

23rd Annual Meeting

August 7-11, 2023 Bangkok, Thailand



Genomic Standards for Precision Medicine, Agriculture, Comparative Genomics, and Metabolomics

Hosted by the Faculty of Medicine Siriraj Hospital at Mahidol University and
the National Center for Genetic Engineering and Biotechnology (BIOTEC/NSTDA)



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AGENDA

23rd Genomic Standards Consortium Annual Meeting

Location: SiMR room 101. Siriraj Hospital Mahidol University

Monday, August 7, 2023	Education Day GSC Board Meeting (<i>Board members only</i>) GSC23 Reception
Tuesday, August 8, 2023	Session 1: Genomic Standards for Precision Medicine Keynote: Leslie Biesecker (NIH/NHGRI, USA) Personalized Medicine Session 2: Standards Perspectives from Publishing and Databases
Wednesday, August 9, 2023	Session 3: Genomic Standards for Metabolomics Keynote: Claire O'Donovan - EMBL-EBI, UK Standards in Metabolomics Session 4: New Sequencing Technologies & Genome Sequencing Standards
Thursday, August 10, 2023	Session 5: GSC Current & Evolving Standards Session 6: Genomic Standards for Comparative Genomics
Friday, August 11, 2023	GSC23 workshops MGnify Oxford Nanopore

Monday, August 7, 2023

9:00 am - 1:00 pm

Student Education Day

Education Day Agenda: moderator Sakda Khoomrung

9:00 am Manop Pithukpakorn (Faculty of Medicine Siriraj Hospital, Mahidol University)

9:20 am Lynn Schriml (University of Maryland School of Medicine): ML/AI-ready biomedical and genomic data

9:40 am Susanna-Assunta Sansone (University of Oxford) - Standards across life Science

10:00 am Ramona Walls (Critical Path Institute) - Biomedical data models and standards (OMOP, CDISC, FHIR) and how data are used for regulatory science.

10:20 am Scott Edmunds (GigaScience) - Open Access publishing

10:40 am Rob Finn (EMBL-EBI European Bioinformatics Institute) - Challenges (and benefits) in depositing multiomics datasets

11:00 am **Break**

11:10 am Joao Setubal (University of São Paulo) - Metagenomics

11:30 am Tanja Woyke (Joint Genome Institute, DOE) - Single cell genomics

11:50 am Phil Hugenholtz (Queensland University) - Human gut microbiome derived Therapeutics

12:10 pm Kasthuri J Venkateswaran (Jet Propulsion Laboratory, NASA) - Lessons learned from “Space Genomics”

2:00 pm - 5:00 pm GSC Board meeting

7:00 pm - 10:00 pm Welcome to GSC23 Reception

Tuesday, August 8, 2023

9:00 - 9:15 am	Welcome, Open GSC22, Introduction to the GSC
9:15 - 10:45 am	<u>Session 1: Genomic Standards for Precision Medicine</u> Session Chair: Manop Pithukpakorn Keynote: Leslie Biesecker (NIH/NHGRI) - Personalized Medicine
9:45-10:45 am	Session Speakers: <u>Manop Pithukpakorn</u> (Faculty of Medicine Siriraj Hospital, Mahidol University) Curating Clinical Genomes: Real World Practice of Human Genome Variant Interpretation <u>Varodom Charoensawan</u> (Faculty of Science, Mahidol University) Human Cell Atlas: Putting together reference transcriptomes of all human cells <u>Susanna-Assunta Sansone</u> (University of Oxford, UK) FAIR Principles <u>Tip Wongsurawant</u> (Platinum Sponsor: Oxford Nanopore Technologies) Game-changer in Epigenetics
10:45 - 11:30 am	Tea and Coffee Break Networking
11:30 - 12:45 pm	<u>Session 2: Standards Perspectives from Publishing and Databases</u> Session Chair: Chris Hunter Session Speakers: <u>Chris Hunter</u> (GigaScience Press) Journal perspective: Gigascience <u>Peter Woollard</u> (ENA, EMBL-EBI, UK) Database perspective: EBI Resources <u>Kyung-Bum Lee</u> (DDBJ) Database perspective: The DDBJ resources based on standards <u>Yiming Bao</u> (National Genomics Data Center) Database perspective: National Genomics Data Center [virtual] <u>Zuwei Qian</u> (Platinum Sponsor: PacBio) Changing the sequencing game: Unlocking the promise of genomics through HiFi sequencing
12:45pm	Group Photo
1:00 - 2:00 pm	Lunch and Networking
2:00 - 5:00 pm	Working Group Sessions - Afternoon Networking 1. GSC Compliance and Interoperability Working Group 2. Developing Standards in Personalized Medicine



Wednesday, August 9, 2023

9:00 - 9:15 am	Welcome & Daily announcements
9:15 - 10:45 am	<u>Session 3: Genomic Standards for Metabolomics</u>
	Session Chair: Sakda Khoomrung

9:15-9:45 am **Keynote:** Claire O'Donovan (EMBL-EBI, UK) - **Standards in Metabolomics [virtual]**

9:45-11:00 am **Session Speakers:**

Sakda Khoomrung (Siriraj Hospital, Mahidol University)

Metabolomics standards in clinical research

Jutarop Phetcharaburanin (Khon Kaen University)

Advancements in NMR Metabolomics: Enhancing Sample Handling and Metabolite Identification

Umaporn Uawisetwathana (National Center for Genetic Engineering and

Biotechnology-BIOTEC.)

Metabolomics for agriculture applications

Yuri Corilo (PNNL, USA)

Metadata Informed Metabolomics

Yan Ting Lim (Platinum Sponsor: Waters)

Waters' solutions for Omics Analysis



11:00 - 11:45 am Tea, Coffee and Networking

11:45 - 1:00 pm **Session 4: New Sequencing Technologies and Genomic Standards**

Session Chair: **Scott Jackson**

Session Speakers:

Wanilada Rungrassamee (National Center for Genetic Engineering and Biotechnology, Thailand)

Standardization of next-generation sequencing method to study gut microbial diversity in shrimp

Tanja Woyke (JGI, USA)

Obligate insect symbionts: insights from new genome sequences and possible standards for these tiny genomes

Kasturi Venkataswarans (Jet Propulsion Lab/NASA, USA)

Intergalactic Microbes: Uncovering the Invisible Co-Pilots of Space Exploration

Kris Locken (Platinum Sponsor: Zymo)

A Practical Guide to Microbiome Reference Standards



1:00 - 2:00 pm **Lunch and Networking**

2:00 - 5:00 pm **Working Group Sessions - Afternoon Networking**

3. NMDC workshop
4. Metabolomics Standards
5. Standardized Protocols

Thursday, August 10, 2023

9:00 - 9:15 am	Welcome & Daily announcements
9:15 - 10:45 am	<u>Session 5: GSC Current & Evolving Standards</u> Session Chair: Ramona Walls
9:15 - 9:30 am	The Dawn Field Award invited speaker <u>Montana Smith</u> (PNNL, USA) Accomplishing FAIR: For data generators and users
	Session Speakers: <u>Ramona Walls/Chris Hunter</u> (CIG Co-Chairs) GSC Compliance and Interoperability Working Group Update <u>Mark Miller</u> (LBNL, USA) A Social and Technical implementation of LinkML by GSC and NMDC <u>Lisa Karstens</u> (Oregon Health & Science University) UroBiome new MIxS extension [virtual] <u>Scott Jackson</u> (National Institute of Standards and Technology (NIST)) Measurement Assurance for Innovation in Microbiome Science <u>Svetta Kwan</u> (Platinum Sponsor: ThermoFischer/GenePlus) The Impact of Rapid Next-Generation-Sequencing on Precision Oncology
10:45 - 11:30 am	Tea, Coffee and Networking
11:30 - 1:00 pm	<u>Session 6: Genomic Standards for Comparative Genomics</u> Session Chair: Nikos Kyripides (JGI) Session Speakers: <u>Chuck Cook</u> (Global Biodata Coalition, UK) The Global Biodata Coalition: towards a sustainable biodata infrastructure <u>Naraporn Somboonna</u> (Chulalongkorn University, Thailand) Gut microbiota alteration associated with SLE (systemic lupus erythematosus) development <u>Nikos Kyripides</u> (JGI, USA) Microbial Comparative Genomics-MGE Standards: viruses and plasmids <u>Rob Finn</u> (MGnify, EMBL-EBI, UK) Towards producing representative genome catalogues for microbial communities <u>Phil Hugenholtz</u> (University of Queensland, Aus.) The Genome Taxonomy Database (GTDB) initiative to establish a standardised microbial taxonomy
1:00 - 1:15 pm	Close of Meeting and handoff to GSC24 Tucson, Arizona
1:15 - 2:15 pm	Lunch and Networking
2:15 - 3:30 pm	Working Group Sessions - (and/or Afternoon Networking) 6. Standardizing Open Microbiome Data analysis and workflow sharing to further federated resource development

Friday, August 11, 2023

GSC Workshop 1

MGNify (whole day) - Rob Finn and EBI staff: Lorna Richardson and Tanya Gurbich
Location: 1st Floor, SiMR Building, Siriraj Hospital Mahidol University



MGNify is a freely available resource, provided by EMBL-EBI, for the analysis and archiving of microbiome-derived sequencing data. Users can request assembly of raw-read data, and taxonomic and functional analysis of publicly available (or pre-publication) datasets available in ENA. MGNify also provides microbiome-derived genome catalogues for multiple biomes, and a non-redundant protein database of over 3 billion sequences.

Two half-day workshops (a mixture of hands-on practicals and lectures) are available on Friday 11th August, provided by the MGNify resource. In the morning there will be an introductory session for users new to the field and/or the resource, and in the afternoon a more advanced session. More details are provided below about what will be covered in each session. Registration will be for both sessions, but delegates should specify which sessions they will attend - either one or both of the sessions, as they prefer based on their experience.

Please note that attendees will be expected to bring along their own laptop computer for use within the practical sessions. All practical aspects will be browser-based and so the only software required for the course will be access to a recent version of either Chrome, Firefox, or Safari.

Introductory session (9:00-12:00)

Topics covered:

- What is MGNify?
- Analysis and results of metabarcoding and metagenomic/metatranscriptic datasets
- Assembly of metagenomic data - the advantages and disadvantages
- Submitting data to ENA for analysis by MGNify
- MGNify quality control and data cleaning steps
- Why metadata is important
- Searching MGNify and accessing results

Advanced session (14:00-17:00)

Topics covered:

- Generating MAGs from metagenomic data
- Biome-specific genome catalogues
- Comparing new MAGs against genome catalogues
- Programmatic access to MGNify
- Downstream analysis in Jupyter notebooks, e.g. comparing across datasets

Friday, August 11, 2023

GSC Workshop 2

Oxford Nanopore (whole day) - Tip Wongsurawant and Nanopore Staff

Location: Room 304-305, SiMR Building, Siriraj Hospital Mahidol University

[Oxford Nanopore Technologies Limited](#) is a UK-based company which develops and sells nanopore sequencing products (including the portable DNA sequencer, MinION) for the direct, electronic analysis of single molecules.



This hands-on workshop will provide attendees with a critical understanding of Oxford Nanopore sequencing technology through a comprehensive laboratory and analysis workflow approach. The workshop will provide a background understanding of the sequencing technology, QC, and sequencer set up with laboratory training including “hands-on” individually generated sequence ready barcoded libraries, loading flow cells, and operating the MinKNOW sequencing software. Basic data analysis with EPI2ME software will allow participants to analyze and view the results. The program will consist of a morning session where participants will prepare actual sequencing libraries, load the MK1C MinION flow cell and initiate sequencing. An afternoon session will include understanding the QC outputs and Introduction to basecalling and Epi2ME analysis workflows.

Prerequisites include basic laboratory safety procedures, low volume pipette operations, and basic understanding of DNA sequencing.

Topics covered:

- Introduction and Overview of Technology
- Library Synthesis and Quality Control
- Loading the Nanopore Flow Cell
- Operating MinKNOW Software
- Sequence Operations of MK1C
- Sequence Monitoring Metrics
- Post Sequencing Data Analysis

Keynote Speaker Abstracts

Leslie Biesecker (NIH/NHGRI, USA) [virtual]

Genotype First – in Research and In Clinical Care

The rapidly falling cost of genome sequencing necessitates rethinking about how we care for patients and perform research. The clinical research paradigm has been dominated by a phenotype-first approach. In this paradigm it is the patient's manifestations or disease that initiate the process of care, through the mechanism of the differential diagnosis. It is essential to recognize that a key function of the differential diagnostic paradigm was to narrow the range of potential diagnoses to match the limits of the then available genetic tests. Now that large panel tests can be had for \$250, the upstream narrowing of the diagnoses is less critical to success in achieving the diagnosis. In fact, it can be argued that the broad testing is superior to a narrow differential as the test interrogates genes for disorders that the clinician may not have thought to consider in her differential diagnosis. Thinking more broadly than panels, clinical genome sequencing is becoming affordable and has the potential to provide a life-long health care resource for a patient.

In the research realm, a similar paradigm has held – that of the assembly of a patient cohort or a case-control cohort for the purpose of elucidating the cause of a disorder affecting the people of that cohort. This is an arduous and expensive process and sometimes has limited long term utility, after the primary questions have been addressed. Instead, because a research genome can cost as little as \$250, one can begin to consider the establishment of genetically characterized cohorts, which can subsequently be interrogated by a genomic attribute. In this mode of research, the clinical investigator searches the genomic database to identify individuals harboring genetic variants of interest. These individuals can be recalled to the research center on the basis of their genetic change and the researcher then tests a hypothesis of that variant being associated with a phenotype. A large advantage of this approach is the reduction in bias as a part of the recruitment and eligibility review process and the reduced cost.

These examples and principles will be reviewed and discussed with pertinent examples.

Claire O'Donovan (EMBL-EBI, UK) [virtual]

Standards in Metabolomics

The aim of this talk is to present the current status of standards in Metabolomics with its diverse communities including academia and industry. It will highlight some of the collaborative efforts and challenges that the community faces and how we are evolving resources to respond to its needs. It will also include how metabolomics is learning from the other omics communities' experiences and how all the omics need to interact together going forward.

Dawn Field Award for Outstanding Contributions to Genomic Standards



Dawn was instrumental in founding the GSC in 2005 pulling together a group of like-minded people to form the consortium, and she was the founding president from 2009 through 2014. The GSC award a honorarium to one outstanding person in the field of genomic standards each year in Dawns' name to reflect and remember the great work she performed in creating and building the GSC. This years award goes to:

Montana Smith

(PNNL, USA)

Montana is a Data Scientist at the Department of Energy's Environmental Molecular Sciences Laboratory (EMSL), a division within Pacific Northwest National Laboratory. She leads the metadata collection and standardization efforts in close collaboration with the EMSL user community. Her work in the EMSL user program has included incorporated the GSC standards for sample metadata collection and is working to improve interoperability and expand use, familiarity, and development of these standards. She also serves as the Product Owner for the National Microbiome Data Collaborative's Submission Portal and interfaces directly with the broader microbiome research community to implement the GSC's standards into the Submission Portal. Smith received a B.S. in Biology from Iowa State University and has a strong research background in soil microbial ecology, and data and sample management. She is deeply committed to supporting FAIR metadata standards and improving community adoption through her involvement in the GSC's CIG and TWG. She is currently leading efforts with the research community and the GSC to propose community standards for quantitative stable isotope probing experiments and provides user insight for improvements and expansion of the GSC standards. Smith engages with users across multiple programs to expose researchers to FAIR metadata, standards, and ontologies, and improve community adoption and usability.

Montana will give a presentation on her work:

Accomplishing FAIR: For data generators and users

FAIR data can be hard to accomplish and implementing standards can be a challenge. User feedback and usability testing provides valuable information that enables us to improve standards and increase community adoption. In my talk, I'll share some of the feedback we've received and changes that have been suggested to improve adoption and understanding as well as development of the new MISIP (Minimal information for Stable Isotope Probing) checklist currently in development. Providing FAIR data will improve and expand scientific discovery, and providing an easily adopted and comprehensive standard will make FAIR data obtainable.

Speaker Abstracts

Session 1: Genomic Standards for Precision Medicine

1.1 Manop Pithukpakorn (Faculty of Medicine Siriraj Hospital, Mahidol University)

Curating Clinical Genomes:

Real World Practice of Human Genome Variant Interpretation

With the rapid advances in genomic sequencing technology and increased knowledge of genomics in medicine, the integration of genomic data into clinical practice is becoming increasingly important.

The ACMG 2015 Standards and Guidelines for the Interpretation of Sequence Variants provide a valuable framework for variant interpretation, but they also have limitations. Real-world data on the clinical significance of many variants is limited, and there are knowledge gaps in our understanding of how variants affect the disease pathogenic mechanisms and clinical phenotypes.

The ClinVar database is a valuable resource for variant interpretation, but it is not without its limitations. The database is not comprehensive, and the quality of the data varies. ClinGen specific gene guidelines provide additional information on the interpretation of variants in specific genes but add more complexity to interpretation process. Interpretation of copy number variants (CNV) and splicing variants is particularly challenging. Changes in the number of copies or splice sites of coding sequences do not always alter the gene function. There are no clear guidelines for the interpretation of these variants, and they are often classified as "uncertain significance."

This presentation will discuss the challenges of curating clinical genomes, and the resources available to help with variant interpretation. The presentation will also discuss the future of clinical genomics, and how we can use genomic data to improve patient care.

1.2 Varodom Charoensawan (Faculty of Science, Mahidol University)

Human Cell Atlas:

Putting together reference transcriptomes of all human cells.

Regulation of gene expression is an important biological process that gives rise to phenotypic diversity from the same genetic information. In addition to reference genomes and variations between individuals, reference transcriptomes among tissues and cell types are crucial to understanding fundamental units of life and serve as a platform for developing targeted therapy in the precision medicine era.

Human Cell Atlas (HCA) consortium aims to 'create comprehensive reference maps of all human cells – as a basis for both understanding human health and diagnosing, monitoring, and treating diseases'. As part of the HCA, we are establishing the reference immune and gene expression repertoires of the Asian population under the 'Asian Immune Diversity Atlas (AIDA)' project, together with collaborators from eight countries. Asian genomic data are heavily underrepresented as compared to genomic data of people of European ancestry that accounts for 80 percent of the current data worldwide. In the AIDA project, we will rectify this imbalance by characterizing the nature and extent of variation in immune cell types from diverse Asian populations. Within Thailand, we will investigate the conserved and unique patterns in the immune diversity of the Thai populations in different parts, which are historically, culturally, and genetically unique. These data will serve as a healthy baseline for characterizing cell state changes in various immune-related diseases. This project is a collaborative effort involving researchers from Mahidol University, Chiang Mai University, Khon Kaen University, and Prince of Songkla University in Thailand.

1.3 Susanna-Assunta Sansone (University of Oxford, UK)

FAIR (Findable Accessible Interoperable and Reusable) Principles

The FAIR Principles (<https://doi.org/10.1038/sdata.2016.18>), I have co-authored, have propelled the global debate in all disciplines on the importance of Findable, Accessible, Interoperable, and Reusable data, by humans and machines, and the need for better research data management, transparent and reproducible data worldwide. FAIR has united stakeholders world-wide behind a common concept: good data management under common standards. It is no longer optional. However, the FAIR Principles are aspirational, and putting FAIR into practice is work in progress; it "takes a village"! Starting with a brief history of the Principles, Susanna will paint the landscape of key initiatives and community activities for FAIR data, with a focus in the Life Science, including resources like FAIRsharing (<https://fairsharing.org>), and the FAIR Cookbook (<https://faircookbook.elixir-europe.org>).

Session 2: Standards Perspectives from Publishing and Databases

2.1 Chris Hunter (GigaScience Press, UK)

Journal perspective: Gigascience

GigaScience Press (<https://www.gigasciencepress.org/>) has the goal of achieving true open science by embracing the UNESCO Open Science Recommendation as the primary goal for its publications and activities. A major part of those efforts is the creation and use of GigaDB, the open access repository of datasets directly associated with all *GigaScience* journal articles. GigaDB datasets are created by the authors with expert guidance from highly experienced data curation staff, making use of relevant standards and ensuring deposition of all relevant data in public repositories. Our curation staff keep themselves abreast of many on-going standards efforts including the GSC MIxS as well as relevant ontologies, and guide authors on their appropriate use in dataset metadata. Here I will present GigaDB and highlight some of the ways we utilise standards in our curation work.

2.2 Peter Woollard, Josie Burgin, Guy Cochrane (ENA, EMBL-EBI, UK)

Database perspective: EBI Resources

Core and diverse sample metadata has been explicitly captured with checklist templates for a number of years, by the European Nucleotide Archive(ENA) and other INSDC partners. There is now a broader and more complex spread of sequencing experiment related metadata that could usefully be collected too, due to the increasing use of sequencing technologies to study the general biological world, particularly for human health and the environment. Capturing experiment metadata information more accurately and consistently will increase the usefulness of the data, by making it more FAIR. We are exploring experimental checklists conceptually similar to existing sample level checklists to tailor metadata provided for different 'types' of sequencing experiments. We have integrated learnings from sample checklists, including the need to have checklist versioning and dependency validation. To do the initial validation for the experiment checklists, we are using: JSON, JSON schema and ELIXIR bio validation technologies. These can rapidly catch most validation issues and provide immediate feedback to users. Deeper automated validation will still be performed to ensure INSDC standards.

Currently, we have a dozen 'experiment type' checklists ranging from metabarcoding to spatial transcriptomics. These experiment type checklist JSON and accompanying JSON schema files are all driven from a single JSON configuration file. It will be straightforward and sustainable to add further experiment types.

A pilot use and submission of experiment type checklists is planned for later this year. All code and documentation is publicly accessible: github.com/enasequence/ena-experiment-checklist/. In this talk, we will outline what we are doing and illustrate how it will improve the standardisation of sequence experimental metadata.

2.3 Kyung-Bum Lee (DDBJ)

Database perspective: The DDBJ resources based on standards

The DDBJ (DNA Data Bank of Japan) Center is a global biological database serving as a comprehensive repository for diverse biological information. We collect and handle a variety of data types, including raw, assembled, and annotated nucleotide sequence data, as well as functional genomics data (GEA: Genomic Expression Archive), metabolomics data (MetaboBank), and human genetic and phenotypic data (JGA: Japanese Genotype-Phenotype Archive).

The DDBJ seeks to broaden its collaboration with other national-class data providers like KOBIC (Korea Bioinformation Center) and BRIN (National Research and Innovation Agency, Indonesia) as a founding member of the International Nucleotide Sequence Database Collaboration (INSDC) with NCBI and EBI. Key components of science are inclusion and FAIR-ness. I will outline the DDBJ databases based on such principles.

2.4 Yiming Bao (National Genomics Data Center, China) [virtual]

Database perspective: National Genomics Data Center

National Genomics Data Center (NGDC) at Beijing Institute of Genomics, Chinese Academy of Sciences was established by the Ministry of Science and Technology and Ministry of Finance of China in 2019. NGDC is a national platform for archiving, managing and processing a wide range of genomics related data. These include the BioProject and BioSample databases, Genome Sequence Archive (GSA=SRA) family, Genome Warehouse (GWH=WGS), GenBase (=GenBank), Gene Expression Nebulas (GEN=GEO), Genome Variation Map (GVM=dbSNP), and many others. Following largely the data structure and standards of the corresponding databases in INSDC, NGDC is ready and has started to smoothly exchange its data with those of INSDC, therefore is working hard towards becoming a partner of INSDC. Additionally, NGDC has developed several procedures and tools to facilitate the implementation of various standards

Session 3: Genomic Standards for Metabolomics

3.1 Sakda Khoomrung (Siriraj Hospital, Mahidol University, Thailand)

Mass spectrometry-based metabolomics standards in clinical research

Owing to growing interest in personalized medicine, the combination of clinical data, multi-omics data, and systems analysis has been increasingly studied. Mass spectrometry (MS)-based metabolomics is an ideal technology for identifying and quantifying metabolites in various biological samples.

Experimental MS workflow, along with advanced bioinformatics, is considered one of the fundamental tools in systems biology for phenotype characterization and the development of precision medicine. In fact, the standardized methods/protocols for metabolomics, including sample preparation, metabolite identification, data processing and analysis, and quantification, represent crucial steps for the translation of research outcomes into clinical practice. Quantitative metabolomics allows the accumulation and comparison of metabolomics data across different studies, which eventually can lead to the establishment of a critical resource for biomarker research, precision medicine, and bridging scientific outcomes with clinical applications. In this talk, I will discuss several fact-based challenges, including metabolite identification, data processing and analysis, and the measurement precision of absolute quantification in clinical research.

3.2 Jutarop Phetcharaburanin (Khon Kaen University)

Advancements in NMR Metabolomics: Enhancing Sample Handling and Metabolite Identification

Metabolomics, a powerful tool in systems biology, enables the characterization of phenotypes. Nuclear magnetic resonance (NMR) spectroscopy plays a pivotal role in capturing dynamic changes in metabolites as a response to gene-environment interactions, diets, diseases, and stimuli. However, the NMR metabolomics pipeline is susceptible to confounding factors stemming from improper handling of biospecimens, including inadequate containers, storage conditions, and durations. These inconsistencies can introduce consequential effects that undermine accurate analysis. By utilizing NMR metabolomics, an optimized fecal sample handling strategy has been developed, leading to improved fecal metabolic phenotyping-based diagnoses. Additionally, the identification of metabolites presents another critical step in the NMR metabolomics pipeline. Misassignments of metabolites, caused by factors like human errors and the complexity of spectral data, can result in data misinterpretation and misunderstanding. To mitigate these challenges, a computational approach, specifically artificial intelligence-guided metabolite identification, is being developed. This approach aims to enhance the accuracy and precision of NMR metabolite identification, enabling more reliable data interpretation.

3.3 Umaporn Uawisetwathana (National Center for Genetic Engineering and Biotechnology-BIOTEC.)

Metabolomics for agriculture applications

Metabolomics is increasingly employed to comprehensively study of all metabolites of organisms. It has been proven to be instrumental to unravel important metabolites as well as molecular pathways governing genotypes and phenotypes of an organism. Several metabolomics applications in agricultural research focus on identification of biomarkers and discovery of metabolic mechanism underlying particular traits to rationally develop an agricultural practice as well as new varieties with better yields, resistance to biotic/abiotic stresses and desirable nutritional contents. With challenging of untargeted metabolomics in plant research, standardization at each step of metabolomics pipeline is considered. Examples of metabolomics standard for agricultural research will be presented. In summary, metabolomics have been successfully proved as a high-throughput screening method in functional genomics to generate more fundamental knowledge of the biochemical process underlying crucial traits.

3.4 Yuri Corilo (PNNL, USA)

Metadata Informed Metabolomics

This presentation will discuss the importance of metabolomics standards, data models, and metadata. The metabolome is a complex molecular system; multiple sample preparation methods and analytical techniques are required to identify its vast molecular compositional space. We will provide an overview of how we aim to capture and describe a complete and unified experimental design for metabolomics, including metadata on the samples, sample processing, instruments used, and metabolites. This comprehensive metadata is crucial for establishing standardized workflows for molecular annotation and promoting data reuse. Additionally, we will share our vision for utilizing metadata to enhance the accuracy of metabolite annotation and facilitate new molecular discoveries in metabolomics.

Session 4: New Sequencing Technologies & Genome Sequencing Standards

4.1 Wanilada Rungrassamee (National Center for Genetic Engineering and Biotechnology, Thailand)

Standardization of next-generation sequencing method to study gut microbial diversity in shrimp

The gut microbiota plays an essential role in animal health and production, and its study has become increasingly important in the aquaculture sector. However, standard protocols and best practices for measuring shrimp microbiota are not well established, leading to uncertainty in the accuracy of results and complicating cross-study analyses.

In this study, we investigated the influence of key methodological variables on the gut microbiota profiles of black tiger shrimp, an important product of the aquaculture industry in Asia Pacific region. We used pooled gut samples and synthetic DNA spike-in standards to evaluate four commercial kits for DNA extraction with primer sets for the V1-V2, V3-V4, and V6-V8 hypervariable regions of the 16S rRNA gene. We evaluated the performance of the kits and PCR primers based on the diversity of observed microbiota profiles, taxon-specific biases, and accuracy of quantification of spike-in standards. We also evaluated different bioinformatics pipelines for data analysis, focusing on the advantages of using denoising algorithms compared to traditional clustering methods to resolve sequence diversity in dominant species.

Our results showed that the variables assessed had a significant impact on shrimp gut microbiota profiles. This highlights the need for standard protocols and best practice guidelines for measuring shrimp microbiota. Our study provides several recommendations to improve the accuracy and reproducibility of shrimp microbiota measurements and should be of great benefit to aquaculture microbiome research.

4.2 Tanja Woyke (JGI)

Obligate insect symbionts: insights from new genome sequences and possible standards for these tiny genomes

Insects, the most species-rich animal group, comprise over one million depicted species. Insect-bacteria partnerships are widespread and significantly impact the evolutionary success and diversification of this animal group. Bacterial endosymbionts supplement essential nutrients absent in insects' diets, aid in the degradation of tough food sources, defend against natural enemies, and enhance resistance to stress and insecticides. Additionally, certain 'selfish' insect endosymbionts manipulate insect reproduction to facilitate their own spread. This talk will explore newly discovered obligate insect symbiont genomes and potential criteria for standards for these small genomes.

4.3 Kasthuri Venkateswaran (JPL, NASA)

Standards that apply to space genomics, detection, planetary protection

In our quest to comprehend space, understanding the invisible travelers accompanying us is crucial. These microscopic entities are key to astrobiology, astronaut health, and the maintenance of space habitats. Our research has portrayed a vibrant microbial ecosystem on the International Space Station (ISS) and NASA cleanrooms where spacecraft are assembled.

Through advanced molecular techniques and traditional culture methods, our Microbial Tracking initiatives have extensively catalogued the dynamic microbial populations adapting to extraterrestrial conditions. These adaptable communities can be both allies and threats to human health.

Our investigations delved into the microbial universe, uncovering complexities through amplicon sequencing, metagenomics, and resistomes. We've identified about 3,000 bacterial and fungal strains, some new species, along with their potential virulence traits and beneficial metabolites.

Yet, our pioneering Environmental 'Omics' project isn't simply an academic exercise - it's a practical guide for space exploration. The main objective of our microbial tracking research is to leverage these insights to enhance human health within the ISS and other similar closed systems. We're dedicated to transforming our fundamental research into tangible benefits, such as pathogen detection and the development of health countermeasures.

In keeping with our commitment to scientific transparency and innovation, we've made our 'omics' datasets accessible on NASA's GeneLab bioinformatics platform. This platform houses a robust database and advanced computational tools. We warmly invite the wider scientific community to make use of these resources, contributing to the ongoing exploration and understanding of life - seen and unseen - throughout the cosmos.

Session 5: GSC Current & Evolving Standards

5.1 Ramona Walls (Critical Path Institute/CIG co-chair) & Chris Hunter (GigaScience Press/CIG co-chair)

GSC Compliance and Interoperability Working Group

The GSC *Compliance and Interoperability Working Group* (CIG) is responsible for defining (with input from the GSC Board) and implementing GSC policies around standards, with a primary focus on the Minimum Information about any (x) Sequence (MIxS) standards, and for maintaining the GSC website. The last year has been very busy for the CIG. We are reviewing three new MIxS checklists or extensions (Urobiome, MISIP, and MiNAS) for the upcoming MIxS v.7 release.

After converting MIxS hosting to Github and LinkML (see the following talk) with the 2022 release of MIxS v.6, we have spun off a *Technical Working Group* (TWG) that oversees all technical aspects of MIxS, including Github issue and project tracking, LinkML coding, and creating releases. The TWG is currently working to stabilize our LinkML implementation. The CIG and TWG are working together to update GSC policies to make MIxS more sustainable and serve MIxS in a form that is machine validatable. The main GSC website (gensc.org) has undergone a major overhaul, with new content being added regularly, including links to the formal MIxS documentation generated automatically by LinkML.

Both CIG and TWG are open and active working groups, and we are seeking new members from the GSC community who are willing to provide a few hours of work per month.

5.2 Mark Miller (LBNL)

A Social and Technical implementation of LinkML by GSC and NMDC

The Genomic Standards Consortium published the first version of the Minimum Information about any (X) Sequence standard in May 2011 (DOI:10.1038/nbt.1823). Since then, it has since been adopted, in part, into the INSDC's requirements for describing Biosample and sequence submissions. That means that all of those records have a minimum set of required attributes and that submitters are provided with a standardized vocabulary of other terms they can use to describe their submissions.

The National Microbiome Data Collaborative was launched by the US Department of Energy in ~2020 with the intent of aggregating standardized metadata and results from numerous environmental omics projects (DOI:10.1038/s41579-020-0377-0). NMDC is distinctive in the fact that multiple omics modalities are supported (metagenomics, metaproteomics, metabolomics, etc.) and that the metadata conform to a formal schema with semantic web and linked data compatibility.

NMDC adopted many MIxS terms to describe biosamples but recognized the potential for higher levels of enforceable formality in the MIxS standard. This led to a 2020 effort to express MIxS in RDF, which has since evolved into a campaign to express MIxS in the LinkML language, which can be converted into RDF. Multiple stakeholders from NMDC and the wider GSC community have contributed to this new implementation of MIxS, which stands as a formal specification independent of any organization that may wish to implement portions of it. This was only possible because the contributors had a combination of skills and principles that included sustainable software development, schema testing with valid and invalid examples, the ability to gather user input, and an appreciation for predictable release processes.

5.3 Lisa Karstens (Oregon Health & Science University)[virtual]

Urobiome new MiXS extension

Over the past decade, complementary sequence-based and culture-based approaches have provided clear, reproducible evidence that the human urinary bladder has a microbial community (the urobiome) that includes bacteria, fungi and viruses. The urobiome appears to be associated with several urological disorders in the absence of clinically identifiable infections, including kidney disease, overactive bladder, and bladder cancers. However, urobiome research has had inconsistent reporting of sampling conditions and participant-related factors, which will substantially limit reuse and secondary analyses of data collected from urobiome studies. To enable urobiome research to be Findable, Accessible, Interoperable and Reusable (FAIR), consensus amongst researchers on information collected and metadata standards are needed. Towards this goal, the urobiome research community has generated a consensus statement (published in 2021) and has worked with the GSC to develop an extension of Minimum Information about any (X) Sequence (MiXS) standards.

5.4 Scott Jackson (National Institute of Standards and Technology)

Measurement Assurance for Innovation in Microbiome Science

Appreciation for the role of microbes in our lives has been growing rapidly, but the measurement science needed to understand and fully exploit microbial systems has developed at a much slower pace than the industries dependent on them demands. In all applications involving complex microbial communities, the research is hampered by the lack of standards, protocols, and technical infrastructure to allow confidence in the data and comparability. At NIST, we are developing tools to enable measurement assurance of complex microbial systems for applications in the clinic, agriculture, and the environment.

Session 6: Genomic Standards for Comparative Genomics

6.1 Chuck Cook (Global BioData Coalition, UK)

The Global Biodata Coalition: towards a sustainable biodata infrastructure

Collectively, life science data resources around the world form a vast, distributed, and interconnected infrastructure that is critical for life science and biomedical research. Unlike other scientific infrastructures, biodata resources are globally distributed and lack any kind of central coordination. The distributed nature of the infrastructure supports innovation, but lends itself poorly to the long-term sustainability of individual biodata resources and the infrastructure as a whole. The Global Biodata Coalition (GBC) brings together life science research funding organisations that recognise these challenges and acknowledge the threat that the lack of sustainability poses. They agree to work together to find ways to improve sustainability.

In the presentation I will provide an overview of the global biodata resource infrastructure, focusing in particular on challenges to providing sustained long-term funding to the resources that comprise the infrastructure. Covering some of the work that GBC has carried out to understand and classify biodata resources and the entire biodata resource infrastructure, we will outline the Global Core Biodata Resource programme and Inventory project and also introduce the stakeholder consultation processes around approaches to sustainability and open data. Finally, we will lay out the path GBC is taking to engage researchers, informaticians, funding organisations and other stakeholders in moving towards greater sustainability for these critical resources.

6.2 Naraporn Somboonna (Chulalongkorn University, Thailand)

Gut microbiota alteration associated with SLE (systemic lupus erythematosus) development

Gut microbiota play an important role in nutritional, metabolic and immunological systems. The balance of microbiological ecology in the intestine (homeostasis) is crucial for health maintenance. Dysbiosis of gut microbiota and diversity can lead to immunological disorders and associated with many diseases, including systemic lupus erythematosus (SLE); a chronic autoimmune disease effecting at least five million people worldwide.

To study gut microbiota alteration associated with SLE, fecal samples were collected from lupus-prone mice, by chemically induced (pristane) and genetically induced (FcRIIb knockout) SLE development. We found that gut microbiota of lupus-prone mice began to change at 4 months of age, and some clinical symptoms were found significantly different compared with healthy mice beginning at 6 months after microbiota changes. Differences of physicochemical conditions in each section of feces can affect the composition of gut microbiota, we investigated gut microbiota composition along mouse GI tract among different groups of mice at various ages.

Here I present our preliminary data, and suggested that gut microbiota composition was associated with SLE by increasing or decreasing of some bacteria, hence the modification of the gut microbiota, using diet, prebiotics and probiotics, might function to re-balance the composition of gut microbiota and, hopefully relieve the SLE progression in a safe manner than the immunosuppressed drug. Our ongoing experiment is to perform fecal transplant of fecal portion that was statistically denoted important to the non-SLE, as preventive and therapeutic strategies, and determine gut microbiota along clinical SLE progression, in pristane and FcRIIb knockout mice.

6.3 Nikos Kyrpides (JGI)

Microbial Comparative Genomics (MGE Standards: viruses and plasmids)

Mobile genetic elements (MGEs), which comprise viruses and plasmids, are highly abundant across all life forms, showcasing a remarkable array of genetic diversity. Their exceptional capability to mobilize is pivotal in facilitating horizontal gene transfer, a mechanism that enables the acquisition of genetic information through means other than vertical inheritance. This process fosters the exchange of genetic material among distantly related lineages, profoundly influencing evolution, ecological innovation, and the dynamics of biological communities and biogeochemical cycles. Despite their crucial role, the functional repertoire of MGEs remains largely unexplored, primarily due to a significant portion of their genes lacking known functions. Additionally, the dynamics of gene gain, loss, and exchange have not been systematically studied across large datasets.

I will summarize recent large scale efforts to characterize the diversity of MGEs, and discuss some ideas for the development of new genomic standards for these genetic elements. By setting up new genomic standards for MGEs, researchers can enhance our understanding of their functional significance and unravel their potential impact on various aspects of life, including evolution, ecological dynamics, and biogeochemical cycles. The development of such standards promises to open up exciting avenues for future research, making strides towards unlocking the full potential and implications of MGEs in the world of genetics and beyond.

6.4 Rob Finn (EMBL-EBI, UK)

Towards producing representative genome catalogues for microbial communities

An increasingly common output arising from the analysis of metagenomic datasets is the generation of metagenome-assembled genomes (MAGs). However, the discovery of MAGs is problematic and there are no requirements to archive them in an INSDC database, and comparison of these MAG collections is hampered by the lack of uniformity in their generation, annotation and storage. To address this, we have developed MGnify Genomes, a growing collection of biome-specific microbial genome catalogues generated using MAGs and publicly available isolate genomes from a common biome. Strategies for improving these catalogues and subsequent expansion will be discussed, as well as an outline of how these can be used to contextualise new datasets.

6.5 Phil Hugenholtz (University of Queensland)

The Genome Taxonomy Database (GTDB) initiative to establish a standardised microbial taxonomy

Naming of microorganisms (nomenclature) is highly standardized through nomenclatural codes such as the International Code of Nomenclature of Prokaryotes (ICNP), which have rules governing how names are formed and used. By contrast, classification of microorganisms (taxonomy) is completely unregulated to ensure freedom of taxonomic opinion. This is due in part to the recognition that taxonomy could become methodologically outdated if fixed in time. However, we have entered the age of genome-based taxonomy, and genomes are the most fundamental blueprints of life making it unlikely that a widely accepted alternative methodology resulting in a radically different and improved taxonomy will be developed. I suggest that now is the time to adopt a standardized taxonomic framework based on comparative analysis of genome sequences, or at least we should establish a primary taxonomic reference ('Tassonomia Franca') that will facilitate unambiguous scientific communication of microbial diversity.

Platinum Sponsor Speakers Abstracts

S.1 Oxford Nanopore Thidathip Wongsurawat

Game-changer in Epigenetics

The application of DNA methylation patterns holds a considerable potential in cancer care, notably improving the processes of classification, diagnosis, prognosis, and treatment response prediction. Oxford Nanopore Technologies (ONT) stands uniquely positioned in this regard, capable of detecting DNA methylation directly from the raw data without additional chemical treatments and offering 'real-time methylation analysis'. This represents an advantage over most next-generation sequencing technologies, which cannot directly distinguish between methylated and unmethylated nucleotides in native DNA. In the GSC23, I will present the application of nanopore technology, focusing on the methylation detection in a key biomarker in cancer samples.

S.2 PacBio Zuwei Qian

Shifting paradigms with PacBio HiFi sequencing

The combination of high accuracy, long read length, kinetic information and evenness of coverage makes PacBio HiFi long read sequencing a unique sequencing technology platform, providing best-in-class performance in de novo assembly, calling of all variant types, long-range allelic phasing, 5mC methylation calling, full-length transcriptomes (including single cell level), and many other applications. This technology is poised to set the standard for genomics research where comprehensive detection of all variant type is a must. In this presentation, highlights of application examples and opportunities leveraging these features for human, plant and animal genomic researches and associated computational solutions will be presented.

S.3 Waters Yan Ting Lim

Waters' solutions for Omics Analysis

Omics as a scientific discipline identifies, describes, and quantifies the biomolecules and molecular pathways that contribute to the form and function of cells and tissues. This can be done in a targeted or untargeted way or alternatively using imaging techniques to map these molecules to their distribution in tissue. This presentation provides an overview of Waters' portfolio for Omics Analysis and explore how the latest innovations are being used to help scientists better characterize their samples.

S.4. Zymo Kris Locken

A Practical Guide to Microbiome Reference Standards

Microbiome data is being generated at an unprecedented pace. In many cases, a lack of proper controls or comparison to microbiome reference materials means that important and high-impact conclusions cannot be reproduced or reliably compared to similar data sets. Microbiome standards are imperative for microbial community profiling and analysis. Whereas the microbial compositions of experimental samples are variable and often unknown, microbiome standards provide an accurate, and consistent measurement as a basis for comparison. Different Types of microbial reference materials including both whole cell and DNA mock communities, spike in controls and microbiome reference materials are useful for assessing bias at different steps of the microbiome analysis workflow.

S.5. GenePlus Svetta Kwan (ThermoFisher)

The Impact of Rapid Next-Generation-Sequencing on Precision Oncology.

Over the last two decades, with the evolving targeted therapies in the Oncology and Haematology-Oncology landscape, more cancer patients can live longer, as demonstrated in many clinical trial data on improvement in the overall survival of cancer patients. The evolution of targeted therapies has shifted the paradigm in clinical molecular testing, leading to more sequential tests and a longer waiting time and the need for more tissues. Tissue scarcity, longer turn-around-time, and rebiopsy patients are now problems for many clinicians and pathologists around the globe, impacting the overall survival of real-world cancer patients in their treatment journey. In addition, in most developing countries, patients discovered cancers are usually in Stages III and IV onwards, which is a real battle against time. The need to democratize In-House Next-Generation-Sequencing (NGS) to improve actual personalized medicine and allows the development of local expertise in biomarker testing to support the future of precision medicine. Because every patient deserves informed therapy and treatment, and every patient deserves a chance to fight for their life.



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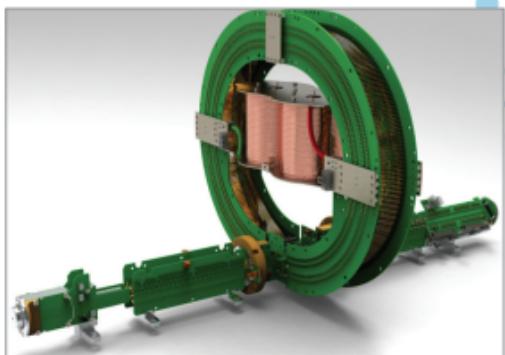
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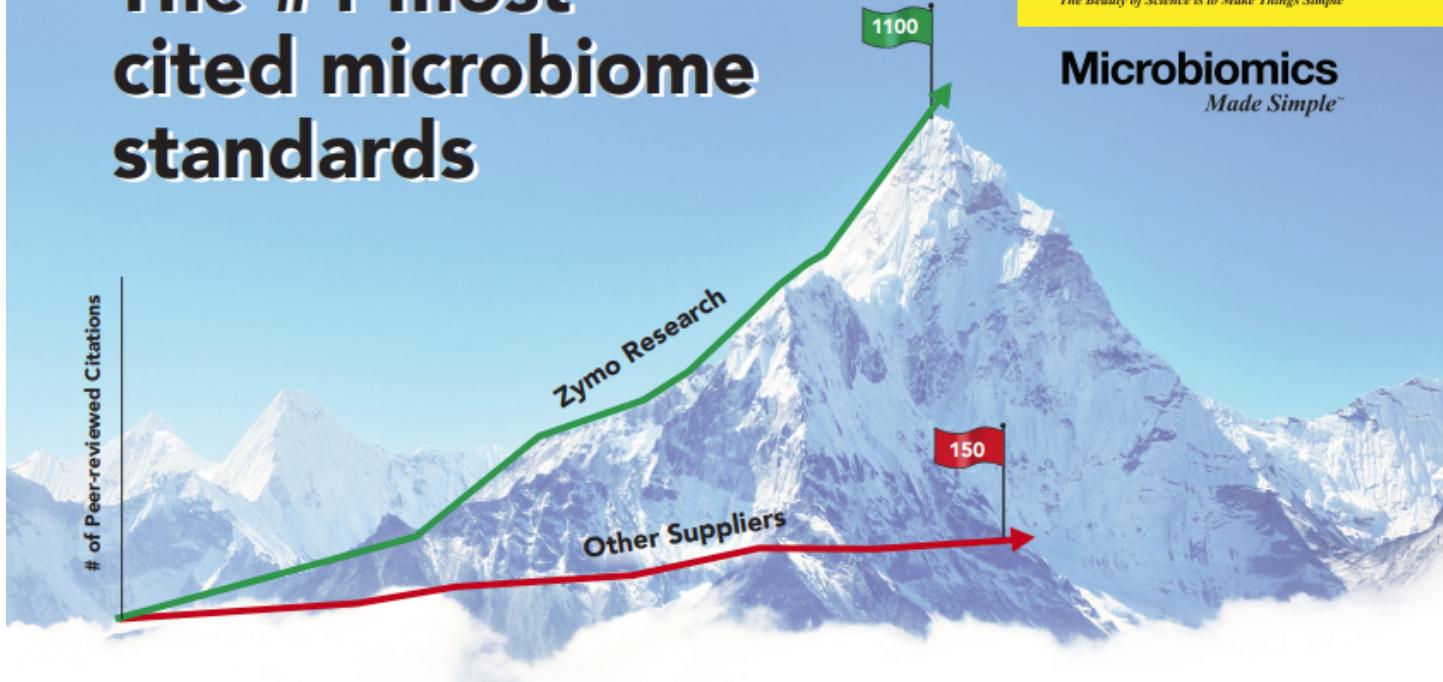
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