



**HUMAN
GENOME
MEETING**

**21-24
April
2026**

ZAPPEION AIGLI
Athens, Greece

www.hugo-hgm2026.org



FINAL PROGRAM



HUMAN GENOME MEETING

GREECE





WELCOME ADDRESS

Dear colleagues & friends,

On behalf of the local organizing committee and the **Human Genome Organization (HUGO)**, it is our great pleasure to welcome you to the **Human Genome Meeting 2026** which is taking place on April **21st to 24th, 2026** in **Athens, Greece**.

The theme of the HGM2026 – “Bringing our Genomes to Medicine” reflects the focus on various aspects of genome-stratified medicine and therapeutic interventions, including but not limited to pharmacogenomics, polypharmacy, population genomics, large-scale sequencing projects, clinical implementation of personalized medicine interventions, genome informatics, public health genomics, ethics in genomics, health economics, and others. In addition, HGM 2026 will bring in educational workshops and sessions for students and early-career scientists, combined with opportunities to present their works, including mentoring sessions, combined with corporate lectures and workshops.

HGM 2026 will bring together renowned geneticists, clinicians, and doctors along with promising early-career professionals. We will have prominent keynote lectures, scientific sessions addressing recent advances in genomic research with a focus on diversity, and fruitful discussions to promote exchange of data and ideas. Drawing on a truly broad-reaching and international line up of experts, HGM 2026 promises to be both a dynamic forum for scientific exchange and a unique opportunity to bring together global expertise all focused on the subject of “Personalized Medicine and Therapeutics – Improving health”.

We would like to thank you all for your participation!

Yours sincerely,

Piero Carninci
President, HUGO

George P. Patrinos
Chair, Organizing Committee





COMMITTEES

INTERNATIONAL ORGANISING COMMITTEE

Chair

George P. Patrinos

*Director General, Hellenic Pasteur Institute, Athens
Professor, University of Patras Department of Pharmacy, Patras, Greece*

Vice Chair

Juergen Reichardt

Professor, Australian Institute of Tropical Health and Medicine, Australia

Members

Zilfalil Bin Alwi

Professor, Universiti Sains Malaysia, Kuala Lumpur, Malaysia

Fabiana Arzuaga

*Professor, Ministry of Science, Technology and Productive Innovation Argentina,
Buenos Aires, Argentina*

Piero Carninci

Head of Genomics Research Centre, Human Technopole, Milan, Italy

Ada Hamosh

Johns Hopkins University, US

Partha Majumder

Professor, National Institute of Biomedical Genomics, New Delhi, India

Vasilis Vasiliou

Professor, Yale University, New Haven CT, USA

Joris Veltman

Professor, The University of Edinburgh, Edinburgh, UK





21-24 April



LOCAL SCIENTIFIC COMMITTEE

Mihail Aivaliotis

Aristotle University of Thessaloniki, Faculty of Medicine, Thessaloniki, Greece

Pantelis Bagos

Department of Computer Science and Biomedical Informatics, Lamia, Greece

Sotiria Boukouvala

Democritus University of Thrace, Department of Molecular Biology and Genetics, Alexandroupolis, Greece

Artemis Chatzigeorgiou

University of Thessaly, Department of Bioinformatics, Lamia, Greece

Dimitra Dafou

Aristotle University of Thessaloniki, Department of Biology, Thessaloniki, Greece

Antigone Dimas

Biomedical Sciences Research Center Alexander Fleming, Athens, Greece

Giannoulis Fakis

Democritus University of Thrace, Department of Molecular Biology and Genetics, Alexandroupolis, Greece

Maria Gazouli

National and Kapodistrian University of Athens, School of Medicine, Athens, Greece

Antonios Giakountis

University of Thessaly, Department of Biochemistry and Biotechnology, Larissa, Greece

Nikolaos Karamanos

University of Patras, Department of Chemistry, Patras, Greece

Timokratis Karamitros

Hellenic Pasteur Institute, Athens, Greece

Dimitris Kletsas

National Center for Scientific Research "Democritus", Athens, Greece

Athanassios Kotsinas

National and Kapodistrian University of Athens, School of Medicine, Athens, Greece

Christos Kroupis

National and Kapodistrian University of Athens, School of Medicine, Athens, Greece

George Lagoumintzis

University of Patras, Department of Pharmacy, Patras, Greece

Evi Lianidou

National and Kapodistrian University of Athens, Department of Chemistry, Athens, Greece

Ioanna Maroulakou

Democritus University of Thrace, Department of Molecular Biology and Genetics, Alexandroupolis, Greece

Theologos Michailidis

University of Ioannina, Department of Biomedical Applications and Technology, Ioannina, Greece

Alexandros Onoufriadis

Aristotle University of Thessaloniki, School of Medicine, Thessaloniki, Greece

Spiros Papakostas

International Hellenic University, Department of Science and Technology, Thessaloniki, Greece

Pavlos Pavlidis

University of Crete, Department of Biology, Heraklion, Greece

Vassilios Plagianakos

University of Thessaly, Department of Bioinformatics, Lamia, Greece

Nikos Poulakakis

University of Crete, Department of Biology, Heraklion, Greece

Maria Samara

University of Thessaly, Department of Medicine, Larissa, Greece

Argyro Sgourou

Hellenic Open University, Patras, Greece

Antonios Stamatakis

National and Kapodistrian University of Athens, Department of Nursing, Athens, Greece

Konstantinos Stathopoulos

University of Patras, Department of Medicine, Patras, Greece

Evgenia Stefanou

University of Patras, Department of Medicine, Patras, Greece

Dimitrios Stravopodis

National and Kapodistrian University of Athens, Department of Biology, Athens, Greece

Dimitrios Thanos

Biomedical Research Foundation of the Academy of Athens, Athens, Greece

Nikolaos Vakirlis

Hellenic Pasteur Institute, Athens, Greece

Yiannis Vasilopoulos

University of Patras, Department of Biology, Patras, Greece

Ioannis Vizirianakis

Aristotle University of Thessaloniki, Department of Pharmacy, Thessaloniki, Greece

Christos Yapijakis

National and Kapodistrian University of Athens, School of Medicine, Athens, Greece

Aris Vrahatis

Ionian University, Department of Informatics, Corfu, Greece



SCIENTIFIC PROGRAM

VENUE:
**HELLENIC
PASTEUR
INSTITUTE**

Tuesday, April 21st 2026

HALL: AMPHITHEATER

09:00 - 10:30 EDUCATION WORKSHOP 1 - ALL ABOUT TOOLS

Ada Hamosh, *Johns Hopkins University, US*

GnomAD, seqr, and other genomic resources

Anne O'Donnell-Luria, *Broad Institute of MIT and Harvard, USA*

10:30 - 11:30 EDUCATION WORKSHOP 2 - HOW TO COMPILE A SCIENTIFIC ARTICLE: FROM THE AUTHOR'S, REFEREE'S AND EDITOR-IN CHIEF'S PERSPECTIVE

Co-ordinator: *George P. Patrinos, Editor-In-Chief, The Pharmacogenomics Journal*

Supported by **SPRINGER** The Pharmacogenomics Journal
NATURE

11:30 - 13:00 EDUCATION WORKSHOP 3 - HOW TO WRITE A GRANT

Co-ordinators: *Juergen Reichardt, Australian Institute of Tropical Health and Medicine, Australia*

Ada Hamosh, *Johns Hopkins University, US*

Piero Carninci, *Human Technopole, Milan, Italy*

Vasilis Vasiliou, *Yale University, USA*

13:00 - 13:30 Snack break

15:00 - 18:00 EDUCATION WORKSHOP 4 - VARIANT INTERPRETATION VARIANT CLASSIFICATION AND THE ACMG/AMP CLASSIFICATION SYSTEM

Andreas Laner, *MGZ, Germany*

UCSC Genome Browser in Variant Interpretation

Robert Kuhn, *University of California, Santa Cruz, USA*





Tuesday, April 21st 2026

HALL: PAVILLION

10:00 - 13:30 ELSI WORKSHOP: THE HUMAN GENOME PROJECT 2 AND PRECISION PUBLIC HEALTH

Welcome and Opening Remarks – Introducing the Human Genome Project 2

Chair: Liu Weibin, BGI China

Session 1: Collecting Genomes

What Does One Health tell us About Genomics?

Benjamin Capps, Dalhousie University, Canada

Session 2: Sharing Genomes

Diversity and inclusion of different paradigm thinking towards HGP2

Deborah Mascalonzi, Uppsala University, Sweden & Eurac, Italy

Break: Informal Meetups

Session 3: Accessing Genomes

Reflections on Data Visitation

Francis P. Crawley, CODATA IDPC, Belgium

Session 4: Precision Public Health

What Does a HGP2 Biobank Look Like?

Johnathan LoTempio Jr., University of Pennsylvania, USA

Future Directions: Open Discussion

Moderator: Dawn Chen, Harvard University, USA

Concluding Remarks

13:30 - 14:30 MEET THE PROFESSORS SESSION

Co-ordinators:

George P. Patrinos, Hellenic Pasteur Institute & University of Patras, Greece

Juergen Reichardt, Australian Institute of Tropical Health and Medicine, Australia

Ada Hamosh, Johns Hopkins University, US

Piero Carninci, Human Technopole, Milan, Italy

Zilfalil Bin Alwi, University Sains, Malaysia



VENUE:
**ZAPPEION
AIGLI**

Wednesday, April 22nd 2026

HALL: **PARTHENON**

09:00 - 10:30 PANGENOME WORKSHOP

PART 1

**HUMAN PANGENOME RESOURCE: EMERGING APPLICATIONS
AND METHODS FOR ANALYZING HAPLOTYPE DIVERSITY**

Co-ordinator: Karen Miga, University of California, Santa Cruz, USA

10:30 - 11:00 Coffee Break & Visit to e-Posters / Exhibition

11:00 - 12:30 PANGENOME WORKSHOP

PART 2

12:30 - 13:45 Light Lunch, Visit to Exhibition & Networking

Career Mentoring & Coaching (Academic)

13:45 - 14:15 Opening Ceremony

George P. Patrinos, Piero Carninci, Invited Greek Authorities

14:15 - 15:00 PLENARY LECTURE 1

Chair: George P. Patrinos, Hellenic Pasteur Institute & University of Patras, Greece, National Genomes Projects Consortia,

National Genomes Projects Consortia

Piero Carninci, Human Technopole, Milan, Italy

15:00 - 15:30 Coffee Break & Visit to e-Posters / Exhibition

15:30 - 18:00 SYMPOSIUM 1: NATIONAL GENOME INITIATIVES

Chairs: Piero Carninci, Human Technopole, Milan, Italy

Juergen Reichardt, Australian Institute of Tropical Health and Medicine, Australia

MyGenom: Establishing a Reference Genome for Precision Medicine in Malaysia

Syahril Abdullah (Malaysia)

Genomics Thailand: Five Years, 50,000 Genomes, and Beyond
Jakris Eu-ahsunthornwattana (Thailand)





VENUE:
**ZAPPEION
AIGLI**

Wednesday, April 22nd 2026

HALL: **PARTHENON**

**Developing a National Genomic Ecosystem in Indonesia:
Early Experiences and Challenges**
Indri Rooslamiati (*Indonesia*)

Genome of Europe
Andre Uitterlinden (*The Netherlands*)

Fabiana Arzuaga (*Argentina*)

The Genome of Greece Initiative
George P. Patrinos, (*Greece*)

**The Emirati T2T-Pangenome Project: From Population Genomics
to Precision Medicine**
Habiba Al Safar (*Emirates*)

**Population history, Genetic Variation and it's Medical relevance
in India**
Analabha Basu (*India*)

**Human Genome Project II (HGP2): Sequencing 1% of humanity
for global precision health**
Wei-bin Liu (*China*)

18:00 - 18:15 **The 5000genomi@VdA Project – From Patients To Genomic Data /
Joint Lab with GenomSys on MPEG-G standard adoption test**
Prof. Stefano Gustincich, *Coordinator of the Center for Clinical
and Computational Genomics - C3G – Istituto Italiano di Tecnologia*
Dr. PhD Guido Walter Di Donato, *CTO of GenomSys
and CEO of GenoGra*
Industry Session by GENOMSYS

18:30 **Welcome Reception**





VENUE:
ZAPPEION
AIGLI

Wednesday, April 22nd 2026

HALL: MACEDONIA

09:00 - 10:30 EDUCATION WORKSHOP 5 - NEW TECHNOLOGIES FOR GENETICISTS EDUCATION

PART 1

Understanding cutting-edge genomic technologies in research and diagnostics

Ed Tobias, *University of Glasgow, UK*

10:30 - 11:00 Coffee Break & Visit to e-Posters / Exhibition

11:00 - 12:30 EDUCATION WORKSHOP 5 - NEW TECHNOLOGIES FOR GENETICISTS EDUCATION

PART 2

12:30 - 13:45 Light Lunch, Visit to Exhibition & Networking

Career Mentoring & Coaching (Academic)

15:00 - 15:30 Coffee Break & Visit to e-Posters / Exhibition

15:30 - 16:45 SYMPOSIUM 2: ENVIRONMENTAL GENOMICS

Chairs: Giuseppe Novelli, University of Rome Tor Vergata, Italy

George Lagoumintzis, University of Patras, Greece

ExposoGenomics: Integrating the Genome and Exposome in the Era of AI and Data Science

Vasilis Vasiliou, *Yale University, USA*

Cancer risk at the genome-exposome interface: toward translational composite scoring

Denis Sarigiannis, *National Hellenic Research Foundation*

Bridging Exposome and Genome: Gene-Environment Interactions in PFAS-Associated Liver Disease

Lida Chatzi, *University of Southern California, USA*

O.P.

The non-genetic non-environmental origins of the missing 50% of disease risk

J. Nadeau

O.P.

Relationship between female reproductive traits, exercise, and bone health

B. Chen





Wednesday, April 22nd 2026

HALL: MACEDONIA

16:45 - 18:00 SYMPOSIUM 3: RARE DISEASES AND OTHER ABSTRACTS

*Chairs: Charles Lee, The Jackson Laboratory, US
Christos Yapjakis, National & Kapodistrian University of Athens, Greece*

O.P.

Toward Synthetic Human Genomes: High-Fidelity Chromosome Transplantation

G. Petris

O.P.

Metabolic therapy rescues neuroexcitability and neuroinflammation in GRIN2D GOF

D. Galber

O.P.

Screening newborn conditions using a Probe-based DNA method in Filipino neonates

T.D. Fabella

O.P.

N-of-1 personalized therapy for CMT2S: From patient-specific preclinical models

S. Smieszek

O.P.

Designing equitable benefit sharing for pathogen genomics in pandemics

L. Prinsen

O.P.

One-carbon pathway and cognitive development in trisomy 21

B. Vione

18:00-18:15 Unleash the power of Predictive Genomics: How can we innovate diseases prevention?

*Cristina Valsecchi, Sr. Technical Sales Specialist- Human Predictive Genomics - Italy, Iberia, Israel and South East Europe
Industry Session by **THERMO FISHER***

18:30 Welcome Reception





VENUE:
**ZAPPEION
AIGLI**

Thursday, April 23rd 2026

HALL: **PARTHENON**

**09:00 -10:30 SYMPOSIUM 4 - AI AND MACHINE LEARNING APPLICATIONS
IN HUMAN GENOMICS**

Chairs: Partha Majumder, National Institute of Biomedical Genomics, India

Timokratis Karamitros, Hellenic Pasteur Institute, Athens, Greece

Why Networks Matter: Embracing Biological Complexity

John Quackenbush, Harvard University, USA

O.P.

Using Machine Learning and Germline Pharmacogenomics to Personalize Combined mRNA

P. Khalili

O.P.

ARGformer: learning on ancestral recombination graphs with transformers

D. Bonet

10:30 -11:15 Coffee Break & Visit to e-Posters / Exhibition

11:15 -11:50 PLENARY LECTURE 2:

Chair: Ada Hamosh, Johns Hopkins University, US

Genomics and Public Health Genomics and Public Health

Sonja Rasmussen, Johns Hopkins University

11:50-13:00 Light Lunch, Visit to Exhibition & Networking

Career Mentoring & Coaching (Academic)

13:00-14:30 SYMPOSIUM 6 - GENOME REGULATION

Chair: Anne Bowcock, Mount Sinai, US

Ioanna Maroulakou, Democritus University of Thrace, Alexandroupoli, Greece

Cell type-specific intronic RNAs shape genome architecture during neuronal lineage specification

Magda Bienko, Human Technopole, Italy





VENUE:
ZAPPEION
AIGLI

Thursday, April 23rd 2026

HALL: PARTHENON

O.P.

Genetic regulation across germline and somatic variation on the Y chromosome

G. Sato

O.P.

Leveraging cis and trans variants to improve protein expression level prediction

S. Leal

O.P.

Genome Regulation by NFAT During Endothelial-Like Differentiation

A. Azlina

New genes enable protein structural innovation and function in the brain

V. Luria

14:30-15:00 Enabling precision medicine with Illumina multi-omic technologies

Stef Kupers, *Strategic Segment Manager*

Industry Session by ILLUMINA

15:00-16:00 Coffee Break & Visit to e-Posters / Exhibition

16:00 - 17:15 **SYMPOSIUM 8 - ADVANCES IN PSYCHIATRIC GENOMICS**

Chairs: George P. Patrinos, Hellenic Pasteur Institute & University of Patras, Greece

Christina Mitropoulou, The Golden Helix Foundation, UK

Human-specific small non-coding RNAs link genomic architecture to the transcriptome and proteome of brain disorders

Isidore Rigoutsos, *Thomas Jefferson University Philadelphia, USA*

Personalized psychiatry: pharmacogenetic insights

Alessio Squassina, *University of Cagliari, Italy*

O.P.

Neuroplasticity-Related Genetics and Microbiome Profiles as Determinants of AUD

E. Legaki

O.P.

Tandem Repeat Expansions Modify and Predict Schizophrenia in 22q11.2 Deletion

R. Yuen

19:30

Gala Dinner





VENUE:
ZAPPEION
AIGLI

Thursday, April 23rd 2026

HALL: MACEDONIA

09:00 -10:30 SYMPOSIUM 5 - RARE DISEASES FROM GENOMICS TO PATIENTS

Chairs: Ada Hamosh, Johns Hopkins University, US

Maria Gazouli, National & Kapodistrian University of Athens, Greece

DECIPHERD: an interdisciplinary approach to Rare and Undiagnosed Diseases in Chile

Gabriela Repetto, Universidad del Desarrollo, Chile

Entwined and complex cellular mechanisms underlying monogenic diseases caused by mutations in TGF-beta signalling pathway components

Bassam Ali, United Arab Emirates University, UAE

Rare Diseases, Rare Variants: What changes when Africa is included
Aime Lumaka, University of Kinshasa, DR Congo

10:30 -11:15 Coffee Break & Visit to e-Posters / Exhibition

11:50-13:00 Light Lunch, Visit to Exhibition & Networking

Career Mentoring & Coaching (Academic)

13:00-14:30 SYMPOSIUM 7 - PUBLIC POLICY AND HEALTH ECONOMICS IN GENOMIC MEDICINE

Chairs: Zilfalil Bin Alwi, University Sains, Malaysia

Sotiria Boukouvala, Democritus University of Thrace, Alexandroupoli, Greece

The Health Economics of Genomic Newborn Screening

James Buchanan, Queen Mary University of London, UK

Economic evaluation of genome-guided therapeutic interventions: Results from the PREPARE study in Europe

Christina Mitropoulou, The Golden Helix Foundation, UK

The economic value of human genomics

Ilias Goranitis, University of Melbourne, Australia





VENUE:
ZAPPEION
AIGLI

Thursday, April 23rd 2026

HALL: MACEDONIA

O.P.

GEN-IMPACT: Aligning African Leadership, Genomics & Longitudinal Data for Equity

E. Bolukbasi

15:00 - 16.00 Coffee Break & Visit to e-Posters / Exhibition

16:00 - 17:15 SYMPOSIUM 9 - LARGE-SCALE GENOME PROJECTS AND THE HUMAN PANGENOME

Chair: Vasilis Vasiliou, Yale University, US

Giannoulis Fakis, Democritus University of Thrace, Greece

The Human Pangenome Project: A New Foundation for Human Genomics

Karen Miga, University of California, Santa Cruz, USA

O.P.

Long-read sequencing reveals extensive structural variation across India

D.T. Sowpati

O.P.

A multi ancestry evaluation of Polygenic Scores across global populations

L. Repetto

19:30

Gala Dinner



VENUE:
ZAPPEION
AIGLI

Friday, April 24th 2026

HALL: PARTHENON

09:00 - 10:15 SYMPOSIUM 10 - ANCIENT GENOMES AND HUMAN ANCESTRY

Chair: Anne Bowcock, Mount Sinai, US

How Environment and Migration Shape Human Population Genetic Diversity

Hie Lim Kim, *NTU, Singapore*

High quality human genome assemblies (pangenome) from the island of Crete and the genomic landscape of the Cretan population
Pavlos Pavlidis, *University of Crete, Greece*

O.P.

Deciphering complete archaic introgression sequences in modern human genomes

M. Suo

O.P.

Graph transformer for ancient local ancestry inference

C. Shanks

10:15 - 11:15 Coffee Break & Visit to e-Posters / Exhibition

11:15 - 12:00 PLENARY LECTURE 3

Chair: Ada Hamosh, Johns Hopkins University, US

Federated gnomAD, GA4GH, Standards and Sharing

Heidi Rehm, *Broad Institute, USA*

12:00 - 13:30 Light Lunch, Visit to Exhibition & Networking

Career Mentoring & Coaching (Academic)

13:30 - 14:45 HUGO CHEN AWARDS

Introduction: Piero Carninci

HUGO CHEN AWARDS FOR DISTINGUISHED ACADEMIC ACHIEVEMENT IN HUMAN GENETIC AND GENOMIC RESEARCH:

The genetics and phenotyping of complex disease

Mike Snyder, *Stanford University, USA*





Friday, April 24th 2026

HALL: PARTHENON

HUGO CHEN AWARD OF EXCELLENCE:

A genomics revolution to decode the tissue ecosystems of the human body

Muzlifah Haniffa, Wellcome Sanger Institute, UK

HUGO CHEN AWARD OF EXCELLENCE:

An AI Design Framework For Efficient Manufacturing of mRNA Therapeutics

Yue Wan, A*STAR Genome Institute, Singapore

14:45 - 15:10 PLENARY LECTURE 4: HUGO AFRICAN PRIZE

Chair: Stylianos Antonarakis

From Local Genomes to Global Health: Pioneering Human Genetics Research in Senegal

Rokhaya Ndiaye, Cheikh Anta Diop University, Senegal

15:10 - 15:45 Coffee Break & Visit to e-Posters / Exhibition

15:45 - 17:00 SYMPOSIUM 12 - PRECISION ONCOLOGY: GENOMICS AND THERAPEUTICS OF CANCER

Chairs: Juergen Reichardt, Australian Institute of Tropical Health and Medicine, Australia

Yiannis Vasilopoulos, University of Patras, Greece

Precision Oncology: Personalised Genomic Insights in the Era of Neoadjuvant Immunotherapy

Kai-Keen Shiu, University College London, UK

O.P.

ONCOsORF: Predicting Oncogenic sORFs in Non-coding RNAs
A. Paschoal

O.P.

Novel Stage-Specific microRNA Panel for the Saliva-Based Detection of Oral Squamous Cell Carcinoma and Premalignant Dysplasia

I. Gintoni

17:00-17:45 Poster Prize

17:45 -18:00 Introduction to HGM2027

18:00 -18:15 Closing remarks - End of Meeting





VENUE:
**ZAPPEION
AIGLI**

Friday, April 24th 2026

HALL: **MACEDONIA**

09:00 - 10:15 SYMPOSIUM 11 - POLYPHARMACY AND PHARMACOGENOMICS

(Supported by the SafePolyMed Project)

Chairs: George P. Patrinos, Hellenic Pasteur Institute & University of Patras, Greece

Christina Mitropoulou, The Golden Helix Foundation, UK

Thorsten Lehr, Saarland University, Germany

From variants to protein networks to cure:

The missing therapeutic layers

Harald Schmidt, Maastricht University, The Netherlands

O.P.

The Genetic Architecture of the Hellenes

N. Katsanis

O.P.

MEG3-miRNA Regulatory Axis Modulates Response to bDMARDs in Psoriatic Arthritis

G. Novelli

10:15 - 11:15 Coffee Break & Visit to e-Posters / Exhibition

12:00 - 13:30 Light Lunch, Visit to Exhibition & Networking

Career Mentoring & Coaching (Academic)

15:10 - 15:45 Coffee Break & Visit to e-Posters / Exhibition

15:45 - 17:00 SYMPOSIUM 13 - RNA AND DISEASE

Chairs: Piero Carninci, Human Technopole, Italy

Aris Vrahatis, Ionian University, Greece

Circular RNA aptamers act as RNA chaperones to prevent cytosolic TDP-43 aggregation

Ling-Ling Chen, Shanghai Institute of Biochemistry and Cell Biology, China





VENUE:
**ZAPPEION
AIGLI**

Friday, April 24th 2026

HALL: **MACEDONIA**

Discovery of snRNA and lncRNA associated neurodevelopmental disorders

Anne O'Donnell-Luria, *Broad Institute of MIT and Harvard, USA*

O.P.

Generative Design of Cell Type-Specific RNA Splicing for Gene Regulation

D. Chen

O.P.

Spatial isoforms at sub-micrometer resolution show novel variability patterns

H. Tilgner

illumina®

AUTHORISED
CHANNEL PARTNER



NovaSeq X Plus



NextSeq 2000

Driving the multiomics revolution

TruPath Genome:
Unprecedented simplicity.
Reveal complexity.

Illumina 5-base solution:
One assay.
Dual insights.

M

M



BioAnalytica
BIOTECHNOLOGY SYSTEMS



Oral Presentations



THE NON-GENETIC NON-ENVIRONMENTAL ORIGINS OF THE MISSING 50% OF DISEASE RISK

Joe Nadeau¹, PERMUTE Working Group¹⁻⁷, Andrew Pospisilik²

¹ MaineHealth Institute for Research, Maine Medical Center, Portland, Maine

² Center for Epigenetics, Van Andel Institute, Grand Rapids, Michigan

³ Roux Institute of Northeastern University, Portland, Maine

⁴ Geisinger Health System, Danville, Pennsylvania

⁵ A*STAR Singapore Institute for Clinical Sciences, Singapore

⁶ Center for Informed Futures, University of Auckland, Auckland

⁷ Dept. of Computational and Quantitative Medicine, City of Hope, Duarte, California

GENETIC PERSPECTIVE ON THE RELATIONSHIP BETWEEN FEMALE REPRODUCTIVE TRAITS, EXERCISE, AND BONE HEALTH OUTCOMES: EVIDENCE FROM A NATIONAL DATABASE

Bo-Kai Chen¹, Meng-Che Tsai^{2,3,4}

¹ School of Medicine, College of Medicine, National Cheng Kung University, Tainan, Taiwan

² Department of Pediatrics, National Cheng Kung University Hospital, College of Medicine, National Cheng Kung University, Tainan, Taiwan

³ Department of Genomic Medicine, National Cheng Kung University Hospital, College of Medicine, National Cheng Kung University, Tainan, Taiwan.

⁴ Department of Medical Humanities and Social Medicine, School of Medicine, College of Medicine, National Cheng Kung University, Tainan, Taiwan.

TOWARD SYNTHETIC HUMAN GENOMES: HIGH-FIDELITY CHROMOSOME TRANSPLANTATION

Gianluca Petris^{1,2‡}, Simona Grazioli¹, Linda van Bijsterveldt¹, Pierre Murat^{1§}, Kim C. Liu¹, Jakob Birnbaum¹, Julian E. Sale^{1,2}, Jason W. Chin^{1,2}

¹ Medical Research Council Laboratory of Molecular Biology, Cambridge, UK.

² Wellcome Sanger Institute, Wellcome Trust Genome Campus, Hinxton, UK. ‡ Present address: Università degli Studi di Udine, Udine, Italy, and Fondazione Italiana Fegato, Basovizza, Trieste, Italy. § Present address: Wellcome Sanger Institute, Wellcome Trust Genome Campus, UK

METABOLIC THERAPY RESCUES NEUROEXCITABILITY AND NEUROINFLAMMATION IN GRIN2D GOF

Danielle Galber¹, Mor Yam², Roni Gal², Wayne N. Frankel³, Moran Rubinstein², Karen B. Avraham¹

¹ Laboratory of Neural & Sensory Genomics, Department of Human Molecular Genetics and Biochemistry, Faculty of Medicine and Sagol School of Neuroscience, Tel Aviv University, Tel Aviv, Israel

² Goldschleger Eye Research Institute, Department of Human Molecular Genetics and Biochemistry, Faculty of Medicine and Sagol School of Neuroscience, Tel Aviv University, Tel Aviv, Israel

³ Department of Genetics & Development, Institute for Genomic Medicine, Columbia University Irving Medical Center, New York, NY, USA





SCREENING OF NEWBORN CONDITIONS USING A TARGETED, MULTIPLEXABLE, DNA-BASED METHOD IN FILIPINO NEONATES

Terence Diane Fabella^{1,2,3,5}, Erik A. Sisternans, PhD^{1,2}, Lidewij Henneman, PhD^{1,2}, Gerard Pals, PhD¹, Joery den Hoed, PhD⁵, Carmencita D. Padilla, MD, MAHPS^{3,4}, Eva Maria Cutiongco-de la Paz, MD^{3,4}, Jan Schouten, PhD⁵

- ¹ Department of Human Genetics, Amsterdam UMC, Vrije Universiteit Amsterdam, Amsterdam, the Netherlands,
- ² Amsterdam Reproduction and Development Research Institute, Amsterdam, the Netherlands,
- ³ Institute of Human Genetics, National Institute of Health, University of the Philippines, Manila, Philippines,
- ⁴ Newborn Screening Reference Center, Manila, Philippines,
- ⁵ Research and Development Unit, MRC Holland, Amsterdam, the Netherlands

N-OF-1 PERSONALIZED THERAPY FOR CMT2S: FROM PATIENT-SPECIFIC PRECLINICAL MODELS TO FIRST-IN-HUMAN DOSING

Sandra Smieszek, PhD¹, Bart Przychodzen, PhD¹, Christina Tyner, MS¹, Caroline Johnson, BS¹, Christos M. Polymeropoulos, MD¹, Gunther Birznieks, MS¹, Jennifer M. Kwon⁴, Mihael H. Polymeropoulos, MD¹

Vanda Pharmaceuticals

DESIGNING EQUITABLE BENEFIT SHARING FOR PATHOGEN GENOMICS IN PANDEMICS

L. Prinsen

University of the Free State

ONE-CARBON PATHWAY AND COGNITIVE DEVELOPMENT IN TRISOMY 21

Beatrice Vione¹, Giuseppe Ramacieri^{2,3}, Sara Onnivello⁴, Michela Cicilloni¹, Maria Caracausi¹, Giacomo Sperti⁵, Pietro Paradisi⁶, Chiara Marcolin⁴, Elisa Rossi⁴, Sara Colaianni⁴, Aurora Platè⁴, Francesca Pulina⁴, Francesca Antonaros¹, Maria Chiara Pelleri¹, Allison Piovesan¹, Lorenza Vitale¹, Pierluigi Stripoli¹, Gian Luca Pirazzoli⁷, Luigi Tommaso Corvaglia^{1,5}, Francesca Catapano², Chiara Locatelli⁵, Silvia Lanfranchi⁴.

- ¹ Department of Biomedical and Neuromotor Sciences (DIBINEM), University of Bologna, Bologna, Italy.
- ² Department of Medical and Surgical Science (DIMEC), University of Bologna, Bologna, Italy.
- ³ Speciality School of Child Neuropsychiatry - Alma Mater Studiorum, University of Bologna, Via Massarenti 9, 40138, Bologna, Italy.
- ⁴ Department of Developmental and Socialization Psychology, University of Padua, Padua, Italy.
- ⁵ Neonatal Intensive Care Unit, IRCCS Azienda Ospedaliera-Universitaria di Bologna (A.O.U.BO), Italy.
- ⁶ Speciality School of Paediatrics - Alma Mater Studiorum, University of Bologna, Via Massarenti. 9, 40138, Bologna, Italy.
- ⁷ IRCCS Istituto delle Scienze Neurologiche di Bologna, U.O.C. Clinica Neurologica Rete Metropolitana, Bologna, Italy.



ARGFORMER: LEARNING ON ANCESTRAL RECOMBINATION GRAPHS WITH TRANSFORMERS

David Bonet, Cole Shanks, Marçal Comajoan Cara, Jordi Abante, Alexander G. Ioannidis
University of California, Santa Cruz; Stanford University; Universitat de Barcelona; University of California, Berkeley

GEN-IMPACT: ALIGNING AFRICAN LEADERSHIP, GENOMICS, AND LONGITUDINAL DATA FOR EQUITABLE DISCOVERY

Ekin Bolukbasi
Wellcome

GENETIC REGULATION ACROSS GERMLINE AND SOMATIC VARIATION ON THE Y CHROMOSOME CONTRIBUTES TO TYPE 2 DIABETES

Go Sato¹⁻³, Yuji Yamamoto², Kyuto Sonehara¹, Ryunosuke Saiki⁴, Takafumi Ojima^{1-3,5}, Masahiro Kanai⁶, Aoxing Liu⁶, Ryuya Eda^{2,3}, Yuya Shirai^{2,3}, Shinichi Namba¹⁻³, Ho Namkoong⁷, Atsushi Kumanogoh², Shigeki Nakagome⁸, Gen Tamiya⁵, Mark J. Daly⁶, James F. Wilson⁹, Masayuki Yamamoto⁵, Koichi Matsuda¹, Seishi Ogawa⁴, Toshimasa Yamauchi¹, Takashi Kadowaki¹⁰, and Yukinori Okada¹⁻³

1. *The University of Tokyo, Tokyo, Japan*
2. *The University of Osaka, Suita, Japan*
3. *RIKEN Center for Integrative Medical Sciences, Yokohama, Japan*
4. *Kyoto University, Kyoto, Japan*
5. *Tohoku University, Sendai, Japan*
6. *Massachusetts General Hospital, Boston, MA, USA*
7. *Keio University School of Medicine, Tokyo, Japan*
8. *Trinity College Dublin, Dublin, Ireland*
9. *The University of Edinburgh, Edinburgh, Scotland*
10. *Toranomon Hospital, Tokyo, Japan*

LEVERAGING CIS AND TRANS VARIANTS TO IMPROVE PROTEIN EXPRESSION LEVEL PREDICTION

Rui Dong¹, Derek Lam^{b1}, Gao T. Wang¹, Andrew T. DeWan², Suzanne M. Leal¹
 1. *Center for Statistical Genetics, Gertrude H. Sergievsky Center, and the Department of Neurology, Columbia University, New York, NY 10032, USA*
 2. *Department of Chronic Disease Epidemiology and Center for Perinatal, Pediatric and Environmental Epidemiology, Yale School of Public Health, 1 Church Street, New Haven, CT 06510, USA*

GENOME REGULATION BY NFAT DURING ENDOTHELIAL-LIKE DIFFERENTIATION

Mohadesse Mozafari¹, Siti Nurnasihah Md Hashim¹, Khairul Bariah Ahmad Amin Noordin¹, Siti Aishah Zainal¹, TP Kannan^{1,2}, Suzina Sheikh Abd Hamid³ & Ahmad Azlina^{1,2,3}
 1. *School of Dental Sciences, Universiti Sains Malaysia, Health Campus, 16150 Kubang Kerian, Kelantan, Malaysia*
 2. *Human Genome Centre, School of Medical Sciences, Universiti Sains Malaysia, Health Campus, 16150 Kubang Kerian, Kelantan, Malaysia*
 3. *Tissue Bank, School of Medical Sciences, Universiti Sains Malaysia, Health Campus, 16150 Kubang Kerian, Kelantan, Malaysia*





NEW GENES ENABLE PROTEIN STRUCTURAL INNOVATION AND FUNCTION IN THE BRAIN

Victor Luria^{1,2,5}, Amir Karger³, John W. Cain⁴, Marc Kirschner², Anne O'Donnell-Luria⁵, Nenad Sestan¹

¹ Yale University, Department of Neuroscience, New Haven, CT, USA

² Harvard Medical School, Department of Systems Biology, MA, USA

³ Harvard Medical School, IT-Research Computing, Boston, MA, USA

⁴ Harvard University, Department of Mathematics, Cambridge, MA, USA

⁵ Broad Institute of MIT and Harvard, Program in Medical and Population Genetics, Cambridge, MA, USA

NEUROPLASTICITY GENETICS AND MICROBIOME AS DETERMINANTS OF AUD TREATMENT RESPONSE

Evangelia Legaki¹, Nikolas Dovrolis¹, Ilias Koutromanos^{2,3}, Efthimios Vassilopoulos², Arthur Stem⁴, Vasilis Vasiliou⁴, Elias Tzavellas², Maria Gazouli¹

¹ Laboratory of Biology, Department of Basic Biological Science, School of Medicine, National and Kapodistrian University of Athens, 11527, Athens, Greece.

² First Department of Psychiatry, "Aiginition" Hospital, School of Medicine, National and Kapodistrian University of Athens, 11528, Athens, Greece.

³ Department of Psychiatry and Psychotherapy, Psychiatric Services Aargou AG, 5210, Brugg-Windisch, Switzerland.

⁴ Department of Environmental Health Sciences, Yale School of Public Health, Yale University, New Haven, CT, 06510, USA

TANDEM REPEAT EXPANSIONS MODIFY AND PREDICT SCHIZOPHRENIA IN 22Q11.2DS

Muyang Cheng^{1,2}, Yue Yin², Worrawat Engchuan^{2,3}, Tracy Heung^{4,5}, Bernice E. Morrow^{6,7}, International 22q11.2 Brain Behavior Consortium (IBBC), Anne S. Bassett^{4,5,8,9}, Ryan K.C. Yuen^{1,2,3,9}

¹ Department of Molecular Genetics, University of Toronto

² Genetics and Genome Biology, The Hospital for Sick Children

³ The Centre for Applied Genomics, The Hospital for Sick Children

⁴ Clinical Genetics Research Program, Centre for Addiction and Mental Health

⁵ The Dalglish Family 22q Clinic for Adults with 22q11.2 Deletion Syndrome, Toronto General Hospital, University Health Network

⁶ Department of Genetics, Albert Einstein College of Medicine

⁷ Departments of Obstetrics and Gynecology and Pediatrics, Albert Einstein College of Medicine

⁸ Department of Psychiatry, University of Toronto, Toronto General Hospital Research Institute, and Campbell Family Mental Health Research Institute

⁹ These authors contribute equally



LONG-READ SEQUENCING REVEALS EXTENSIVE STRUCTURAL VARIATION ACROSS INDIA

Sofia Banu^{1,2}, Rhea Jana¹, Sreelekshmi MS¹, A Sreenivas¹, Rakesh Bhagat¹, Suman Govindaraju³, Deepak Jena⁴, Sudeshna Datta⁴, Mamuni Swain⁴, Lalengkimi Ralte⁵, Nishat Ashrafi⁶, Genomelndia Consortium, Mohammed Faruq⁷, Ganganath Jha⁶, Senthil Kumar Nachimuthu⁵, Sunil Raghav⁴, Sasikala Mitnala³, Karthik Bharadwaj Tallapaka^{1,2}, Divya Tej Sowpati^{1,2,*}

1. CSIR Centre for Cellular and Molecular Biology, Hyderabad
2. Academy of Scientific and Innovative Research, Ghaziabad
3. Asian Healthcare Foundation, AIG Hospitals, Hyderabad
4. BRIC Institute of Life Sciences, Bhubaneswar
5. Department of Biotechnology, Mizoram University, Aizawl
6. Vinoba Bhave University, Hazaribagh
7. CSIR Institute of Genomics and Integrative Biology, New Delhi

A MULTI-ANCESTRY EVALUATION OF POLYGENIC SCORES ACROSS GLOBAL POPULATIONS

Linda Repetto¹, Brooke Wolford², Elia Tiso¹, Veronica Tozzo³, Kristina Zguro⁴, Masahiro Kanai⁵, Ying Wang⁶, Pedro M. De Macedo Gomes⁷, Muhammad Khir A. Kohilan⁸, Radja Badji⁸, Kuang Lin⁹, Kristi Läll¹, Shuifeng Huang¹⁰, Benjamin Wingfield¹¹, Max Tamlander¹², Aoife McMahon¹¹, Yue Ji¹¹, Sarah Finer¹³, Zhengming Chen⁹, Kristian Hveem¹⁴, Yukinori Okada¹⁵, Bogdan Pasaniuc³, Robin G. Walters, Derrick Bennett⁹, David A. Van Heel¹³, Ren-Hua Chung¹⁶, Henrike O. Heyne¹⁷, Alicia Martin¹⁸, Nina Mars⁴, Andrea Ganna⁴, Samuli Ripatti⁴, Stavroula Kanoni¹³

1. University of Tartu, Tartu, Estonia,
2. Norwegian University of Science and Technology, Trondheim, Norway,
3. David Geffen School of Medicine at UCLA, Los Angeles, CA, USA
4. University of Helsinki, Helsinki, Finland,
5. Massachusetts General Hospital & Broad Institute of MIT and Harvard, Boston, MA, USA,
6. Massachusetts General Hospital, Boston, MA, USA,
7. University of Potsdam, Potsdam, Germany,
8. Qatar Precision Health Institute, Ar-Rayyan, Qatar
9. University of Oxford, Oxford, United Kingdom,
10. Institute of Population Health Sciences, National Health Research Institutes, Zhunan, Taiwan,
11. European Bioinformatics Institute, Hinxton, United Kingdom,
12. Institute for Molecular Medicine Finland (FIMM), HiLIFE, University of Helsinki, Helsinki, Finland,
13. Queen Mary University of London, London, United Kingdom,
14. NTNU, Trondheim, Norway,
15. Osaka University, Osaka, Japan,
16. National Health Research Institutes, Zhunan, Taiwan,
17. Hasso Plattner Institute, Potsdam, Germany,
18. Massachusetts General Hospital, Medford, MA, USA

DECIPHERING COMPLETE ARCHAIC INTROGRESSION SEQUENCES IN MODERN HUMAN GENOMES

Mingyu Suo, Aoyue Bi, Quanyu Chen, Dan Zhang, Dongya Wu, Guojie Zhang
 Zhejiang University School of Medicine, Hangzhou, China





GRAPH TRANSFORMER FOR ANCIENT LOCAL ANCESTRY INFERENCE

Cole Shanks¹, David Bonet¹, Marçal Comajoan Cara², Alexander Ioannidis¹

¹ Department of Biomolecular Engineering, University of California, Santa Cruz,

² Department of Computer Science and Electrical Engineering, University of California, Berkeley

THE GENETIC ARCHITECTURE OF THE HELLENES

Nicholas Katsanis on behalf of the GeneSIs study

Galatea Bio, IMBB-FoRTH

MEG3-miRNA REGULATORY AXIS MODULATES RESPONSE TO bDMARDs IN PSORIATIC ARTHRITIS

Giada De Benedittis¹, Chiara Morgante¹, Andrea Latini², Arianna D'Antonio³, Paola Conigliaro³, Maria Sole Chimenti³, Cinzia Ciccacci², Paola Borgiani¹, Giuseppe Novelli¹

¹ Department of Biomedicine and Prevention, Section of Genetics, University of Rome "Tor Vergata", 00133 Rome, Italy;

² UniCamillus-Saint Camillus International University of Health Sciences, 00131 Rome, Italy;

³ Rheumatology, Allergology and Clinical Immunology, Department of System Medicine, University of Rome "Tor Vergata", 00133 Rome, Italy.

ONCOSORF: PREDICTING ONCOGENIC SORFS IN NON-CODING RNAs

Fabiana Rodrigues de Goes^{1*}, Makanaka Mazheke^{1,4}, Aparajita Karmakar¹, Amanda Piveta Schnepfer³, Nayane de Souza², Robson Francisco Carvalho³, Mark Basham¹, and Alexandre Rossi Paschoal^{1,2*}

¹ Rosalind Franklin Institute, Didcot, OX11 0QS, UK

² Departamento Acadêmico de Computação (DACOM), Universidade Tecnológica Federal do Paraná (UTFPR), Campus Cornélio Procopio, 86300-000, Paraná, Brazil

³ Sao Paulo State University, UNESP, Brazil, Botucatu, Brazil

⁴ University College London (UCL), London, UK

* {fabiana.goes,alexandre.paschoal}@rfi.ac.uk

NOVEL STAGE-SPECIFIC MICRORNA PANEL FOR THE SALIVA-BASED DETECTION OF ORAL SQUAMOUS CELL CARCINOMA AND PREMALIGNANT DYSPLASIA

Iphigenia Gintoni^{1,2,3}, Stavros Vassiliou³, George Chrousos², Christos Yapijakis^{1,2,3}

¹ Unit of Orofacial Genetics, 1st Department of Pediatrics, National Kapodistrian University of Athens, "Aghia Sophia" Children's Hospital, Athens, Greece

² University Research Institute for the Study of Genetic and Malignant Disorders in Childhood, Choremion Laboratory, "Aghia Sophia" Children's Hospital, Athens, Greece

³ Department of Oral and Maxillofacial Surgery, National Kapodistrian University of Athens, "Attikon" Hospital, Athens, Greece



GENERATIVE DESIGN OF CELL TYPE-SPECIFIC RNA SPLICING ELEMENTS FOR PROGRAMMABLE GENE REGULATION

Xi Dawn Chen^{1,2,3†}, Maile Jim^{1,2,3†}, Mounica Vallurupalli^{1,4†}, Kai Cao^{1†}, Andrea Navarro Torres¹, Jing Wesley Leong¹, Yifan Zhang¹, David Wollensak¹, Qiyu Gong¹, Jing Sun¹, Mehdi Borji¹, Gili Schor¹, Sofia Mrowka¹, Margaret Hu¹, Anisha Laumas¹, Jennifer Roth¹, Todd Golub^{1,4}, Fei Chen^{1,2*}

¹ Broad Institute of MIT and Harvard, Cambridge, MA 02142, USA

² Department of Stem Cell and Regenerative Biology, Harvard University, Cambridge, MA 02138, USA

³ Systems, Synthetic and Quantitative Biology Ph.D. Program, Harvard University, Cambridge, MA 02138, USA

⁴ Dana-Farber Cancer Institute, Boston, MA, 02215, USA

SPATIAL ISOFORMS AT SUB-MICROMETER RESOLUTION SHOW NOVEL VARIABILITY PATTERNS.

Lieke Michielsen¹, Andrey D. Prjibelski², Careen Foord¹, Wen Hu¹, Julien Jarroux¹, Justine Hsu¹, Alexandru I. Tomescu², Iman Hajirasouliha¹, Hagen U. Tilgner¹

¹ Weill Cornell Medicine,

² University of Helsinki





e-Posters



EP001**UNIQUE GENETIC SIGNATURES IN HIV-1 SUBTYPE A1 AND A1D RECOMBINANT ENVELOPE GLYCOPROTEIN DISTINGUISH CONTEMPORARY TRANSMITTED /FOUNDER VIRUSES FROM HISTORICAL STRAINS IN EAST AFRICA**

Frank Kato^{1,2}, Anne Kapaata¹, Ronald Galiwango^{3,4}, Angella Nakyanzi⁵, Christian Ndekezi^{1,2}, Fortunate Natwijuka^{1,2}, Denis Omara^{1,2}, Andrew Ekii Obuku¹, Brian Foley⁶, Pontiano Kaleebu^{1,2}, Eunice Nduati⁷, Sheila Nina Balinda^{1,2}.

1. MRC/UVRI & LSHTM Uganda Research Unit, Department of Viral Pathogens, Entebbe, Uganda.
2. Department of Immunology and Molecular Biology, College of Health Sciences, Makerere University, Kampala, Uganda
3. The African Centre of Excellence in Bioinformatics and Data Intensive Sciences, Kampala, Uganda
4. The Infectious Diseases Institute, Makerere University, Kampala, Uganda
5. Uganda Virus Research Institute, Entebbe, Uganda
6. Theoretical Biology and Biophysics, Los Alamos National Laboratory, Los Alamos, NM, United States
7. KEMRI Wellcome Trust Research Programme, Kilifi, Kenya

EP002**AWARENESS AND PERCEPTIONS OF PREMARITAL GENETIC SCREENING AMONG INDIAN YOUTH**

Ss Katpattil

Mother Hospital

EP003**AI-POWERED PANGEN-LM REVEALS ANCESTRY-SPECIFIC VARIANT IN LMIC GENOMES**

Alaa Ramadan

Faculty of Medicine, South Valley University, Qena, Egypt

EP004**CLINICIAN'S KNOWLEDGE, ATTITUDES, AND PERCEPTIONS OF GENETIC TESTING IN INDIA**

Azziz Azbin

Al iqbal hospital

EP005**GENOMIC ANALYSIS OF HYPOXIA RESPONSE IN HUMAN TOF AND NAKED MOLE RATS**

David Oduola, Sarah Adeyemi, Judah Jegede

Department of Biochemistry and Molecular Biology, Obafemi Awolowo University, Ile-Ife, Nigeria;

Department of Bio-informatics, Morgan State University, Maryland, USA ; Department of Medicine and Surgery, Obafemi Awolowo University, Ile-Ife, Nigeria





EP006

FOUNDER EFFECT MAINTAINS RARE GENETIC DISORDERS VARIANTS IN PAKISTANI PUNJABI POPULATION

Iqra Tabassum¹, Muhammad Shafique¹ and Muhammad Shoaib Akhtar^{2,3}

- ¹. National Center of Excellence in Molecular Biology, University of the Punjab, 87 West canal road Lahore
- ². Department of Cell Biology and Human Anatomy, School of Medicine, University of California Davis, Davis, CA 95616, USA
- ³. Department of Integrated Biosciences, Graduate School of Frontier Sciences, The University of Tokyo, Kashiwa, Japan

EP007

A SPTAN1 P.ARG19TRP VARIANT IN A KOREAN FAMILY WITH AUTOSOMAL DOMINANT PURE HEREDITARY SPASTIC PARAPLEGIA

Cha Gon Lee

Department of Pediatrics, Ewha Womans University Seoul Hospital, Ewha Womans University College of Medicine, Seoul, Republic of Korea

EP008

EVALUATING MACHINE-LEARNING DERIVED POLYGENIC SCORES OF PSYCHOLOGICAL RESILIENCE: ACROSS PSYCHIATRIC AND BEHAVIORAL PHENOTYPES IN THE TAIWANESE POPULATION

Meng-Che Tsai^{1,2}, Sakshi Saraswat¹, Mahmood Alhlftee¹

- ¹. Department of Pediatrics, National Cheng Kung University Hospital, College of Medicine, National Cheng Kung University, Tainan, Taiwan
- ². Department of Genomic Medicine, National Cheng Kung University Hospital, College of Medicine, National Cheng Kung University, Tainan, Taiwan

EP009

IN-SILICO IDENTIFICATION OF GENETIC MARKERS LINKED TO ADVERSE REACTIONS FROM ANTI-TUBERCULOSIS THERAPY

Kamal Kishor

IIPS

EP010

THE GENETIC BASIS OF TUBERCULOSIS IN SOUTH AFRICA: A FOCUS ON THE TYK2 GENE

Marlo Möller^(1,2,3,4), Desiree C.Petersen^(1,3), Caitlin Uren^(1,2,3,4), and Vuyo Mavumengwana⁽¹⁾

- ¹. South African Medical Research Council (SAMRC) Centre for Tuberculosis Research (CTR), Division of Molecular Biology and Human Genetics (MBHG), Department of Biomedical Sciences, Faculty of Medicine and Health Sciences, Stellenbosch University, Cape Town
- ². Centre for Bioinformatics and Computational Biology, Stellenbosch University, Cape Town
- ³. Genomics for Health in Africa (GHA), Africa-Europe Cluster of Research Excellence (CoRE)
- ⁴. National Institute for Theoretical and Computational Sciences (NITheCS) South Africa



EP011**LEPTIN RECEPTOR POLYMORPHISM LINKED TO LOWER GUT BIFIDOBACTERIUM IN INFANTS**

Kotryna Simonyte Sjödin, Jonas Hodneland Sundfjord, Stefan Johansson, Torbjörn Lind

Department of Clinical Sciences, Pediatrics, Umeå University, Sweden and Mohn Center for Diabetes Precision Medicine, Department of Clinical Science, University of Bergen, Bergen, Norway

EP012**LEVERAGING ENVIRONMENTAL GENOMICS TO STRENGTHEN INFECTIOUS DISEASE SURVEILLANCE AND PUBLIC HEALTH DECISION-MAKING IN FRAGILE HEALTH SYSTEMS: INSIGHTS FROM SOMALIA**

Dr Dini mohamud, Dr abdullahi abdi ahmed

Somalia Health & Environment Alliance (SHEA), Mogadishu, Somalia, King fasail hospital Rwanda

EP013**PERSONALIZED COLORECTAL CANCER THERAPY USING DROSOPHILA MELANOGASTER**

Clement Ogar Elam, Henry Demian Oyoyo

College of Medicine, University of Ibadan, Nigeria; College Research and Innovation Hub, Ibadan; Missions: Brain Ibadan

EP014**ZI-MENDELIAN: A BIOINFORMATICS WORKFLOW FOR THE ANALYSIS OF MENDELIAN DISORDERS**

(1,2) Nyasha Lorraine Mapira ; (1) Patience Kuona; (2) Zedias Chikwambi; (2) Collen Masimirembwa

¹. *University of Zimbabwe, Faculty of Medicine and Health Sciences*

². *African Institute of Biomedical Science and Technology*

EP015**SYSTEMS BIOLOGY APPROACHES FOR THE IDENTIFICATION OF NOVEL BIOMARKERS IN DIABETIC FOOT ULCER MANAGEMENT**

Manoj Kumar Manish Singh, Rubi Solanki, Ravins Dohare

Jawaharlal Nehru University, New Delhi India

Centre for Interdisciplinary Research in Basic Sciences Jamia Millia Islamia, New Delhi India

EP016**INTEGRATED GENOME SEQUENCING AND LINKAGE ANALYSIS FOR DISEASE GENE DISCOVERY IN DUCHENNE MUSCULAR DYSTROPHY**

Rohit Rajput, Umesh Gangadhar, Sagar Verma

Jawaharlal Nehru University, New Delhi India

Department of Biochemistry, F H Medical College and Hospital, India





EP017

MACHINE LEARNING BASED GENOMIC AND MICROBIOME INTEGRATION FOR GASTRIC CANCER

Rohit Rajput, Umesh Gangadhar Sagar Verma

Department of Biochemistry, F H Medical College and Hospital, India

EP018

INTEGRATIVE MULTI-OMICS PREDICTS IMMUNOTHERAPY RESPONSE IN NON-SMALL CELL LUNG CANCER

Mahdis Monadi

School of Pharmacy, Charles University, Czech Republic

EP019

MACHINE LEARNING INTEGRATION OF TUMOR GENOMICS PREDICTS CHEMOTHERAPY RESPONSE IN TRIPLE-NEGATIVE BREAST CANCER

Shirin Katebi

Khosro Medisa Teb Company

EP020

DIAGNOSED CASES OF SARS-COV-2 PANDEMIC IN PARTS OF NIGERIA BETWEEN MARCH–SEPTEMBER 2020, IRRUA, NIGERIA

Adomeh, Donatus I.^{1,2}, Odia Ikpomnwosa.¹, Aire, Chris O.¹, Omiunu, Racheal¹, Ehikhametalor, Solomen¹, Ebo Benevolence O.¹, Okonofua, Naregose G.¹, Giwa, Rosemary E.¹, Aiyepada, John O.¹, Agbukor, Jacqueline.¹, Ebhodaghe, Paulson O.¹, Yerumoh, Edna O.¹, Airende, Michael¹, Gold, Blessing¹, Bright-Edoh, Egbedufa P¹, Akhilomen, Patience¹, Esumeh, Rita O.¹, Giwa, Olabode¹, Ayodele, Oluwatosin¹, Edeko, Henry E.¹, King James Ebhomhene¹, Umoera, Winifred E.¹ Njigha, Precious O.¹, Subor, Ohunwu, C.¹, Sule, Pius Omakoji¹, Orukpe, Anthony¹, Eremah, Agnes¹, Akhabue, Ose F.¹, Ebhojie, Rahab¹, Ilonah, Emmanuel¹, Isekialuona Grace¹, Asogun, Chris¹, Omoigiade-Udefiagbon A.¹, Imoebe, Frank¹, Isiramen, Ehinomen¹, Oboh, Collins I.¹, Ogbaini-Emovon, Ephraim¹, Omoregbe, Isaac², Tobin, Ekaete², William, Wisdom³, Edeawe Osahogie⁴, Okonofua, Martha⁴, Atafo, Rebecca⁴, Oestereich, Lisa⁵, Duraffour, Sophie⁵, Gabriel, Martin⁵ and Pahlmann, Meike⁵

¹ Diagnostics and Research Laboratory, Institute of Viral and Emergent Pathogens Control and Research, Irrua Specialist Teaching Hospital, Edo State, Nigeria.

² Public Health Department, Irrua Specialist Teaching Hospital, Edo State, Nigeria.

³ SOMERS, NCDC, Abuja, Nigeria.

⁴ Isolation Ward, Irrua Specialist Teaching Hospital, Edo State, Nigeria.

⁵ Department of Virology, Bernhard Nocht Institute for Tropical Medicine, Hamburg, Germany.

EP021

NOVEL PATHOGENIC ATP7B GENE VARIANT IN WILSON DISEASE

Javeria Tanveer, Ayisha Aman Ullah

Department of Biotechnology, Jinnah University for Women, Karachi, Pakistan



EP022**EXPLORING SINDHI POPULATION GENETICS: FROM ANCIENT ANCESTRY TO RECENT DIVERGENCE**

Chanchal Devnani^{1*}, Khushboo Gautam¹⁻², Rakesh Rawal²⁻³, Gyaneshwer Chaubey^{1*}

¹. Gyan Lab, Cytogenetics Unit, Department of Zoology, Banaras Hindu University, Varanasi, Uttar Pradesh 221005, India.

². Department of Biochemistry and Forensic Science, Gujarat University, Ahmedabad, India.

³. Department of Medical Biotechnology, Gujarat Biotechnology University, Gandhinagar, India.

EP023**UNCOVERING POPULATION-SPECIFIC TLR4 VARIANTS AND CYTOKINE PROFILES: A REGIONAL GENETIC INSIGHT INTO OSTEOPOROSIS RISK IN PAKISTANI WOMEN**

Rozeena Baig, Rifat Ahmed, Rehana Rehman

University of Karachi, Aga Khan University

EP024**THE IMPACT OF VACCINATION IN THE PRESENCE OF HOST GENETIC RISK FACTORS LINKED TO COVID-19 SEVERITY**

Yentl Lamprecht¹, Gert van Zyl^{2,3}, André G. Loxton¹, Gian van der Spuy^{1,4}, Marlo Möller^{1,4,5,6}, Desiree C. Petersen^{1,4,5,6}

¹. South African Medical Research Council (SAMRC) Centre for Tuberculosis Research (CTR), Division of Molecular Biology and Human Genetics (MBHG), Department of Biomedical Sciences, Faculty of Medicine and Health Sciences, Stellenbosch University, Cape Town, South Africa

². National Health Laboratory Services (NHLS), Cape Town, South Africa

³. Division of Medical Virology, Stellenbosch University, Cape Town, South Africa

⁴. Centre for Bioinformatics and Computational Biology (CBCB), Stellenbosch University, Cape Town, South Africa

⁵. Genomics for Health in Africa (GHA), Africa-Europe Cluster of Research Excellence (CoRE), Cape Town, South Africa

⁶. National Institute for Theoretical and Computational Sciences (NITheCS), Cape Town, South Africa

EP025**GENOMIC CORRELATES OF TMB-HIGH STATUS IN SOLID TUMORS ASSESSED BY TSO500**

Jianli Dong

The University of Texas Medical Branch, Galveston, Texas, USA





EP026

COMPUTATIONAL PREDICTION AND VALIDATION OF SHIKONIN, MYRICETIN, AND KAEMPFEROL AS MULTI-TARGET INHIBITORS OF PARP1, BRCA1, AND RAD51 IN TRIPPLE-NEGATIVE BREAST CANCER

Abdulrahman Olamilekan Raji; Aisha Eniola Olayiwola; Habeeb Akorede Lawal; Amos Abigail Abosedo; Adebayo Temitope Aderonke; Ede, Dorathy Amuche; Nwodo Amaka Marvellous; Maduka Amuche Stella; Golagha Regina Oghenevwede; Alake, Aanuoluwapo Olamide; Sunmola Adebola Latifat; Okoroma Bright; Agbi Mayowa

1. *Departments of Biochemistry, Kwara State University, Maleta, Nigeria.*
2. *Eagle Research Laboratory, Ladoke Akintola University of Technology, Neuroscience Group, Ogbomosho, Oyo State, Nigeria*
3. *Division of Vaccine and Pharmacotherapies Design and Development, Helix Biogen Institute, Ogbomosho, Oyo state, Nigeria.*
4. *Department of Molecular Genetics and Infectious Diseases, Abubakar Tafawa Balewa University Teaching Hospital, Bauchi, Nigeria.*
5. *Department of Microbiology, Federal University of Agriculture, Abeokuta. P. M. B. 2240, Abeokuta, Ogun State, Alabata Road, Abeokuta, Ogun State, 110111, Nigeria*
6. *Department of Microbiology (Industrial Microbiology and Biotechnology) University of Ibadan, Nigeria*
7. *Department of Science Laboratory Technology, Redeemer's College of Technology and Management Mowe, Ogun State, Nigeria*
8. *Department of Applied Biology and Biotechnology, Enugu State University of Science and Technology Agbani, Enugu State, Nigeria*
9. *Department of science education, Nnamdi Azikiwe University Awka, Anambra state Nigeria*
10. *Department of Microbiology, Federal University of Agriculture, Abeokuta, Nigeria*
11. *Federal Medical Centre Abeokuta Ogun State, Nigeria*
12. *Covenant University, Ota, Ogun State, Nigeria*

EP027

GENETIC LIABILITY TO PSYCHIATRIC DISORDERS, ADDICTION & MENARCHE: A TWB MR STUDY

Jia Fong Tsai¹ Meng-Che Tsai^{2,3}

1. *School of Medicine, College of Medicine, National Cheng Kung University, Tainan, Taiwan*
2. *Department of Pediatrics, National Cheng Kung University Hospital, College of Medicine, National Cheng Kung University, Tainan, Taiwan*
3. *Department of Medical Humanities and Social Medicine, College of Medicine, National, Cheng Kung University, Tainan, Taiwan*

EP028

CRISPR/CAS9 KNOCKOUT OF MPV17 GENE: INVESTIGATING MITOCHONDRIAL DYSFUNCTION AND CELLULAR HOMEOSTASIS

Tendai Makwikwi, Maryke Schoonen, Francois van der Westhuizen

Biochemistry & Biomedical and Molecular Metabolism Research (BioMMet), North-West University, Potchefstroom, South Africa



EP029**PRIME EDITING AS A POTENTIAL THERAPEUTIC STRATEGY FOR β -THALASSEMIA**

Wai Teng Cheng¹, Zilfalil Bin Alwi^{3,5}, Sek Chuen Chow¹, Azlan Husin^{2,5}, Muhammad Farid Johan⁴, Ashish Dutt¹, Qasim Ayub^{1,6}

- School of Science, Monash University Malaysia, Bandar Sunway 47500, Selangor Darul Ehsan, Malaysia.*
- Department of Internal Medicine, School of Medical Sciences, Universiti Sains Malaysia, Kubang Kerian 16150, Kelantan, Malaysia.*
- Human Genome Centre, School of Medical Sciences, Universiti Sains Malaysia, Kubang Kerian 16150, Kelantan, Malaysia.*
- Department of Haematology, School of Medical Sciences, Universiti Sains Malaysia, Kubang Kerian 16150, Kelantan, Malaysia.*
- Hospital Pakar Universiti Sains Malaysia, Kubang Kerian, Kelantan, 16150, Malaysia.*
- Monash University Malaysia Genomics Platform, School of Science, Monash University, Bandar Sunway 47500, Selangor Darul Ehsan, Malaysia.*

EP030**A REFRACTORY LIPID STORM: VUS IN APOC2**

Rubiano CJ^(1,2), Forero NE^(3,4)

- BioGenEtica Research Group, faculty of Medicine, Universidad Militar Nueva Granada, Bogot , Colombia*
 - GENOSIG Genetics and Genomics Interest Group, faculty of Medicine, Universidad Militar Nueva Granada, Bogot , Colombia*
 - Hospital Militar Central, Bogot , Colombia*
 - CLINALTEC Research and Diagnostic Center, Ibagué, Colombia.*
- Contact for the abstract: est.cristianj.rubi1@unimilitar.edu.co

EP031**TRANSCRIPTOMIC RESPONSES TO AZITHROMYCIN AND TEMOZOLOMIDE IN U87 CELLS**

Siti Nazihahasma Hassan^{a,b} and Farizan Ahmad^{a,b*}

- Department of Neurosciences, School of Medical Sciences, Universiti Sains Malaysia, 16150 Kubang Kerian, Kelantan, Malaysia.*
- Hospital Pakar Universiti Sains Malaysia, 16150 Kubang Kerian, Kelantan, Malaysia.*

EP032**ENABLING POPULATION-SCALE GENOMICS IN ITALY: THE NATIONAL FACILITY FOR GENOMICS**

N. Alfano¹, F. Simeoni¹, L. Lamparelli¹, A. Pirola¹, D. Carrillo¹, E. Ricciardelli¹, J. Cibella¹, M. De Cesare¹, P. Ferrari¹, G. Sabbatinelli¹, R. Piccioni¹, E. Ferracci¹, S. Esposito¹, C. Conca Dioguardi¹, F. Giannese¹, L. Rotta¹, C. Peano¹

- Human Technopole, Via Rita Levi Montalcini 1, 20157, Milano*





EP033

UTILISING NEXT-GENERATION SEQUENCING TO IDENTIFY THE GENETIC CAUSES OF FETAL CONGENITAL ABNORMALITIES IN A SOUTH AFRICAN RARE-DISEASE COHORT

Jade Ramini, Shahida Moosa

- 1. Division of Molecular Biology and Human Genetics, Stellenbosch University, Cape Town, South Africa*
- 2. Medical Genetics, Tygerberg Hospital, Cape Town, South Africa*
- 3. SAMRC Centre for Tuberculosis Research, Faculty of Medicine and Health Sciences, Stellenbosch University, Cape Town, South Africa*
- 4. Genomics for Health in Africa (GHA), Africa-Europe Cluster of Research Excellence (CoRE), Cape Town, South Africa*

EP034

INTEGRATING NEXT-GENERATION PHENOTYPING INTO THE UDP'S DIAGNOSTIC PIPELINE

Montana Robinson, Prof Shahida Moosa

- 1. Division of Molecular Biology and Human Genetics, Stellenbosch University, Cape Town, South Africa*
- 2. Medical Genetics, Tygerberg Hospital, Cape Town, South Africa*
- 3. SAMRC Centre for Tuberculosis Research, Faculty of Medicine and Health Sciences, Stellenbosch University, Cape Town, South Africa*
- 4. Genomics for Health in Africa (GHA), Africa-Europe Cluster of Research Excellence (CoRE), Cape Town, South Africa*

EP035

DNA-INDEPENDENT FLUORESCENCE IN IDR-DRIVEN CONDENSATES

Aldardasawi, Mas Ra'ed; Costantino, Michele (2025),
BBMB Department, Whitman College

EP036

A COMPREHENSIVE REPORT OF ALLELE AND GENOTYPE FREQUENCIES OF 313 BREAST CANCER ASSOCIATED SNPS IN THE IRANIAN BEH COHORT: A FOUNDATIONAL STEP TOWARD POLYGENIC RISK SCORE CALIBRATION

Farima Farsi¹, Nafiseh Tavasoli², Atefeh Amrollahi bioky², Mohammad Bidkhori⁵, Danoosh Esmaeili³, Noushin Fahimfar⁵, Mandana Hasanazad^{4*}

- 1. Obesity and Eating Habits Research Center, Endocrinology and Metabolism Clinical Sciences Research Institute, Tehran University of Medical Sciences, Tehran, Iran.*
- 2. Endocrinology and Metabolism Research Center, Endocrinology and Metabolism Clinical Sciences Institute, Tehran University of Medical Sciences, Tehran, Iran.*
- 3. Medical Genomic Research Center, TeMS.C., Islamic Azad University, Tehran, Iran*
- 4. Personalized Medicine Research Center, Endocrinology and Metabolism Clinical Sciences Research Institute, Tehran University of Medical Sciences, Tehran, Iran.*
- 5. Osteoporosis Research Center, Endocrinology and Metabolism Clinical Sciences Institute, Tehran University of Medical Sciences, Tehran, Iran.*



EP037

UNRAVELLING TOX3 AND FGF ASSOCIATIONS IN PATIENTS WITH NONSYNDROMIC CLEFT LIP AND/OR PALATE: A MALAY COHORT STUDY IN KELANTAN, MALAYSIA

Nurul Syazana Mohamad Shah¹, Noor Areefa Ameerah Mohd Ma'amor¹, Sarina Sulong², Nazia Abdul Majid³, Wan Azman Wan Sulaiman¹

- 1. Reconstructive Sciences Unit, School of Medical Sciences, Health Campus, Universiti Sains Malaysia, 16150 Kubang Kerian, Kelantan, Malaysia*
- 2. Human Genome Center, School of Medical Sciences, Health Campus, Universiti Sains Malaysia, 16150 Kubang Kerian, Kelantan, Malaysia*
- 3. Institute of Biological Sciences, Faculty of Sciences, Universiti Malaya, 50603 Kuala Lumpur, Malaysia*

EP038

ZFX3 DELETION DETECTED BY WES IN A CHILD WITH DEVELOPMENTAL DELAY: A CASE REPORT

Nurul Aain Ahmad Fauzi¹, Mohd Ridzuan Hamid¹, Aziati Azwari Annuar¹, Rowani Mohd Rawi², Wan Norizati Wan Mohamad Zamri³, Nur Fatin Syahirah Rasudin¹, Nurul Alia Mohd Nawi¹, Ninie Nadia Zulkipi¹, Sarina Sulong¹

- 1. Human Genome Centre, School of Medical Sciences, Universiti Sains Malaysia Health Campus, Kelantan, Malaysia*
- 2. Department of Paediatrics, Specialist Hospital Universiti Sains Malaysia, Kelantan, Malaysia*
- 3. Genetic Laboratory, Pathology Department, Hospital Tunku Azizah, Kuala Lumpur, Malaysia*

EP039

GENOMIC AND BIOINFORMATICS APPROACHES TO ELUCIDATE THE MOLECULAR BASIS OF MARFAN SYNDROME IN THE TUNISIAN POPULATION

Imen ABDALLAH^{1,2}, Nesrine MEZZI¹, Wajdi ARFA³, Ella LAHMER⁵, Thouraya BEN YOUNES⁶, Ilhem TURKI-YOUSSEF⁶, Ichraf KRAOUA⁶, Fatma OUARDA⁵, Sami BOUCHOUCHA⁵, Mourad JENZRI³, Lilia ROMDHANE^{1,7*}

- 1. Laboratory of Biomedical Genomics and Oncogenetics (LR20PT05), Institut Pasteur de Tunis, Université Tunis El Manar, El Manar I, 2092 Tunis, Tunisia*
- 2. Université de Tunis El Manar, Faculté de Médecine de Tunis, 1007, Tunis, Tunisie*
- 3. Pediatric Orthopedic Department, Kassab Institute, El Manar University, Tunis, Tunisia*
- 4. Children's Hospital Béchir Hamza, Tunis Tunisia Pediatric Orthopedic Surgery Department, Tunisia*
- 5. Pediatric Cardiology Department, La Rabta Hospital, Tunis, Tunisie*
- 6. Pediatric Neurology Department. Mongi Ben Hmida National Institute of Neurology. Tunis. Tunisie*
- 7. Department of Biology, Faculty of Science of Bizerte, Université Carthage, 7021 Jarzouna*

EP040

CONTEXT-DEPENDENT M⁶A MODIFICATION CONTROL OF MRNA FATE IN INVASIVE BREAST CANCER

Azime Akçaöz-Alasar^{1,2,3}, Gökalp Ero ul¹, Beyzanur Albayrak¹, Claudio R. Alarc n^{2,3}, Bünyamin Akgül^{1*}

- 1. Department of Molecular Biology and Genetics, Izmir Institute of Technology, Urla, Izmir, Türkiye*
- 2. Department of Pharmacology, Yale University School of Medicine, New Haven, CT 06520, USA*
- 3. Yale Cancer Biology Institute, Yale University, West Haven, CT 06516, USA.*





EP041

GENETIC-INFORMED MACHINE LEARNING FOR IN SILICO PHARMACOKINETIC MODELING

Navid kaboudi

Faculty of pharmacy, Tabriz university of medical sciences

EP042

COMPUTATIONAL GENOMIC PROFILING OF SLC46A3 REVEALS EPIGENETIC AND IMMUNE-ASSOCIATED PATTERNS ACROSS HUMAN CANCERS

Auwal Ibrahim Tanko, Imran Kazmi

Hamad Bin Khalifa University; King Abdulaziz University

EP043

PHARMACOGENETICS LANDSCAPE IN IRANIANS: FREQUENCY OF 608 VARIANTS IN 3000 PEOPLE

Negar Sarhangi¹, Negar Niknam^{2,3}, Farshad Sharifi⁴, Shekoufeh Nikfar¹, Bagher Larijani⁵, Mandana Hasanzad^{1,6}

- ¹ Personalized Medicine Research Center, Endocrinology and Metabolism Clinical Sciences Institute, Tehran University of Medical Sciences, Tehran, Iran
- ² Department of Pharmacoeconomics and Pharmaceutical Administration, Faculty of Pharmacy, Tehran University of Medical Sciences, Tehran, Iran
- ³ LifeandMe, Inc., Tehran, Iran
- ⁴ Elderly Health Research Center, Endocrinology and Metabolism Population Sciences Institute, Tehran University of Medical Sciences, Tehran, Iran
- ⁵ Endocrinology and Metabolism Research Center, Endocrinology and Metabolism Clinical Sciences Institute, Tehran University of Medical Sciences, Tehran, Iran
- ⁶ Medical Genomic Research Center, TeMS.C., Islamic Azad University, Tehran, Iran

EP044

HOST TRANSCRIPTOMIC SIGNATURES UNDERLYING THE PROGRESSION FROM LATENT TO ACTIVE TUBERCULOSIS

Youssra Boumait¹, and Rajaa Chahboune¹

- ¹ Biology Molecular Unit, Life and Health Sciences Laboratory, Faculty of Medicine and Pharmacy, Abdelmalek Essaâdi University, Tangier

EP045

SINGLE LARGE-SCALE MTDNA DELETION SYNDROME DIAGNOSED BY WES: A CASE REPORT

^{1,4}Ruzanaz Syafira Ruzman Azlee, ^{2,4}Fashihah Sherina Abdul Hadi Sabri, ^{2,4}Noor Azlin Azraini Che Soh, ^{3,4}Rowani Mohd Rawi, ^{3,4}Muhammad Farisham Mohd Khanafiah, ^{1,4}Mohd Ridzuan Hamid

- ¹ Human Genome Centre, School of Medical Sciences, Universiti Sains Malaysia, 16150, Kubang Kerian, Kelantan, Malaysia
- ² Department of Chemical Pathology, School of Medical Sciences, Universiti Sains Malaysia, 16150, Kubang Kerian, Kelantan, Malaysia
- ³ Department of Paediatrics, School of Medical Sciences, Universiti Sains Malaysia, 16150, Kubang Kerian, Kelantan, Malaysia
- ⁴ Hospital Pakar Universiti Sains Malaysia (Universiti Sains Malaysia Specialist Hospital), 16150, Kubang Kerian, Kelantan, Malaysia



EP046

ETHICS OF PANGENOMIC DATA SHARING FOR RARE DISEASE RESEARCH IN SOUTH AFRICA

Lexie Timina¹, Jantina De Vries¹, Melissa Nel², Heidi Matisonn¹

- Ethics Lab, University of Cape Town, South Africa*
- Neuroscience Institute, University of Cape Town, South Africa*

EP047

GENOMIC ADAPTATIONS OF MICROORGANISMS TO HEAVY METAL CONTAMINATION

Irimiya Benjamin Haruna^{1*}, Udoh Stephen Nsikak¹, Abdulazeez Hanif², Nuhu Mandi Istifanus², Jamiu Bello Ogarima¹, and Suleiman Yusuf Alhaji¹

- Molecular Genetics & Infectious Diseases Research Laboratory, Abubakar Tafawa Balewa University & Teaching Hospital Bauchi, Nigeria.*
- Abubakar Tafawa Balewa University Bauchi, Nigeria.*

EP048

AUTOMATED FUNCTIONAL CHARACTERIZATION OF VARIANTS IN MICRORNA BINDING SITES

Thodoris Daskalopoulos^{1,2}, Marios Miliotis^{1,2}, Izabella Gogaeva^{1,2}, Artemis G. Hatzigeorgiou^{1,2}

- DIANA-Lab, Department of Computer Science and Biomedical Informatics, University of Thessaly, Greece*
- Hellenic Pasteur Institute*

EP049

COMMUNICATING PHARMACOGENOMIC TEST RESULTS TO PEDIATRIC PATIENTS

Abdullah Al Maruf^{1,3,6,7}, Natalia Morcilla¹, Nuzhat Tabassum¹, Anju Sareen¹, Myles Le¹, Jessica Hartley^{2,6}, Sarah Malik⁷, Mahin Hasan¹, Laurence Katz^{2,3}, Paul Arnold^{4,5}, Chad Bousman^{4,5,8}

- College of Pharmacy, Rady Faculty of Health Sciences, University of Manitoba, Winnipeg, MB, Canada*
- Children's Hospital Research Institute of Manitoba, Winnipeg, MB, Canada*
- Department of Psychiatry, University of Manitoba, Manitoba, MB, Canada*
- The Mathison Centre for Mental Health Research & Education, Hotchkiss Brain Institute, Cumming School of Medicine, University of Calgary, Calgary, AB, Canada*
- Department of Psychiatry, University of Calgary, Calgary, AB, Canada*
- Department of Biochemistry and Medical Genetics, University of Manitoba, Winnipeg, MB, Canada*
- High School Student, Strathcona-Tweedsmuir School, Calgary, AB, Canada*
- Department of Medical Genetics, University of Calgary, Calgary, AB, Canada*

EP050

SOMATIC CLONAL HAEMATOPOIESIS MUTATIONS IN MULTIPLE SYSTEM ATROPHY BRAINS

Ben Thompson, Dominic Horner, Caoimhe Morley, Zane Jaunmuktane, Christos Proukakis
Queen Square Institute of Neurology, University College London, UK





EP051

DDI2 LINKS THE PROTEASOME AND AUTOPHAGY PATHWAYS

Nayerehalsadat Hosseini, Ahmed M. Elshazly, Holly A. Byers, Madison A. Ward, Amy N. Brooks, Janakiram R. Vangala, Senthil K. Radhakrishnan

Department of Pathology, Virginia Commonwealth University, Richmond, VA, USA

EP052

A BRIDGE BETWEEN GENOMICS AND REAL-WORLD CANCER CARE IN LOW-RESOURCE SETTINGS INDIA

Aditya Manna

MAS Clinic & Hospital

EP053

ASSEMBLY OF NON-REFERENCE SEQUENCES FROM WGS ON A BRAZILIAN COHORT

Pedro Sebe¹, Jaqueline Y. T. Wang², Guilherme L. Yamamoto^{2,4,5,6}, Victor H. C. Toledo^{1,2}, Maria Rita Passos Bueno^{1,2}, Yeda A. O. Duarte^{7,8}, Mayana Zatz^{1,2}, Michel Naslavsky^{1,2,3}

- ¹ *Department of Genetics and Evolutionary Biology, Biosciences Institute, University of S o Paulo, S o Paulo, SP, Brazil*
- ² *Human Genome and Stem Cell Research Center, University of S o Paulo, S o Paulo, SP, Brazil*
- ³ *Hospital Israelita Albert Einstein, S o Paulo, SP, Brazil*
- ⁴ *Instituto da Criança, Faculdade de Medicina da Universidade de S o Paulo, S o Paulo, SP, Brazil*
- ⁵ *Orthopedic Research Labs, Boston Children's Hospital and Department of Genetics, Harvard Medical School, Boston, MA, USA*
- ⁶ *Laboratório DASA, S o Paulo, Brazil*
- ⁷ *Medical-Surgical Nursing Department, School of Nursing, University of S o Paulo, S o Paulo, SP, Brazil*
- ⁸ *Epidemiology Department, Public Health School, University of S o Paulo, S o Paulo, SP, Brazil*

EP054

MULTIVARIATE GWAS MAPS SHARED BIOLOGY OF CARDIOVASCULAR COMORBIDITY IN PSORIASIS

Charalabos Antonatos, Yiannis Vasilopoulos

Laboratory of Genetics, Section of Genetics, Cell Biology and Development, Department of Biology, University of Patras

EP055

SHARED GENETIC ARCHITECTURE BETWEEN PSORIASIS AND THE LIPIDOME

Alexandros Papadopoulos, Charalabos Antonatos, Yiannis Vasilopoulos

Laboratory of Genetics, Section of Genetics, Cell Biology and Development, Department of Biology, University of Patras



EP056**FUNCTIONAL ANALYSIS OF THE OLFACTORY SYSTEM IN AN IN VITRO MODEL OF PSORIASIS**

Adam Akritidis¹, Charalabos Antonatos¹, Katerina Grafanaki^{2,3}, Constantinos Stathopoulos², Yiannis Vasilopoulos¹

- Laboratory of Genetics, Section of Genetics, Cell Biology and Development, Department of Biology, University of Patras, 26504 Patras, Greece,*
- Department of Biochemistry, School of Medicine, University of Patras, 26504 Patras, Greece,*
- Department of Dermatology-Venereology, School of Medicine, University of Patras, 26504 Patras, Greece*

EP057**GENETIC AND FUNCTIONAL EVIDENCE FOR THE NEUROPSYCHIATRIC COMORBIDITY IN ECZEMA**

Alexandros Pontikas¹, Eleftheria Georgalaki¹, Charalabos Antonatos¹, Katerina Grafanaki^{2,3}, Marios Mouzouris¹, Constantinos Stathopoulos², Yiannis Vasilopoulos¹

- Laboratory of Genetics, Section of Genetics, Cell Biology and Development, Department of Biology, University of Patras, 26504 Patras, Greece,*
- Department of Biochemistry, School of Medicine, University of Patras, 26504 Patras, Greece,*
- Department of Dermatology-Venereology, School of Medicine, University of Patras, 26504 Patras, Greece*

EP058**PHARMACOGENETIC MAPPING OF IL6 SIGNALING AND TREATMENT RESPONSE IN ECZEMA**

Dimitra Triantafyllidi¹, Vasiliki Tziouvara¹, Alexandros Pontikas¹, Adam Akritidis¹, Aikaterini Zacharopoulou², Aikaterini Tsiogka², Ileana-Afroditi Kleidona², Katerina Grafanaki³, Kleoniki Chaidaki⁴, Olga Kouvarou⁴, Kalliopi Gerogianni⁴, Efterpi Zafiriou⁴, Grigoriou Stamatias², Vasilopoulos Yiannis¹

- Laboratory of Genetics, Section of Genetics, Cell Biology and Development, Department of Biology, University of Patras,*
- Department of Dermatology-Venereology, Faculty of Medicine, Andreas Sygros Hospital, National and Kapodistrian University of Athens,*
- Department of Dermatology-Venereology, School of Medicine, University of Patras, 26504 Patras, Greece*
- Department of Dermatology, University General Hospital Larissa, University of Thessaly, 41334 Larissa, Greece*

EP059**MAPPING THE IMPACT OF IL17 PATHWAY VARIANTS ON TREATMENT RESPONSE IN PSORIASIS**

Ioanna Karantali¹, Nicky Thanopoulou¹, Aikaterini Nikolaidou¹, Adam Akritidis¹, Katerina Grafanaki³, Kleoniki Chaidaki², Olga Kouvarou², Kalliopi Gerogianni², Efterpi Zafiriou², Yiannis Vasilopoulos¹

- Laboratory of Genetics, Section of Genetics, Cell Biology and Development, Department of Biology, University of Patras, 26504 Patras, Greece,*
- Department of Dermatology, University General Hospital Larissa, University of Thessaly, 41334 Larissa, Greece,*
- Department of Dermatology-Venereology, School of Medicine, University of Patras, 26504 Patras, Greece*





EP060

GREPL: GENOMICS OF RECURRENT PREGNANCY LOSS

Yan Eve Tan¹, Madhuri Pulijala¹, Aswini Leela Loganathan², T Valliammai Jayanthi Thirunavuk Arasoo³, Ah Onn Chu⁴, SK Ong⁵, Wei Yee Wee¹, Mukhri Hamdan^{6,7}, Qasim Ayub^{1,2}

- ¹ School of Science, Monash University Malaysia, Bandar Sunway, Selangor, Malaysia.
- ² Monash University Malaysia Genomics Platform, Bandar Sunway, Selangor, Malaysia.
- ³ Clinical School Johor Bahru, Jeffrey Cheah School of Medicine and Health Sciences, Monash University, Johor Bahru, Johor, Malaysia.
- ⁴ Century Specialist Maternity Sdn Bhd, Johor Bahru, Johor, Malaysia.
- ⁵ Khor and Loh Maternity Clinic, Johor Bahru, Johor, Malaysia.
- ⁶ Universiti Malaya, Kuala Lumpur, Malaysia.
- ⁷ Universiti Malaya Medical Centre, Petaling Jaya, Kuala Lumpur, Malaysia

EP061

CASE STUDY: INTRONIC MUTATIONS AS A HIDDEN FACTOR IN ALPURT SYNDROME RISK ASSESSMENT

Diana Basharova, Elena Zholdybaeva
LLP «National Center for Biotechnology»

EP062

IN SILICO DESIGN OF HPV 35 MULTI-EPI TOPE PEPTIDE VACCINE CANDIDATE FOR AFRICAN POPULATION

Maclay. C Muzhuzha¹, Wonderful T. Choga², Reginald. T Mandizvidza¹, Alltalents. T Murahwa³, Exnevia Gomo¹, Justen Manasa¹, Vurayai Ruhanya¹

- ¹ Medical Microbiology Unit, Faculty of Medicine and Health Sciences, University of Zimbabwe, Box A178, Avondale, Harare, Zimbabwe
- ² Botswana Harvard AIDS Institute Partnership, Gaborone Botswana
- ³ Institute for Infectious Disease and Molecular Medicine, Department of Pathology, Division of Medical Virology, Faculty of Health Sciences, University of Cape Town

EP063

GENETIC CHARACTERISTICS OF CHILDREN WITH SUSPECTED MODY: WHOLE-EXOME SEQUENCING RESULTS

Aiganym Toleuzhanova, Elena Zholdybayeva, Aliya Romanova
National Center for Biotechnology, Astana, Kazakhstan

EP064

HEARING IMPAIRMENT GENETICS IN RWANDA REVEALS A POTENTIAL TMC1 FOUNDER EFFECT

Esther Uwibambe, Leon Mutesa, Ambroise Wonkam
University of Rwanda, University of Cape Town



EP065

PRECISION MEDICINE FOR DEAFNESS: GENE DISCOVERY AND THERAPY

Karen B. Avraham¹, Lama Khalaily¹, Inbar Blech¹, Roni Hahn¹, Lara Kamal¹, Katherine Domb¹, Buwei Shao¹, Eyal Marton¹, Alice Mahameed¹, Shahar Taiber¹, Zippora Brownstein¹, Danielle Oz¹, Gal Ofir¹, Shahar Kasirer², Yazeed Zoabi³, Shadi J. Khoury³, Varda Shalev⁴, Asaf Peretz⁵, Tal Patalon⁵, Juan Fernandez-Recio⁶, Xavier de La Cruz⁷, Fabian Glaser⁸, Litao Tao⁹, Ran Elkon¹, Noam Shomron³, David Sprinzak²

- Department of Human Genetics & Computational Medicine, Gray Faculty of Medical and Health Sciences and Sagol School of Neuroscience, Tel Aviv University, Tel Aviv, Israel;*
- School of Neurobiology, Biochemistry & Biophysics, Wise Faculty of Life Sciences, Tel Aviv University, Tel Aviv, Israel;*
- Department of Cell & Developmental Biology, Gray Faculty of Medical and Health Sciences, Tel Aviv University, Tel Aviv, Israel;*
- School of Public Health, Gray Faculty of Medical and Health Sciences, Tel Aviv University, Tel Aviv, Israel;*
- KSM-Kahn-Sagol-Maccabi Research and Innovation, Maccabi Healthcare Services, Tel Aviv, Israel;*
- Instituto de Ciencias de la Vid y del Vino (ICVV), CSIC-UR-Gobierno de La Rioja, 26007 Logroño, Spain;*
- Research Unit in Clinical and Translational Bioinformatics, Vall d'Hebron Institute of Research (VHIR), Universitat Autònoma de Barcelona, Barcelona, Spain & Institut Català per la Recerca i Estudis Avançats (ICREA), Barcelona, Spain;*
- THHI, Technion Human Health Initiative, Israel Institute of Technology, Haifa, Israel;*
- Department of Biomedical Sciences, School of Medicine, Creighton University, Omaha, NE, USA.*

EP066

CONSERVED MIR-141-3P IN SARS-COV-2 DRIVES CYTOSKELETON REMODELING IMMUNE EVASION

Michela Murdocca¹, Gerardo Pepe², Andrea Latini¹, Annalisa Botta¹, Paola Spitalieri¹, Lara Guerrieri¹, Manuela Helmer Citterich², Federica Sangiuolo¹, Giuseppe Novelli¹

- Department of Biomedicine and Prevention, University of Rome, Italy*
- Department of Biology, University of Rome, Italy*

EP067

PHARMACOGENOMICS OF METHADONE-INDUCED SEXUAL DYSFUNCTION IN MALE OPIOID USERS

Shahzad Riahi¹, Muhammad Fazril Mohamad Razif², Loh Huai Seng³, Anne Yee⁴ and Sim Maw Shin^{1,*}

- Department of Pharmaceutical Life Sciences, Faculty of Pharmacy, University Malaya, 50603 Kuala Lumpur, Malaysia*
- Department of Molecular Medicine, Faculty of Medicine, Universiti Malaya, Kuala Lumpur, Malaysia*
- Clinical Academic Unit, Newcastle University Medicine Malaysia, Iskandar Puteri, Johor, Malaysia*
- Jeffrey Cheah School of Medicine & Health Sciences, Monash University Malaysia, Johor Bahru, Johor, Malaysia*





EP068

ALBINISM IN ANGOLA: INTEGRATING CULTURE, GENETICS AND SKIN CANCER PREVENTION

S Ivia Germany Pires Manuel, Nkembi Matilde Miguel Ferraz¹, Maria Madalena Paulo Chimpolo^{1,2}

¹ *Katyavala Bwila University of Medical Faculty,*

² *Agostinho Neto University of Medical Faculty*

EP069

FUNCTIONAL FE2O3/COOX HYBRID NANOCOMPOSITES FOR ANTIBIOTIC DEGRADATION AND EVALUATION OF ANTICANCER EFFECTS

Mohammad S. Alanazi^{1,2*} · Ibrahim O. Alanazi³ · Mohamed S. Elroh² · Zahid Khan¹ · Abdullah Alamri^{1,2} · Jilani Purusottapatnam²

¹ *Department of Biochemistry, College of Science, King Saud University, P.O. Box 2455, 11451 Riyadh,*

² *Genome Research Chair, Department of Biochemistry, College of Science, King Saud University, Riyadh.*

³ *Healthy Aging Research Institute, King Abdulaziz City for Science and Technology (KACST), Riyadh. Saudi Arabia.*

EP070

TIBETAN T2T PANGENOME REVEALS COMPLEX VARIANTS OF HIGH-ALTITUDE ADAPTATION

Yaoxi He, Yaifei Mao, Bing Su

State Key Laboratory of Genetic Evolution and Animal Models, Kunming Institute of Zoology, Chinese Academy of Sciences, Kunming, China; Bio-X Institutes, Shanghai Jiao Tong University, Shanghai, China

EP071

MULTIPLE PRIMARY CANCERS: A RARE CASE STUDY AND LITERATURE REVIEW

Ewa Czukiewska^{1,2}, Tymoteusz Czukiewski³, Radosław Mencfel⁴, Ilona Pasięka⁶, Kinga Kuranty², Agnieszka Kurniar

¹ *Department of Cancer Genetics with Cytogenetic Laboratory, Medical University of Lublin, Poland;*

² *The Center of Oncology of the Lublin Region St. Jana z Dukli; ul. Lublin, Poland;*

³ *Faculty of Medicine; Medical University of Lublin, Poland;*

⁴ *Department of Basic Medical Sciences, The John Paul II Catholic University of Lublin, Poland;*

⁵ *Laboratory of Genomics and Genetics; Faculty of Medicine; The John Paul II Catholic University of Lublin, Poland;*

⁶ *Department of Medical Analytics, 1st Military Clinical Hospital with Polyclinic, Independent Public Healthcare Institution (SPZOZ), Lublin, Poland;*

EP072

NOVEL BIOMARKER OF CHRONIC INFLAMMATION IN ATHEROSCLEROSIS

Svetlana Lukina¹, Natalia Elizova^{1,2}, Mariam Bagheri Ekta², Vasily Sukhorukov¹

¹ *Institute of General Pathology and Pathophysiology, Moscow, Russia*

² *Petrovsky National Research Centre of Surgery, Moscow, Russian Federation*



EP073**TRANSCRIPTIONAL ADAPTATIONS TO KRAS-G12C INHIBITORS IN LUNG CANCER CELL LINES**

Paraskevi Lagkada^{1,2}, Mariachiara Grieco^{2,3,4}, Debora Caprella^{1,2}, Chiara Ambrogio^{1,2,*}, Matteo Cereda^{2,3,4,*}

- ¹ Department of Molecular Biotechnology and Health Sciences, University of Turin, Turin, Italy
- ² Italian Institute for Genomic Medicine, c/o IRCCS, Candiolo, Turin, Italy
- ³ Department of Oncology and Hematology-Oncology, University of Milan, Milan, Italy
- ⁴ IFOM ETS, The AIRC Institute of Molecular Oncology, Milan, Italy

EP074**CRACKING THE GLOBIN CODE IN THALASSAEMIA USING NEXT-GENERATION SEQUENCING**

Ezalia Esa¹, Mohd Khairul Nizam Mohd Khalid², Syahzuwan Hassan³, Faidatul Syazlin Abdul Hamid³, Nor Soleha Mohd Dali³, Wahyuni Sofia Foo Mohammad Salleh³, Mohd Shaiful Jefri Mohd Nor Sham Kunusagaran⁴, Nor Zam Azihan Mohd Hassan⁴, Zulaiha Muda⁵, Hishamshah Mohd Ibrahim⁵, Nur Aisyah Aziz³, Norafiza Mohd Yasin³, Noraesah Mahmud⁶, Ezzanie Suffya Zulkefli³, Hafiza Alauddin⁷, Yuslina Mat Yusoff³.

- ¹ Institute for Medical Research (IMR), National Institutes of Health (NIH), Selangor, Malaysia.
- ² Inborn Errors of Metabolism Unit, Nutrition Metabolic and Cardiovascular Research Centre (NMCRC), IMR, NIH, Selangor, Malaysia.
- ³ Haematology Unit, Cancer Research Centre (CaRC), IMR, NIH, Selangor, Malaysia.
- ⁴ Center for Health Economics Research, Institute for Health Systems Research, NIH, Selangor, Malaysia.
- ⁵ Haematology Unit, Department of Paediatric, Hospital Tunku Azizah, Kuala Lumpur, Malaysia.
- ⁶ Haematology Unit, Department of Pathology, Hospital Kuala Lumpur, Kuala Lumpur, Malaysia.
- ⁷ Department of Pathology, Faculty of Medicine, Universiti Kebangsaan Malaysia, Kuala Lumpur, Malaysia.

EP075**IMPLEMENTING PRECISION ONCOLOGY IN RURAL INDIA: A REAL-WORLD GENOMIC CARE MODEL**

Manna Aditya

MAS Clinic & Hospital

EP076**IL-1B TOLERANCE LNCRNAs IN THP-1 CYBRIDS WITH ATHEROSCLEROSIS MTDNA MUTATIONS**

Vasily Sukhorukov, Daria Borodko

Institute of General Pathology and Pathophysiology, Moscow, Russia





EP077

GENOMIC LANDSCAPE OF SINGLE-STRAND DNA BREAKS IN PLS-PATIENT-DERIVED IPSCS NEURONS

Laverde-Paz, Mayra Juliana^{a,b}; McCartan, Robyn^{a,b}; Margetts, Alex^{a,b}; Ricciardi, Natalie^{a,b}; Maki, Cassidy^{a,b}; Melo-Escobar, Maria Isabel^{a,b}; Wu, Joanne^{c,d}; Benatar, Michael^{c,d}; Cao, Huifen^e; Kapranov, Phillip^e; Tuesta, Luis^{a,b}; Wahlestedt, Claes^{a,b}; Volmar, Claude-Henry^{a,b}; Zeier Zane^{a,b*}

^a Department of Psychiatry and Behavioral Sciences, Sylvester Comprehensive Cancer Center

^b Center for Therapeutic Innovation, University of Miami Miller School of Medicine, Miami, FL 33136

^c Department of Neurology, University of Miami Miller School of Medicine, Miami, FL 33136

^d ALS Center, University of Miami Miller School of Medicine, Miami, FL 33136

^e Institute of Genomics, School of Biomedical Sciences, Huaqiao University, 668 Jimei Road, Xiamen 361021, China

EP078

CURRICULUM REFORM IN MEDICAL EDUCATION IN ANGOLA: INTEGRATING DIGITAL HEALTH AND GENOMICS COMPETENCIES AT THE FACULTY OF MEDICINE OF THE UNIVERSIDADE KATYAVALA BWILA

Nkemi Matilde Miguel Ferraz¹; Maria Madalena Chimpolo^{1,2}

Katyavala Bwila University Faculty of Medicine,

Agostinho Neto University Faculty of Medicine

EP079

EXPLORING LINKAGE DISEQUILIBRIUM IN GWAS USING MULTIPLE PANELS

Georgios A. Manios, Panagiota I. Kontou, Pantelis G. Bagos¹

University of Thessaly

EP080

EFFICACY OF INDIVIDUALIZED ESCITALOPRAM DOSING BASED ON PLASMA LEVEL MONITORING

A. Jeremic, P. Vukovic, M. Vezmar, D. Pesic, J. Drakulic, F. Milosavljevic, B. Pejuskovic, N. Milinkovic, C. Miljevic, B. Markovic, M. Ingelman-Sundberg, N. Maric Bojovic, M. Jukic

University of Belgrade - Faculty of Pharmacy, Belgrade, Serbia; Institute of Mental Health, Belgrade, Serbia; University of Belgrade - Faculty of Medicine, Belgrade, Serbia; Karolinska Institutet, Department of Physiology and Pharmacology, Stockholm, Sweden

EP081

BEYOND CYP2C19: THE ROLE OF TRANSPORTER POLYMORPHISMS AND SEX IN VORICONAZOLE PHARMACOKINETIC VARIABILITY

Allegra Sarah¹, Francesco Chiara^{1,2}, Valeria Ballero¹, Giuliana Abbadessa¹, Maura Caudana¹, Silvia De Francia¹

¹ Department of Clinical and Biological Sciences, University of Turin, Orbassano, Italy;

² Department of Physics, University of Trento, Povo, Italy



EP082

GENOMIC DRIVERS AND THERAPEUTIC VULNERABILITIES IN INDIAN GALLBLADDER CANCERPuja Sinha¹, Amit Kumar¹, M. Ramachandran¹, Shobit Gupta¹, [Giulliana Tessarin](#)^{1*}¹. Genes2Me Pvt. Ltd., Gurugram, Haryana, India

EP083

TRACKING FGFR3 AND PIK3CA MUTATIONS IN BLADDER CANCER VIA PLASMA AND URINE DDPCREleni Thanou^a, Nikos Gavalas^b, Foteini Grigoriou^b, Meropi Galari^b, Vasiliki Malamatini^b, Keisi Mara^b, Konstantina Kakogianni^b, Aristotle Bamias^b, Athina Markou^a^a. *Analysis of Circulating Tumor Cells, Lab of Analytical Chemistry, Dept. of Chemistry, National and Kapodistrian University of Athens, 15771, Athens, Greece*^b. *2nd Propaedeutic Dept of Internal Medicine, National and Kapodistrian University of Athens, ATTIKON University Hospital, Chaidari, Greece*

EP084

SOMATIC RECOMBINATION OF REPEAT ELEMENTS: CHALLENGES AND OPPORTUNITIESGiovanni Pascarella¹, Martin Frith^{2,3,4}, Piero Carninci^{1,5}¹. *RIKEN Center for Integrative Medical Sciences (IMS), Yokohama 230-0045, Japan*². *Artificial Intelligence Research Center, National Institute of Advanced Industrial Science and Technology (AIST), Tokyo 135-0064, Japan*³. *Graduate School of Frontier Sciences, University of Tokyo, Chiba 277-8562, Japan*⁴. *Computational Bio Big-Data Open Innovation Laboratory (CBBD-OIL), AIST, Tokyo 135-0064, Japan*⁵. *Human Technopole, Milan 20157, Italy*

EP085

DEVELOPMENT OF A CUSTOM ASSAY UTILISING MINIHAPOLOTYPES FOR MIXTURE DECONVOLUTIONKao Chin [Ngeow](#)^{*1}, Jaren [Sia](#)^{*1}, Nayana [Prabhu](#)¹, Chin Chin [Lim](#)¹¹. *Home Team Science and Technology Agency, Forensics Centre of Expertise, Singapore*

EP086

TRANSCRIPTOMIC/FUNCTIONAL ANALYSES HIGHLIGHT AN INNATE IMMUNE FACTOR IN MASLD-HCCSujin Kim¹, Gaeul Park¹, TaeJin Ahn², Yong Sun Lee³ and Yeon-Su Lee¹¹. *National Cancer Center, Goyang, Gyeonggi-do, Rep. of Korea*². *Handong Global University, Pohang, Rep. of Korea*³. *Graduate School of Cancer Science and Policy, National Cancer Center, Goyang, Gyeonggi-do, Rep. of Korea*



EP087

POPULATION DISTRIBUTION OF PHARMACODYNAMIC PGX VARIANTS IN CNS

Eleni Ntoutoumou^{1*}, Effie Salata^{1*}, Dimitris Miltiadis Vrachnos¹, Thanos Fotis¹, Sevastiani Papailia¹, Angeliki Kapellou¹ and Spiros Vittas¹

¹. *iDNA Laboratories*

EP088

PRENATAL TRIO WHOLE-EXOME SEQUENCING IN LOW-RISK PREGNANCIES

Katia Margiotti, Marco Fabiani, Costanza Zangheri, Antonella Cima, Francesca Monaco, Chiara Ali', Antonietta Barone, Antonella Viola, Alvaro Mesoraca, Claudio Giorlandino

Human Genetics Lab, Altamedica Rome, Italy

EP089

LARGE-SCALE STUDY OF ERNA-CHROMATIN INTERACTIONS DURING NEURAL DIFFERENTIATION

Jessica Armida Bertolini¹, Chi Wai Yip¹, Rodrigo Pracana¹, Roberta Bosotti², Kayoko Tanaka¹, Giulia Scotti², Eugenia Ricciardelli³, Marco Gaviraghi¹, Chiara Medaglia¹, Clelia Peano³, Alberto Riva², Hazuki Takahashi⁴, Masaki Kato⁴, Takeya Kasukawa⁴, Piero Carninci^{1,4}

¹. *Genomics Research Centre - Functional Genomics, Human Technopole, Milan, Italy.*

². *National Facility for Data Handling and Analysis, Human Technopole, Milan, Italy.*

³. *National Facility for Genomics, Human Technopole, Milan, Italy.*

⁴. *Center for Integrative Medical Sciences, Laboratory for Transcriptome Technology, RIKEN, Yokohama, Japan.*

EP090

EASIGEN-DS: DESIGNING A RESEARCH INFRASTRUCTURE FOR ADVANCED GENOMICS

Mireia Vaca-Dempere¹, Maria Xandri¹, Steven McGinn², Jean-François Deleuze², Michael Forster³, Andre Franke³, Philine Warnke⁴, Liliya Pullmann⁴, Louis Colnot⁵, Jessica Catalano⁵, Orlando Contreras⁶, Tuuli Lappalainen⁶, Tuuli Reisberg⁷, Kristiina Tambets⁷, Hegel Rafael Hernández-López⁸, Joris Vermeesch⁸, Filip Rokić⁹, Oliver Vugrek⁹, Christian Conrad¹⁰, Mònica Bayés^{1*}, Ivo Gut^{1*}

¹. *Centro Nacional de An lisis Gen mico (CNAG), Barcelona, Spain;*

². *Centre National de Recherche en Génomique Humaine (CNRGH), Institut de biologie François Jacob, CEA, Evry, France;*

³. *Institute of Clinical Molecular Biology, Kiel University, Kiel, Germany;*

⁴. *Fraunhofer Institute for Systems and Innovation Research ISI, Karlsruhe, Germany*

⁵. *CSIL Scrl, Milan, Italy; Science for Life Laboratory, Department of Gene Technology,*

⁶. *KTH Royal Institute of Technology, Stockholm, Sweden;*

⁷. *Institute of Genomics, University of Tartu, Tartu, Estonia;*

⁸. *Department of Human Genetics, KU Leuven, Leuven, Belgium;*

⁹. *Rudjer Boskovic Institute, Zagreb, Croatia;*

¹⁰. *Berlin Institute of Health at Charité, University of Medicine Berlin, Corporate Member of the Free University of Berlin, Humboldt-University of Berlin, Berlin, Germany;*



EP091**CASE STUDY OF A PATIENT WITH SOMATIC MUTATION IN POLE EXONUCLEASE DOMAIN**

Nika Rihar¹, Tijana Samardžić¹, Alenka Bombač¹, Vita Šetrajčič Dragoš^{1,2}, Gregor Hertl¹, Gašper Klančar¹, Lorena Kunc¹, Petra Škerl^{1,2}, Anja Zagožen¹, and Vida Stegel^{1,3}

¹ Department of Molecular Diagnostics, Institute of Oncology Ljubljana, Ljubljana, Slovenia

² Faculty of Medicine, University of Ljubljana, Ljubljana, Slovenia

³ Biotechnical Faculty, University of Ljubljana, Ljubljana, Slovenia

EP092**SEQUENCING & VARIATION PROFILING OF THE MULTI-ETHNIC MALAYSIAN POPULATION: THE MYGENOM PROJECT**

Syahril Abdullah, Ryia Illani Mohd Yunos, Yusuf Muhammad Noor & The MyGenom Project Consortium

¹ Malaysia Genome and Vaccine Institute, National Institutes of Biotechnology Malaysia;

² National Institutes of Health, the Ministry of Health

EP093**MULTI-OMIC PHENOME-WIDE MENDELIAN RANDOMIZATION IN HUMAN DISEASES**

Shun-Chun Yu¹, Che-Yu Chou¹, Chun Chieh Fan², Chia-Yen Chen³, Hung-Hsin Chen¹

¹ Institute of Biomedical Sciences, Academia Sinica

² Laureate Institute for Brain Research

³ Merck & Co., Inc

EP094**GENETIC FACTORS ASSOCIATED WITH TACROLIMUS PHARMACOKINETIC VARIABILITY IN GREEKS**

Anna Tsironi¹, Effrosyni Mendrinou², Stavroula Siamoglou¹, Kyriaki Kydonopoulou³, Anne John⁴, Alexandra Gerou³, Spyridon Gerou³, Bassam R. Ali⁴, Marios Papisotiriou⁵ and George P. Patrinos^{1,4,6,7}

¹ Department of Pharmacy, Laboratory of Pharmacogenomics and Individualized Therapy, University of Patras School of Health Sciences, Patras, Greece.

² Molecular Diagnostics Laboratory, INRASTES, National Centre for Scientific Research 'Demokritos', Athens, Greece.

³ ANALYSI S.A., Thessaloniki, Greece.

⁴ College of Medicine and Health Sciences, Department of Genetics and Genomics, United Arab Emirates University, Al-Ain, Abu Dhabi, UAE.

⁵ Department of Nephrology and Kidney Transplantation, University Hospital of Patras, Patras, Greece.

⁶ Faculty of Medicine and Health Sciences, Department of Pathology, Clinical Bioinformatics Unit, Erasmus University Medical Center, Rotterdam, the Netherlands.

⁷ Hellenic Pasteur Institute, Athens, Greece





EP095

VALIDATION OF B-THALASSEMIA GENOTYPING ON PORTAGEN

Ioanna Konstantina Routsis¹, Ioanna Poulida¹, George Patrinos^{1,2,3,4,*}

- ¹ University of Patras, School of Health Sciences, Department of Pharmacy
- ² Department of Genetics and Genomics, College of Medicine and Health Sciences, United Arab Emirates University, Al-Ain, United Arab Emirates,
- ³ Hellenic Pasteur Institute, Laboratory of Innovative Therapeutics and Personalized Medicine, Athens, Greece
- ⁴ Clinical Bioinformatics Unit, Department of Pathology, Faculty of Medicine and Health Sciences, Erasmus University Medical Center, Rotterdam, Netherlands

EP096

STUDY OF CLINICALLY SIGNIFICANT PHARMACOGENES IN IMMUNOSUPPRESSION

Zoi-Maria Thermou¹, Stefania Tornosaki¹, Kariofyllis Karamperis^{1,2}, George P. Patrinos^{1,3,4,5}

- ¹ University of Patras, School of Health Sciences, Department of Pharmacy, Patras, Greece
- ² The Golden Helix Foundation, London, UK
- ³ Department of Genetics and Genomics, College of Medicine and Health Sciences, United Arab Emirates University, Al-Ain, United Arab Emirates,
- ⁴ Hellenic Pasteur Institute, Laboratory of Innovative Therapeutics and Personalized Medicine, Athens, Greece
- ⁵ Clinical Bioinformatics Unit, Department of Pathology, Faculty of Medicine and Health Sciences, Erasmus University Medical Center, Rotterdam, the Netherlands

EP097

STUDY OF CLINICALLY SIGNIFICANT PHARMACOGENES IN CARDIOVASCULAR DISEASE TREATMENT

Stefania Tornosaki¹, Zoi-Maria Thermou¹, Kariofyllis Karamperis^{1,2}, George P. Patrinos^{1,3,4,5}

- ¹ University of Patras, School of Health Sciences, Department of Pharmacy, Patras, Greece
- ² The Golden Helix Foundation, London, UK
- ³ Department of Genetics and Genomics, College of Medicine and Health Sciences, United Arab Emirates University, Al-Ain, United Arab Emirates,
- ⁴ Hellenic Pasteur Institute, Laboratory of Innovative Therapeutics and Personalized Medicine, Athens, Greece
- ⁵ Clinical Bioinformatics Unit, Department of Pathology, Faculty of Medicine and Health Sciences, Erasmus University Medical Center, Rotterdam, the Netherlands

EP098

BEYOND LOW-COVERAGE: A COMPREHENSIVE COMPUTATIONAL PIPELINE FOR ACCURATE ANALYSIS OF ANCIENT DNA

Youssef Abili^{1*}, Nicola Rambaldi Migliore^{2*}, Sara Terzoli¹, Roberta Bosotti¹, Alberto Riva¹, Alessandro Achilli², Luca Pagani³

* Equal contributor

- ¹ National Facility for Data Handling and Analysis, Human Technopole, Milan, Italy.
- ² Dipartimento di Biologia e Biotecnologie "Lazzaro Spallanzani", University of Pavia, Pavia, Italy.
- ³ Department of Biology, University of Padova, Padova, Italy.



EP099**CRISPR/Cas13a DETECTION OF microRNA BIOMARKERS FOR SEPSIS STRATIFICATION**

Despina Panteleon¹, Maria-Nikolina Kerpinioti², Andreas Katsaros³, Grigoris Tzortzakis³, Konstantinos Anastasopoulos³, Irene Pierrakea², Nefeli Zormpa Sfaellou², Maria Electra Pauli², Myrto Xydi², Spiros Giannopoulos², Kariofyllis Karamperis¹, Athina Latsi¹, Kostantinos Bazinis⁴, Nikoleta Galanakou⁵, Thodoris Skondras⁶, George P. Patrinos¹, George Lagoumintzis¹

1. University of Patras, Department of Pharmacy, 26504, Patras, Greece
2. University of Patras, Department of Biology, 26504, Patras, Greece
3. Department of Computer Engineering and Informatics, 26504, Patras, Greece
4. University of Piraeus, Department of Business and Administration, Piraeus, Greece
5. National and Kapodistrian University of Athens, Department of Chemistry, Athens, Greece
6. Massachusetts Institute of Technology, Department of Business Analytics, MIT, USA

EP100**LONGITUDINAL FULL-LENGTH 16S REVEALS MICROBIOME COLLAPSE AFTER ALLOGENEIC HSCT**

Gerasimos Iordanis^{1,2}, Maria Asmi¹, Chara Papaioannou², Maria Liga^{3,4}, Alexandros Spyridonidis^{3,4}, Costas Batargias², Fotini Paliogianni⁵, and George Lagoumintzis¹

1. University of Patras, Department of Pharmacy, 26504, Patras, Greece
2. University of Patras, Department of Biology, 26504, Patras, Greece
3. Bone Marrow Transplantation Unit, Department of Internal Medicine, University Hospital of Patras, Rion, Greece
4. University of Patras, Department of Medicine, Hematology Stem Cell Transplant Unit, Patras, Greece
5. University of Patras, Department of Medicine, Lab of Microbiology, 26504 Patras, Greece

EP101**QUANTIFYING TF MOTIF EFFECTS IN A METHYLATION-CONDITIONED DEEP LEARNING MODEL**

Ayesha Bajwa, Oberon Dixon-Luinenburg, Aaron Streets, Nilah Ioannidis

UC Berkeley

EP102**GENETIC COUNSELLING IN MEDICAL EDUCATION IN ANGOLA: CHALLENGES, OPPORTUNITIES AND STRATEGIES FOR THE IMPLEMENTATION OF BASIC UNITS**

Elka Kay

Faculty of Medicine, Agostinho Neto University

EP103**ASSESSMENT OF GENOTYPE-DIET CONCORDANCE IN NUTRITIONAL REQUIREMENTS**

Marija Staninova-Stojovska¹, Aleksandra Kapedanovska-Nestorovska¹, Tanja Petreska¹, Zoran Zhivikj¹, Ana Marija Bajatovska¹, Angeliki Kapellou², Spiros Vittas²

1. Faculty of Pharmacy, University "Ss. Cyril and Methodius" in Skopje, RN Macedonia
2. IDNA Laboratories, Athens, Greece





EP104

PREDICTORS OF KNOWLEDGE AND PERCEPTIONS OF GENOMICS AMONG MEDICAL STUDENTS

Jolly Thomas, Igdaliah Otitoola, Veronica Obidoyin
College of Medicine, University of Ibadan, Nigeria

EP105

FOCAL PPK WITHOUT NAIL INVOLVEMENT IN A PATIENT WITH A KRT6B MUTATION

Dimitra Kiritsi¹, Stavritsa Taxiarchoula Varvara¹, Athina Ververi², Alexandros Onoufriadis³, Elena Sotiriou¹

1. First Department of Dermatology, Aristotle University of Thessaloniki, Thessaloniki, Greece
2. Department of Genetics for Rare Diseases, "Papageorgiou" General Hospital, Thessaloniki, Greece
3. Laboratory of Medical Biology and Genetics, School of Medicine, Aristotle University of Thessaloniki, Thessaloniki, Greece

EP106

TWO NOVEL GERMLINE H3-3A VARIANTS WITH DIVERGENT NEURODEVELOPMENTAL PHENOTYPES

M. Bakshi⁽¹⁾, B Nash⁽²⁾, UDN-AUS Network⁽³⁾, A Johnson⁽⁴⁾, J Steadson⁽⁵⁾

1. Clinical Genetics, Royal North Shore Hospital, St Leonards, NSW, Australia, 2119
2. Sydney Genome Diagnostics, Sydney Children's Hospital Network, Westmead, NSW, Australia, 2145
3. Department of Neurology, Sydney Children's Hospital Network, Randwick, NSW, Australia, 2031
4. UDN-Aus, Murdoch Children's Research Institute, Victoria, Australia 3052 (5). Department of Paediatrics, Hornsby Ku-Ring-Gai Hospital, Hornsby, NSW Australia 2077

EP107

CLINICAL AND MOLECULAR GENETIC SPECTRUM IN A COHORT OF 18 ECTODERMAL DYSPLASIA PATIENTS

Christos Yapijakis^{1,2}, Nickolas Ziakas^{1,2}, Myrsini Chamakioti^{1,2}, Iphigenia Gintoni^{1,2}, George P Chrousos²

1. Unit of Orofacial Genetics, First Department of Pediatrics, School of Medicine, National Kapodistrian University of Athens, "Aghia Sophia" Children's Hospital, 11527 Athens, Greece
2. University Research Institute of Maternal and Child Health and Precision Medicine, School of Medicine, National Kapodistrian University of Athens

EP108

GENETIC EPIDEMIOLOGY OF FEMALE INFERTILITY AND REPRODUCTIVE SUBTYPES

Maryam Almarzooqi^{1,2}, Mira Mousa^{2,3}, Amira Alaamri², Reem Bakullah², Michael Olbrich², Habiba Al-safar^{1,2}

1. Department of Biomedical Engineering & Biotechnology, Khalifa University of Science and Technology, Abu Dhabi, United Arab Emirates
2. Center for Biotechnology (BTC), Khalifa University of Science and Technology, Abu Dhabi, United Arab Emirates
3. Department of Epidemiology and Public Health, Khalifa University of Science and Technology, Abu Dhabi, United Arab Emirates



EP109**GENOMIC LITERACY AND COMPETENCE AMONG NURSES**

Kyriaki Anyfantaki¹, Eirini Stratidaki², Konstantinos Giakoumidakis³, Athina Patelarou³, Evridiki Patelarou⁴

- 1. PhD(C), Laboratory of Evidence-Based Healthcare, Education and Clinical Protocols, Department of Nursing, Hellenic Mediterranean University, Greece*
- 2. Laboratory of Evidence-Based Healthcare, Education and Clinical Protocols Department of Nursing Hellenic Mediterranean University, Greece*
- 3. Associate Professor, Laboratory of Evidence-Based Healthcare, Education and Clinical Protocols, Department of Nursing, Hellenic Mediterranean University, Greece*
- 4. Professor, Laboratory of Evidence-Based Healthcare, Education and Clinical Protocols, Department of Nursing, Hellenic Mediterranean University, Greece*

EP110**GENOMIC CONTINUING EDUCATION FOR CLINICAL NURSES**

Kyriaki Anyfantaki¹, Evridiki Patelarou⁴, Eirini Stratidaki², Konstantinos Giakoumidakis³, Athina Patelarou³

- 1. PhD(C), Laboratory of Evidence-Based Healthcare, Education and Clinical Protocols, Department of Nursing, Hellenic Mediterranean University, Greece*
- 2. Laboratory of Evidence-Based Healthcare, Education and Clinical Protocols Department of Nursing Hellenic Mediterranean University, Greece*
- 3. Associate Professor, Laboratory of Evidence-Based Healthcare, Education and Clinical Protocols, Department of Nursing, Hellenic Mediterranean University, Greece*
- 4. Professor, Laboratory of Evidence-Based Healthcare, Education and Clinical Protocols, Department of Nursing, Hellenic Mediterranean University, Greece*

EP111**GENOMIC INTEGRATION INTO NURSING CURRICULA**

Kyriaki Anyfantaki¹, Evridiki Patelarou⁴, Eirini Stratidaki², Konstantinos Giakoumidakis³, Athina Patelarou³

- 1. PhD(C), Laboratory of Evidence-Based Healthcare, Education and Clinical Protocols, Department of Nursing, Hellenic Mediterranean University, Greece*
- 2. Laboratory of Evidence-Based Healthcare, Education and Clinical Protocols Department of Nursing Hellenic Mediterranean University, Greece*
- 3. Associate Professor, Laboratory of Evidence-Based Healthcare, Education and Clinical Protocols, Department of Nursing, Hellenic Mediterranean University, Greece*
- 4. Professor, Laboratory of Evidence-Based Healthcare, Education and Clinical Protocols, Department of Nursing, Hellenic Mediterranean University, Greece*

EP112**BLOOD TRANSCRIPTOME IN OSTEOPOROTIC VERTEBRAL FRACTURES IN POSTMENOPAUSAL WOMEN**

Ondrej Seda, Josef Vcelak, Blanka Chylikova, Adam Kral, Lucie Sedova

Institute of Biology and Medical Genetics, First Faculty of Medicine, Charles University and the General University Hospital in Prague; Department of Orthopaedics, First Faculty of Medicine, Charles University, University Hospital Na Bulovce, Prague, Czech Republic





EP113

RECONSTRUCTING ANCIENT HBV FROM HUMAN GENOMES ACROSS EURASIA

Maria Bousali¹, Pavlos Tsiatras¹, Magdalini Bletsas¹, Dimitrios Paraskevis², and Timokratis Karamitros¹

- ¹ *Bioinformatics and Applied Genomics Unit, Department of Microbiology, Hellenic Pasteur Institute, Athens, Attica, Greece*
- ² *Department of Hygiene Epidemiology and Medical Statistics, School of Medicine, National and Kapodistrian University of Athens, Athens, Attica, Greece*

EP114

PREDICTING EPIGENETICALLY-DRIVEN REGULATION WITH CONDITIONED SEQUENCE EMBEDDINGS

Oberon Dixon-Luinenburg, Ayesha Bajwa, Michal Rozenwald, Aaron Streets, Nilah Ioannidis
UC Berkeley Bioengineering, UC Berkeley Electrical Engineering and Computer Science, UC Berkeley Center for Computational Biology

EP115

NUCLEOTIDE TRANSFORMER MODELS ENABLE PRECISE CLASSIFICATION OF HUMAN MIRTRONS

Alisson G. Chiquitto^{1,2*}, Fabiana R. de Goes⁴, Liliane S. Oliveira¹, Aparajita Karmakar⁴, Mark Basham³, Roberto T. Raittz³, Alexandre R. Paschoal^{1,4*}

- ¹ *Department of Computer Science, Federal University of Technology of Paran —UTFPR, Cornélio Procópio, 86300-000, Brazil*
- ² *Department of Informatics, Federal Institute of Education, Science and Technology of Mato Grosso do Sul - IFMS, Navira, 79947-334, Brazil*
- ³ *Laboratory of Bioinformatics, Professional and Technological Education Sector, Federal University of Paran —UFPR, São Paulo, 81520-260, Brazil*
- ⁴ *The Rosalind Franklin Institute, Didcot, OX11 0QX, United Kingdom*

EP116

AUTOMATED FUNCTIONAL CHARACTERIZATION OF VARIANTS IN MICRORNA BINDING SITES

Thodoris Daskalopoulos^{1,2}, Marios Miliotis^{1,2}, Izabella Gogaeva^{1,2}, Artemis G. Hatzigeorgiou^{1,2}

- ¹ *DIANA-Lab, Department of Computer Science and Biomedical Informatics, University of Thessaly, Greece*
- ² *Hellenic Pasteur Institute*

EP117

THREE STAGES OF BUILDING A NATIONAL DATABASE OF GENOMIC VARIATION IN POLAND

Luiza Handschuh^{1,✉}, Ma gorzata Marcinkowska-Swojak¹, Natalia Koralewska¹, Ireneusz Stolarek¹, Paweł Wojciechowski^{1,2}, Magdalena Rakoczy¹, Jan Podkowinski¹, Sylwia Nawrocka¹, Klementyna Marciniak¹, Lucyna Budźko¹, Piotr Łukasik², Aleksandra Świercz^{1,2}, Jacek Błażewicz², Robert Pękal¹, Marek Figlerowicz¹

- ¹ *Institute of Bioorganic Chemistry PAS*
- ² *Poznań University of Technology*



**EP118**
GENOMIC MODULATION OF SEASONAL VITAMIN D RESPONSE: A PREDICTIVE EASTERN-EUROPEAN

Carmina Mariana Stroia¹, Maria Vranceanu², Annamaria Pallag¹

¹ University of Oradea, Romania;

² "Aurel Vlaicu" University of Arad, Romania

EP119
STRENGTHENING ONE HEALTH ANTIMICROBIAL RESISTANCE SURVEILLANCE: INSIGHTS FROM ENVIRONMENTAL AND ANIMAL HEALTH SECTOR COLLABORATION IN KADUNA STATE, NIGERIA

Isiaq Hadji Shehu, Angela Binta², Amina Birma³, Jeremiah Daikwo⁴, Abdullahi Musa Garba⁵, Haliru Usman⁵

¹ Nigeria Centre for Disease Control and Prevention, Kaduna, Nigeria

² Ministry of Environment and Natural Resources, Kaduna State, Nigeria

³ Ministry of Agriculture, Kaduna State, Nigeria

⁴ Ministry of Health, Kaduna State, Nigeria

⁵ Primary Health Care Board, Kaduna State, Nigeria

EP120
SINGLE-CELL RNA SEQUENCING OF AIRWAY RESPONSES TO TEMPERATURE AND AIR POLLUTION

Firdian Makrufardi¹, Annisa Fairuz Nur Azizah²

¹ Department of Medicine, Faculty of Medicine, Universitas Negeri Yogyakarta, Indonesia

² Faculty of Medicine, Universitas Diponegoro, Indonesia

EP121
GENE-DIET INTERACTIONS AND EPIGENETIC AGING IN OLDER ADULTS

Parisa Gholamabreshi¹, Alireza Saadati²

¹ Department of Nutrition, Mashhad University of Medical Sciences, Mashhad, Iran

² Faculty of Medicine, Mashhad University of Medical Sciences, Mashhad, Iran

EP122
GENOMIC AND BIOMARKER PROFILING IN MULTIPLE SCLEROSIS

Alireza Saadati¹, Parisa Gholamabreshi²

¹ Department of Nutrition, Mashhad University of Medical Sciences, Mashhad, Iran

² Faculty of Medicine, Mashhad University of Medical Sciences, Mashhad, Iran

EP123
ENVIRONMENTAL GENOMICS AND CHRONIC DISEASE RISK IN AFRICAN COMMUNITIES

Yusufu Mohamed Filemon, MD¹

¹ Dr Nature Lifestyle Center, Dar es Salaam, Tanzania





EP124

COMPUTATIONAL DISCOVERY OF *PILIOSTIGMA THONNINGII* PHYTOCOMPOUNDS AS MULTITARGET ACETYLCHOLINESTERASE AND AKT1 INHIBITORS FOR ALZHEIMER'S DISEASE

Onwu, Daniel Ocha^{1*}, Moses, Dele Adams², Rofiat, Abdulkareem³, Habiganuchi Adele⁴, Tobiloba Isaac Bajepade⁵, Onekhena Victor Ogie⁶, Mayowa, Agbi⁷, Dasofunjo Kayode⁸, Uraku Anayo Joseph⁹, Ishaya Longdet Yohanna¹⁰, Nwachukwu Gerald Chimaobi¹¹, Israel Ogwuche Ogra¹², Olubukola Catherine Martins¹³, Olaniyan Babatunde Felix¹⁴, Adedara Joshua Favour¹⁵, Adaji Jeremiah Zaphnath-paaneah¹⁶, Oyewole Oluwadamilare Goodness¹⁷, Ihinmikaye Busayo Christiana¹⁸, ¹⁹Esther M. Pius

^{1,8,11}Department of Medical Biochemistry, University of Cross River State (UNICROSS), Calabar, Faculty of Basic Medical Sciences, Okuku Campus, PMB 1123, Cross River State, Nigeria.

² Clinical Biochemistry, Phytopharmacology and Biochemical Toxicology Research Laboratory (CBPBT-RL), Department of Biochemistry, Baze University, Abuja 900108, Nigeria.

³ Bayero University Kano, Department of Pharmacology.

⁴ Department of Biochemistry, Faculty of Science, University of Port Harcourt.

⁵ Covenant University, Ota, Ogun State, Nigeria.

⁷ GeneLab Bioscience, Nigeria.

⁶ Department of Physiology, Ekiti State University, Ado-Ekiti, Ekiti State.

⁸ Department of Medical Biochemistry, Ebonyi State University Abakaliki.

¹⁰ Department of Medical Biochemistry, University of Jos.

¹² Biological Institute, Tomsk State University, 36 Lenin Avenue, Tomsk, 634050, Russian Federation.

¹³ Lagos State University, Faculty of Pharmacy Department of Pharmaceutical Chemistry.

¹⁴ Chemical pathology department, Federal Medical Center, Owo, Ondo State.

¹⁵ Department of Microbiology, Ekiti State University.

¹⁶ School of Health Sciences, Maduka University, Ekwegbe -Nsukka, Enugu State

¹⁸ Bioinformatics and Molecular Biology Unit, Department of Biochemistry, Federal University of Technology, Akure, Ondo State, Nigeria.

¹⁹ Cancer Research and Molecular Biology Laboratories, Department of Biochemistry, Faculty of Basic Medical Sciences, University of Ibadan, Ibadan, Oyo State, Nigeria.

EP125

GENOMIC MINING OF *BACILLUS SAFENSIS* NWU MK_WT AND *ENTEROCOCCUS LACTIS* ENT7/ENT3

Daniel Jesuwenu Ajose¹, Christ-Donald Kaptchouang Tchatchouang²

¹ School of Biology and Environmental Sciences, Faculty of Agriculture and Natural Sciences, University of Mpumalanga, Private Bag X11283, Mbombela, 1200, Mpumalanga, South Africa.

² Eduvos, Applied Sciences, 44 Alsatian Road, Glen Austin AH, Midrand, 1685 Gauteng, South Africa.



EP126**AI-ASSISTED TRACKING OF ESBL-PRODUCING *ESCHERICHIA COLI* IN WEST AFRICAN ABATTOIRS**

Tofio B.F.^{1,2}, Adeyeye B.R.¹, Odeniyi O.A.²

- ¹ Department of Biological Sciences, Faculty of Natural Sciences, Redeemer's University, Ede, Osun State, Nigeria.
- ² Department of Microbiology, University of Ibadan, Ibadan, Oyo State

EP127**COMPARATIVE ENVIRONMENTAL GENOMICS TO STUDY XENOBIOTIC AND SECONDARY METABOLISM IN AN IN-HOUSE COLLECTION OF BACTERIAL ISOLATES**

Zekkas S¹, Basdani D¹, Kylonis A¹, Tzimotoudis D¹, Tsiami E¹, Varela M¹, Stavraki C¹, Devaud A¹, Kontomina E¹, Garefalaki V¹, Evmorfidou D¹, Fakis G¹, Felföldi T², Márialigeti K², Fylaktakidou KC³, Samouris G⁴, Katikou P⁴, Boukouvala S¹

- ¹ Democritus University of Thrace, Department of Molecular Biology and Genetics, Alexandroupolis, Greece
- ² Department of Microbiology, ELTE Eötvös Lor nd University, Budapest, Hungary
- ³ Department of Chemistry, Aristotle University of Thessaloniki, Thessaloniki, Greece
- ⁴ Veterinary Research Institute, Greek Agricultural Organization Demeter, Thessaloniki, Greece

EP128**DIET-RESPONSIVE GENETIC EFFECTS FOLLOWING SHORT-TERM ANIMAL PRODUCT RESTRICTION**

Alexandros Simistiras¹, Georgia Katsoula^{2,3}, Christina Emmanouil¹, Ozvan Bocher^{2,4}, Stavros Glentis^{1,5}, Eleftheria Zeggini^{2,3}, Konstantinos Rouskas^{1,6}, Antigone S Dimas^{1,2*}

- ¹ Institute for Bioinnovation, Biomedical Sciences Research Center 'Alexander Fleming', Fleming 34, 16672, Vari, Greece
- ² Institute of Translational Genomics, Helmholtz Zentrum München – German Research Center for Environmental Health, 85764 Neuherberg, Germany
- ³ Technical University of Munich (TUM) and Klinikum Rechts der Isar, TUM School of Medicine and Health, Munich, Germany
- ⁴ Univ Brest, Inserm, EFS, UMR 1078, GGB, F-29200 Brest, France
- ⁵ Pediatric Hematology/Oncology Unit (POHemU), First Department of Pediatrics, University of Athens, Aghia Sophia Children's Hospital, Athens
- ⁶ Institute of Applied Biosciences, Centre for Research & Technology Hellas, Thessaloniki, Greece





EP129

GUT MICROBIOME CHANGES UPON DIETARY RESTRICTION AND LINKS WITH HOST GENOTYPE

Christina Emmanouil¹, Eleni Aplakidou^{2,3}, Fotis A. Baltoumas², Alexandros Simistiras¹, Maria Anezaki¹, Eleni Katsoni², Maria-Nefeli Stoupa², Stavros Glenti^{1,4}, Evangelos Karatzas⁵, Konstantinos Rouskas^{1,6}, Georgios A. Pavlopoulos², Pantelis Hatzis², Antigone S. Dimas^{1,7}

- ¹ Institute for Bioinnovation, Biomedical Sciences Research Center 'Alexander Fleming', Fleming 34, 16672, Vari, Greece
- ² Institute for Fundamental Biomedical Research, Biomedical Sciences Research Center 'Alexander Fleming', Fleming 34, 16672, Vari, Greece
- ³ Division of Basic Sciences, University of Crete Medical School, 71110 Heraklion, Greece
- ⁴ Pediatric Hematology/Oncology Unit (POHemU), First Department of Pediatrics, University of Athens, Aghia Sophia Children's Hospital, Athens
- ⁵ European Molecular Biology Laboratory, European Bioinformatics Institute (EMBL-EBI), Wellcome Genome Campus, Cambridge, United Kingdom
- ⁶ Institute of Applied Biosciences, Centre for Research & Technology Hellas, Thessaloniki, Greece
- ⁷ Institute of Translational Genomics, Helmholtz Zentrum München – German Research Center for Environmental Health, 85764 Neuherberg, Germany

EP130

FERRETIN PREDICTS CYTOKINE RELEASE SYNDROME IN CAR T-CELL THERAPY: A SYSTEMATIC REVIEW

Sangam Shah

Institute of Medicine, Tribhuvan University

EP131

CHIMERIC ANTIGEN RECEPTORS (CAR) T CELL THERAPY IN SYSTEMIC LUPUS ERYTHEMATOSUS

Bhatta Nikita

Institute of Medicine, Nepal

EP132

MACHINE LEARNING-DRIVEN GENOMIC ANALYSIS OF CLIMATE ADAPTATION IN NIGERIAN RAIN-FOREST ZONES

¹Oladapo Oladoja*, ²Abosede Adepoju

¹Department of Mathematics and Statistics, Abiola Ajimobi Technical University, Ibadan, Nigeria

² Department of Statistics, University of Ibadan, Nigeria



EP133

TOWARD PRECISION ANTIRETROVIRAL THERAPY FOR CHILDREN AND ADOLESCENTS LIVING WITH HIV IN CONFLICT-AFFECTED EASTERN DEMOCRATIC REPUBLIC OF THE CONGO: A GENOMIC IMPLEMENTATION STUDYOlivier Mukuku¹, Kaymarlin Govender²

- ¹ Institut Supérieur des Techniques Médicales de Lubumbashi, Lubumbashi, Democratic Republic of Congo
- ² Health Economics and HIV/AIDS Research Division (HEARD), University of KwaZulu-Natal, Durban, South Africa

EP134

PREDICTING OFF-TARGET TOXICITIES OF LNP-MRNA THERAPEUTICS VIA INTEGRATIVE GENOMIC AND PHARMACOGENOMIC MODELING

Erfan Mousavi

School of Pharmacy, Cyprus International University

EP135

HOST GENOMICS AND THE GUT MICROBIOME CROSS-TALK INTEGRATION IN ADVANCED PRECISION APPROACHES IN COLORECTAL CANCERHadi Feizi^{1*}, Gholamhassan Shahbazi¹, Azizeh ShadiDizaji^{1,2}

- ¹ Research and Innovation Hub, Ostad Aali-Nasab Hospital, Iranian Social Security Organization, Tabriz, Iran
- ² Vaccine Development Application and Research Center, Ataturk University, Erzurum, Turkey

EP136

AUTOAI HUB: FROM DATA TO SOCIETY, DEMOCRATIZING MACHINE LEARNING FOR EVERYONE

Robson Bonidia, Bruno Florentino, Marina Carvalho Arruda Barreto, Breno L. S. de Almeida, João Lucas R. Constantino, Denis Tavares da Silva, Ulisses Rocha, Guilherme Goedert, Claudio Struchiner and André de Carvalho

- ¹ Institute of Mathematics and Computer Sciences, University of São Paulo, São Carlos 13566-590, Brazil
- ² Federal University of Technology – Paran (UTFPR), Campus Cornélio Procopio
- ⁴ Department of Computational Biology and Chemistry, Helmholtz Centre for Environmental Research – UFZ GmbH, Leipzig, Saxony, Germany,
- ⁵ Escola de Matemática Aplicada, Fundação Getúlio Vargas, Rio de Janeiro, Brazil
- ⁵ Global South Artificial Intelligence for Pandemic and Epidemic Preparedness & Response Network (AI4PEP)

EP137

ASSESSING THE METHODOLOGICAL RIGOR AND CLINICAL READINESS OF MACHINE LEARNING MODELS PREDICTING OUTCOMES IN PATIENTS WITH BRAIN METASTASES TREATED WITH RADIOSURGERY: A SYSTEMATIC REVIEW AND CRITICAL APPRAISAL

Muili Abdul basit, Godswill Uzoechina, Olayiwola Qudus, Oluwapelumi Solagbade, Oladahunsi Mercy, Adedire Priscilla, Ibeh Sylvia

University of Nigeria, Ladoke Akintola University of Technology, Obafemi Awolowo University, University of Ilorin, Pavlov Saint Petersburg State Medical University





EP138

ML INTEGRATION OF TUMOR AND GERMLINE GENOMICS TO PREDICT CHEMO-MRNA RESPONSE

Negar Zahirmirdamadi

Faculty of pharmacy, Yazd shahid sadoughi university of medical sciences, Yazd, Iran

EP139

GERMLINE PHARMACOGENOMICS TO PREDICT LNP MRNA VACCINE SAFETY & EFFICACY

Mahsa Hamooni

Faculty of pharmacy, Tehran university of medical sciences, Tehran, Iran

EP140

PREDICTING LONG-TERM MRNA EXPRESSION FROM PRE-TREATMENT OMICS VIA DEEP LEARNING

Mahsa Hamooni

Faculty of pharmacy, Tehran university of medical sciences, Tehran, Iran

EP141

MULTI-OMICS DISCOVERY OF MODIFIERS IN A RARE MITOCHONDRIAL DISORDER

Nikoo Ghasabe

Islamic azad university, Tehran medical branch, Tehran, Iran

EP142

THE GENETIC LANDSCAPE OF KELOIDS

Iasonas Dermitzakis¹, Alexandros Onoufriadis²

¹ Department of Histology-Embryology, School of Medicine, Aristotle University of Thessaloniki, 54124 Thessaloniki, Greece

² Laboratory of Medical Biology and Genetics, School of Medicine, Aristotle University of Thessaloniki, 54124 Thessaloniki, Greece

EP143

CHARACTERIZATION OF COMPLETE HUMAN PEGIVIRUS TYPE 2 (HPGV-2) GENOMES IN INDIVIDUALS CO-INFECTED WITH HEPATITIS C VIRUS (HCV) IN CAMEROON

Aristide Mounchili-Njifon^{1,3}, Vireak Heang², Leakhena Pum², Loique Landry E Messanga¹, Moise Henri Moubeket-Yifomnjou¹, Abdou Fatawou Modiyinji¹, Desmon Toutou Tsafack¹, Ladifatou Nzi Mbouo-Njoya¹, Simon Frederic Lissoc¹, Pretty Rosereine Mbouyap¹, Jean Paul Assam Assam³, Erik A. Karlsson², Janin Nouhin², and Richard Njouom¹

¹ Department of Virology, Centre Pasteur of Cameroon, Yaoundé, Cameroon.

² Virology Unit, Institut Pasteur du Cambodge, Pasteur Network, Phnom Penh, Cambodia.

³ Department of Microbiology, Faculty of Sciences University of Yaounde I, Yaoundé, Cameroon.



EP144**PPARS IN CANCER THERAPY: DRIVING THE METABOLIC REVOLUTION IN ONCOLOGY (A REVIEW)**

Nuhu Mandi Istifanus^{1,*}, Irmiya Benjamin Haruna¹, Abdulazeez Hanif¹

¹. Abubakar Tafawa Balewa University, 740272, Bauchi, Nigeria

EP145**GENOMIC PROFILING OF MATERNAL AND CHILD HEALTH RISKS IN THE LAKE CHAD BASIN: A PATHWAY TO PRECISION PUBLIC HEALTH IN THE SAHARA**

Michael Markus Kutte, M. M. Wesley, B. Wayeh, F. Bitrus

Yobe State University, Damaturu; Modibbo Adama Teaching Hospital Yola, Adamawa State University, Yola

EP147**AN INTEGRATED MACHINE LEARNING GUIDED THERAPEUTIC APPROACH TO FERROPTOSIS: A NEW FRONTIER IN PRECISION ONCOLOGY**

Sana Shaik¹, Zohra Rao², Zoi Man³, Sakshi Kumar⁴, Leon Samuel Barboza⁵, Afra Wasama Islam⁶, Salwa Asif⁷, Muhammad Umer Farooq Mujahid⁸, Abdelrahman Albahtiti⁹ Hafiz Muhammad Haris¹⁰

¹. Faculty of Medicine, Tbilisi State Medical University, Tbilisi, Georgia

². Faculty of Medicine, Tbilisi State University, Tbilisi, Georgia

³. University of Nicosia, Nicosia, Cyprus

⁴. Shimoga Institute of Medical Sciences, India

⁵. Faculty of Medicine, Ivane Javakhishvili Tbilisi State University, Tbilisi, Georgia

⁶. Faculty of Medicine, Tbilisi State Medical University, Tbilisi, Georgia

⁷. Faculty of Medicine, Intern, Thumbay Hospital, UAE

⁸. University of Health Sciences, Lahore, Pakistan

⁹. Faculty of Medicine, Ivane Javakhishvili Tbilisi State University, Tbilisi, Georgia

¹⁰. King Edward Medical University, Lahore

Pakistan

EP148**SELF-SUPERVISED LEARNING IMPROVES FUNCTIONAL ANNOTATION OF RARE VARIANTS**

NEDA SEDGHNEJAD

School of pharmacy, Eastern Mediterranean university

EP149**AI-ENHANCED TRANSCRIPTOMIC-BASED DRUG REPURPOSING PIPELINE FOR LIMB-GIRDLE MUSCULAR DYSTROPHY**

Amal GASMI^{1,2,*}, Lilia ROMDHANE^{1,3,+}

¹. Laboratory of Biomedical Genomics and Oncogenetics, Pasteur Institute of Tunis, Tunis, Tunisia

². Higher Institute of Biotechnology of Sidi Thabet (ISBST), Tunis, Tunisia

³. Department of Life Sciences, Faculty of Sciences of Bizerte, University of Carthage, Tunisia





EP150

DECODING ANTIBIOTIC RESISTANCE IN ESCHERICHIA COLI: MULTIPLEX PCR PRECISION FOR TEM B-LACTAMASE & TEQ A RESISTANT GENES

Qaisar Akram¹, Muhammad Ahsan Naeem², Waqas Ahmad³

¹ Department of Basic Sciences (Pharmacology), University of Veterinary and Animal Sciences, Lahore (Narowal Campus) Narowal 51600, Pakistan

^{2,3} Department of Pathobiology (Microbiology), University of Veterinary and Animal Sciences, Lahore (Narowal Campus) Narowal 51600, Pakistan

EP151

MACHINE LEARNING FOR EARLY DETECTION OF CHRONIC KIDNEY DISEASE

Abdulazeez Hanif^{1*}, Irmiya Benjamin Haruna¹ and Nuhu Mandi Istifanus¹

Abubakar Tafawa Balewa University, 740272, Bauchi, Nigeria

EP152

PREDICTIVE, PREVENTIVE AND PERSONALIZED MEDICINE OF RESPIRATORY SYNCYTIAL VIRUS

Ousman Bajinka

University of The Gambia

EP153

RATIONAL COMPUTATIONAL DESIGN AND DEVELOPMENT OF AN IMMUNOGENIC MULTIEPITOPE VACCINE INCORPORATING TRANSMEMBRANE PROTEINS OF STAPHYLOCOCCUS LUGDUNESIS

Aswad Khan¹

Department of Biotechnology, Faculty of Sciences, International Islamic University, Islamabad, Pakistan

EP154

MOLECULAR CHARACTERIZATION OF HEPATITIS B VIRUS ISOLATE FROM A VACCINATED CHILD IN OGBOMOSO, OYO STATE, NIGERIA

Busayo Kayode Akomolafe^{1,2,3}, Etuvie Favour Akomolafe^{3,6}, Olawale Sunday Animasaun^{1,3,4}, Nengak Precious Danladi², & Rosemary Ajuma Audu^{1,5}

¹ Department of Biological Sciences, Lead City University, Ibadan, Nigeria,

² National Reference Laboratory, Nigeria Center for Disease Control, Abuja, Nigeria

³ Department of Medical Laboratory Services, Oyo State Primary Health Care Board, Ibadan, Nigeria,

⁴ Georgetown Global Health Nigeria, Abuja, Nigeria,

⁵ Centre for Human Virology and Genomics, Nigerian Institute of Medical Research, Lagos, Nigeria,

⁶ Bowen University Teaching Hospital, Ogbomosho, Nigeria



EP155

STRUCTURAL VARIATIONS OF ALU ELEMENTS IN KUWAITI POPULATION: APPLICATIONS IN FORENSIC MEDICINE

Seham S. A. Abumarzouq¹, Abdel Halim Deifalla²

- ¹ Molecular Medicine Department, College of Medicine and Health Sciences, Arabian Gulf University, Bahrain;
- ² Postgraduate Studies and Research, ²Department of Anatomy, College of Medicine and Health Sciences, Arabian Gulf University, Bahrain.

EP156

REFRAMING THE HIMALAYA: POPULATION DYNAMICS, MIGRATIONS, AND MALARIA ADAPTATION

Mark A. Penjueli¹, Rita Fahmy^{1,2}, Aashish Gyawali³, Obed Garcia⁴, Yoshina Gautam¹, Dinesh Bhandari⁵, Sarmila Tandukar⁵, Eaaswarkhanth Muthukrishnan¹, Shirley Sutton⁶, Kaitlyn Spees⁶, Genevieve L. Wojcik⁶, Guru Prasad Gautam⁷, Jeevan Bahadur Sherchand⁵, Carlos D. Bustamante⁶, Alexandar G. Ioannidis⁶, Aashish R Jha^{1,2,6,8}

- ¹ Genetic Heritage Group, Program in Biology, New York University Abu Dhabi, Abu Dhabi, UAE
- ² Center for Genomics and Systems Biology, New York University Abu Dhabi, Abu Dhabi, UAE
- ³ Sabitri Sciences Pvt. LTD. Kathmandu, Nepal
- ⁴ Department of Ecology and Evolutionary Biology, University of California, Santa Cruz
- ⁵ Public Health Research Laboratory, Institute of Medicine, Maharajgunj, Kathmandu, Nepal
- ⁶ Department of Biomedical Data Science, Stanford University, Palo Alto, California, USA
- ⁷ Department of Geography, Tribhuvan University Nepalgunj, Nepalgunj, Nepal
- ⁸ Public Health Research Center, New York University Abu Dhabi, Abu Dhabi, UAE

EP157

INVESTIGATING THE POTENTIAL OF BTD7 COMPOUND AS A NOVEL ANTI-MYCOBACTERIAL TUBERCULOSIS AGENT: AN IN VITRO STUDY

Haftay Abrahama,b, c#, Zhangon Liua,b c#, Buhari Yusufa,b, c, Gui-Yang Zhaoe, Md Shah Alama,b, c, Belachew Awekea,b, c, Abdul Malika,b, c, H.M. Adnan Hameeda,b,c*, and Zhang Tianyua,b,c,d

State Key Laboratory of Respiratory Disease, Guangzhou Institutes of Biomedicine and Health, Chinese Academy of Sciences, Guangzhou 510530, China





EP158

THE NOVEL (TCTG)_N MOTIF IN CNBP EXPANDED ALLELES: COMPOSITION, DYNAMICS AND GENOTYPE-PHENOTYPE CORRELATION IN DM2

Federica Centofanti¹, Virginia Veronica Visconti¹, Maria Rosaria D'Apice², Marco Carlomagno³, Simone Maestri³, Dario Ciabini¹, Mario Bengala², Enrica Marchionni², Erica Frezza⁴, Roberto Massa⁴, Antonio Petrucci⁵, Francesca Lupidi⁶, Elena Pegoraro⁷, Gabriele Siciliano⁸, Matteo Garibaldi^{9,10}, Massimo Delledonne³, Marzia Rossato³, Annalisa Botta¹, Giuseppe Novelli¹

- ¹ Department of Biomedicine and Prevention, Medical Genetics Section, University of Rome Tor Vergata, Rome, Italy
- ² Laboratory of Medical Genetics, Tor Vergata Hospital, Rome, Italy.
- ³ Department of Biotechnology, University of Verona, Verona, Italy.
- ⁴ Department of Systems Medicine (Neurology), University of Rome Tor Vergata, Rome, Italy.
- ⁵ Center for Neuromuscular and Neurological Rare Diseases, S. Camillo Forlanini Hospital, Rome, Italy
- ⁶ Neurological Clinic, Azienda Ospedaliera Universitaria delle Marche, Ancona, Italy.
- ⁷ Department of Neurosciences, DNS Clinica Neurologica, Azienda Ospedale University of Padova, Padua, Italy
- ⁸ Department of Clinical and Experimental Medicine, University of Pisa, Pisa, Italy.
- ⁹ Neuromuscular and Rare Disease Centre, Neurology Unit, Sant'Andrea Hospital, Rome, Italy
- ¹⁰ Department of Neuroscience, Mental Health and Sensory Organs (NESMOS), SAPIENZA University of Rome, Rome, Italy

EP159

MULTI-OMICS REVEAL UROGENITAL SCHISTOSOMIASIS-SPECIFIC DRIVERS OF BLADDER CANCER

¹Adeboye Adedoyin T., ²Anumudu Chiaka I., ³Adebayo Adewale S.

- ¹ Cell Biology and Genetics Unit, Department of Zoology, University of Ibadan, Nigeria
- ² Cellular Parasitology Programme, Department of Zoology, University of Ibadan, Nigeria
- ³ School of Medicine and Population Health, University of Sheffield UK

EP160

AI-DRIVEN GENETIC ANALYSIS OF CARDIOMYOPATHY IN THE OMANI POPULATION

A. Al Amri¹, N. Al Mahrami², F. Al Hattali¹, A. Al Balushi¹, N. Al Hashmi¹, M. Al Araiimi¹, M. Al Rawahi³, T. Al Lawatia³, M. Al Balushi¹, B. Al Shamsi¹

- ¹ National Genetics Centre of the Royal Hospital
- ² Oman College of Health Sciences
- ³ National Heart Centre of the Royal Hospital



EP161

THE MYH9 E-1 HAPLOTYPE IS ASSOCIATED WITH CHRONIC KIDNEY DISEASE AMONG SOUTH AFRICAN PATIENTS WITH HYPERTENSION

Bianca Obadic^{1,2}, Nosipho Mabizela^{1,2}, Jonathan Katsukunya^{1,2}, Bianca Davidson^{1,2,3}, Erika S. W. Jones^{1,2,3}, Collet Dandara^{1,2} and Khuthala Mnika^{1,2}

- 1. Division of Human Genetics, Department of Pathology, Faculty of Health Sciences, & Institute of Infectious Disease and Molecular Medicine, University of Cape Town, South Africa*
- 2. Platform for Pharmacogenomics Research and Translation, South African Medical Research Council, Cape Town, South Africa*
- 3. Division of Nephrology & Hypertension, Department of Medicine, Faculty of Health Sciences, University of Cape Town, South Africa*

EP162

AFRICAN-SPECIFIC PATHOGENIC VARIANTS OF NEPHROTIC SYNDROME IN SOUTH AFRICAN PAEDIATRIC PATIENTS: PRELIMINARY FINDINGS

Thina Gcobo^{1,2}, Arinao Ndadza,³ Lindiwe Lamola⁴, Denis Awany⁵, Kim Whitehead⁴, Nilesh Lala Shannon⁴, Collet Dandara^{1,2}, Amanda Krause⁴, Karen Petersen⁴, and Khuthala Mnika^{1,2}

- 1. Division of Human Genetics, Department of Pathology, Faculty of Health Sciences, & Institute of Infectious Disease and Molecular Medicine, University of Cape Town, South Africa*
- 2. Platform for Pharmacogenomics Research and Translation, South African Medical Research Council, Cape Town, South Africa*
- 3. Department of Molecular Medicine and Haematology, School of Pathology, Faculty of Health Sciences, University of the Witwatersrand, Johannesburg, South Africa*
- 4. Department of Human Genetics, National Health Laboratory Service, and School of Pathology, Faculty of Health Sciences, University of the Witwatersrand, Johannesburg, South Africa*
- 5. South African Tuberculosis Vaccine Initiative, Institute of Infectious Disease and Molecular Medicine and Division of Immunology, University of Cape Town*

EP163

GENOME REGULATION BY NFAT DURING ENDOTHELIAL-LIKE DIFFERENTIATION

Mohadese Mozafari¹, Siti Nurnasihah Md Hashim¹, Khairul Bariah Ahmad Amin Noordin¹, Siti Aishah Zainal¹, TP Kannan^{1,2}, Suzina Sheikh Abd Hamid³ & Ahmad Azlina^{1,2,3}

- 1. School of Dental Sciences, Universiti Sains Malaysia, Health Campus, 16150 Kubang Kerian, Kelantan, Malaysia*
- 2. Human Genome Centre, School of Medical Sciences, Universiti Sains Malaysia, Health Campus, 16150 Kubang Kerian, Kelantan, Malaysia*
- 3. Tissue Bank, School of Medical Sciences, Universiti Sains Malaysia, Health Campus, 16150 Kubang Kerian, Kelantan, Malaysia*

EP164

GENOTYPE VS PHENOTYPE DRIVEN WGS/WES ANALYSES

Maria Horianopoulou¹, Marios Kambouris^{1,2}

- 1. MedCare Polyclinics & Diagnostic Center, Athens, Greece;*
- 2. Sidra Medicine, Doha, Qatar*





EP165

POPULATION-SPECIFIC VARIATION IN MMF-RELATED PHARMACOGENETIC VARIANTS AND EXPLORATORY ASSOCIATIONS WITH RENAL RESPONSE IN SOUTH AFRICAN PATIENTS WITH LUPUS NEPHRITIS

Migael Mouton^{1,2}, Bianca Davidson⁴, Erika Jones⁴, Robert Freercks³, Khuthala Mnika^{1,2}, Collet Dandara^{1,2}

- 1. Division of Human Genetics, Department of Pathology and Institute of Infectious Disease and Molecular Medicine (IIDM), Faculty of Health Sciences, University of Cape Town, Cape Town, South Africa.*
- 2. SAMRC/UCT Platform for Pharmacogenomics Research and Translation, South African Medical Research Council, Cape Town, South Africa.*
- 3. Faculty of Health Sciences, Department of Medicine, Nelson Mandela University, Gqeberha, South Africa*
- 4. Division of Nephrology and Hypertension, University of Cape Town, Cape Town, South Africa*

EP166

MULTIDIMENSIONAL VARIATION ACROSS 8000 COMPLETE HUMAN CENTROMERES

Dongya Wu, Yanqing Sun, Xiaofei Yang, Guojie Zhang
Zhejiang University, China; Xi'an Jiaotong University, China

EP167

MGRN1 IS IMPLICATED IN RECESSIVE CONGENITAL HEART AND LATERALITY DEFECTS IN HUMANS

Laura Kasak¹, Kristiina Rull^{2,3}, Anu Valkna¹, Maris Laan¹

- 1. Chair of Human Genetics, Institute of Biomedicine and Translational Medicine, University of Tartu, Tartu, Estonia*
- 2. Women's Clinic, Tartu University Hospital, Tartu, Estonia*
- 3. Institute of Clinical Medicine, University of Tartu, Tartu, Estonia*

EP168

STRUCTURAL VARIANTS IN IDIOPATHIC INFERTILITY UNCOVERED BY EXOME SEQUENCING

Kikas, Triin¹; Valkna, Anu¹; Jakovlev, Ülle²; Pomm, Kristjan³; Rull, Kristiina^{4,5}; Punab, Margus^{1,3,4}; Laan, Maris¹

- 1. Institute of Biomedicine and Translational Medicine, University of Tartu, Tartu, Estonia*
- 2. Centre of Endocrinology, Clinic of Internal Medicine, East-Tallinn Central Hospital, Tallinn, Estonia*
- 3. Andrology Clinic, Tartu University Hospital, Tartu, Estonia*
- 4. Institute of Clinical Medicine, University of Tartu, Tartu, Estonia*
- 5. Women's Clinic, Tartu University Hospital, Tartu, Estonia*



EP169**FOLATE DOSE AND EPIGENETIC VARIATION: A GENOME-WIDE METHYLATION STUDY**

Marília Körbes Rockenbach^{1,3}, Jéssica Moreira de Menezes², Lailson Melgueira Navarro³, Bárbara Pilger dos Santos³, Lucas Petitembert de Souza⁴, Laís Gonçalves⁴, William Domingues⁴, Vinícius Farias Campos⁴, Maria Teresa Vieira Sanseverino^{1,5} and Thayne Woycinc Kowalski^{1,2,3,5}

1. Graduate Program in Genetics and Molecular Biology (PPGBM), Genetics Department, Universidade Federal do Rio Grande do Sul, Porto Alegre, Brazil
2. Graduate Program in Medicine: Medical Sciences, Medicine Faculty, Universidade Federal do Rio Grande do Sul, Porto Alegre, Brazil
3. Laboratory of Genomic Medicine (LMG), Center of Experimental Research, Hospital de Clínicas de Porto Alegre, Porto Alegre, Brazil
4. Laboratory of Structural Genomics, Universidade Federal de Pelotas, Pelotas, Brazil
5. Medical Genetics Service (SGM), Hospital de Clínicas de Porto Alegre, Porto Alegre, Brazil

EP170**EXOME SEQUENCING FOR NDD DIAGNOSIS: A BRAZILIAN PUBLIC HOSPITAL EXPERIENCE**

Thayne Woycinc Kowalski^{1,2,3}, Mariana Rodrigues Botton^{1,2,4}, Gabriela Flores Gonçalves^{1,3}, Renan Cesar Sbruzzi¹, Leonardo Simão Medeiros^{1,2}, Osvaldo Artigal S^{1,2}, Ursula Matte^{1,3}, Temis Maria Felix^{1,2}, Jonas Alex Morales Saute^{1,2}, Fernanda Sales Luiz Vianna¹

1. Program of Genomic and Precision Medicine, Hospital de Clínicas de Porto Alegre;
2. Medical Genetics Service, Hospital de Clínicas de Porto Alegre;
3. Bioinformatics Core, Hospital de Clínicas de Porto Alegre;
4. Laboratory Diagnostics Service, Hospital de Clínicas de Porto Alegre

EP171**META-ANALYSIS OF TETRALOGY OF FALLOT: IDENTIFYING NEW CANDIDATE GENES**

Fabyanne Guimarães de Oliveira^{1,2}; João Vitor Pacheco Foletto^{2,3}; Júlia Rei Pires²; Lavínia Schuler-Facchini^{1,2}; Thayne Woycinc Kowalski^{1,2,3,4}

1. Laboratory of Medical Genetics and Evolution, Graduate Program in Genetics and Molecular Biology, Genetics Department, Universidade Federal do Rio Grande do Sul (UFRGS), Porto Alegre 91501-970, Brazil.
2. Teratogen Information System (SIAT), Medical Genetics Service, Hospital de Clínicas de Porto Alegre (HCPA), Porto Alegre 90035-903, Brazil.
3. Laboratory of Genomic Medicine, Center of Experimental Research, Hospital de Clínicas de Porto Alegre (HCPA), Porto Alegre 90035-903, Brazil.
4. Bioinformatics Core, Hospital de Clínicas de Porto Alegre (HCPA), Porto Alegre 90035-903, Brazil.





EP172

GENE EXPRESSION PROFILE OF NEWBORNS EXPOSED PRENATALLY TO ANTIPILEPTIC DRUGS

João Vitor Pacheco Foletto^{1,2,3,4,5}; Fabianne Guimarães de Oliveira^{1,3,5}; Marília Körbes Rockenbach^{1,3,5}; Fernanda Sales Luiz Vianna^{1,2,3,4,5}; Lavínia Schuler-Faccini^{1,3,5}; Thayne Woycinck Kowalski^{1,2,3,4,5}.

1. Graduate Program in Genetics and Molecular Biology, Genetics Department, Universidade Federal do Rio Grande do Sul (UFRGS), Porto Alegre 91501-970, Brazil;
2. Laboratory of Genomic Medicine, Center of Experimental Research, Hospital de Clínicas de Porto Alegre (HCPA), Porto Alegre 90035-903, Brazil;
3. Laboratory of Medical Genetics and Evolution, Genetics Department, Universidade Federal do Rio Grande do Sul (UFRGS), Porto Alegre 91501-970, Brazil;
4. Bioinformatics Core, Hospital de Clínicas de Porto Alegre (HCPA), Porto Alegre 90035-903, Brazil;
5. Teratogen Information System (SIAT), Medical Genetics Service, Hospital de Clínicas de Porto Alegre, Porto Alegre, Brazil.

EP173

FOUNDER EFFECT FOR PAH C.441+5G>T IN AN ISOLATED ECUADORIAN HIGHLAND TOWN

Paul Leon-Gomez / Vanessa Romero
Universidad San Francisco de Quito

EP174

INTEGRATING PRS WITH SCORE2 FOR CARDIOVASCULAR RISK STRATIFICATION

Alberto Budillon¹; Maria Pina Concas¹; Paolo Gasparini^{1,2}

1. Institute for Maternal and Child Health – IRCCS “Burlo Garofolo”, Trieste, Italy
2. Department of Medicine, Surgery and Health Sciences, University of Trieste, Trieste, Italy

EP175

DIVERSITY OF VARIATION DATA HELPS CLASSIFY VARIANTS IN INHERITED RETINAL DISEASE

Matthijs Leenders, Tarik Luisman, Mathijs Gaastra, Karen E. Malone
GeneScape

EP176

CLINICAL GENOMIC DETECTION OF COPY NUMBER AND STRUCTURAL VARIANTS IN MISCARRIAGE

Pardeep Kumar, Jagjit Singh, V K Sharma
Biochemistry, Shakuntla Hospital And Research Center, Delhi, India



EP177**INVESTIGATING THE RELATIONSHIP BETWEEN SCHIZOPHRENIA, THE RETINA, AND BRAIN VOLUMES**

Michaela Papangeli¹, Lisa Roberts¹, Dan Stein^{2,3}, and Shareefa Dalvie⁴

- ¹ Division of Human Genetics, Department of Pathology, University of Cape Town, South Africa
- ² Department of Psychiatry and Mental Health, University of Cape Town, Cape Town, South Africa
- ³ South African Medical Research Council Unit on Risk and Resilience in Mental Disorders, University of Cape Town, Cape Town, South Africa
- ⁴ Division of Human Genetics, Department of Pathology, University of Cape Town, South Africa; and UCT Neuroscience Institute

EP178**WES DIAGNOSTIC YIELD FOR MONOGENIC DISORDERS: INSIGHTS FROM AN ITALIAN CENTER**

Stefania Zampieri¹, Domizia Pasquetti^{1,2}, Anna Facchini¹, Elisa Rubinato¹, Elisa Paccagnella^{1,2}, Beatrice Spedicati^{1,2}, Vanessa Rebecca Gasparini², Anna Morgan¹, Stefania Lenarduzzi¹, Alessandro Pecori¹, Giorgia Giroto^{1,2}, Federica Perino², Giulia Pianigiani^{1,2}, Paolo Gasparini^{1,2}

- ¹ Institute for Maternal and Child Health - IRCCS "Burlo Garofolo" – Trieste, Italy
- ² Department of Medicine, Surgery and Health Sciences, University of Trieste, Trieste, Italy

EP179**FROM GENOMIC DISCOVERY TO BEDSIDE PRACTICE: A SYSTEMATIC REVIEW OF CLINICAL IMPLEMENTATION OF PHARMACOGENOMICS IN PERSONALIZED THERAPEUTICS**

Opeyemi Sodiq Hammed^{1,2,*}, Dorcas Ayooluwa Alabi³

- ¹ Genomic and Pharmacotherapy Division, Helix Biogen Institute, Ogbomoso Oyo State, Nigeria.
- ² Collaboration and Research, Slum and Rural Health Initiative Network (SRHIN), Ibadan, Oyo State, Nigeria.
- ³ Acute Assessment Unit, Royal Sussex County Hospital, Brighton, United Kingdom

EP180**ETHICAL AND SOVEREIGN AI DATA VISITATION IN GENOMIC MEDICINE**

Francis P. Crawley

HGP2-ELSI

EP181**DESIGN OF A DUAL EPISOMAL CRISPR SYSTEM FOR ZBTB7A REPRESSION AND HBF INDUCTION**

Irene Dereki¹, Vasiliki Chondrou¹, Eleftherios Bochalis², Ilias Georgakopoulos-Soares², George Lagoumintzis³, Argyro Sgourou¹

- ¹ Biology Laboratory, School of Science and Technology, Hellenic Open University, Patras, Greece
- ² Department of Pharmacology and Toxicology, College of Pharmacy, University of Texas at Austin, Dell Pediatric Research Institute, USA
- ³ Division of Pharmacology and Biosciences, Department of Pharmacy, School of Health Sciences, University of Patras, Patras, Greece





EP182

HARNESSING LONG-READ SEQUENCE DATA OF THE GA4K INITIATIVE TO INFORM PHARMACOGENE VARIATION

Andrea Gaedigk^{1,2}, Shobana John¹, Erin, C Boone¹, Byunggil Yoo³ and Laura B Ramsey^{1,3}

- ¹ *Division of Clinical Pharmacology, Toxicology & Therapeutic Innovation, Children's Mercy Research Institute, Kansas City, MO USA*
- ² *School of Medicine, University of Missouri-Kansas City, Kansas City, MO USA.*
- ³ *Department of Pathology Research, Children's Mercy Research Institute, Kansas City, MO USA*

EP183

FETAL KARYOTYPING: EXPERIENCE IN A REFERENCE CENTER IN SOUTHERN BRAZIL

Hellen Nicole Keller^{1,2}, Bruna Herrmann Janssen da Silva¹, Juliana Vargas Campiol^{1,2}, Maria Clara de Freitas Pinho¹, Rejane Gus¹, Thayne Woycinc Kowalski^{1,2}, Caroline Paula Mescka¹

- ¹ *Medical Genetics Service, Hospital de Clínicas de Porto Alegre – HCPA*
- ² *Graduate Program in Medicine: Medical Sciences, Faculty of Medicine, Universidade Federal do Rio Grande do Sul (UFRGS)*

EP184

POLYGENIC RISK AND ASSOCIATION BEYOND LINEARITY

¹Christophe Thomassin, ²Marc Franquesa Monés, ^{1,3}David Bonet, ⁴Peter A. Gerlach, ^{1,5}Marçal Coma-joan Cara, ¹Daniel Mas Montserrat, and ^{1,3,4}Alexander G. Ioannidis

- ¹ *Dept. of Biomedical Data Science, Stanford University, Stanford, CA, USA*
- ² *Centre de Formació Interdisciplinària Superior, Universitat Politècnica de Catalunya, Barcelona, Spain*
- ³ *Genomics Institute, University of California, Santa Cruz, Santa Cruz, CA, USA*
- ⁴ *Dept. of Genetics, Stanford University, Stanford, CA, USA*
- ⁵ *Dept. of Electrical Engineering and Computer Science, University of California, Berkeley, Berkeley, CA, USA*

EP185

MHC CONSERVED EXTENDED HAPLOTYPES IN FAMILIES: IMPLICATIONS FOR DISEASE ASSOCIATION AND TRANSPLANTATION IN AN ADMIXED POPULATION FROM THE MIDDLE EAST.

Halima Alnaqbi^{1,2}, Guan Tay⁴, Habiba Alsafar²

- ¹ *Center for Biotechnology, Khalifa University of Science and Technology, Abu Dhabi, UAE.*
- ² *Department of Biomedical Engineering, Khalifa University of Science and Technology, Abu Dhabi, United Arab Emirates.*
- ³ *Division of Psychiatry, UWA Medical School, The University of Western Australia, Perth, WA, Australia.*
- ⁴ *School of Medical and Health Sciences, Edith Cowan University, Joondalup, WA, Australia.*

EP186

SELECTIVE MASKING OF LOW-COMPLEXITY REGIONS IMPROVES VARIANT CALLING PRECISION IN CLINICAL NGS WORKFLOWS

C. Kaligerou, A. Tsagkalidou, D. Tremoulis, V. Pogka, T. Karamitros

- ¹ *Bioinformatics and Applied Genomics Unit, Department of Microbiology, Hellenic Pasteur Institute, Athens, Greece*




EP187
MtDNA COPY NUMBER AND RNA EPIGENETIC REMODELING DURING AZA TREATMENT IN HR-MDS

Argyri Chroni^{1,2}, Theodoros Nikolopoulos¹, Irene Dereki¹, Vasiliki Chondrou¹, Theodora Alexiou¹, Katerina Athanasopoulou¹, Eleftherios Bochalis³, Theodora Chatzilygeroudi⁴, John Zafeiropoulos⁵, Ilias Georgakopoulos-Soares³, Kyriakos Bourikas⁵, Argyro Sgourou¹, Argiris Symeonidis^{2,6}

- ¹ *Biology Laboratory, School of Science and Technology, Hellenic Open University, Patras, Greece.*
- ² *School of Health Sciences, Faculty of Medicine, Department of Internal Medicine, Hematology Division, University of Patras, Patras, Greece.*
- ³ *Division of Pharmacology and Toxicology, College of Pharmacy, The University of Texas at Austin, Dell Paediatric Research Institute, Austin, TX, USA*
- ⁴ *Second Department of Internal Medicine and Research Unit, Hematology Unit, National and Kapodistrian University of Athens School of Medicine, Attikon University Hospital, 12462 Athens, Greece.*
- ⁵ *Chemistry Laboratory, School of Science and Technology, Hellenic Open University, Patras, Greece.*
- ⁶ *Olympion General Clinic & Rehabilitation Center, 264.43 Patras, Greece*

EP188
WHEN HOMOLOGOUS RECOMBINATION DEFICIENCY LACKS MUTATION IN OVARIAN CANCER

Sakshi Mattoo, Anurag Mehta, Himanshi Diwan, Manoj Panigrahi, Sheeja Gopalan, Chaitanya Verma
Rajiv Gandhi Cancer Institute and Research Centre, Sharda University

EP189
POINT CLOUD LOCAL ANCESTRY INFERENCE (PCLAI): COORDINATE-BASED ANCESTRY ALONG THE GENOME

Margarita Geleta^{1,2}, Daniel Mas Montserrat¹, Nilah M. Ioannidis^{2,3}, Alexander G. Ioannidis^{1,3}

- ¹ *Department of Biomedical Data Science, Stanford University School of Medicine, Stanford, CA 94305, United States of America*
- ² *Department of Electrical Engineering and Computer Science, University of California, Berkeley, Berkeley, CA 94720, United States of America*
- ³ *Genomics Institute, University of California at Santa Cruz, Santa Cruz, CA 95060, United States of America*

EP190
ARYLAMINE N-ACETYLTRANSFERASE (NAT) PHARMACOGENE NOMENCLATURE AND GENOTYPE-PHENOTYPE CORRELATION: THIRTY YEARS OF EXPERT CURATION TO SERVE THE GLOBAL COMMUNITY

Boukouvala S¹ and Hein DW²

- ¹ *Democritus University of Thrace, Department of Molecular Biology and Genetics, Alexandroupolis, Greece*
- ² *University of Louisville School of Medicine, Department of Pharmacology & Toxicology, Louisville, KY, USA*





EP191

INVESTIGATION OF NAT1, NAT2 AND CYP2D6 GENETIC VARIATION IN THE GREEK POPULATION

Papanikolaou G¹, Mitropoulou S², Gaedigk A³, Poloni ES⁴, Boukouvala S¹, Fakis G¹

- ¹ Department of Molecular Biology and Genetics, Democritus University of Thrace, Alexandroupolis, Greece
- ² Department of Medicine, Democritus University of Thrace, Alexandroupolis, Greece
- ³ Division of Clinical Pharmacology, Toxicology & Therapeutic Innovation, Children's Mercy Research Institute, Kansas City, Missouri, USA
- ⁴ Department of Genetics and Evolution, Institute of Genetics and Genomics Geneva, University of Geneva, Geneva, Switzerland

EP192

INTERPRETABLE GRAPH NEURAL NETWORKS FOR LEARNING DRUG-GENE RELATIONSHIPS AND INFERRING PHENOTYPE OUTCOMES IN PHARMACOGENOMICS

Kgabe Ronald Molepo¹, Hocine Bendou¹, Khuthala Mnika²

- ¹ Department of Integrative Biomedical Sciences, Division of Computational Biology, University of Cape Town
- ² Department of Pathology, Division of Human Genetics, University of Cape Town

EP193

PHENOTYPE PREDICTION USING STRING KERNEL FEATURE EXPANSION

Egor Lappo, David Bonet, Daniel Mas Montserrat, Alexander G. Ioannidis

EL: Stanford University, Stanford, CA, US; DB and AGI: Stanford University, Stanford, CA, US and Genomics Institute, University of California, Santa Cruz, Santa Cruz, CA, US; DMM: Stanford University, Stanford, CA, US

EP194

GENOMIC AND TRANSCRIPTOMIC SIGNATURES OF OSTEOPOROTIC VERTEBRAL FRACTURES

Lucie Sedova, Josef Vcelak, Blanka Chylikova, Adam Kral, Ondrej Seda

Institute of Biology and Medical Genetics, First Faculty of Medicine, Charles University and the General University Hospital in Prague; Department of Orthopaedics, First Faculty of Medicine, Charles University, University Hospital Na Bulovce, Prague, Czech Republic

EP195

LONGITUDINAL EPIGENETIC VARIATION AND ENVIRONMENTAL INFLUENCES AMONG THE EFIK PEOPLE OF NIGERIA

Osim Peace, Nelson Anita, Ekpenyong Blessing, Mary E. Kooffreh
University of Calabar



EP196**DEEPNE, A DEEP LEARNING APPROACH TO INFERENCE OF RECENT EFFECTIVE POPULATION SIZE**

Samuel Sacco, David Bonet Sole, Alex Mentzer, Andres Moreno Estrada, and Alexander Ioannidis
University of California, Santa Cruz, Department of Biomolecular Engineering, Santa Cruz CA, Centre for Human Genetics, Nuffield Department of Medicine, University of Oxford, Oxford, UK., Advanced Genomics Unit, Center for Research and Advanced Studies (Cinvestav), Mexico City, Mexico, Department of Anthropology, University of California, San Diego, San Diego, USA, Stanford University, Stanford, California, United States

EP197**EPIGENETIC AGING IN INDIVIDUALS WITH SUBSTANCE USE DISORDER: SEX AND ANCESTRY**

Patricia N. Torres-Rivera^{1,2,3,6}, Sheila Nagamatsu^{1,2,3}, José J. Martínez-Magaña^{1,2,3}, Lea Zillich⁴, Diana L. Núñez-Ríos,^{1,2,3} Adnel Figueroa¹, Henry Kranzler⁵, Renato Polimanti^{1,2}, Daniel Levey^{1,2}, Joel Gelenter^{1,2,3}, Janitza L. Montalvo-Ortiz^{1,2,3}

- Department of Psychiatry, Yale University School of Medicine, New Haven, CT*
- VA Connecticut Healthcare System, West Haven, CT*
- U.S. Department of Veterans Affairs National Center for Post-traumatic Stress Disorder, Clinical Neurosciences Division, West Haven, CT*
- Department of Genetic Epidemiology in Psychiatry, Central Institute of Mental Health Mannheim, Medical Faculty Mannheim, Heidelberg University*
- Perelman School of Medicine University of Pennsylvania*
- Universidad Central del Caribe, School of Medicine, Bayam n, PR*

EP198**KLEEFSTRA SYNDROME IN A RWANDAN PATIENT WITH NEURODEVELOPMENTAL DISORDERS**

Norbert Dukuze, Prof. Leon Mutesa, Dr Esther Uwibambe
University of Rwanda

EP199**TOWARDS AN OPTIMIZED CFDNA EXTRACTION PROTOCOLS IN LOW RESOURCES SETTING**

Zeinab Sayed Abdelkhalek¹, Rasha Elkaffas¹, Radwa M. Abdel Halim¹, Sarah A. Lotfy¹, Samar Saad¹, Dalia Abd El-Kareem², Gihan Saad Abo Elwafa³, Saeed M. Shaaban⁴, Samar Ahmed Fouad⁵, Loay Kassem⁶, Hebatallah Hany Assal³, Sahar Abdelatty Sharaf¹

- Clinical and Chemical Pathology Department, Faculty of Medicine, Cairo University, Cairo, Egypt*
- Anatomic Pathology Department, Faculty of Medicine, Cairo University, Cairo, Egypt*
- Department of Chest Diseases, Faculty of Medicine, Cairo University, Cairo, Egypt*
- Clinical Oncology Department, Faculty of Medicine, Beni-Suef University, Egypt.*
- Chest Department, Fayoum University, Cairo, Egypt*
- Clinical Oncology Department, Faculty of Medicine, Cairo University, Egypt*





EP200

COMPREHENSIVE WHOLE GENOME SEQUENCING-BASED PHARMACOGENOMICS PROFILING USING A UNIVERSAL PERSONALIZED GENOME INTERPRETATION SOFTWARE

Aikaterini Patrino^{1,#}, Alexandros Kanterakis^{2,#}, Gerasimos Vonitsanos³, Peter J. van der Spek⁴, George P. Patrinos^{4,5,6,7,*}

- ¹ University of Thessaly, Department of Computer Science and Biomedical Informatics, Lamia, Greece
- ² Foundation for Research and Technology – Hellas, Institute of Computer Science, Heraklion, Crete, Greece
- ³ University of Patras, Polytechnic School, Computer Engineering and Informatics Department, Patras, Greece
- ⁴ Erasmus University Medical Center, Faculty of Medicine and Health Sciences, Department of Pathology, Clinical Bioinformatics Unit, Rotterdam, the Netherlands
- ⁵ Hellenic Pasteur Institute, Laboratory of Innovative Therapeutics and Personalized Medicine, Athens, Greece
- ⁶ University of Patras, School of Health Sciences, Department of Pharmacy, Patras, Greece
- ⁷ United Arab Emirates University, College of Medicine and Health Sciences, Department of Genetics and Genomics, Al-Ain, Abu Dhabi, United Arab Emirates

EP201

GERMLINE WHOLE-GENOME SEQUENCING WITH HIGH ACCURACY AND IMPROVED COVERAGE USING THE ULTIMA GENOMICS SEQUENCING PLATFORM AND SOLARIS 2.0 CHEMISTRY

Sarah Pollock*, Mickael Ploquin, Geoff Lowman, Omer Barad, Gilad Almogy
Ultima Genomics

EP202

BEYOND ROUTINE TESTING – INTEGRATION OF KARYOTYPE, OPTICAL GENOME MAPPING AND GENOME SEQUENCING UNCOVERS A BALANCED TRANSLOCATION

Nurit Assia Batzir, Lily Bazak, Naama Orenstein, Lina Basel Salmon, Shiri Yacobson, Ilya Soifer, Mickael Ploquin, Reut Matar, Sarah Pollock

EP203

GENOMIC DIVERSITY OF URINE MICROBIOME IN OVERACTIVE BLADDER: A SYSTEMATIC REVIEW AND META-ANALYSIS

Argyro Kalogeropoulou¹, Maria Papaefthimiou¹, Maria Kampa¹, Panagiota I. Kontou², Pantelis G. Bagos¹, Michael Samarinis³, Georgia G. Braliou¹

- ¹ Department of Computer Science and Biomedical Informatics, University of Thessaly, Lamia, Greece;
- ² Department of Mathematics, University of Thessaly, Lamia, Greece;
- ³ 2nd Urology Department, Papageorgiou Hospital, Aristotle University of Thessaloniki, Thessaloniki, Greece.



EP204

PORTAGEN: A PORTABLE POINT-OF-CARE ANALYZER FOR NEAR-PATIENT PHARMACOGENOMIC TESTING

Ioanna Poulida¹, Kariofyllis Karamperis^{1,2}, George Patrinos^{1,3,4,5,*}

¹ University of Patras, School of Health Sciences, Department of Pharmacy

² The Golden Helix Foundation, London, UK

³ Department of Genetics and Genomics, College of Medicine and Health Sciences, United Arab Emirates University, Al-Ain, United Arab Emirates,

⁴ Hellenic Pasteur Institute, Laboratory of Innovative Therapeutics and Personalized Medicine, Athens, Greece

⁵ Clinical Bioinformatics Unit, Department of Pathology, Faculty of Medicine and Health Sciences, Erasmus University Medical Center, Rotterdam, the Netherlands

EP205

CFC-SEQ: FULL-LENGTH CAPPED RNAS UNVEILS STRUCTURAL DETERMINANTS OF ERNAS

Chi Wai Yip^{1,2}, Hazuki Takahashi², Kayoko Yasuzawa^{1,2}, Callum J.C. Parr², Masaki Kato², Takeya Kasukawa², Ivano Legnini¹, Jay W. Shin^{1,3}, Chung-Chau Hon², Piero Carninci^{1,2}

¹ Human Technopole, Milan, Italy.

² RIKEN Center for Integrative Medical Sciences, Yokohama, Kanagawa 230-0045, Japan.

³ Genome Institute of Singapore, Agency for Science, Technology and Research, Singapore 138672, Singapore.

EP206

SINGLE-NUCLEUS TRANSCRIPTOMICS OF DIABETIC ADIPOSE TISSUE ACR

Noura Al Nuaimi^{1,2}, Lena Löblein², Thanumol Abdul Khader², Mira Mousa^{2,3}, Amayllis Van Craenenbroek^{4,5}, Syed Ashraf¹, Peter Carmeliet⁵, Habiba Alsafar^{2,6}

¹ Department of Biological Sciences, Khalifa University, Abu Dhabi, UAE

² Center for Biotechnology, Khalifa University of Science and Technology, Abu Dhabi, UAE.

³ Department of Public Health and Epidemiology, Khalifa University, Abu Dhabi, UAE

⁴ Department of Nephrology, University Hospitals UZ Leuven, Leuven, Belgium

⁵ Laboratory of Angiogenesis and Vascular Metabolism, Department of Oncology, Center for Cancer Biology, Leuven, Belgium.

⁶ Department of Biomedical Engineering, Khalifa University, Abu Dhabi, UAE

EP207

GENOMIC PROFILING OF BREAST CANCER IN ECUADORIAN PATIENTS USING NEXT-GENERATION SEQUENCING

Patricia Guevara-Ram rez, Viviana A. Ruiz-Pozo, Santiago Cadena-Ullauri, Elius Paz Cruz, Rafael Tamayo Trujillo y Ana Karina Zambrano

Centro de Investigación Genética y Genómica, Facultad de Ciencias de la Salud Eugenio Espejo, Universidad UTE, Quito, Ecuador





EP208

EXPLORING THE GENOMIC ARCHITECTURE OF TRIM32 TO UNDERSTAND ITS FUNCTIONAL PLEIOTROPY

Tsigkas I.^{*1,2}, Chapouton P.³, Gagos S.⁴, and Michaelidis T. M.^{1,2}

- ¹ Dept. of Biological Applications & Technologies, School of Health Sciences, University of Ioannina, Ioannina, Greece.
- ² Biomedical Research Institute, Foundation for Research and Technology-Hellas (FORTH), Ioannina, Greece.
- ³ Helmholtz Zentrum München, German Research Center for Environmental Health, Munich, Germany
- ⁴ Biomedical Research Foundation of the Academy of Athens (BRFAA), Athens, Greece.

EP209

VARIANT RESULTS FROM BREAST CANCER MULTI-GENE PANEL TESTING IN MALAYSIA

Claudia Richard Beginda¹, Nur Tiara Hassan², Vivian Yi-Mun Lee², Heamanthaa Padmanabhan², Yong Quan Lee², Daphne Shin-Chi Lee², Joanna Mei-Ch'wan Lim², Siti Norhidayu Hasan², Meow Keong Thong^{3,4}, Sook-Yee Yoon^{1,2}

- ¹ GENETIX Cancer Research Malaysia, Selangor, Malaysia
- ² Cancer Research Malaysia, Selangor, Malaysia
- ³ University Tunku Abdul Rahman, Selangor, Malaysia
- ⁴ University Malaya Medical Centre, Kuala Lumpur, Malaysia

EP210

GENETIC ARCHITECTURE OF SUDDEN CARDIAC DEATH IN A KAZAKHSTANI COHORT

Ainur Akilzhanova^{1,2}, Ayaulym Chamoieva¹, Madina Zhalbinova¹, Zhanel Mirmnokairov Tomiris Shakhmarova¹, Diana Samatkyzy¹, Saule Rakhimova¹, Ulan Kozhamkulov¹, Gulbanu Akilzhanova³, Ulykbek Kairov⁴, Kenes Akilzhanov³, Tolyn Zhakupova^{5,6}, Makhabbat Bekbosynova⁷, Dos Sarbassov^{1,8}

- ¹ Laboratory of Genomic and Personalized Medicine, Center for Life Sciences, National Laboratory Astana, Nazarbayev University, Astana, Kazakhstan
- ² Heart Rhythm Scientific Research Institute, Medical University Astana, Astana, Kazakhstan
- ³ Semey Medical University, Pavlodar Branch, Pavlodar, Kazakhstan
- ⁴ Laboratory of Bioinformatics and Computational Systems Biology, Center for Life Sciences, National Laboratory Astana, Nazarbayev University, Astana, Kazakhstan
- ⁵ Medical University Astana, Astana, Kazakhstan,
- ⁶ Center for Forensic Medicine, Astana, Kazakhstan,
- ⁷ Heart Center, University Medical Center, Nazarbayev University, Astana, Kazakhstan
- ⁸ Nazarbayev University, School of Science and Humanities, Astana, Kazakhstan



EP211**INTEGRATING GENOMIC AND EPIGENOMIC LANDSCAPES OF BREAST CANCER TO INVESTIGATE BLOOD-BASED BIOMARKERS**

Halima Alnaqbi¹, Noora Zayed¹, Michael Olbrich¹, Faris Meqdam¹, Salsabeel Juneidi¹, Anju Thomas¹, Dalia Barayan¹, Zeinab Mahmoud¹, Habiba Alsafar^{1,2*}

- ¹ Department of Biomedical Engineering and Biotechnology, College of Medicine and Health Sciences, Khalifa University of Science and Technology, Abu Dhabi, United Arab Emirates
- ² Center for Biotechnology, Khalifa University of Science and Technology, Abu Dhabi, United Arab Emirates.

EP212**IMPUTATION METHODS ON HARDY WEINBERG EQUILIBRIUM FOR MISSING GENOME WIDE EXPRESS**

Dennis Makambi¹, Dr. Fred Monari¹, Dr. Robert Nyabwanga²

- ¹ Kisii University
- ² CUEA

EP213**IPSC-DERIVED CARDIOMYOCYTES FOR UNDERSTANDING OF MECHANISMS RYR2-ASSOCIATED ARRHYTHMIA.**

S. Rakhimova, N. Satvaldina, Zh.Abilova, A. Akilzhanova.

National Laboratory Astana, Nazarbayev University, Astana, Kazakhstan

EP214**GENOMIC LANDSCAPE OF KAZAKH INDIVIDUALS FROM UNDERREPRESENTED WORLD REGION**

Serikzhan A, Daniyarov A, Molkenov A, Akhmetova A, Abilova Z, Sharip A, Yerezhepov D, Rakhimova S, Kozhamkulov U, Kushugulova A, Askarova S, Sarbassov D, Akilzhanova A, Kairov U.

CLS, NLA, Nazarbayev University

EP215**PHARMACOGENETICS AND PHARMACOGENOMICS STUDY ON WHOLE GENOME SEQUENCING DATA OF OROFACIAL CLEFTS CASE-PARENTS TRIOS**

Poku-Adusei Elvis¹, Gideon Okyere Mensah¹, Christian Opoku Asamoah¹, Bruce Tsri¹, Hafsa Akeyaa¹, Abass Shaibu Danbaki¹, Solomon Obiri-Yeboah^{2,3}, Azeez Butali⁴, Peter Donkor⁵, Lord Jephthah Joojo Gowans^{1,3}

- ¹ Department of Biochemistry and Biotechnology, College of Science, KNUST, Kumasi, Ghana
- ² Department of Oral and Maxillofacial Sciences, School of Dentistry, KNUST, Kumasi, Ghana
- ³ Cleft-Craniofacial Clinic, Komfo Anokye Teaching Hospital, Kumasi, Ghana
- ⁴ Department of Oral Pathology, Radiology and Medicine, The University of Iowa, Iowa City, Iowa, USA
- ⁵ Department of Surgery, School of Medical Sciences, KNUST, Kumasi, Ghana





EP216

DRUG-RESISTANT TUBERCULOSIS AND WGS OF *M.TUBERCULOSIS* IN KAZAKHSTAN

U. Kozhamkulov, Z. Abilova, A. Akhmetova, S. Rakhimova, D. Yerezhepov, U. Kairov, A. Akilzhanova
National Laboratory Astana, Nazarbayev University, Astana, Kazakhstan

EP217

MUTATION-LEVEL HETEROGENEITY SHAPES NEURODEVELOPMENTAL RISK IN RAS/MAPK DISORDER

Lina Ghanmiyin , Elaine Clark , Kier Shiel , Lara Masad , Erwin Pauws
University College London /United kingdom , Great Ormond st.Hospital / London , United Kingdom , Great Ormond st.Hospital / London , United Kingdom ,Queen Mary University ,London/ University College London

EP218

ADMIXTURE HISTORY AND TROPICAL ADAPTATION OF THE CAMBODIAN INDIGENOUS PEOPLES

Jianxin Guo, Manman Wu, Kai Liu, Yaoxi He, Bing Su
Kunming Institute of Zoology, Chinese Academy of Sciences

EP219

GENEAID: AN INTERPRETABLE RANDOM FOREST FRAMEWORK FOR ACMG-ALIGNED GENETIC VARIANT CLASSIFICATION USING CLINVAR

Lara Abdul Kareem Yosef Masa'ad , Dimitrios Kollias
Queen Mary University of London , Queen Mary University of London



EP220

IMPLEMENTATION OF HEALTH ECONOMICS INTO CLINICOGENETIC MODELS TO ASSESS THE COST AND THE QUALITY OF LIFE OF PATIENTS: THE CASE OF THE GENETIC SYNTAX SCORE (GESS) STUDY

Dimitrios Trygoniari^{1*}, Nikolaos Mittas¹, Fani Chatzopoulou², Efstratios Karagiannidis³, Andreas S. Papazoglou⁴, Vassilis Fragoulakis⁵, Dimitrios Chatzidimitriou⁶, George Giannakoulas⁷, Ioannis S. Vizirianakis^{8,9}, Christina Mitropoulou^{5,10}

1. *Hephaestus Laboratory, School of Chemistry, Faculty of Sciences, Democritus University of Thrace, Kavala, Greece*
2. *Department of Biomedical Sciences, School of Health Sciences, International Hellenic University*
3. *Department of Emergency Medicine, AHEPA University Hospital, School of Medicine, Faculty of Health Sciences, Aristotle University of Thessaloniki, Greece*
4. *Athens Naval Hospital, Athens, Greece*
5. *The Golden Helix Foundation, London, UK*
6. *Laboratory of Microbiology, School of Medicine, Aristotle University of Thessaloniki, Thessaloniki, Greece*
7. *First Department of Cardiology, AHEPA University Hospital, School of Medicine, Faculty of Health Sciences, Aristotle University of Thessaloniki, Greece*
8. *Laboratory of Pharmacology, School of Pharmacy, Aristotle University of Thessaloniki, Thessaloniki, Greece*
9. *Department of Health Sciences, School of Life and Health Sciences, University of Nicosia, Nicosia, Cyprus*
10. *College of Medicine and Health Sciences, Department of Genetics and Genomics, Zayed Center for Health Sciences, United Arab Emirates University, Al-Ain, Abu Dhabi, UAE.*

EP221

DEEP LEARNING-ENHANCED POLYGENIC RISK SCORING FOR EARLY-ONSET TYPE 2 DIABETES

Erfan Mousavi

School of Pharmacy, Cyprus International University

EP222

NOD1 AND NOD2 GENETIC VARIANTS: IMPACT ON HEPATOCELLULAR CARCINOMA SUSCEPTIBILITY AND PROGRESSION IN MOROCCAN POPULATION

Chaimaa Zerrad^{a,b}, Mustapha Lkhider^b, Meryem Bouqdayr^c, Abdelkader Belkouchi^d, Wafaa Badre^e, Mohamed Tahir^{e,f}, Pascal Pineau^g, Soumaya Benjelloun^a, Sayeh Ezzikouri^{a,*}

^a *Virology Unit, Viral Hepatitis Laboratory, Institut Pasteur du Maroc, Casablanca, Morocco*

^b *Laboratoire de Virologie, Oncologie, Biosciences, Environnement et Energies Nouvelles, Hassan II, Casablanca Facult e des Sciences et Techniques, Mohammedia, Morocco*

^c *Virology Unit, Immunovirology Laboratory, Institut Pasteur du Maroc, Casablanca, Morocco*

^d *Surgery Department A, Ibn Sina University Hospital, Rabat, Morocco*

^e *Service d'Hepato-Gastro-Entérologie, CHU Ibn Rochd, Casablanca, Morocco*

^f *Facult'e de M'edecine et de Pharmacie, Universit'e Hassan II, Casablanca, Morocco*

^g *Unit'e "Organisation Nucl'eaire et Oncog'ense", INSERM U993, Institut Pasteur, Paris, France*





EP223

CDX2 AND KI-67 EXPRESSION AS POTENTIAL PROGNOSTIC MARKERS IN METASTATIC COLORECTAL CANCER

Akbota Aitkulova^{1*}, Saule Rakhimova¹, Tatiana Rogounovitch², Ainur Akilzhanova¹, Dos Sarbassov¹

¹ National Laboratory Astana, Nazarbayev University, Astana, Kazakhstan

² Nagasaki University, Nagasaki, Japan

EP224

CIRCULATING KRAS CODON 12 CTDNA AS A MOLECULAR MARKER OF TREATMENT RESPONSE IN COLORECTAL CANCER PATIENTS TREATED WITH STANDARD NEOADJUVANT THERAPY VERSUS ATO/D-VC

Ulan Kozhamkulov¹, Saule Rakhimova¹, Ainur Akilzhanova¹, Dos Sarbassov¹, Akbota Aitkulova^{1*}

¹ National Laboratory Astana, Nazarbayev University, Astana

EP225

IDENTIFICATION OF RECURRENT FUSION GENES IN ESOPHAGEAL SQUAMOUS CELL CARCINOMA PATIENTS FROM KAZAKHSTAN

Aibek Karimov, Rakhimbek Bektayev, Ruslan Kalendar, Aigul Sharip, Saule Rakhimova, Nazerke Satvaldina, Yuri Zhukov, Mukhtar Tuleutaev, Ainur Akilzhanova, Dos Sarbassov and Ulykbek Kairov

Center for Life Sciences, National Laboratory Astana, Nazarbayev University

EP226

WHOLE GENOME SEQUENCING OF 1500 CASES OF PARKINSON'S DISEASE FROM ITALY.

Trova S.¹, Di Donato GW.², Musacchia F.¹, Chiartano G.¹, Treves G.¹, Fant A.¹, Gaudio A.³, Landuzzi F.⁴, Pandolfini L.⁵, Monfrini E.⁶, Trevisan L.³, Cavalli A.⁴, Di Giovanni M.⁷, Lillia N.⁷, Caligiana L.⁷, Cordera S.⁷, Giardini G.⁷, Vecchi M.¹, Mandich P.³, Di Fonzo A.⁸, Aitoro A.², Gustincich S.^{1,5}

¹ Non-coding RNAs and RNA-based therapeutics, Italian Institute of Technology, C3G, Aosta, Italy.

² GenomSys MPEG-G Competence Center, Milano, Italy.

³ IRCCS Ospedale Policlinico San Martino, UOC Genetica Medica, Genova, Italy;

⁴ Computational and Chemical Biology, Italian Institute of Technology, C3G, Aosta, Italy.

⁵ Center for Human Technologies, Non-coding RNAs and RNA-based therapeutics, Italian Institute of Technology, Genova, Italy

⁶ Dino Ferrari Center, Neuroscience Section, Department of Pathophysiology and Transplantation University of Milan, Milano, Italy;

⁷ Neurologia e Stroke Unit, Ospedale Regionale "Umberto Parini", Aosta, Italy;

⁸ Foundation IRCCS Ca' Granda Ospedale Maggiore Policlinico, Neurology Unit, Milano, Italy.



EP227**WGS MEETS AI: AN INITIATIVE IN CLINICAL AND COMPUTATIONAL GENOMICS IN ITALY**

Manuela Vecchi¹, Guido Walter Di Donato², Sara Trova¹, Alessandro Coppe³, Fabio Landuzzi³, 5000genomi@VdAConsortium, Antonio Amoroso¹, Andrea Cavalli^{3,4,5}, Antonio Aitoro², Stefano Gustincich^{1,6}

- ¹ Non-coding RNAs and RNA-based therapeutics, Italian Institute of Technology, C3G, Aosta, Italy.
- ² GenomSys MPEG-G Competence Center – Milan, Italy.
- ³ Computational and Chemical Biology, Italian Institute of Technology, C3G, Aosta, Italy.
- ⁴ Center for Human Technologies, Computational and Chemical Biology, Italian Institute of Technology, Genoa, Italy.
- ⁵ Centre Européen de Calcul Atomique et Moléculaire (CECAM), Ecole Polytechnique Fédérale de Lausanne, Lausanne, Switzerland.
- ⁶ Center for Human Technologies, Non-coding RNAs and RNA-based therapeutics, Italian Institute of Technology, Genova, Italy.

EP228**INTEGRATING EVOLUTIONARY GENOMICS INTO GENOMIC MEDICINE TRAINING**

Konstantina Drosou and Ursula Paredes-Esquivel

City St George's University of London

EP229**ADVANCING EQUITABLE PRECISION MEDICINE THROUGH AN ASEAN GENOME CONSORTIUM**

Dr AUNG THAN OO

Makerere University, Humanities And Social Science Department

EP230**TO DISCLOSE, OR NOT TO DISCLOSE GENETIC RESULTS WITH FAMILY MEMBERS IN UGANDA**

Sylvia Nabukenya^{1,2}, David Kyaddondo¹, Adelline Twimukye², Catriona Waitt², Erisa S. Mwaka¹

- ¹ Makerere University College of Health Sciences, Kampala Uganda
- ² Infectious Diseases Institute, Kampala Uganda





EP231

A META-ANALYSIS OF PRE-ECLAMPSIA GENOMIC SUSCEPTIBILITY FACTORS IN POPULATIONS OF AFRICAN ANCESTRY: POOLED EFFECT ESTIMATES OF SUSCEPTIBILITY LOCI IN 19 GENES ACROSS FOUR PATHWAYS

Jonathan N. Katsukunya^{1,2}, Bianca Davidson^{2,3}, Khuthala Mnika^{1,2}, Nyarai D. Soko^{1,2,4}, Ayesha Osman⁵, Mushi Matjila⁵, Erika Jones^{2,3} and Collet Dandara^{1,2*}

- ¹ Division of Human Genetics, Department of Pathology and Institute of Infectious Disease and Molecular Medicine (IDM), Faculty of Health Sciences, University of Cape Town, Cape Town, South Africa.
- ² SAMRC/UCT Platform for Pharmacogenomics Research and Translation, South African Medical Research Council, Cape Town, South Africa.
- ³ Kidney and Hypertension Research Unit, Division of Nephrology and Hypertension, Department of Medicine, Groote Schuur Hospital and Faculty of Health Sciences, University of Cape Town, Cape Town, South Africa.
- ⁴ Biomedical Sciences Unit, School of Allied Health Sciences, Harare Institute of Technology, Harare, Zimbabwe.
- ⁵ Department of Obstetrics and Gynaecology, Faculty of Health Sciences, University of Cape Town, Groote Schuur Hospital, Cape Town, South Africa.

EP232

PANGENOME-RESOLVED STRUCTURAL VARIATION OF THE PGA CLUSTER UNDERSCORES HUMAN ADAPTATION TO AGRICULTURAL PROTEIN SOURCES

Quanyu Chen, Anguo Liu, Guojie Zhang, Dongya Wu
Zhejiang University

EP233

DEFINING ANCESTRY-SPECIFIC GENETIC AND EPIGENETIC ARCHITECTURE OF FAMILIAL MULTIPLE SCLEROSIS

Halima Alnaqbi^{1,2}, Maryam Albairaq²

- ¹ Center for Biotechnology, Khalifa University of Science and Technology, Abu Dhabi, UAE.
- ² Department of Biomedical Engineering, Khalifa University of Science and Technology, Abu Dhabi, United Arab Emirates.

EP234

HUGH GLUCOSE MODULATES GLYCORNAs IN CULTURED GLOMERULAR EPITHELIAL CELLS

Esther Jung, Nik Tsotakos
Dept. of Biological Sciences, Penn State Harrisburg



EP235

AI-DRIVEN *IN SILICO* PHARMACOGENOMIC MODELLING FOR PRECISION DOSING IN PSYCHIATRY

Kariofyllis Karamperis ^{1,2,3}, Federico Melograna ^{4,5}, Bastien Martin ³, Alexander Kulesza ³, George P. Patrinos ^{1,6,7,8}

- ¹ Department of Pharmacy, School of Health Sciences, University of Patras, Patras, Greece
- ² The Golden Helix Foundation, London, UK
- ³ Modeling & Simulation, Nova In Silico, Lyon, France
- ⁴ Department of Human Genetics, KU Leuven, Leuven, Belgium
- ⁵ GIGA-R Molecular and Computational Biology, University of Liège, Liège, Belgium
- ⁶ Department of Genetics and Genomics, College of Medicine and Health Sciences, United Arab Emirates University, Al-Ain, United Arab Emirates
- ⁷ Hellenic Pasteur Institute, Athens, Greece
- ⁸ Clinical Bioinformatics Unit, Department of Pathology, Faculty of Medicine and Health Sciences, Erasmus University Medical Center, Rotterdam, the Netherlands

EP236

REAL-WORLD APPLICATION OF WES IN RARE DISEASES IN A SOUTH AFRICAN SETTING

Patricia Nevondwe¹, Maryke Schoonen¹, Leeré Scott¹, Ilse Du Preez¹, Chris Vorster¹

- ¹ Centre for Human Metabolomics, Desmond Tutu School of Medicine, Faculty of Health Sciences, North-West University, Potchefstroom, South Africa

EP237

INTEGRATING GENOMIC INSIGHTS INTO THE MANAGEMENT OF RARE DISEASES AND NON-COMMUNICABLE DISEASES IN VULNERABLE POPULATIONS IN SOMALIA

Abdirezak Mawlid Abdi. RN, BSC, MPH.^{1, 2, 3} Hamdi Osman¹, Mustafe Awil¹

- ¹ Somalia NCD Alliance (SNCA), Mogadishu, Somalia.
- ² Ministry of Health and Human Services, Mogadishu, Somalia.
- ³ Department of Medicine and Health Science, Benadir University, Mogadishu, Somalia





Faculty



**ABDULLAH SYAHRIL**

Associate Professor, Director, Malaysia Genome and Vaccine Institute (MGVI), National Institutes of Biotechnology Malaysia (NIBM)

ALI BASSAM

Professor - Chair, Department of Genetics and Genomics, UAE University, Dept of Genetics and Genomics, United Arab Emirates University, UAE

ANTONARAKIS STYLIANOS

Professor Emeritus (active), Department of Genetic Medicine, University of Geneva Medical School, Geneva, Switzerland, Former President of HUGO

ARZUAGA FABIANA

Professor of Regulation of Biotechnology and Patent Law, Chair of the Advisory Commission in Cellular Therapies and Regenerative Medicine at the Ministry of Science, Technology and Productive Innovation of Argentina, HUGO, HCA (Board member).

BASU ANALABHA

Professor, BRIC - National Institute of Biomedical Genomics, India

BIENKO MAGDA

Research Group Leader, Functional Genomics Research Centre, Human Technopole, Milan, Italy; Karolinska Institutet / SciLifeLab, Stockholm, Sweden

BIN ALWI ZIFALIL

Senior Consultant Pediatrician & Clinical Geneticist, Professor of Medical Genetics, Human Genome Center, University Sains, Malaysia

BOUKOUVALA SOTIRIA

Democritus University of Thrace, Alexandroupoli, Greece

BOWCOCK ANNE

PhD, is the Norman Orentreich, MD Professor of Dermatology Research at the Icahn School of Medicine at Mount Sinai, and is also Professor, Oncological Sciences, and Genetics and Genomic Sciences., Mount Sinai, US

BUCHANAN JAMES

Queen Mary University of London

CAPPS BENJAMIN

Associate Professor, Department of Bioethics Faculty of Medicine, Dalhousie University, Halifax, Canada

CARNINCI PIERO

Head of Genomics Research Centre-Functional Genomics Programme, Human Technopole (Italy), Team Director, RIKEN IMS (Japan)

CHATZI LIDA

Professor of Population & Public Health Sciences, Director for USC Center on Translational Exposomics Research, Director for USC Superfund Research & Training Program for PFAS Assessment, Remediation & Prevention, Deputy Director of the Southern California Environmental Health Sciences Center, University of Southern California, USA

CHEN DAWN

Ph.D., Professor Institute of Biochemistry and Cell Biology, Shanghai Institutes for Biological Sciences, Chinese Academy of Sciences, 320 Yue-yang Road, Shanghai 200031, China

CHEN LING-LING

Ph.D., Professor Institute of Biochemistry and Cell Biology, Shanghai Institutes for Biological Sciences, Chinese Academy of Sciences, 320 Yue-yang Road, Shanghai 200031, China

CRAWLEY FRANCIS P.

Philosopher, Research Ethics & Integrity, Global Data and AI Ethics, Policy, and Governance Leuven Belgium

DI DONATO GUIDO WALTER

CTO of GenomSys and CEO of GenoGra

EU-AHSUNTHORNWATTANA JAKRIS

Lecturer in the Department of Community Medicine at Faculty of Medicine Ramathibodi Hospital, Mahidol University

FABELLA T.D**FAKIS GIANNOULIS**

Democritus University of Thrace, Greece





GAZOULI MARIA

National & Kapodistrian University of Athens,
Greece

GORANITIS ILIAS

Professorial Fellow, Health Economics, School
of Population and Global Health, University of
Melbourne, Australia

GUSTINCICH STEFANO

Coordinator of the Center for Clinical and
Computational Genomics - C3G – Istituto
Italiano di Tecnologia

HABIBA AL SAFAR

Associate Professor, Department of Genetics
and Molecular Biology; Associate Dean of
Student Affairs; & Director of the KU Center for
Biotechnology, Emirates

HAMOSH ADA

American pediatrician and geneticist, Sutland
Professor of Genetics in the Departments of
Genetic Medicine and Pediatrics at the Johns
Hopkins University, US

HANIFFA MUZLIFAH

Head of the Cellular Genomics Programme and
Deputy Director of the Wellcome Sanger
Institute and Professor of Clinical Dermatology
at the University of Cambridge. She is also a
Wellcome Senior Clinical Research Fellow in
Clinical Science, and a Senior Research Fellow
at Churchill College, University of Cambridge,
Wellcome Sanger Institute, UK

JUERGEN REICHARDT

Adjunct Professor, Australian Institute of
Tropical Health and Medicine, James Cook
University, Australia

KARAMITROS TIMOKRATIS

Hellenic Pasteur Institute, Athens, Greece

KIM HIE LIM

Associate Professor, Centre for Environmental
Life Sciences Engineering, Nanyang
Technological University (NTU), Singapore

KUHN ROBERT

Associate Director with the UCSC Genome
Browser team at the UC Santa Cruz Genomics
Institute, Santa Cruz, USA

KUPERS STEF

Strategic Segment Manager

LAGOUMINTZIS GEORGE

Assistant Professor, Biochemistry-Molecular
Biology, Department of Pharmacy, Division of
Pharmacology-Biosciences, Lab of Molecular
Biology and Immunology, Patras, Greece

LANER ANDREAS

PhD Head of Department at Medical Center of
Genetics

LEE CHARLES

The Jackson Laboratory, US

LEHR THORSTEN

Professor of Clinical Pharmacy, Saarland
University, Germany

LIU WEIBIN

Vice President, BGI Group, China

LO TEMPIO JR. JONATHAN

Professor of Pediatrics and Biological
Chemistry, University of California, Irvine, USA

LUMAKA AIME

Professor of Genetics, Centre for Human
Genetics - Department of Pediatrics, University
of Kinshasa, University of Kinshasa, DR Congo

MAJUMBER PARTHA

Professor, National Institute of Biomedical
Genomics, New Delhi, India

MAROULAKOU IOANNA

Democritus University of Thrace,
Alexandroupoli, Greece

MASCALZONI DEBORAH

Associate Professor in Biomedical Ethics, ELSI
Research Group Institute of Biomedicine,
Bolzano, Italy

MIGA KAREN

Associate professor of Biomolecular
Engineering at the University of California,
Santa Cruz, and Associate Director at the UC
Santa Cruz Genomics Institute.





MITROPOULOU CHRISTINA

Adjunct Asst. Professor, United Arab Emirates University, College of Medicine and Health Sciences, Department of Genetics and Genomics, Al-Ain, Abu Dhabi, UAE, Managing Director and Senior Health Economist, The Golden Helix Foundation, London, UK

NDIAYE ROKHAYA

Professor in Human Genetics, Faculty of Medicine, Pharmacy & Odontology, University Cheikh Anta Diop, Senegal

NOVELLI GIUSEPPE

Head of the Human Genetics Research Unit at the Tor Vergata University of Rome and President of Lorenzini Foundation, Milan, Italy

O'DONNELL-LURIA ANNE

Co-Director of the Center for Mendelian Genomics (CMG) at the Broad Institute of MIT and Harvard, Attending Physician in Genetics and Metabolism, Boston Children's Hospital, Associate Professor, Harvard Medical School, USA

PATRINOS GEORGE P.

Director General and Director of Research, Hellenic Pasteur Institute, Professor of Pharmacogenomics and Personalized Therapy, Department of Pharmacy, University of Patras

PAVLIDIS PAVLOS

[Bioinformatics], Institute of Computer Science, ICS-FORTH, University of Crete, Greece

QUACKENBUSH JOHN

Professor of Computational Biology and Bioinformatics Biostatistics, Harvard T.H. Chan School of Public Health, Harvard University, USA

RASMUSSEN SONJA

Professor in the Department of Genetic Medicine at Johns Hopkins School of Medicine

REHM HEIDI

Professor of Pathology, Massachusetts General Hospital; Broad Institute of MIT and Harvard, USA

REPETTO GABRIELA

Clinical Geneticist, Professor of Genetics, Facultad de Medicina, Clinica Alemana Universidad del Desarrollo, Santiago, Chile

RIGOUTSOS ISIDORE

Richard W. Hevner Professor of Computational Medicine, Computational Medicine Center, Thomas Jefferson University

SARIGIANNIS DENNIS

Prof., PhD, Director and President of the Board, National Hellenic Research Foundation

SCHMIDT HARALD

Maastricht University, The Netherlands

SHIU KAI-KEEN

Kai-Keen Shiu PhD MRCP Consultant Medical Oncologist Gastrointestinal Oncology Service, Clinical Lead for Cancer of Unknown Primary Clinical Lead for the Acute Oncology Service, Deputy Chair NCRI CUP Working Group University College London Hospital and UCL Cancer Institute, UK

SNYDER MICHAEL

Stanford W. Ascherman Professor, Department of Genetics, Director, Center for Genomics and Personalized Medicine, School of Medicine, Stanford University, USA

SQUASSINA ALESSIO

Associate Professor of Pharmacology, Chief of the Laboratory of Pharmacogenomics of the Department of Biomedical Sciences, University of Cagliari, Italy

SUPRIADI INDRI ROOSLAMIATI

Director, Center for Biomedical and Health Genomics, Ministry of Health, Indonesia

TOBIAS EDWARD

Professor of Genetic Medicine (Medicine) consultant clinical geneticist in the NHS and is an active invited member of several international genomics committees. BSc (1st class Hons), MBChB, MRCP, PhD (Molecular Biology), FRCP, FHEA, FAoP (Assoc. of Physicians), University of Glasgow, UK





UITTERLINDEN ANDRE

Em. Professor Complex Genetics, The Netherlands

VALSECCHI CRISTINA

Sr. Technical Sales Specialist- Human Predictive Genomics - Italy, Iberia, Israel and South East Europe

VASILIOU VASILIS

Department Chair and Susan Dwight Bliss Professor of Epidemiology (Environmental Health Sciences) and of Ophthalmology and Visual Science and of Environment Director, Yale Superfund Research Center, Affiliated Faculty, Yale Cancer Center, Affiliated Faculty, Yale Institute for Global Health; Co-Director, Environmental Health Sciences Track, Executive MPH, Yale University, USA, Yale University, USA

VASILOPOULOS IOANNIS

University of Patras, Greece

VELTMAN JORIS

Principal investigator, University of Edinburgh

VRAHATIS ARIS

Ionian University, Greece

WAN YUE

Executive Director, A*STAR Genome Institute of Singapore

YAPIJAKIS CHRISTOS

National & Kapodistrian University of Athens, Greece





Take the 3D tour

Confidence at scale for modern genomics laboratories. **Scan to explore the SwiftArrayStudio solution in 3D.**

Next-day human genotyping starts here

Automated microarray processing

For pharmacogenomics, population studies, and large-scale genotyping programs, the Applied Biosystems™ SwiftArrayStudio™ solution is the only automated microarray platform built specifically to help laboratories deliver reliable, high-throughput human genotyping—fast.

Why use the SwiftArrayStudio solution?

- **Next-day turnaround**—sample to report in as little as 30 hours
- **True walk-away automation**—help minimize variability and staffing burden
- **High throughput**—up to 1,536 samples per week on a single system
- **Built for reliability**—durable design with intelligent software monitoring
- **Flexible human genotyping assays**—including Applied Biosystems™ Axiom™ PharmacoPro™ and Axiom™ PangenomePro™ array

Learn more at thermofisher.com/swiftarraystudio

applied biosystems



Oxford
Nanopore
Technologies



Bringing innovation in genetic diagnostics

Proud Sponsor of HUMAN GENOME MEETING 2026

Human Genomics and Sequencing Innovation

Oxford Nanopore enables researchers to explore all forms of human genetic variation — genomic, epigenomic, and transcriptomic — in real time.

- Ultra-long reads (>4 Mb) for complex regions
- Detect SNVs, STRs, and Structural Variants
- Direct methylation detection
- Devices: MinION to PromethION 24
- Real-time sequencing & adaptive sampling



Applications: rare disease genomics, RNA sequencing, epigenetic profiling.



ELTA90 MGR representing Oxford Nanopore
office@elta90mgr.gr | www.elta90mgr.gr | +30 2310483100

BioLegend

From Revvity



P. Zafiropoulos s.a.

Diagnostica

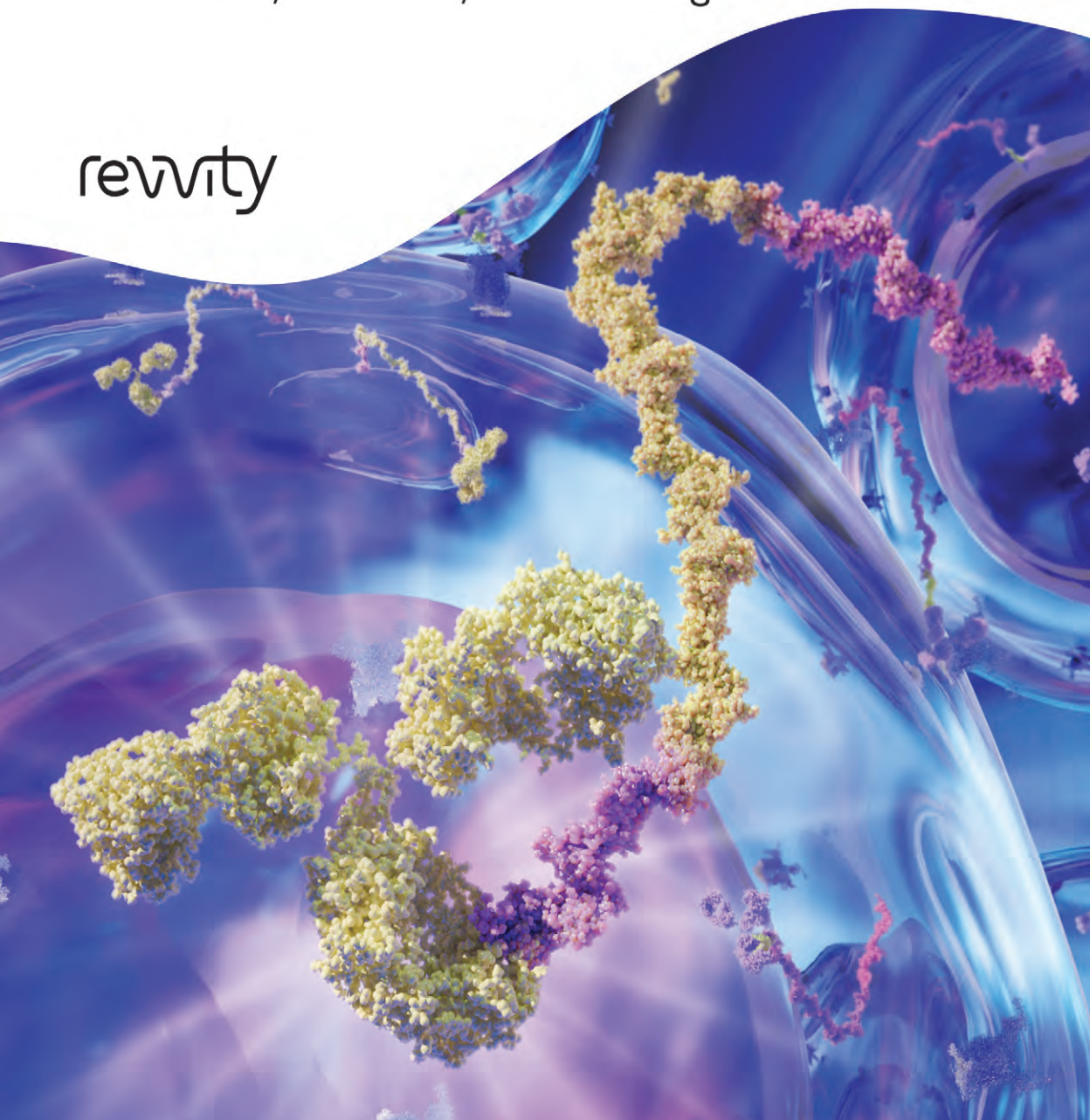
Advancing Life Sciences - Innovating Health Care™

www.pzafiropoulos.gr

Multiomics with TotalSeq reagents

Antibodies, cocktails, and hashtags

revvity



THE FULL STACK FOR NGS DATA



GenomSys provides ISO-standard genomic data management and variant analysis; GenoGra brings pangenomic graph technology for accurate, scalable secondary analysis. Together, they cover the entire NGS workflow, from data to insight.

www.genomsys.com

www.genogra.com

ACKNOWLEDGEMENTS

The Organizing Committee of the **HGM 2026**, wishes to acknowledge the following companies for their significant contribution.

CO-ORGANIZING ENTITIES



GOLD SPONSORS



SILVER SPONSORS



SPONSORS



SUPPORTED BY

