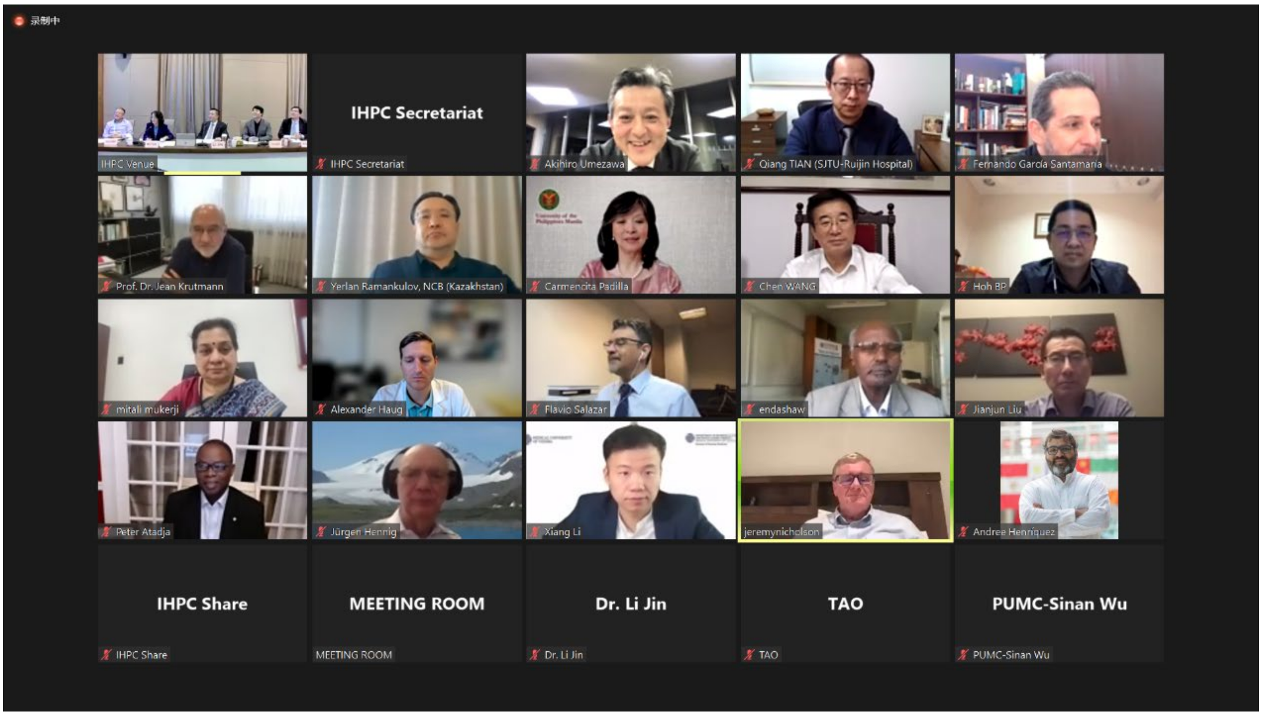
# Newsletter of the International Human Phenome Consortium

Issue 1 | February 2022

International Human Phenome Institutes(Shanghai)

## In this Issue:

# WELCOME NEW MEMBERS



The 3rd board meeting of the IHPC was held online on November 19th. Over 30 IHPC board members and invited scholars have participated in the conference to discuss the next move for the Human Phenome Project (HPP). The first preliminary Human Phenome Navigation Atlas was made public at the meeting. Through deep conversations among board members and invited scholars, all participants have reached a consensus on the Principles of Trans-Border Data Sharing and Openness of the Human Phenome Project. At the meeting, three outstanding experts were elected as new members of the board. With Professor Marcus Hacker, Professor Hoh Boon-Peng and Professor Flavio Salazar Onfray joining the IHPC board, we have now reached 24 board members of IHPC from 20 different countries across 6 continents. Brief introductions of our new members are provided as follows:

## Professor Marcus Hacker

Full Professor/Director, Division of Nuclear Medicine, Medical University of Vienna, Austria
Deputy Chair, Department of Biomedical Imaging and Image-guided Therapy, Medical University of Vienna, Austria

Professor Hacker is a nuclear medicine specialist who has completed his medical studies in 1998 at University of Erlangen-Nuremberg and University of Munich, Germany. In 2004, He received a degree as a specialist in nuclear medicine. Since 2013, He has become a full professor for nuclear medicine, director of Division of Nuclear Medicine, and deputy chair of the Department of Biomedical Imaging and Image-guided Therapy at the Medical University of Vienna. Professor Hacker has the highest level of professional skills in nuclear medicine diagnostics and therapy and has published more than 250 Original Manuscripts and Reviews. He also possesses particular expert knowledge in oncological, cardiovascular, and neuropsychiatric diseases. He won the Dagmar-Eißner-Award for Nuclear Medicine in 2006 and Wolfgang-Becker-Award for Nuclear Medicine in 2007.

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## Professor Hoh Boon-Peng

Full Professor, Faculty of Medicine and Health Sciences, UCSI University, Malaysia
Director, Belt-And-Road Joint Research Centre for Human Phenomics, Malaysia

Professor Hoh Boon-Peng is a molecular population geneticist and a trained Biorisk Professional in Malaysia. He obtained his Ph.D. from Universiti Putra Malaysia. Since 2016, He has become a full professor at UCSI University and his research interests include human genome variation and population genetics. Professor Hoh seeks to address the fundamental question of the functions of genomic variation and how its interaction with the environment is shaping the human traits and complex diseases, ultimately to contribute to the approach of precision medicine. He was the chairman of the Institutional Biosafety Committee for UCSI University. Professor Hoh’s laboratory was recognized by the Ministry of Health Malaysia as a laboratory to assist the government for COVID-19 diagnostic test. In 2013, Owing to his outstanding academic achievement, he was awarded with the JCI "Ten Outstanding Young Malaysian" under the category of Academic Achievement & Accomplishment.

## Professor Flavio Salazar Onfray

Full professor and Vice president of research and development, Institute of Biomedical Sciences, Faculty of Medicine, University of Chile, Chile
Deputy Director, Millennium Institute on Immunology and Immunotherapy (MIII), Chile

Professor Flavio Salazar Onfray is a biologist in Chile and his major interest has been focused in several aspects of tumor immunology, from basic research to clinical trials. He obtained his Ph.D. at the Karolinska Institute in Sweden in 1998. He founded two spinoff companies—Oncobiomed and Bionex—dedicated to cellular immunotherapy technology transfer and advice basic scientist in the design and writing of R&D applications. For more than 15 years, Professor Salazar has organized and led a multidisciplinary group of scientists including physicians, specialists in oncology and immunology, biologists and biochemists, to build the first GMP facility for manipulation of human cells and performed the first Phase I and Phase I/II clinical trial of immunotherapy based on dendritic cells for the treatment of melanoma and prostate cancer in Chile. In 2007, Professor Salazar was awarded "The best paper of the decade" by The Disciplinary Program of Immunology. He also won the AVONNI Award "the outstanding innovator of the year" by the Forum for the innovation from the Ministry of Economy and the private sector in both 2007 and 2008.

# HIGHTLIGHTS AND RECENT NEWS

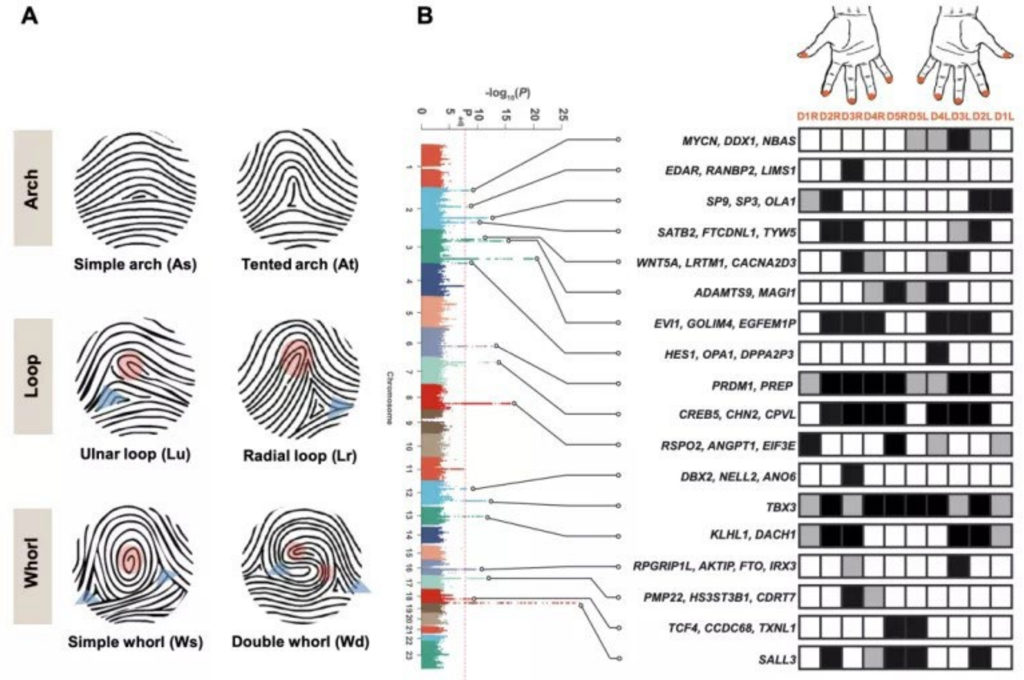
## HIGHLIGHTS

### Cell: Fingerprint Patterns Are Linked to Limb Development Genes

In the most comprehensive analysis to date, researchers found that the shapes of fingerprints—whether they are circular, wavy, or winding—are influenced by the genes responsible for limb development instead of skin patterning. The study, presented January 6 in the journal Cell, could help scientists better understand the association between genes and phenotypical traits in humans.

"The study, as a good example in human phenome research, demonstrates the significant role of human phenomics in the development of life sciences," said Li Jin, CAS academician, president of Fudan University, dean of International Human Phenome Institute (Shanghai), Board Member and Co-founder of IHPC and co-senior author on the paper.

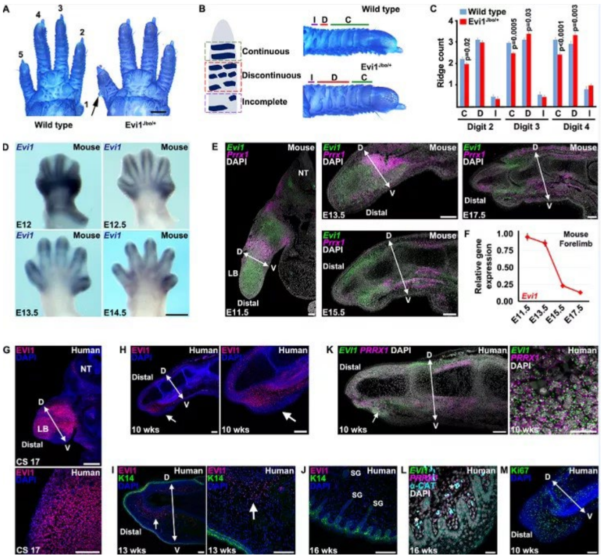
While fingerprints are unique to individuals, they are generally categorized into three types: arch, loop, and whorl. These furrows and ridges begin to form on a fetus' fingers and toes after the third month of pregnancy. Scientists have suspected that fingerprints have potentially evolved to help grab objects and sense their textures, but exactly how these patterns are formed remains unknown.

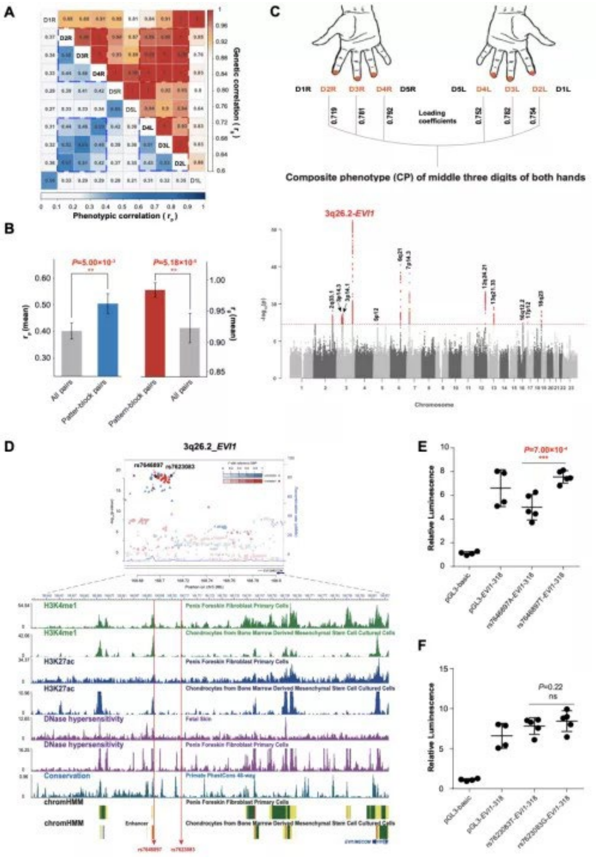


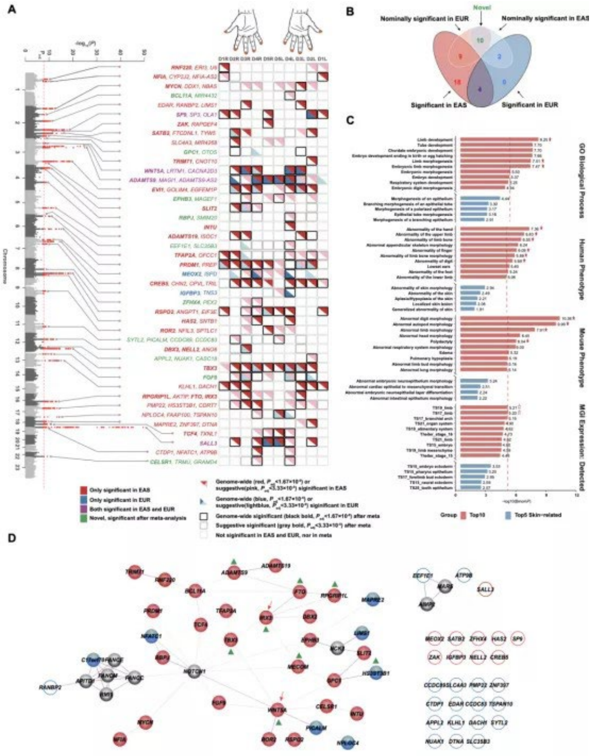
"We started the work purely out of curiosity. But later it turned out fingerprint pattern is associated with genes for limb growth, which are critical for fetal development," said Sijia Wang, a geneticist at the Shanghai Institute of Nutrition and Health, of Chinese Academy of Sciences, and co-senior author on the paper, "This provides another classic example of pleiotropy, when multiple phenotypes are interrelated to each other and are affected by the same genes."

Wang and colleagues scanned the DNA of more than 23,000 people across ethnic groups and found that at least 43 regions on the genome are associated with fingerprint patterns. One of the most influential regions appeared to be regulating the expression of a gene called EVI1, which is known for its role in embryonic limb development.

To test their finding, the team modified the DNA of mice so their expression of EVI1 was turned down. They found that mice with downregulated EVI1 developed abnormal skin patterns on their digits compared with wild-type mice.







Analysis of human data revealed that fingerprint patterns are genetically correlated with finger length. For example, people with whorl-shaped fingerprints on both of their little fingers tend to have longer little fingers than those who do not, and this correlation is strongly linked to genes involved in limb development.

“We don’t know exactly how the genes shape fingerprint patterns, but it could be determined by the amount of strength from growth that’s put on an embryonic tissue called volar pads that plays an important role in the formation of different patterns of fingerprint,” says Jinxi Li, a postdoctoral researcher at the Human Phenome Institute at Fudan University in Shanghai, and co-first author on the paper. She explains that as a fetus’ hands grow, the palms and fingers would stretch and elongate. These forces could turn a whorl into a loop, for example.

Notably, previous research has suggested that EVI1 is linked to risk of leukemia, and some studies have observed that people with more whorl patterns are more susceptible to the disease, Wang says.

The research is part of the Human Phenome Project. Next, the team plans to conduct more research on how dermatoglyphic patterns are related to diseases and the underlying pleiotropic mechanism.

Link of the Article:

Cell, Li, Glover, Zhang, and Peng et al.: “Limb development genes underlie variation in human fingerprint patterns,” https://www.cell.com/cell/fulltext/S0092-8674(21)01446-X DOI: 10.1016/j.cell.2021.12.008

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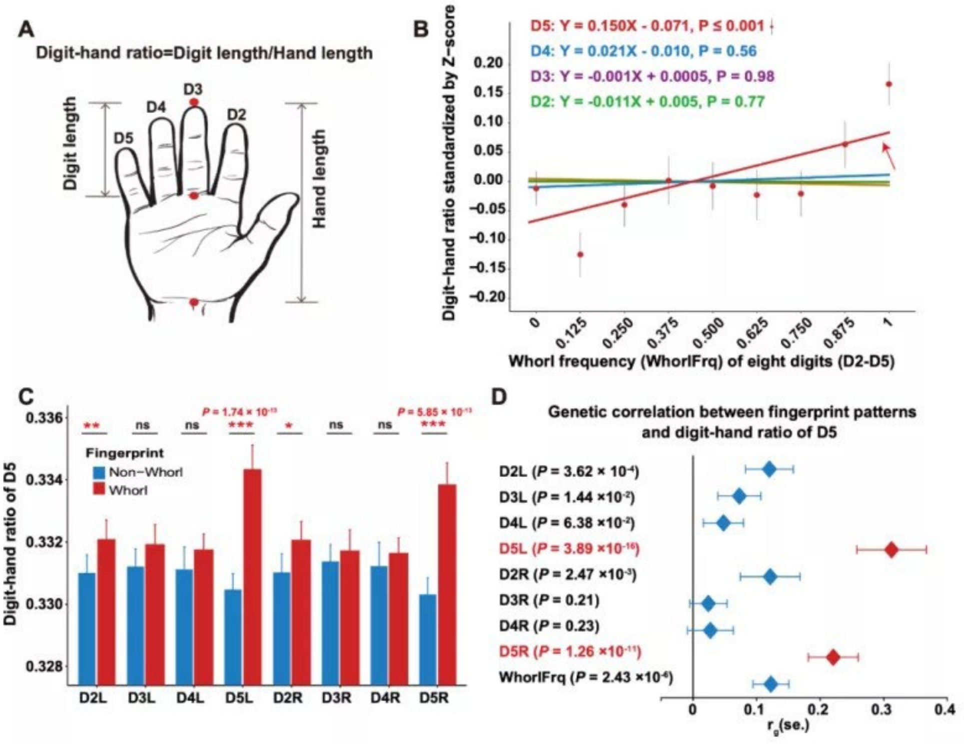
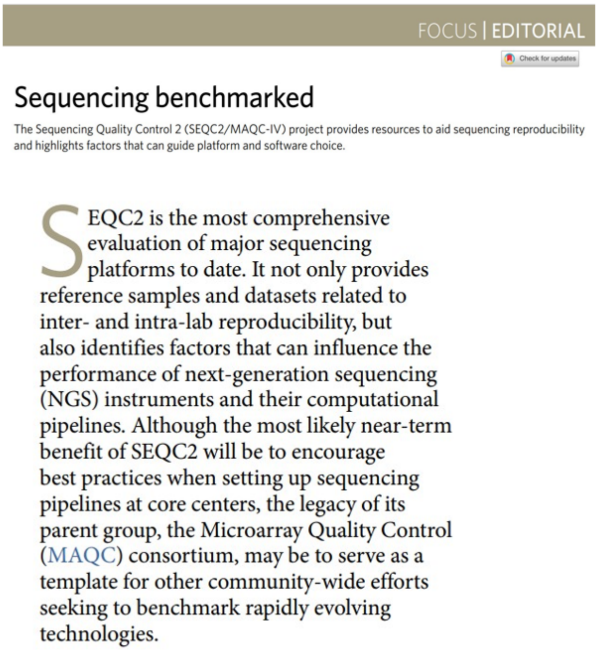


## SEQC2/MAQC-IV Project: The Most Comprehensive Evaluation of Major Sequencing Platforms to Date

The *MicroArray and SEquencing Quality Control (MAQC/SEQC) Project* is helping improve the microarray and next-generation sequencing (NGS) technologies and foster their proper applications in discovery, development and review of FDA regulated products. The MAQC consortium is the US FDA-led community wide consortium effort to address reproducible issues relating to the application of constantly evolving high-throughput genomics technologies to either assess safety and efficacy of FDA regulated products or their safe and effective use in clinical applications as in vitro diagnostic devices. The MAQC consortium completed three projects between 2005-2014 (namely *MAQC I, II and III*), resulting in ~30 publications. Its fourth project, also called the *SEQC2 Project* was completed in 2021. The primary objective of the *SEQC2/MAQC-IV* project is to develop standard analysis protocols and quality control metrics for fit-for-purpose use of NGS data to enhance regulatory science research and precision medicine.

The human phenomics research team from Fudan University (China) has taken a significant part in the *SEQC2/MAQC-IV Project*. The team has participated and led the publication of two out of five papers published the September 2021 issueof *Nature Biotechnology*. The first paper, titled "Establishing community reference samples, data and call sets for benchmarking cancer mutation detection using whole-genome sequencing", aimed at establishing community reference samples, data and call sets for benchmarking cancer mutation detection using whole-genome sequencing. The other paper titled "Toward best practice in cancer mutation detection with whole-genome and whole-exome sequencing" has reported a systematic interrogation of somatic mutations in paired tumor-normal cell lines to identify factors affecting detection reproducibility and accuracy at six different centers.

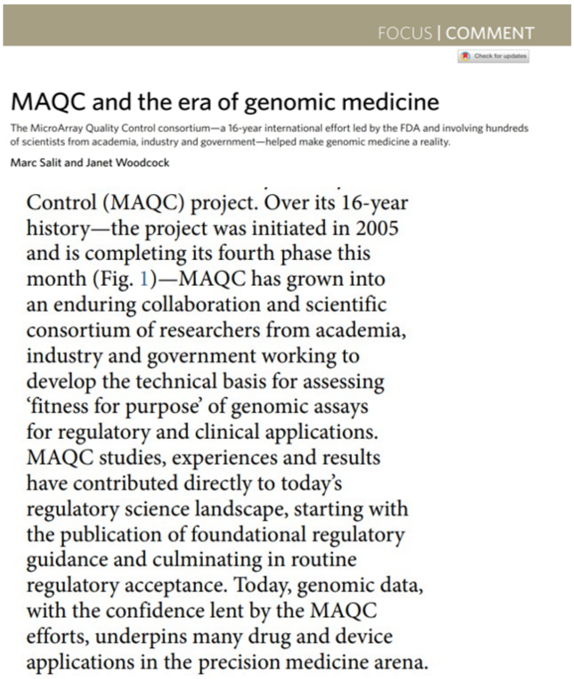
The Editorial described the *SEQC2* project as the most comprehensive evaluation of major sequencing platforms to date. In addition, Dr. Marc Salit, a professor at Stanford University, and Dr. Janet Woodcock, Acting FDA Commissioner, commented that "MAQC’s production of high-quality science and publications has provided a substrate both for regulatory use and in the clinic, moving the entire field forward and enabling widespread adoption of the new technologies." Importantly, they emphasized the unmet need of reference "samples authoritatively characterized at the genome scale … for quantitative properties" to "allow us all to do better genome-scale biology, to better consider hypothesis testing, and to develop better hypotheses and chase fewer (costly) wild geese."



**Sequencing benchmarked**

The Sequencing Quality Control 2 (SEQC2/MAQC-IV) project provides resources to aid sequencing reproducibility and highlights factors that can guide platform and software choice.

SEQC2 is the most comprehensive evaluation of major sequencing platforms to date. It not only provides reference samples and datasets related to inter- and intra-lab reproducibility, but also identifies factors that can influence the performance of next-generation sequencing (NGS) instruments and their computational pipelines. Although the most likely near-term benefit of SEQC2 will be to encourage best practices when setting up sequencing pipelines at core centers, the legacy of its parent group, the Microarray Quality Control (MAQC) consortium, may be to serve as a template for other community-wide efforts seeking to benchmark rapidly evolving technologies.



**MAQC and the era of genomic medicine**

The MicroArray Quality Control consortium—a 16-year international effort led by the FDA and involving hundreds of scientists from academia, industry and government—helped make genomic medicine a reality.

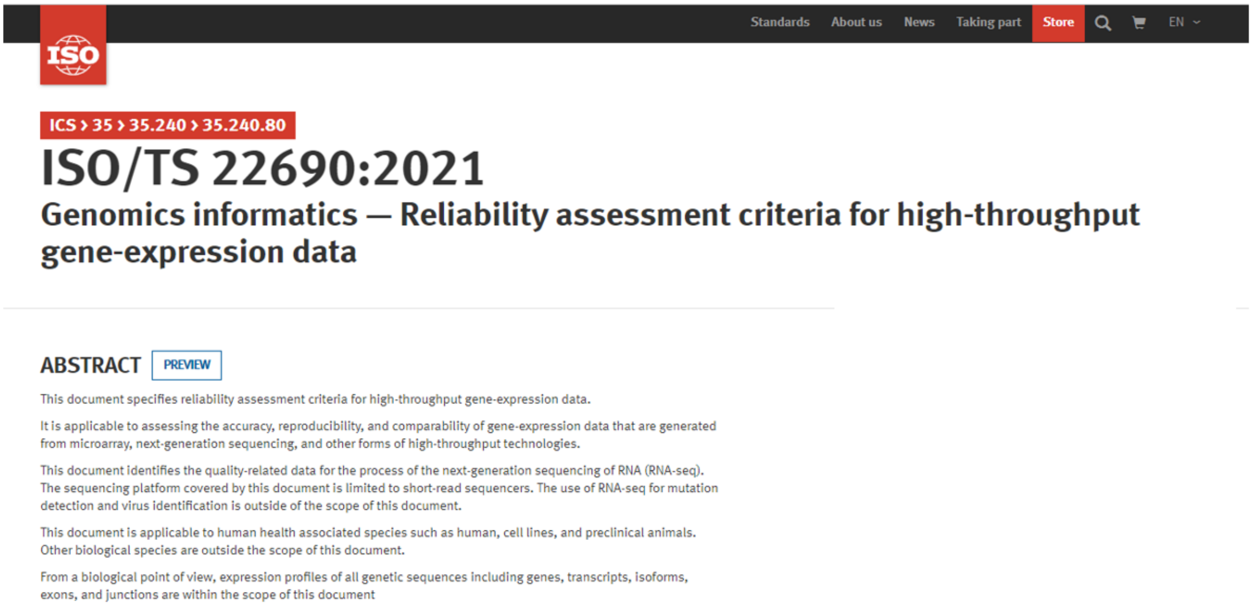
Marc Salit and Janet Woodcock

Control (MAQC) project. Over its 16-year history—the project was initiated in 2005 and is completing its fourth phase this month (Fig. 1)—MAQC has grown into an enduring collaboration and scientific consortium of researchers from academia, industry and government working to develop the technical basis for assessing 'fitness for purpose' of genomic assays for regulatory and clinical applications. MAQC studies, experiences and results have contributed directly to today's regulatory science landscape, starting with the publication of foundational regulatory guidance and culminating in routine regulatory acceptance. Today, genomic data, with the confidence lent by the MAQC efforts, underpins many drug and device applications in the precision medicine arena.

MAQC/SEQC FDA website: https://www.fda.gov/science-research/bioinformatics-tools/microarraysequencing-quality-control-maqcseqc

Nature Biotechnology: https://www.nature.com/nbt/volumes/39/issues/9

## Professor Leming Shi: Initiator of International Standards on Data Quality Control



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**ISO/TS 22690:2021**

**Genomics informatics — Reliability assessment criteria for high-throughput gene-expression data**

**ABSTRACT**

This document specifies reliability assessment criteria for high-throughput gene-expression data.

It is applicable to assessing the accuracy, reproducibility, and comparability of gene-expression data that are generated from microarray, next-generation sequencing, and other forms of high-throughput technologies.

This document identifies the quality-related data for the process of the next-generation sequencing of RNA (RNA-seq). The sequencing platform covered by this document is limited to short-read sequencers. The use of RNA-seq for mutation detection and virus identification is outside of the scope of this document.

This document is applicable to human health associated species such as human, cell lines, and preclinical animals. Other biological species are outside the scope of this document.

From a biological point of view, expression profiles of all genetic sequences including genes, transcripts, isoforms, exons, and junctions are within the scope of this document

Scientists have widely recognized the low reproducibility in omics research. The complexity and massiveness of omics data make the manual inspection of data quality and analysis results impossible. Thus, quality control processes for high-throughput gene-expression experiments are essential for improving the reproducibility of biological results. The international standard published in October 2021, ISO/TS 22690, "Genomics Informatics -- Reliability Assessment Criteria for High-throughput Gene-expression Data", was developed by a group of experts led by Professor Leming Shi from the Human Phenome Institute of Fudan University (China). Professor Shi was the first Secretary-General of the IHPC and the initiator of the MicroArray and Sequencing Quality Control (MAQC/SEQC) project. This standard aims to help remove the barriers in the generation, analysis, and interpretation of high-throughput expression profiling data so as to prevent the production of irreproducible or inconsistent scientific results. Assessment criteria and proficiency tests with well-characterized RNA reference samples are introduced in the standard to evaluate the reliability and reproducibility of the data for quality control. ISO/TS 22690 is underpinned by the research outcomes from projects, especially MAQC/SEQC, that provide a collection of quality metrics for expression data evaluation.

It has been stated in the document that the ISO/TS 22690 can be utilized to:

(i) enhance community's understanding of the technical performance of high-throughput gene expression;

(ii) benefit the interoperability of qualified gene-expression data by researchers, commercial entities and regulatory bodies;

(iii) improve the application of high-throughput gene expression in industry and clinics;

(iv) promote the acceptance of transparent reporting according to the FAIR (findable, accessible, interoperable, and reusable) data principles; and

(v) contribute to the development of precision medicine.

Moreover, Fudan University has developed the Quartet multiomics reference materials of DNA, RNA, proteins, and metabolites simultaneously manufactured from the same batch of cultured immortalized B-lymphoblastoid cell lines of the four members of a Chinese Quartet family from the Fudan Taizhou Cohort. Over 100 international and domestic users have adopted the reference material for data quality control, including European Infrastructure for Translational Medicine (EATRIS). The Quartet Reference Materials suite is intended for quality control and performance assessment of each omics profiling and integration of the resulting multiomics data.

The Quartet Project Official Website: http://chinese-quartet.org/

### Australian National Phenome Centre (ANPC) : Molecular Phenomic Approaches to Human Health

Last year, Professor Jeremy Nicholson decided that the Australian National Phenome Centre (ANPC) should shift focus to deal with the world's emerging health crisis, COVID-19. Since then, the lab has made huge progress in understanding the disease based on samples collected from around the world. Since last October, he and his team have had 15 papers published or accepted for publication on COVID-19, covering novel diagnostics, disease mechanisms, and new ways of assessing Post-acute COVID-19 Syndrome.

Professor Nicholson has also published a review article on Phenomics Journal, titled "Molecular Phenomic Approaches to Deconvolving the Systemic Effects of SARS-CoV-2 Infection and Post-acute COVID-19 Syndrome". The article introduces a multiplatform metabolic phenotyping approach to studying pathology and systemic metabolic sequelae of COVID-19, together with a framework for assessing post-acute COVID-19 Syndrome (PACS) that is a major long-term health consequence for many patients. Professor Nicholson has stated in the article that to fight against the disease in "The COVID-19 War", an important future determinant will be the ability and agility to adapt healthcare policy to changes in the virus structure and biological properties, and metabolic phenotyping can be a vital tool for the molecular investigations and large-scale screening.

## RECENT NEWS

### The 3rd Board Meeting of the International Human Phenome Consortium (IHPC)

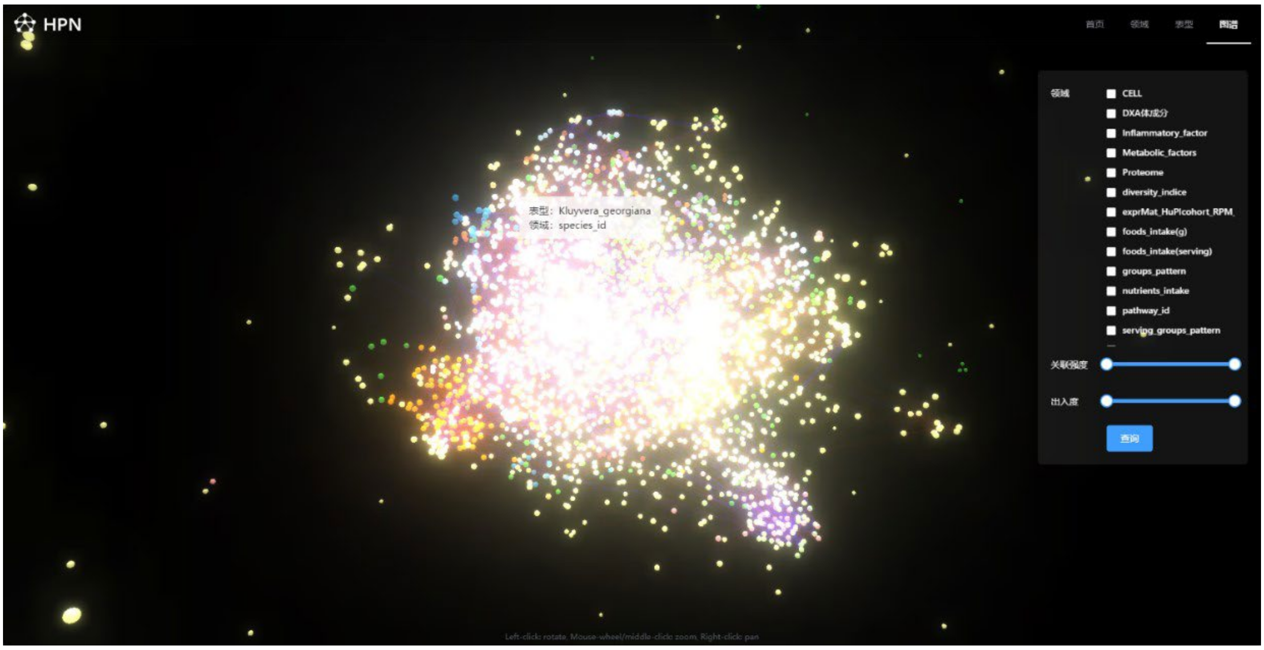
The 3rd board meeting of the International Human Phenome Consortium (IHPC) was held online on November 19, 2021. Over 30 scholars from 19 countries joined the conference to discuss the next move for the Human Phenome Project (HPP).

According to the vote of IHPC board meeting, Professor and Director of Division of Nuclear Medicine at the Medical University of Vienna Marcus Hacker, Professor at Faculty of Medicine and Health Sciences at UCSI University Hoh Boon-Peng, and Vice President at the University of Chile and Professor at the Millennium Institute of Immunology and Immunotherapy Flavio Salazar Onfray were elected as members of the board. With scholars from 24 institutions of 20 countries across 6 continents now, IHPC has become a diverse global community committed to collaborative innovation on human phenome studies

As the IHPC co-founders, Leroy Hood, founder of Institute for Systems Biology, USA, Jeremy Nicolson, executive director of the Australian National Phenome Centre, Australia, and Li Jin, president of Fudan University & director of International Human Phenome Institutes (Shanghai), China made keynote speeches on the latest progress of HPP over the past year.

The draft entitled “Principles of Trans-Border Data Sharing and Openness of the Human Phenome Project”, introduced by Jean Krutmann, director of IUF-Leibniz Research Institute for Environment Medicine, Germany, on behalf of Subcommittee of data sharing was passed as academic consensus by the board meeting unanimously.

President of National Research Institute for Child Health and Development Akihiro Umezawa and Professor at Indian Institute of Technology Jodhpur Mitali Mukerji shared their latest research findings and plans respectively. Professor Umezawa focused his talk on establishment of disease models using human iPS cell-organoids for genetic disorders. Prof. Mukerji introduced “Thar DESIGNS” research project in desert ecosystem sciences promoted by her institute.



At the meeting, Chen Ding, executive editor of the journal Phenomics and deputy dean of Human Phenome Institute at Fudan University, addressed the board on the development of this journal in the past year. According to Ding, Phenomics has received 70 contributions from 10 countries, and has published 6 issues with 25 high-quality academic papers. It has become an important platform for scientists around the world to exchange ideas and latest findings in phenomics. Mei Tian, secretary-general of IHPC, deputy director of International Human Phenome Institute (Shanghai) and distinguished professor at Fudan University presided over the meeting.

### Fudan Takes the Lead in Drawing the World's First "Navigation Atlas" for Human Phenome

Phenotype is the biological trait in a living creature, while phenome is the assembly of the phenotypes at both the micro and macro levels throughout the creature's life. Genes (internal factor) and environment (external factor, including exposure to environment and way of living) jointly determine a creature's phenotypes. Adhering to the principle of 'measure what is measurable', HPP aims to measure human phenotypes on a cross-scale and in a full cycle to analyze the relationship between genome-phenome-environment as well as the relationship between macro-phenotypes and micro-phenotypes, so that scientists can better understand how phenotypes may herald certain health conditions or diseases.

The international science community now shares the understanding that human phenome is a critical cutting-edge area and meta-innovation source, and that HPP will profoundly change the landscape of life sciences and lead a paradigm change in this field.

The primary goal of HPP is to draw the world's first "navigation atlas" for human phenome which illustrates the correlations between tens of thousands of different human phenotypes. By following the atlas, scientists can greatly improve the efficiency of making innovative achievements in life sciences and deepen understanding of various life phenomena. The atlas will also provide new directions and valuable clues for scientists to further analyze the mechanism of complex living systems in the future.

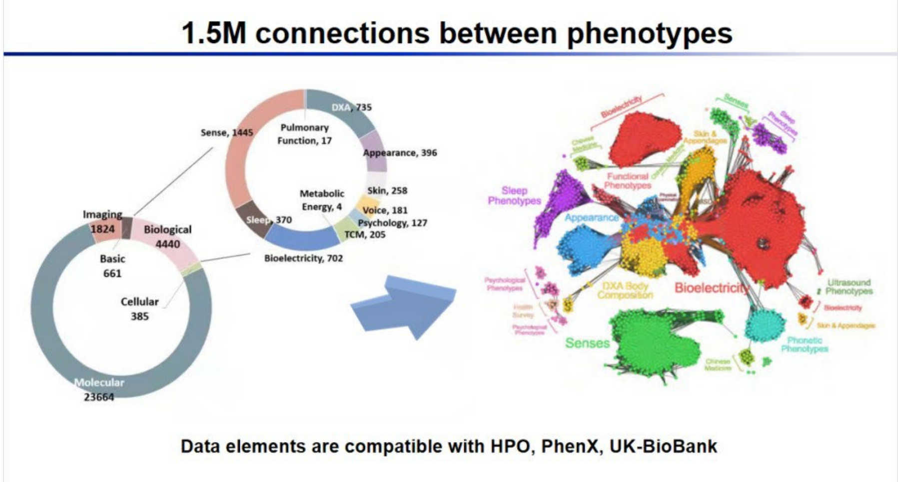
At the 3rd IHPC board meeting, Prof. Li Jin elaborated on the exploratory research on this "navigation atlas" by Chinese scientists. According to Jin, Chinese scientists have drawn a preliminary "navigation map" of human phenome covering and networking more than 1.5 million strong associations among different phenotypes based on Shanghai core cohort studies on residents' health-related phenotypes.

With the help of the world's first cross-scale and multi-dimensional phenotyping platform and the automatic full-process big-data phenome platform built in Zhangjiang Fudan International Innovation Center, as of November 19, 2021, more than 730 volunteers aged 20-60 who have lived in Shanghai for years have participated in the cohort study. Each volunteer had stayed for 2 days in phenotyping platform and contributed their data from more than 30,000 phenotypes per person. This enables scientists to obtain more than 3PB phenotypic baseline data in more than 20 categories.

Through the analysis of existing data by multidisciplinary teams, scientists eventually have found more than 1.5 million strong associations, of which about 39% are cross-scale associations, and most of the associations were discovered for the first time. Networking these strong associations, the world first preliminary "navigation atlas" of human phenome has been achieved.

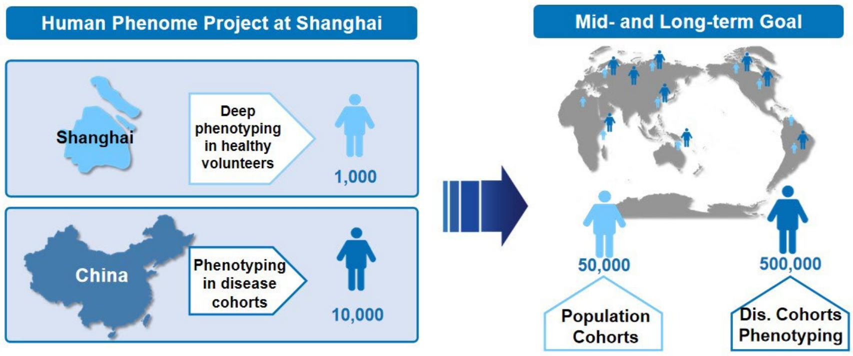
The "navigation atlas", which is now at the beta testing stage, has become available for scientists and researchers. Many researchers are using this map to identify the strong associations of great significance in lab research and practical use.

IHPC Consensus on the Principles of Trans-Border Data Sharing and Openness of the Human Phenome Project



**1.5M connections between phenotypes**

**Data elements are compatible with HPO, PhenX, UK-BioBank**



HPP will carry out phenotypic measurements of 50,000 people from different continents with panoramic measurement of more than 100,000 phenotypic indicators per person as well as a specialized study targeting specific phenotypes of over 500,000 people across the world. It's essential to promote sharing and openness of global scientific research data across borders to achieve this goal, as it is impossible for one team or country to complete this project without collaboration worldwide.

Upholding the principle of security and openness, the members of International Human Phenome Consortium (IHPC) agree at the meeting on the following consensus on principles of trans-border sharing and openness of scientific research data of human phenome.

First, the FAIR Principles. The IHPC members shall follow the "FAIR principles" adopted by the world's major scientific data-sharing institutions—"Findability, Accessibility, Interoperability, and Reusability". The human phenome research data for trans-border sharing shall be guided by the FAIR principles.

Second, security and compliance. The collection, transmission, preservation, use, sharing, publication, and destruction of scientific research data of the human phenome need to comply with laws and regulations in countries/regions as they concern the different stages of the entire data life cycle.

Third, data standardization. Data standardization is the basis for data sharing and joint research in the International Human Phenome Project. Through the cooperation on international standardization of research data, IHPC would like to facilitate the data sharing and open accessibility of International Human Phenome Project.

Fourth, classification and grading. Different types and levels of data are subject to different sharing and openness principles.  In the future, IHPC will contribute to the formation of international standards on human phenome data classification based on consultation and coordination among members.

Fifth, multi-model and scenario-based sharing. IHPC encourages scientific research institutions in various countries to work with a variety of innovative data sharing models in addition to the traditional model of data copying and sharing.

Sixth, follow best practices. IHPC expects scientific research teams to follow the rules and regulations for good scientific practice which are in place in the different countries and regions participating in IHPC.

Click the link below to learn more about Human Phenome Research Progress Report (2020)
https://hupi.fudan.edu.cn/en/content.jsp?urltype=news.NewsContentUrl&wbtreeid=1041&wbnewsid=1154

## Phenome Health (U.S)

In October 2021, Professor Leroy Hood announced the formation of Phenome Health, which is a non-profit organization created to advance a science and data-driven approach to optimizing the brain and body health of individuals. The organization has initiated the Beyond the Human Genome (BHG) project aiming to understand how to optimize the health trajectory of each individual through spending ten years to analyze the genomes and phenomes of one million people across the spectrum of human diversity within the U.S. The phenome of an individual includes all longitudinal measurements in humans, excluding genomic data. This approach provides robust new solutions for the four striking challenges of contemporary healthcare — quality, cost, an aging population, and exploding chronic diseases. BHG is supposed to cause an enormous paradigm shift in healthcare from disease-focused to prevention and wellness-focused vision, based on the concept of P4 Medicine came up by Professor Leroy Hood— predictive, preventive, personalized, and participatory.

Official Website: https://www.phenomehealth.org/

## The 3rd National Conference on Human Phenomics (China)

The Third National Conference on Human Phenomics was held in Chongqing, China. Academicians and experts gathered to discuss the major frontier innovations in the field of life sciences.

On September 24th, the most important frontier academic summit in the field of Phenomics in China - the third National Conference on Human Phenomics was held in Chongqing. The conference was jointly sponsored by International Human Phenome Institutes (Shanghai), Human Phenome Institute Fudan University, China Medical Device Industry Technology Innovation Strategic Alliance and Biophysical Society of China.

Focusing on the theme of "Phenomics and precision medicine", the conference attracted 7 academicians, as well as more than 100 experts, scholars and industry leaders from across the country to gather in Chongqing to share and discuss the latest achievements of Phenomics and precision medicine, and seek future research directions and technological solutions for major human health problems.

Professor Li Jin (Academician of the Chinese Academy of Sciences, President of Fudan University, President of Shanghai Medical College of Fudan University and President of International Human Phenome Institutes (Shanghai)) delivered a welcome speech as the co-chair of the conference. In his speech, he introduced that human phenome is the next strategic commanding point and original innovation source of life sciences after genome, which is expected to stimulate and lead the new paradigm change and industrial revolution of biomedicine. Since 2020, China has made a series of important progress in the scientific research of human phenomics, including the official publication of the world's first international peer-reviewed academic journal Phenomics, the official launch of human phenome data processing cloud platform, and the rapid advancement of the panoramic phenotypic study of the core cohort of healthy people. This conference was an opportunity to lead the scientific community to continuously and rapidly promote the research of human phenome through the collision of ideas among participating experts and scholars.

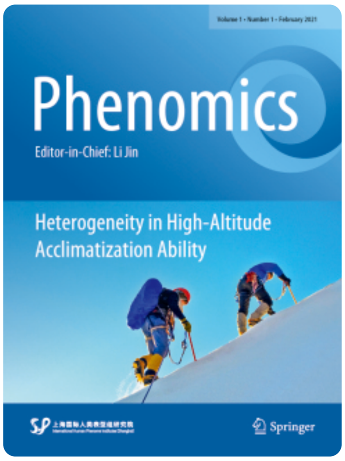
After the conference, Chongqing Liangjiang New Area has signed a strategic cooperation framework agreement with International Human Phenome Institutes (Shanghai) and Chongqing Medical University, which will jointly promote human phenome research in Western China. According to the framework agreement, Chongqing Liangjiang New Area, International Human Phenome Institutes (Shanghai), and Chongqing Medical University will jointly build a precision measurement and research center of human phenome to optimize further the collaborative network and industry-university-research alliance of the International Human Phenome big science Project.

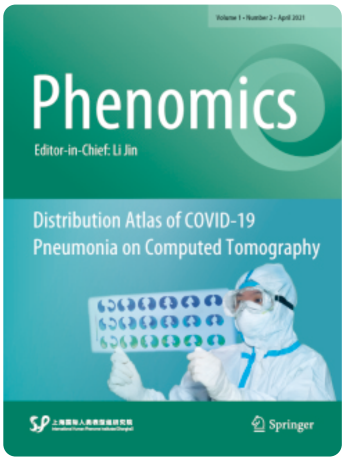
## The "Thar DESIGNS" Project (India)

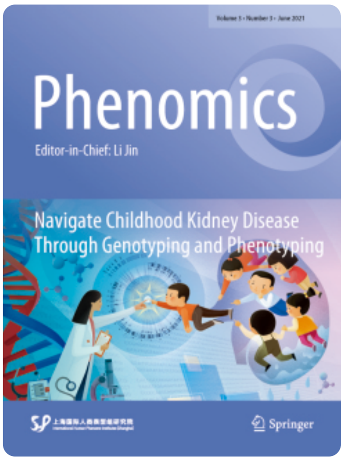
Professor Mitali Mukerji is formerly the Chief Scientist at the Institute of Genomics and Integrative Biology (CSIR-IGIB), Delhi. Recently, she left the CSIR and became a professor at the Indian Institute of Technology (IIT) Jodhpur. On September 5th 2021, under Professor Mukerji's coordination, IIT Jodhpur initiated a transdisciplinary Phenomics Project named "Thar DESIGNS (Desert EcoSystem Innovations Guided by Nature and Selection)". The initiative is launched on the Jodhpur City Knowledge and Innovation cluster (JCKIC) platform and aims to carry out Ecosystem phenomics through a transdisciplinary framework of medical, engineering, environmental and life sciences.

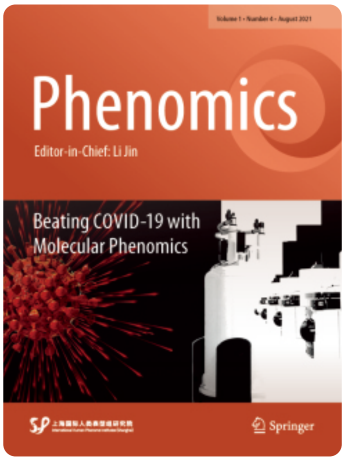
Thar, a hot desert in India, has been one of the largest natural laboratories for evolving innovative 'designs' that ensure the adaptation and survival of its constituent species, their interdependencies, and the conservation of the entire ecosystem. Through IoT-enabled devices, data analytics framework, Machine learning and domain knowledge, the project would infer links between environment, phenotype and genotype at geo-spatiotemporal scales and identify signatures of "Thar DESIGNS" for early actionable intervention strategies. The project would also provide a "Desert Ecosystem Knowledge Grid" that could foster the cycle of engineering-research-development-commercialization. These could range from solutions for the management of diseases common and endemic to desert regions, novel bioprospecting opportunities and innovative bio-inspired engineering designs. In addition, it could help evolve unique strategies for ecological conservation and restoration that ensure sustained livelihood for its inhabitants. "Thar DESIGNS" is supposed to encourage citizen participation, enhance industry capacity and benefit policymakers and diverse stakeholders.

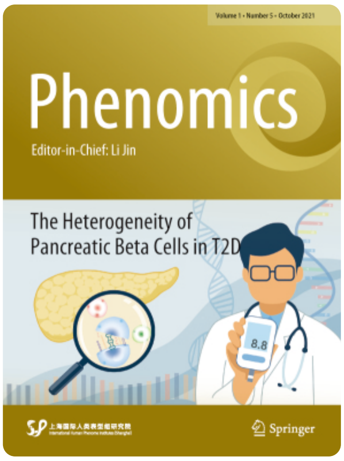
# *Phenomics* Journal

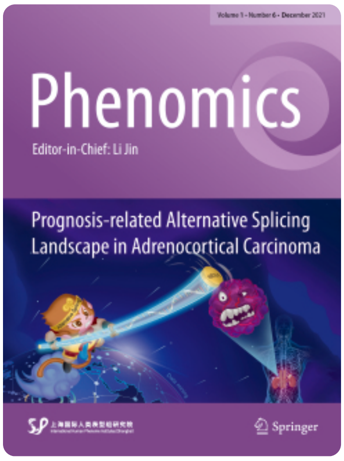












*Phenomics* is a peer-reviewed, international and interdisciplinary research journal run by Springer Nature and International Human Phenome Institutes (Shanghai). *Phenomics* is dedicated to publishing the finest articles and communicating scientific progress in the field of *phenomics*. The topics of interest to Phenomics include but are not limited to high-throughput phenotyping and technological innovations; Linking the genome to the phenome with models, algorithms, databases, etc.; Exploring the relationships between phenotypes and understanding phenotypic variation and responses to the environment; Phenotypic research and its precise application in clinical disease, treatment, prevention and control; Phenome-related multi-omics studies, novel data fusion approaches and integrated analysis; and Phenome-related model organism research, interdisciplinary multiscale research, etc.

The bimonthly journal publishes four articles in each issue starting from January 2021. By January 5th, 2022, *Phenomics* has released 6 issues with a total of 26 articles. The journal was downloaded nearly 22,000 times so far.

Phenomics website: https://link.springer.com/journal/43657/volumes-and-issues

# CONGRATULATIONS TO MEMBERS IN THE NEWS

Congratulations to our IHPC members who have won awards for their outstanding contributions to human societies and distinguished leadership in promoting scientific progress in the past year. (All the following information was collected from the online news and may be incomplete).

**Professor Jie Chen**

- Professor Jie Chen has been elected to the American Institute for Medical and Biological Engineering (AIMBE) College of Fellows for leadership and outstanding contributions to designing and commercializing low-power biomedical ultrasound circuits and impedance-based microfluidic sensor chips.

February 15th 2021

**Professor Jianjun Liu**

- Professor Liu Jianjun was presented with the President's Science Award (PSA) in Singapore for his outstanding contributions to genetic studies of Asian populations, which has advanced biomedical research and precision medicine, and benefited clinical practice in the prevention of diseases and adverse drug responses.

December 18th 2020

**Professor Carmencita Padilla**

- Professor Carmencita Padilla was conferred the 2019 Outstanding Professional in the Field of Medicine Award by the Professional Regulation Commission of the Republic of the Philippines for her distinguished contribution in the field of Medicine and her exemplar service in her profession.

June 20th 2019

**Professor Sir Munir Pirmohamed**

- Professor Sir Munir Pirmohamed has been appointed by the UK Secretary of State for Health and Social Care as Chair of the Commission on Human Medicines (CHM) for 4 years from February 12th 2021.

February 12th 2021

**Professor Chen Wang**

- Professor Chen Wang has been elected to the American National Academy of Medicine as one of the ten international members for his leadership of China's three leading medical bodies and his impactful clinical research and medical reforms.

October 19th 2020