transgenic cotton on soil microbial communities.

P13. BS-Seq analysis pipeline for targeted next generation sequencing data

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DNA methylation is the phenomenon of the addition of a methyl group especially at the fifth position of cytosine in the context of CG dinucleotides that has the potential to alter gene expression. Bisulfite treatment of the DNA, converts the cytosine residues to uracil leaving 5-methyl cytosine residue intact is one of the most persuasive technique to study DNA methylation. This technique has been routinely used in Sanger method of sequencing to determine the methylation levels at the individual CG sites. Presently, next-generation sequencing techniques such as BS-Seq (bisulfite sequencing) are used to determine the extent of methylation at a genomic level. This strategy is suitable for researchers looking at regions of interest who may not have the resource for genome wide sequencing. Targeted BS-Seq, may help researchers working on selective regions from large number of samples, where specific targets are

amplified and sequenced. Currently, most of the analytical techniques and the available tools are designed for whole genome bisulfite sequencing and are unable to efficiently quantify methylation in targeted sequencing. Here, we present a pipeline to efficiently analyze and quantify methylation data generated from single end sequencing of targeted regions. Targeted bisulfite sequencing reads in fastq format is preprocessed to check for quality and corrected. This data is then mapped to the reference sequence using a bisulfite specific mapper, and the resultant data is quantified using Bi-Q analyser HT. The entire workflow is automated using shell scripts and hosted online to make it user-friendly for researchers to analyze and visualize targeted bisulfite sequence data.

P14. Prediction of transcription factor binding sites in differentially expressed genes for water stress in rice mediated by *Pseudomonas fluorescens*

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The regulation of various biological processes in the plant systems, especially during different adverse climatic conditions are brought about by the change in the expression of different genes which in turn is the result of the binding of specific transcription factors in their respective transcription factor binding sites (TFBSs).In this study, an in-silico analysis of the data of differentially expressed genes of rice under water stress mediated by Pseudomonas fluorescens was performed to find out the TFBSs. The AGI codes of differentially expressed genes were utilized for the prediction of TFBSsby performing orthologue search against Arabidopsis genome in RGAP (Rice Genome Annotation Project) database. Further, TFBSs were identified by AthaMap database,a genome-wide map of TFBSs in Arabidopsis thaliana, and STIFDB2 (Stress Responsive Transcription Factor Database V2.0) database which is a comprehensive collection of biotic and abiotic stress responsive genes inArabidopsis and Oryza sativa L.The Matrix score ≥ 10 and Threshold ≥5 were selected as potential TFBSs using “Search” tool in AthaMap and Z-score with above 1.5 were indicated as potential TFBSs in STIFDB2. The significant TFBSs were analyzed based on the parameters provided by databases and were cross validated. The results revealed that the WRKY, MYB, HSF, bZIP, ARF, AP2/EREBP, bHLH, Trihelixand ABI3/VP1 TF families and their respective TFBSs were predicted as functionally significant. These predicted TFBSs would be responsible for the change in expression of genes under water stress. Key words: Water stress, rice, TFBSs, AthaMap, STIFDB2.

P15. Identifying a novel deletion mutation in exon 3 of phenylalanine hydroxylase (PAH) gene in phenylketonuria (PKU) patients of UAE population

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Phenylketonuria (PKU) is an inherited autosomal recessive disorder that increases the levels of an amino acid phenylalanine in the blood. A liver-specific phenylalanine-4-hydroxylase enzyme instructed by phenylalanine hydroxylase (PAH) gene catalyzes the hydroxylation of phenylalanine to tyrosine. The mutations in the PAH gene reduces the activity of phenylalanine-4-hydroxylase enzyme which leads to the accumulation of phenylalanine in the tissues and plasma of PKU patients and causes intellectual disability and other serious health problems. In the present study genetic mutations in the PAH gene were identified in the PKU patients of UAE population. After obtaining informed consent, 46 Blood samples were collected from eight families including 13 PKU patients. DNA extraction, PCR amplification and DNA sequencing was performed to find the genetic mutation present in PAH gene. A major deletion of about 100 bp was observed in exon 3 of PAH gene of PKU patients studied. The results obtained in this study were also compared with published research work. It was observed that this novel deletion mutation is

present only in PKU patients of UAE population. The future work will include the DNA sequencing of remaining PKU patients, and to measure the oxidative stress using various parameters such as Glutathione/glutathione disulfide (GSH/GSSG) using GSSG/GSH Quantification kit and Malondialdehyde (MDA) by using thiobarbituric acid Assay in the plasma from UAE patients suffering from phenylketonuria. This research work will be helpful to find a specific mutations and oxidative stress parameters as an indicative of PKU disease which will ultimately lead to develop a better diagnostic and preventive measures against PKU disease.

P16. Genome-wide analysis revealed that DNA methylation is a dynamic regulator of salt and osmotic stress in rice

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Genome-wide MeDIP-seq was performed to analyze the epigenetic role of DNA methylation for salt and osmotic stress regulation in rice. Overall, we observed a great variation in methylation of the target genes that caused their differential expression and finally regulated the target stress. By comparing the methylation level of CK vs. NaCl treatment and CK vs. PEG treatment it was found that control has more methylation quantity than two stressed samples. At the gene level, the reads were also quantified in different genic regions including CpG islands, upstream2k, five-UTR, CDS, intron, three-UTR and finally investigated in downstream2k. This analysis revealed that upstream2k (promoter) is highly involved (about 43.14% in CK, 42.35% in NaCl and 43.81% in PEG) region and the others significantly involved regions were downstream2k and coding sequences (CDS). Through MeDIP-on-chip hybridization 33,009 different regulatory expressed genes were isolated, and there was a negative correlation between MeDIP-seq data and mRNA data throughout genome. Similarly, four modified forms of genes on the behalf of promoter and gene body methylation were observed throughout the genome. Based on the signal intensities of gene chip microarrays, the expressed genes were divided into five groups: lower, low, medium, high and higher and different level of methylation for each group was analyzed. RT-PCR and qRT-PCR was performed to confirm the expression patterns of targeted genes for stress regulation. Keywords:MeDIP-on-gene chip; DNA methylation/demethylation; abiotic stresses;Oryza sativa.

P17. Identification of superior buffalo bulls for increasing milk production through genome analyses

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Milk production enhancement holds promise for the prosperity and progress of Pakistan. Bull selection is important as compared to dam due to its status of half herd. A superior buffalo bull has capability to produce many thousand off spring in his life span as compared to dam who produces only 5-6. Till to date, the best procedure for bull selection was estimated breeding values (EBVs) using best available computer software. These evaluating method have their own merits and demerits but due to availability of non-authenticated data, their reliability has been questionable. To resolve this problem, bull selection will be proceed exploiting Genotyping by sequencing (GBS) approach. It is flexible, novel, sufficiently high-throughput and capable of providing acceptable genetic markers for genomic selection and genome wide association studies (GWAS). The GBS is simple and highly multiplexed system to construct libraries for next generation sequencing (NGS). It is expected that such selection will be more reliable and help to promote dairy industry when the semen of such superior bulls distributed to

Government and private breeders. So, Identification of superior buffalo bulls through genome analyses may be useful approach to increase milk production in subsequent generation. Blood samples will be collected from physically healthy buffalo bulls. Then DNA will be extracted followed by restriction digestion of whole genome. Genotyping-by-sequencing (GBS) will designed for efficient genotyping of large numbers of samples using NGS platform. Genome complexity will reduce with methylation-sensitive restriction enzyme digestion. The ends of small restriction fragments are sequenced at 96- to 384-plex levels per flow channel on the Illumina HiSeq instrument. GBS allows simultaneous discovery and genotyping of thousands of SNPs. Then genome analysis will be performed to identify the superior buffalo bulls for increasing milk production. This study will help to identify the superior buffalo bulls. Genome analyses of selected animals will help to increase milk production and also help to improve economy and health status.

P18. Morphology, physiology and microenvironment determine the morphogenetic potential of cell types of leaf callus of pearl millet cultivars

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Plant regeneration is a prerequisite for genetic transformation experiments. Pearl millet being an open pollinated crop, micropropagation is mandatory to obtain pure lines of a desired genotype, towards this endeavor, leaf explants of 3 cultivars of pearl millet IP 8182, ICP 501 and Vg 272 were chosen for the present study. Leaf explants of field grown plants failed to callus in MS or N6 medium supplemented with 2,4-D (1-5.0mg/L) or NAA (5.0mg/L), while those of in vitro grown seedlings of all cultivars callused in the same media. Leaf blade and leaf sheath callused in 2 weeks, leaf sheath responded earlier than the blade. Callus initiated from the cut ends or from the injured regions of the blade which is not in contact with the medium. Explants from younger leaves callused better than those of older leaves. All cultivars responded best to MS medium containing 5.0mg/L 2,4-D for callus induction, with different Per cent of success. Four months old embryogenic calli of cv IP8182 and of Vg 272 differentiated to plantlets in MS basal medium in a month; those of ICP 501 failed to differentiate in the same media. The plantlets were transferred to ½ strength MS medium and were hardened in Hoagland' nutrient compositions for 2 weeks before they were transferred to autoclaved soil. Genotypic differences were observed among the three cultivars with respect to callusing and plantlet formation. The chronological series of callus morphological data studied at microscope revealed three distinct types: 1.Multicellular globules, containing bipolar somatic embryoids, 2. Long, multicellular, one-cell thick filaments and 3. Large, club-shaped cells, hosting intra-cellular embryoid like structures in all three cultivars. Different cell types of the leaf explant of pearl millet respond differently to the same culture conditions, a feature shared among the three cultivars. The micro-environment and the physiological condition of cells appear to be responsible for the morphogenetic potential of mesophyll cells.

P19. Whole genome sequencing and gene annotation of almond (*Prunus dulcis*)

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Prunus dulcis (Almond)is a small deciduous trees belonging to the Rosaceae family. The study focuses on presenting the draft genome of P.dulcis along with SNP identification. The raw data, paired-end sequence libraries generated using Illumina Genome Analyzer I and II (Bio project accession SRP). The quality of the reads was checked for low-quality bases, adapters and unwanted nucleotides. All the low-quality reads were trimmed off thus giving better reads. A total of +12.19Gbp trimmed data was subjected to De Novo assembly using DDBJ pipeline and velvet for Linux for various K-mer values. The

optimum K-mer value for P.dulcis was found to be 39 with the total number of contigs and optimum N50 value as 541874 and 1235 respectively. The assembled sequence was compared with reference genome Prunusmume for gene prediction and analysis along with SNP detection. Hereby, the draft genome of P.dulcis was completed along with significant SNPs identification. The P.dulcis genome is expected to provide information on Rosaceae evolution and also can provide important data which can be used for the improvement of fruit trees with much more nutritional and medicinal value.

P20. Evaluation of suitable reference genes in date palm (*Phoenix dactylifera L.*) under drought and salinity stress for quantitative real-time PCR

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Date palm is a socio-economically important crop plant in the arid and semi-arid regions, supporting human population in the Middle East and North Africa. These areas have been largely affected by drought and salinity due to insufficient rainfall and improper irrigation practices. Date palm is a relatively salt- and drought-tolerant plant, in which efforts have been directed to identifying genes and pathways that confer its stress tolerance. Quantitative real-time PCR (qPCR) is a promising technique for the analysis of stress-induced differential gene expression, which involves the use of stable reference genes for normalizing gene expression. In an attempt to find the best reference genes for date palm's drought and salinity research, we evaluated the stability of 12 most commonly used reference genes, using RefFinder which integrates geNorm, NormFinder, BestKeeper and Comparative ΔCT methods. The comprehensive results from these analyses revealed that HEAT SHOCK PROTEIN (HSP), UBIQUITIN (UBQ) and YTH domain-containing family protein (YT521) were more stable in drought-stressed leaves, whereas GLYCERALDEHYDE-3-PHOSPHATE DEHYDROGENASE (GAPDH), ACTIN and TUBULIN were more stable in drought-stressed roots. On the other hand, SMALL SUBUNIT RIBOSOMAL RNA (25S), YT521, 18S RIBOSOMAL RNA (18S); and UBQ, ACTIN, ELONGATION FACTOR 1-ALPHA (eEF1a) were more stable in leaves and roots, respectively, under salt stress. The stability of these reference genes was verified by using the abiotic stress-responsive CYTOSOLIC Cu/Zn SUPEROXIDE DISMUTASE (Cyt-Cu/Zn SOD), ABSCISIC ACID RECEPTOR (ABA), and the PROLINE TRANSPORTER 2 (PRO) genes. These stable reference genes will facilitate studies on gene expression analysis and functional characterization of genes associated with drought and salinity tolerance in date palm. Key words: date palm, drought, salinity, quantitative real-time PCR, housekeeping genes, differential gene expression.

P21. Overexpression, purification & antimicrobial activity of recombinant human ß defensin 3

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Human ß defensin 3 (hBD-3) is a novel member of defensin family that has a broad spectrum of antimicrobial activity. hBD-3 also exhibits cytotoxic and chemotactic activities unlike other defensins. The hBD3 gene (accession number AJ237673) was synthesized from Integrated DNA Technology (IDT®). The gene was cloned in E. coli DH5α using pTZ57R/T vector and restriction sites in gene terminal ends were added by PCR. The cloned hBD-3 gene was subcloned in pET28a (+) expression vector to transform BL21 (DE3) cells for expression of recombinant hBD-3. The optimum conditions for the expression were following: LB media, the induction with 0.5mM IPTG and incubation at 37°C for 6 hours. The molecular weight of recombinant hBD-3 is 15 kDa. Recombinant hBD-3 was purified by electroelution and nickel

affinity chromatography. Antimicrobial activity of hBD-3 was determined by disc diffusion method against Bacillus thuringiensis, Bacillus simplex, Bacillus murallis, Psychrobacter alimentarius and Staphylococcus aureus. hBD-3 was found highly active against Bacillus murallis and less active against Staphylococcus aureus. Recombinant hBD-3 itsels or in combination with other antibiotics could be treat multidrug resistant pathogens.

P22. Construction, expression and purification of thymosin α1-Azu28 fusion protein for targeted cancer therapy

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Thymosin α 1 (Tα 1) has wide variety of therapeutic applications and is currently being used either alone or in combination therapy for the treatment of several diseases such as cancer. It induce the production and differentiation of lymphocytes through increasing CD3+, CD4+ and CD8+ cell proliferation, stimulating the production of different cytokines, increasing the expression of IL-2 receptors, recruiting the pre natural killer cells and increasing the production of antibodies. Azurin is a cuperodoxin protein of Pseudomonas aeruginosa whose peptide fragment from amino acids 50-77 (Azu 28) acts as a potential cell penetrating peptide and preferentially penetrates cancerous cells, stabilizes p53 inside the tumor cells and induces apoptosis through Bax mediated cytochrome c release from mitochondria. In this study, gene encoding Azu 28 was fused with Tα1gene to enhance the anti-cancer effect of Tα 1 and targeting Tα 1 to cancerous tissues. Overlap extension PCR was used to fuse Tα 1 gene and Azu 28 gene and fused gene was cloned into pTZ57/R vector. Transformed clones were screened by colony PCR and restriction analysis. Fusion gene was sub-cloned into pET28 a (+) and recombinant fusion protein was expressed in E. coli BL21 (DE3) after 4 hour induction with 1mM IPTG at 370C. The Fusion protein was purified Nickel chromatography using 150 mM imidazole and characterized with western blot and immune dot blot assays. It is concluded that synergic effect of Tα1and Azu 28 will not only make it more effective anti-cancer drug but tumor penetrating ability of Azu 28 will make it targeted therapeutic tool for safe and secure treatment of patients.

P23. Testimony of rbcL and matK loci for eight United Arab Emirates native plant species

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Plant DNA barcoding is a tool for rapid species identification that consists of a short DNA sequence unique to each species on the planet. An international body of the Plant Working Group (PWG) of the Consortium for the Barcoding of Life (CBOL) studied and validated a few gene loci; recommended two candidate-plastid sequences (rbcL and matK) for species identification. The flora of United Arab Emirates (UAE) is identified based on their morphology and no barcode is available for any of the plant species so far. An attempt is made to develop barcode for a small group of 8 native plants (2 monocotyledons and 6 dicotyledons) with rbcL and matK plastid gene sequences. Genomic DNA was isolated from the 8 plant species, the respective DNA was amplified with rbcL and/or matK primer sets following the standard protocols; the amplified PCR products (query sequences) were matched with the closest match in the National Center for Biotechnology Information (NCBI). MEGA 5 software was used to construct phylogenetic tree analysis by using neighbor joining (NJ) method. The resolving power of

rbcL was accurate among all 8 plants while the discriminatory power of matK matched for 6 out of 8 plant species. We conclude that for the tested group of plants the rbcL region is well conserved than matK, a tentative possibility to be verified for a larger sample of plant species. Further work in this direction will enable conformation of morphological.

P24. Using novel remote sensing approaches and stable isotope to predict drought tolerance in IPT transgenic creeping bentgrass in the field

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Water availability is a significant challenge to plant production in United Arab Emirates and increase drought tolerance of plants is one of the national priority and strategy for the country. In the last 20 years, genomics for drought resistance, including gene discovery and functional genomics, have been increasing exponentially. Since then, many transgenic plants harboring genes that encode for drought resistance have been produced in many model and crops plants. In this work, we have evaluated different transgenic creeping bentgrass lines with Isopentenyltransferase (IPT) gene (under the control of drought induced promoter P_{SARK}) that encodes a rate limiting enzyme in cytokinin biosynthesis that induce a delay in leaf senescence under drought stress. The experiment was carried out in the field with 3 transgenic lines and wild type (non transformed) as control. For the assessment of the green biomass, we have used a novel affordable and easy-use methods based on vegetation indices derived from conventional camera. Our measurements confirm that one of the transgenic lines (IPT-9), keep a green biomass during the period of drought treatments. These results were corroborated with RT-PCR analysis and Carbon Isotope discrimination. These preliminary data have to be confirmed by other lines in different environments and analyze the GxE interaction and gene expression.

P25. Anti-malarial drug discovery using integrative biology and systems biology

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There were an estimated 198 million cases of malaria worldwide (range 124–283 million) in 2013 and an estimated 584 000 deaths (range 367 000–755 000). 90% of all malaria deaths occur in Africa. Emerging parasite resistance to antimalarial medicines and mosquito resistance to insecticides, if left unaddressed, could render some of the current tools ineffective and trigger a rise in global malaria mortality. The P. falciparum, Anopheles gambiae and Homo sapiens genome sequences have been completed in the between 2000-2002 and represent new starting points in the centuries-long search for solutions to the malaria problem. In the past few years, there have been strong demands to generate and integrate molecular, functional and pharmacological data into a common malaria-related chemogenomic knowledge space. In the present work, we have experimented the integrative approach of using in silico study for target finding and molecular docking, pharmacophore based screening in combination with in vitro study for screening novel drug candidate against P. falciparum. The Research Database was developed from the genomics, literature and ligand information. Genome data were incorporated into pathologics software and embedded further with current literature information and made available offline and online for researcher. Search for novel anti P. falciparum was done from the Heterobase database library comprising more than 3000 compounds reported from the Ph. D thesis available in Gujarat state University Library. The consecutive in silico and in vitro screening resulted in 27 lead compounds with potential anti P. falciparum activity. The lead compounds can be explored eventually as drug candidate for antimalarial drug discovery. The consensus matrix analysis for molecular docking, Structure-based pharmacophore and ligand-based pharmacophore based was effective to reduce the

shortlisted compounds from individual screening to consensus lead compound with augmented assurance. The major study was concentered on the Plasmepsin target from the P. falciparum and the top compound recorded from in silico screening was 3001. Consequently compound 3001 and similar compounds were explored with in vitro study and dispalyed comparable trend in activity. However, the active lead compounds of in vitro study were 3004, 3007 and 3010 against both P. falciparum and P. vivax. As in either case compound 3007 have the most versatile ability to bind with the multiple targets including multi-drug resistant targets. Substantially, compound 3007 was appeared as the best lead compound of the present study. The in silico screening results proposed the need of further study on compound 3007 for anti P. vivax. and anti-HIV drug discovery. Polypharmacology in silico screening against seven targets for P. falciparum extracted 22 compounds with exceptional potential. Furthermore, the compound 518(4-(2-chloro-6,8-dimethylquinolin-3-yl)-6-(3-nitrophenyl)-3,3a,4,5-tetrahydro-2H-indazol-3-one) was obtained as the outstanding compound for dual targeting the HIV and P. falciparum in the current study. This can be further explored with in vitro testing.

P26. In-silico design of inhibitors for cyclin dependent kinase 2 molecule in breast cancer: a computational approach

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In Breast cancer disease, the cells in the breast begin to grow out of control and metastasize to invade nearby tissues and even usually spread through the body. In signal cascade mechanisms, including phosphorylation regulate activities within the cell. A change in the level of phosphorylation can have significant effects on cellular activity. Cyclin-dependent kinase 2 (CDK-2) is one of those kinases which plays central roles in regulation of cell cycle progression, transcription and metabolism. Deregulation of CDKs due to amplification, over expression or mutation contributes to proliferation of cancer cells. These kinases therefore constitute biomarkers of proliferation and attractive pharmacological targets for development of anticancer compounds. Hence CDK-2 was used as a potential target molecule in this computer aided drug designing. The in-silico study was used to identify the inhibitors, using three dimensional structures for Breast cancer susceptibility protein CDK-2 structure using Bioinformatics software. Several bioinformatics tools and software were used to determine the properties of target molecule and to develop new and improved lead compounds. 3D structure of CDK-2 was obtained from Protein databank. The structure was validated by Ramachandran plot analysis and the active site of the target molecule was found out using CASTp server. Various chemical libraries containing the 3D structure of approximately five hundred molecules were virtually screened against it employing molecular docking. The computational experiments undertaken resulted in the identification of three molecules, which docked well into the active site of the target. These three molecules obeyed Lipinski's rule of five which have been identified as potential anti-cancer agents in this in-silico study.

P27. Synthesized novel Fmoc-2-aminothiozole: protective against cadmium induced apoptosis, teratogenic effect on zebrafish (*Danio rerio*) embryos and molecular docking studies

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The 1,3Thiozole is an interesting building block in natural and synthetic compound which have been studied as a various peptide based compound. Due to health aspects of thiozole, we selectively synthesized Fmoc-2-aminothiozole-2-phenylalanine and Fmoc-2-aminothiozole-2-trptophan based on Hantzach's protocol via mixed anhydride reaction forming the intermediates diazomethyl ketone, bromomethyl ketone and under sonication thiozoles was formed. We found protective mechanism of Fmoc-2-aminothiozole-2-phenylalanine and Fmoc-2-aminothiozole-2-trptophan against cadmium intoxicated zebrafish embryos (Danio rerio). Embryos exposed to cadmium exhibited significantly reduced survival, delayed hatching, increased cardiac function and phenotypic abnormalities at 24, 48, 72 and 96 hpf. Finally, we evaluated in order to check the drug likeness of the synthesised compound molecular docking was done against the protein involved in control of variety of cellular processes. The 3D x-ray crystallography protein structure was retrieved from the protein databank (PDB) PDB ID: 1BX6 and minimization was carried out. Synthesised compounds were designed and checked for Lipinski rule of 5, ADMET properties to find the druk likeliness. These compounds were allowed to dock with the protein for 200 default iterations and the interactions of the compounds and protein was analysed and also the Hydrogen bond interactions was also obtained by the molecules, selected lead compounds were subjected in to molecular dynamics to understand the structure stability. Our results shown less toxicity compare standard compounds.

P28. Structural insights into the polypharmacological activity of dietary flavonols on serine/threonine kinases

Bincy Baby

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Discovering or designing drug molecules that can simultaneously interact with multiple targets is gaining interest in contemporary drug discovery. Serine/threonine kinases are attractive targets among protein kinases for therapeutic intervention in oncology due to the altered expression in cellular phosphorylation. Quercetin, a naturally occurring flavonol, has attracted attention for its ability to inhibit various cancer cell lines. The biological activity of quercetin glycosides has also received some attention due to their high bioavailability and activity against various diseases including cancer but has been studied to a lesser extent. This study explored the structural insights of the multitarget inhibitory activity of quercetin and its glycoside derivative, isoquercitrin on serine/threonine kinases using molecular modeling. Structural analysis showed that both quercetin and isoquercitrin exhibited good binding energies and interacted with the key aspartate residue in the highly conserved Asp–Phe–Gly motif. The results indicate that isoquercitrin could be a more potent inhibitor of several members of the serine/threonine kinase family. Thus, this study provides a structural picture of the multitarget inhibitory property of quercetin and its potential to be a chemical platform for oncological polypharmacology.

P29. Inhibiting non-receptor tyrosine kinases associated with prostate cancer using polypharmacological natural compounds

Priya Antony

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Prostate cancer is one of the most frequently diagnosed forms of cancer with high global incidence and mortality rate. Protein kinases are attractive therapeutic targets against prostate cancer due to their vital role in regulating various cellular processes. Over expression of several non-receptor tyrosine kinases (NRTKs) have been widely observed in prostate cancer. In this study, a large collection of natural compounds from the InterBioScreen library was virtually screened against three major kinases - Bruton's tyrosine kinase (BTK), focal adhesion kinase (FAK) and Src kinase to identify novel polypharmacological molecules that could inhibit the activity of these proteins. Molecular docking analysis revealed that four natural compounds that are structurally similar, possessed polypharmacological properties by interacting with these three NRTKs in a similar manner by orienting one end towards the hinge region and the other towards the activation loop. Binding score and interactions of these natural compounds were better than currently available kinase inhibitors. Thus, these natural molecules could be a framework for developing novel kinase inhibitors for the treatment of prostate cancer.

P30. Bioinformatic methodologies reveal metagenomic depiction of Indian hot springs' microbial community

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High-throughput sequencing allows quick and inexpensive analysis of econiches and revolutionizing microbial ecology studies of extreme environments. However, the effective analysis of the huge biological data is a key challenge of computational biology. In this study, we analysed 9 metagenomes of Indian hot springs and their comparative studies were conducted using MG-RAST. Furthermore, the phylogenetic and functional characterizations of the microbial communities hot springs were performed to decode the hidden microbial ecosystem. All hot springs were found to dominated by bacteria and viruses with a significant presence of unassigned sequences. The dominating Firmicutes phyla in most of the hot spring was reported due to their thermo-tolerance nature. However, Taptapani, Arti, and Unkeshwar hot springs dominated by Bacteriodetes, Cyanobacteria, and Actinobacteria respectively indicated the geographic variation play the significant role in a distribution of microorganisms. Deinococcus-Thermus group in 5 host spring can be significantly used as a metagenomic biomarker to identifies the radioactive substances. The co-occurrence association between complex microbial communities at taxonomic and functional level were also significant. The detection of stress response genes in hot spring metagenomes reveals the secrets hold by thermophiles for survival at elevated temperature. Uncharacterized genes detection in all metagenomes is the key invention towards the hidden unculturable microbes. The diversity of unculturable bacteria in all hot springs demonstrates a vast gene pool for biotechnological exploration and creates a major face for microbiologists to know the phylogenetic correlation and ecological implication of habitats. The result also enlightens the abundance, diversity, distribution and coexistence of microorganisms.

P31. Advanced statistical methods in genetic association studies

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During the last two decades, many statistical tests have been proposed for genetic associations as well as for next generation sequencing. These tests are useful to analyze genotype and next generation sequencing data and to detect genetic variations that may be responsible for disease progression. In association studies, standard approach is to consider one phenotype at a time but the new approaches during the last few years have focused on multiple correlated phenotypes and genotypes jointly. Many genetic associations are observed through phenotypes and genotypes and it could be affected by covariates like age, sex etc. Variables included in genetic studies are measured on different scales. Recognizing these scales is an important part of any research. Identification of outliers in univariate and multivariate analysis is also an important aspect. Sequential Kernel Association Test (SKAT) for rare and common variants is computationally efficient regression method to test for association between genetic variants (common and rare) in a region and a continuous or dichotomous trait while easily adjusting for covariates. Expression quantitative trait loci (eQTLs) are genomic loci that contribute to variation in expression levels of mRNAs. eQTLs that map to the approximate location of their gene-of-origin are referred to as local eQTLs. In contrast, those that map far from the location of their gene of origin, often on different chromosomes, are referred to as distant eQTLs. Often, these two types of eQTLs are referred to as cis and trans. In this talk, I will be covering some part of research methodology, identification of outliers, different measures of association useful in genetic studies and some part of eQTL analysis.

P32. EHealth applications for clinical genetics

Muhammad Jawad Hashim
United Arab Emirates University, UAE

The field of genetics is advancing at an accelerated pace. Clinical genetics is the practical implementation of genomics and bioinformatics for guiding medical decisions in patient care. Applications (or informally ‘apps’) for mobile electronic devices such as the iPad and Android tablets and smartphones have the potential to assist geneticists at the point of care. In this study, currently available mobile applications (‘apps’) were reviewed for genetic counselling by family physicians and other health professionals providing care to families with inheritable disorders. Methods Genetics apps were searched through MEDLINE via PubMed using the term “genetic mobile app”, without restriction on language or date of publication. A concurrent search was conducted on iTunes [Apple Inc.] and the Google Play Store [https://play.google.com/store/] for apps on these platforms. Apps were reviewed from the publisher’s descriptions and user’s comments. Functionality was assessed for apps based on typical clinical work environment based on the following criteria (1) applicability to clinical genetics, (2) usability defined as intuitiveness and ease-of-use, and (3) content validity. The MEDLINE search yielded 86 total articles, of these only 3 were relevant to the study. Selected articles included reviews of genetics apps as well as guidelines for development. A survey of 166 people on use of mobile apps for persons with genetic disorders such as Down's syndrome, Williams' syndrome, and 22q11 deletion syndrome found an encouraging attitude. Unfortunately, very few studies evaluated the clinical effectiveness of these apps in improving patient outcomes. One exception was an iPhone app that assisted patients with familial dysautonomia for balance and gait training. We found no apps that promised direct-to-consumer genetic testing at home based on single nucleotide polymorphisms (SNPs). Few apps met our criteria for usability, specifically for children with genetic disorders.
A small range of mobile device/smartphone applications (‘apps’) exist for clinical genetics. Most of these apps have not been validated for clinical effectiveness. While promising as useful tools for improving care of persons with genetic disorders, these apps need further refinement.

P33. Artificial Intelligence (AI), Genomics and Personalized Medicine

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The concept of personalized medicine has evolved with current trends in genomics giving the extra bit of personalization, in theory at least. While classical genomics (without using Ml/AI) still is years away from delivering tailored personalized medicine and we are yet to fully explore the solutions - modern & alternates, and the solutions that currently being specialized is majorly in modern medicine. From the recent nature article 0 speaks of fairly direct natural extract for cancer treatment, the natural/alternate forms do find traction in the Indian subcontinent, and have had selective relative impact in treating various ailments, though disputable in some circles. In places like the Middle East have had the Unani form as well, the focus here being different "body types " responding differently to ways of medicine, with renowned hospitals like the Amrita 1 giving Integrated Medical treatment. AI based treatments like AIM 2 is currently set to modern methodology of treatment based of their cases' database derived from modern medical techniques only. Here we face two fronts A. the AI part where in the Machine Learning needs to be oriented inclusive of the proven/documented success cases in Alternative forms be in Ayurveda, Unani and/Homoepathy, encompassing a greater & more challenging deviations B. The Genomics part wherein active efforts to see how the isolated metabolite works at the "omics" levels : Middle eastern/ GCC Countries like the UAE having a sizeable expat population can play host to the myriad of phenotypes present in their mixed population-Arab European Asian mix 3. As for the local UAE population studies, cue can be taken from the Qatar genome 4 where in some families (like Suawaidis) share ancestry across the borders. In matters of transcontinental diversity and well as ease of access to the “local” Indian subcontinent population (of India Pakistan Nepal and Srilanka) efforts in the UAE in the above direction in collaboration with parents countries would bear considerable fruit. Here we are in a race against time, a time wherein ever complex cancer leads the worlds death cases 5 We are at a unique juncture in history wherein highly efficient machine learning is being deeply genomics crunching big data, pacing personalization of medicine- only integrating medicine of all proven forms can do justice to the term “personal”.

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