Asia Pacific Society of Human Genetics

APSHG Newsletter

Volume 03 | Issue 01 | June 2022

# Message from the President

**Dear members, friends and colleagues of the Asia Pacific Society of Human Genetics (APSHG)**

With the third year of the COVID-19 pandemic since its start in early 2020, the scale and speed of the COVID-19 pandemic have presented numerous challenges to people and healthcare professionals. Information related to the pandemic cited from different sources has spread through the social medias. The dissemination of information and misinformation is almost a virus in its own right. In fact, the World Health Organization (WHO) asserted that, 'we're not just fighting an epidemic; we're fighting an 'infodemic'.

Recently, much has been discussed with optimism about the de-escalation of the COVID-19 pandemic to an endemic infection. The contradiction for the discussion is the reports from many countries including countries from our region of an increasing number of infected people with the differed Sar-CoV-2 variants, which might lead to another surge of infection. The highest level of air borne viral precaution and proper vaccination should still be applied to all.

From an academic perspective, studies have provided great insights into explored the human genetic variation in host susceptibility to SARS-CoV-2 and the genetic variants of the organism the world had encountered. These might be one of the critical issues of interest to our young researchers and physicians.

On that note, I would like to invite you to the next APSHG's Summer School in October 2022. The event will be held in Bangkok, Thailand on 21st October 2022.This is one of the society's regular academic activities which held every two years. We had a successful APSHG's 3rd Summer School in Taipei in August 2020 with about 200 registrants from 15 countries.

The 4th APSHG Summer School in Bangkok will be held in hybrid form with the theme of Genetics and Genomics in Medicine. Details of program contents, speakers and registration will be disseminated to members, institutes in Asia-Pacific region and in APSHG website. The registration for the event is free. Due to the current pandemic restriction rules of Thailand, space for participants from abroad is limited. Please register as soon as possible if you would like to attend the onsite meeting.

Finally, with the uncertain factors related to the next phase or the final chapter of COVID-19 pandemic, I would like to express my appreciation to all the hardworking members of this newsletter's editorial board for their contributions in getting this inaugural issue published on time.

To all APSHG members, your views are very valuable. As always, the editorial board will do their best to maintain the quality of the newsletter and strive for improvement with feedback from all members. If you wish to get in touch, email us at apshg@apshg.info.

I wish you all the best in this global hardship and hope to see you in the near future either virtually or in person in Bangkok Thailand on 21st October 2022, the 4th APSHG's Summer School.

Sincerely yours,

Than Sura
Thanyachai Sura MD MRCP
Professor of Medical Genetics
President of Asia Pacific Society of Human Genetics (APSHG)
President of Medical Genetics and Genomic Association (Thailand)

## APSHG Committee Members



## Board of Editors 2022

**Volume 03| Issue 01| Year 2022**



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## Message from the Editor-in- Chief

Dear members,

The time for change is now. I am happy to announce a move from text-based content of the newsletter to a more graphic visual representations in the hope of capturing and keeping our readers' attention, and conveying more academic information, data, or knowledge that present complex information quickly and in an easily digestible manner.

May the aesthetic change affect and impact our members as engaged content consumers where words of past publications alone may fail to accomplish.

In short, make it visual: easy to consume, easy to share.

If you have any comments, information or news that you would like to share in the newsletter, please email it to me at zilfalil@gmail.com.

**Prof. Zilfalil Bin Alwi**
Editor-in-Chief
APSHG Newsletter

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Journal of Personalized Medicine

MDPI

*Article*

## Global Globin Network Consensus Paper: Classification and Stratified Roadmaps for Improved Thalassaemia Care and Prevention in 32 Countries

**Bin Hashim Halim-Fikri [1,†], Carsten W. Lederer [2,†], Atif Amin Baig [3], Siti Nor Assyuhada Mat-Ghani [4], Sharifah-Nany Rahayu-Karmilla Syed-Hassan [1], Wardah Yusof [1], Diana Abdul Rashid [5], Nurul Fatihah Azman [5], Suthat Fucharoen [6], Ramdan Panigoro [7], Catherine Lynn T. Silao [8,9], Vip Viprakasit [10], Norunaluwar Jalil [11], Norafiza Mohd Yasin [12], Rosnah Bahar [13], Veena Selvaratnam [14], Norsarwany Mohamad [5], Nik Norliza Nik Hassan [4], Ezalia Esa [12], Amanda Krause [15], Helen Robinson [16], Julia Hasler [17], Coralea Stephanou [2], Raja-Zahratul-Azma Raja-Sabudin [18], Jacques Elion [19], Ghada El-Kamah [20], Domenico Coviello [21], Narazah Yusoff [22], Zarina Abdul Latiff [23], Chris Arnold [24], John Burn [25], Petros Kountouris [2], Marina Kleanthous [2], Raj Ramesar [26], Bin Alwi Zilfalil [27,*] and on behalf of the Global Globin Network (GGN) [‡]**

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check for updates

Citation: Halim-Fikri, B.H.; Lederer, C.W.; Baig, A.A.; Mat-Ghani, S.N.A.; Syed-Hassan, S.-N.R.-K.; Yusof, W.; Abdul Rashid, D.; Azman, N.F.; Fucharoen, S.; Panigoro, R.; et al. Global Globin Network Consensus Paper: Classification and Stratified Roadmaps for Improved Thalassaemia Care and Prevention in 32 Countries. *J. Pers. Med.* **2022**, *12*, 552. https://doi.org/10.3390/jpm12040552

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https://doi.org/10.1186/s13073-021-01004-8

Genome Medicine

SOFTWARE Open Access

## ClinGen Variant Curation Interface: a variant classification platform for the application of evidence criteria from ACMG/AMP guidelines

Christine G. Preston[1†], Matt W. Wright[1†], Rao Madhavrao[1], Steven M. Harrison[2], Jennifer L. Goldstein[3], Xi Luo[4], Hannah Wand[5], Bryan Wulf[1], Gloria Cheung[1], Mark E. Mandell[1], Howard Tong[1], Shaung Cheng[1], Michael A. Iacocca[1], Arturo Lopez Pineda[6], Alice B. Popejoy[6], Karen Dalton[7], Jimmy Zhen[7], Selina S. Dwight[8], Lawrence Babb[2], Marina DiStefano[2], Julianne M. O'Daniel[3], Kristy Lee[3], Erin R. Riggs[9], Diane B. Zastrow[10], Jessica L. Mester[11], Deborah I. Ritter[4], Ronak Y. Patel[12], Sai Lakshmi Subramanian[12], Aleksander Milosavljevic[12], Jonathan S. Berg[3], Heidi L. Rehm[2,13], Sharon E. Plon[4,12], J. Michael Cherry[14], Carlos D. Bustamante[6,14], Helio A. Costa[1,6*] and on behalf of the Clinical Genome Resource (ClinGen)

**Abstract**

**Background:** Identification of clinically significant genetic alterations involved in human disease has been dramatically accelerated by developments in next-generation sequencing technologies. However, the infrastructure and accessible comprehensive curation tools necessary for analyzing an individual patient genome and interpreting genetic variants to inform healthcare management have been lacking.

**Results:** Here we present the ClinGen Variant Curation Interface (VCI), a global open-source variant classification platform for supporting the application of evidence criteria and classification of variants based on the ACMG/AMP variant classification guidelines. The VCI is among a suite of tools developed by the NIH-funded Clinical Genome Resource (ClinGen) Consortium and supports an FDA-recognized human variant curation process. Essential to this is the ability to enable collaboration and peer review across ClinGen Expert Panels supporting users in comprehensively identifying, annotating, and sharing relevant evidence while making variant pathogenicity assertions. To facilitate evidence-based improvements in human variant classification, the VCI is publicly available to the genomics community. Navigation workflows support users providing guidance to comprehensively apply the ACMG/AMP evidence criteria and document provenance for asserting variant classifications.

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## Attitudes and training needs of oncologists and surgeons in mainstreaming breast cancer genetic counseling in a low-to-middle income Asian country

Yong-Quan Lee [1], Sook-Yee Yoon [1], Tiara Hassan [1], Heamanthaa Padmanabhan [1], Cheng-Har Yip [2], Wee-Teik Keng [3], Meow-Keong Thong [4], Muhammad Azrif Ahmad Annuar [5], Nur Aishah Mohd Taib [6], Soo-Hwang Teo [1]

Affiliations + expand

PMID: 35481858 DOI: 10.1002/jgc4.1579

### Abstract

With the advent of poly-ADP-ribose polymerase inhibitor (PARPi) therapies, the focus of genetic testing for breast, ovarian, and other cancers has shifted from risk management to treatment decision-making in high-resource settings. Due to the shortage of genetic counselors worldwide, alternative ways of delivering genetic counseling have been explored, including training nongenetics healthcare professionals (NGHPs) to provide genetic counseling. However, little is known about the feasibility of adopting such models in healthcare settings with insufficient specialists, where population health literacy is low and where access to new therapies may be limited. In this study, we evaluated the attitudes, considerations, and self-efficacy of oncologists, breast surgeons, and general surgeons in mainstreaming breast cancer genetic counseling in Malaysia, a middle-income Asian country with a universal healthcare system. We developed a 32-item survey via a modified Delphi method, which was then distributed via a purposive and network sampling approach. While 77% of respondents expressed interest in providing breast cancer genetic counseling, 85% preferred to refer patients directly to genetic services for genetic counseling and testing. The main considerations for mainstreaming were the cost of genetic testing and PARPi therapy, as well as the availability of support from genetics professionals. Respondents reported a lack of confidence in communicating genetic risk, particularly to patients with poor health literacy, and in the clinical management of patients with variants of uncertain significance. Our results highlight the urgent need to train more NGHPs in providing genetic counseling and testing in low-to-middle income countries, and suggest that the mainstay for genetic counseling in this setting may be for risk management rather than access to PARPi therapy.

**https://doi.org/ 10.1002/jgc4.1579**

Review Article
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ANNALS OF LABORATORY MEDICINE

## Current Issues, Challenges, and Future Perspectives of Genetic Counseling in Korea

Namhee Kim, M.D.[1], Sun-Young Kong, M.D., Ph.D.[2,3], Jongha Yoo, M.D., Ph.D.[4], Do-Hoon Kim, M.D., Ph.D.[5], Soo Hyun Seo, M.D.[6], and Jieun Kim, M.D., Ph.D.[7]

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Genetic testing has become increasingly integrated into all areas of healthcare, and complex genetic testing usage continues to grow; thus, the demand for genetic counseling (GC) is likely to increase. However, it is unclear whether the current clinical GC capacity is sufficient for meeting the existing demand. This review describes the current issues, challenges, and future perspectives of GC in Korea based on a professional survey conducted among laboratory physicians. In view of the growing GC demand in the clinical setting, participants expressed a concern about the lack of support from the national healthcare insurance policy and legal requirements, such as certification, for GC practice. The implementation of genetic testing in the overall healthcare system in Korea is in an early phase. Proper implementation can be achieved through education and training of specialists, collaboration among healthcare personnel, proper regulatory oversight, genomic policies, and public awareness. Understanding the current GC capacity, issues, and challenges is a prerequisite for effective strategic planning by healthcare systems considering the expected growth in the demand for clinical genetic services over the next few decades.

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**Key Words:** Genetic counseling, Genetic services, Specialists, Healthcare system, Policy



### INTRODUCTION

Genetic testing usage in routine clinical practice has greatly increased over the past decade. As genetic testing expands with the growth of new genetic technologies, there is an emerging need for genetic counseling (GC). Although the demand for GC is increasing, and 94.2% of laboratory physicians concede the advantages of GC performed by professional personnel, several challenges and concerns regarding GC exist, especially regarding the need for support from national healthcare insurance policy and for laying out legal requirements for GC to ensure standardization and quality [1].

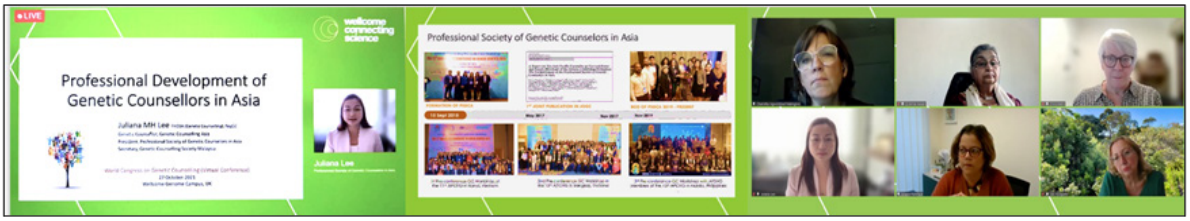
We assessed the status of GC and its present and future challenges in Korean hospitals based on professional surveys. An electronic survey was designed and conducted among 54 certified laboratory physicians associated with Korean Society of Laboratory Medicine between July and September 2020. The involvement of human participants was reviewed and approved by the Institutional Review Board of Soonchunhyang University Seoul Hospital, Seoul, Korea (IRB approval number 2020-07-002). The participants provided written informed consent to participate in the study. The contents of the survey responses were sufficient to complete a descriptive assessment and gain an in-depth understanding of the GC status in Korea. The ques-

**https://doi.org/10.3343/alm.2022.42.3.314**

**PSGCA Activity Report (July 2021- July 2022)**

**1. Participation in the 6th Board of Genetic Counseling – India (BGC-I) (Virtual Conference) 2- 4 July 2021**
In this virtual conference themed "Genomics and Genetic Counseling: Value in Health Care", the organiser invited PSGCA members, Dr Ma-am Joy Tumulak and Breana Cham to present as speakers. Dr. Kunal Sanghavi, Juliana Lee, Sook-Yee Yoon and Niby Elackatt also participated as chairpersons and panelist in the conference.

**2. Participation in the World Congress of Genetic Counselling (Virtual Conference) 27-28 October 2021**
The organiser of the third world congress on genetic counseling invited President of PSGCA, Juliana Lee to present on the "Professional Development of Genetic Counsellors in Asia". The speakers and panel discussions provided a global perspective and valuable information on the practice of genetic counseling in various countries. To encourage a global audience of professionals interested in genetic counseling, registration for this conference was waived. Congress recording is made HERE.




**3. PSGCA 4th General Annual Assembly, 4 December 2021**
In the 4th General Annual Assembly held virtually, the PSGCA members were updated on our past activities, financial and membership reports, reviewed and approved proposed changes to the PSGCA constitution. The outcome of the meeting are as follows:
a. The membership voted to have the current board of directors hold over their positions till the next general meeting.
b. Treasurer Saahil Kejriwal stepped down as Treasurer and Yoyo Chu, Treasurer Elect took over the position as Treasurer.
c. Two new directors, Suraj Manikandan as Secretary Elect and Edbert Jasper Jover as Treasurer Elect were elected.
d. PSGCA revised its vision and mission as approved by the membership.

Vision: Advancing and mainstreaming the genetic counseling profession in Asia for equitable access to quality genetic counseling services.
Mission: Empowering genetic counselors to develop and lead in the provision of genetic counseling services


Country Representatives

Hong Kong – Kelvin Chan
India – Niby Elackatt
Indonesia – Prof Sultana MH Faradz
Japan – Yasue Horiuchi
Malaysia – Rifhan Azwani
Myanmar – Dr Sann Lin Ko
Philippines – Dr Ma-am Joy Tumulak
Singapore –Jiing-Ying Lim

**4. Forum: Professional Development of Genetic Counselors in Asia, 4 December 2021**


This forum was co-organised by PSGCA together with the Board of Genetic Counseling India (BGC-I), Genetic Counselling Society Malaysia (GCSM), Indonesia Society of Genetic Counseling (ISGC) and Philippines Society of Genetic Counselors (PSGC). The international panel covered topics on the current status, professional issues, and career pathway of genetic counselors in the Asia-Pacific region. We hope to collaborate with the professional societies especially to help establish local credentialing pathway for genetic counselors practicing in Asia.

**5. Participation in the 7th Board of Genetic Counseling-India (BGC-I), 1-3 July 2022**

The BGC-I conducted its 7th Annual Conference (International) in a hybrid mode in collaboration with JSS Medical College, JSS Academy of Higher Education and Research, Mysore, India. The conference was insightful with presentations on education and training in genetic counseling and application of clinical genetics across various medical specialities. We thank the organisers for hosting PSGCA member, Sook-Yee Yoon in India and invited speakers, Juliana Lee, Niecy Ganza Bautista and Yoyo Chu as panelist.

**6. Participation in the "International Exchange Committee Program" at the 46th Annual Meeting the Japanese Society for Genetic Counseling (JSGC), 3 July 2022**

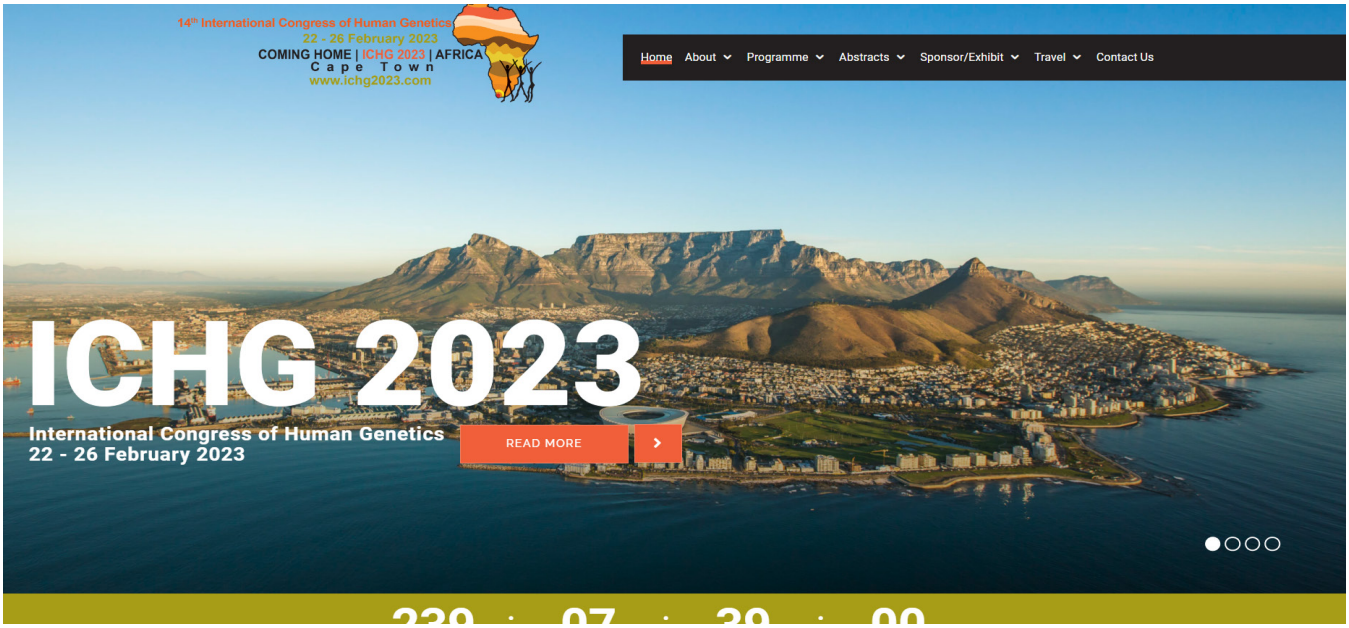
We were delighted to be invited by the Japan Society for Genetic Counseling (JSGC) to participate in a special international program in their annual meeting. The session was moderated by Prof. Masakazu Nishigaki (Professor, Department of Genetic Counseling, International University of Health and Welfare Graduate School of Health and Welfare Sciences, Japan) and Ms. Yasue Horiuchi (Chairperson, International Exchange Committee of JSGC, Japan)


The invited speakers presented the titles as below:

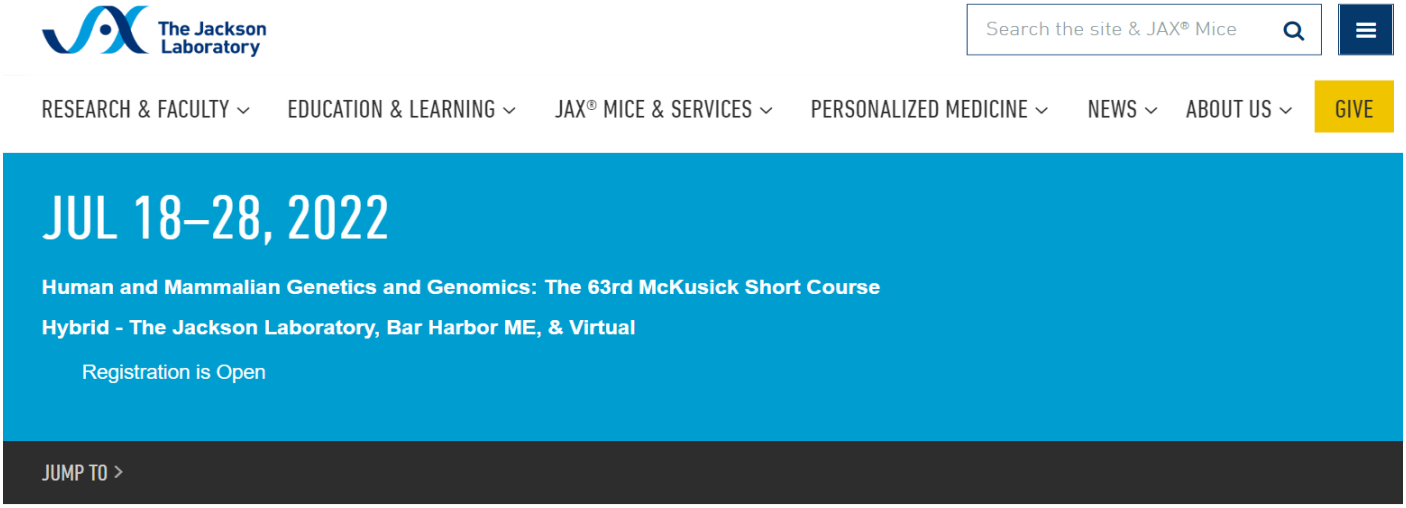
1. "Professional Development of Genetic Counselors in Malaysia" - Ms. Juliana Lee (President, PSGCA)
2. "Opportunities and Future Directions for Genetic Counseling in the Philippines" Mr. Peter J. Abad (President, Philippines Society of Genetic Counselors)
3. "Genetic Counseling Moving Ahead - Indian Scenario" Dr. Annie Hasan (President, Board of Genetic Counselors India)
4. "Current situation and challenges of the genetic counseling system in Taiwan" Prof. Yin-Hsiu Chien (Joint Committee of the Taiwan Human Genetic Society)

Followed by a panel discussion on professional development efforts in respective countries and future collaboration among the societies.

- **14th International Congress of Human Genetics (ICHG2022), Cape Town, South Africa , 22 - 26 February 2023**




**Human and Mammalian Genetics and Genomics: The 63rd McKusick Short Course, 18-28 July, 2022 Bar Harbor, US (Hybrid)**




For nearly 6 decades the 'McKusick Short Course' has been the most influential annual training event in medical genetics. The course was initiated by Dr. Victor McKusick and Jackson Laboratory Faculty and remains a joint effort of Jackson Laboratory and Johns Hopkins University. A large number of the more than 6,000 alumni are now leaders in their fields and have returned to teach as faculty members.

**Viral Genomics and Bioinformatics (Asia), 22 – 26 Aug**
**Virtual**

wellcome connecting science

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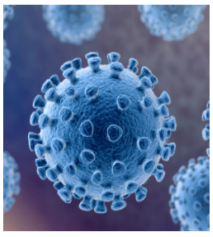


## Viral Genomics and Bioinformatics (Asia)

**22–26 August 2022**

Virtual Course

**Hands-on training in viral genome sequence analysis and interpretation of large-scale sequencing genomics data with a spotlight on SARS-CoV-2.**

Registration and deadlines

Deadlines

Application deadline
Closed

Questions?

Email the organiser

+44 (0)1223 496910

Already registered?
Log into your account.

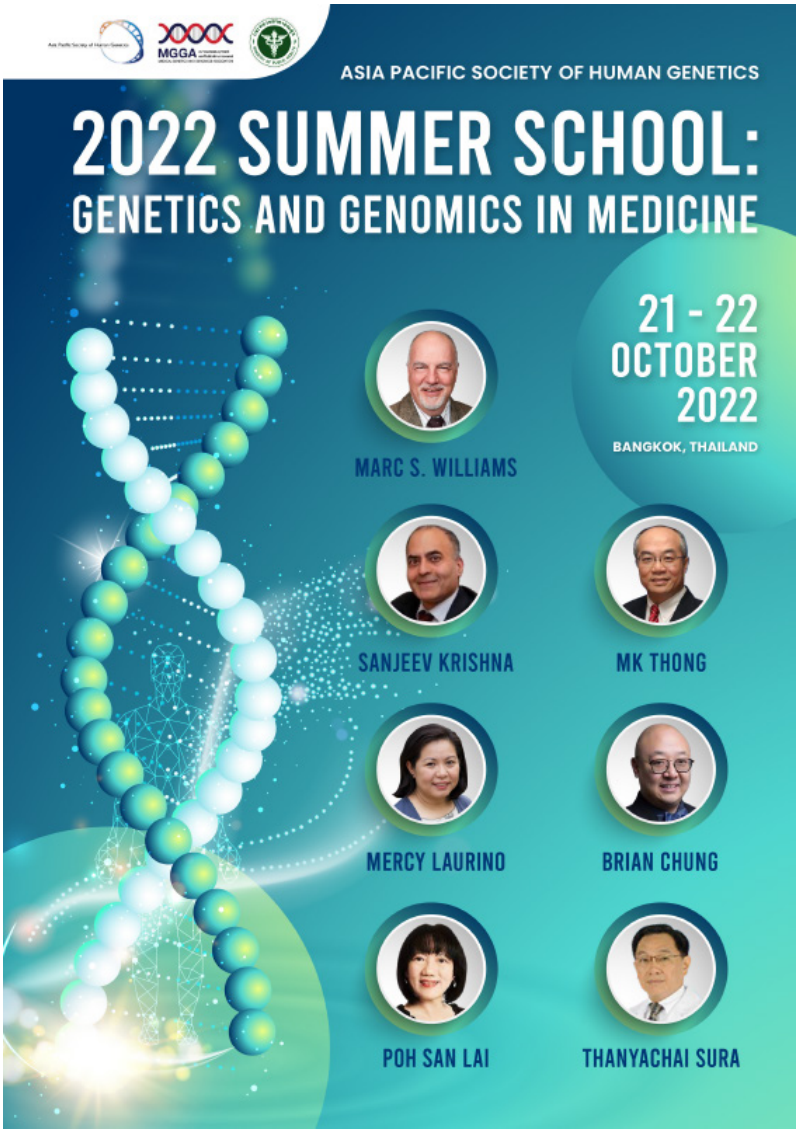
Collaborating with global partners

- **The 3rd Indonesian Society of Human Genetics (InaSHG) Annual Meeting in conjuction with The 1st Indonesian Society of Genetic Counselor (ISGC) Conference, September 9-11, 2022**

Home Committee Program Invited Speaker Free Paper Registration Accomodation Sponsorship Download Contact


Second Announcement
InaSHG
ISGC
Indonesian Society of Genetic Counselors
IDI Accredited*
The 3rd Indonesian Society of Human Genetics (InaSHG) Annual Meeting
in conjuction with
The 1st Indonesian Society of Genetic Counselor (ISGC) Conference
Enhancing Precision Medicine: The Role of Big Data for Genomic Disease Management
September 9th - 11th , 2022
08.00 - 17.00 WIB
Offline Meeting, at
Faculty of Medicine
Universitas Airlangga
REGISTER NOW
ONLINE REGISTRATION
www.3rd-annualmeeting-inashg2022.com
IMPORTANT DATE
Abstract submission (extended): Until July, 15th 2022
Early bird (extended): Until August, 15th 2022
Normal registration: August, 16th - Sept, 8th 2022
1. PRE-CONFERENCE WORKSHOP (PCW)
Friday, September 9th, 2022
2. THE 3rd InaSHG ANNUAL MEETING
Saturday, September 10th, 2022
3. THE 1st ISGC CONFERENCE
Sunday, September 11th, 2022
CALL FOR PAPER COMPETITION ORAL AND POSTER)
Selected papers will have chance to be published in scientific journal. The winner for the best oral/poster presentation will walk home with cash rewards!
REGISTRATION FEE
Symposium Only
General participant
Member InaSHG/ ISGC
Student
Early bird (IDR) 1.200.000 900.000 500.000
Normal (IDR) 1.400.000 1.050.000 750.000
Bundling Symposium & Workshop
General participant
Member InaSHG/ ISGC
Student
Early bird (IDR) 1.500.000 1.125.000 750.000
Normal (IDR) 2.000.000 1.500.000 1.000.000
Friday, Sept 9th, 2022
PRE CONFERENCE WORKSHOP
1 WS BASIC CONVENTIONAL CYTOGENETICS (max 20 persons)
Practice:
Hands on basic conventional cytogenetic from cell culture to karyotyping
Lecture:
1. Introduction lecture: General cytogenetic
2. Cytogenetic protocol
3. Genetic in hematologic malignancy
4. Sex assignment & cytogenetic in infertility
5. Cytogenetic in intellectual disability
6. CMA in microdeletion or cryptic chromosome abnormality
7. Prenatal diagnosis
2 WS GENETIC COUNSELING (max 100 persons)
Lecture:
1. Experiences of Genetic Counseling in Indonesia
2. Genetic Counselor Lecture*
Role play for genetic counseling practice and interactive discussion with international and national panelists in several cases:
1. Intellectual Disability (Fragile X Syndrome)
3 WS BASIC OF NGS BIOINFORMATICS
(max 50 persons, and bring your own laptop)
Lecture:
1. NGS Data Analysis, Filtration Pipeline
2. Application of American College of* Medical Genetics and Genomics (ACMG) Classification*


Second Announcement
InaSHG
ISGC
Indonesian Society of Genetic Counselors
IDI Accredited*
The 3rd Indonesian Society of Human Genetics (InaSHG) Annual Meeting
in conjuction with
The 1st Indonesian Society of Genetic Counselor (ISGC) Conference
Enhancing Precision Medicine: The Role of Big Data for Genomic Disease Management
September 9th - 11th , 2022
08.00 - 17.00 WIB
Offline Meeting, at
Faculty of Medicine
Universitas Airlangga
Saturday, Sept 10th, 2022
THE 3rd InaSHG ANNUAL MEETING
KEYNOTE SPEAKER AND PLENARY LECTURE
Keynote Speaker:
The Role of Government in Precision Medicine
Ir. Budi Gunadi Sadikin, CHFC, CL - Menteri Kesehatan RI
Plenary Lecture:
The Importance of Stem Cell Biobank and Regenerative Medicine
Dr. dr. Ferdiansyah, Sp.OT(K) - Universitas Airlangga
Moderator
Prof. Dr. dr. Tri Indah Winarni, M.Si.Med., PA
Universitas Diponegoro
Best Practice: Precision Medicine in Indonesia (Mendelian)
Prof. Dr. dr. Damayanti Rusli Sjarif, Sp.A(K)
Universitas Indonesia
TOPICS OF PARALEL SYMPOSIUM
1. Emerging and Re-emerging Infectious Diseases
2. Reproductive Health
3. Inborn Error of Metabolism: The Most Treatable Rare Diseases
4. Stem Cells Therapy
5. Chromosomal Disorders
6. Big Data in Human Genetics
Population Genomics (Multifactorial)
Prof. dr. Herawati Sudoyo, M.S., Ph.D.
Mochtar Riady Institute for Nanotechnology
Precision Medicine: Future Direction
Prof. Lai Poh San
Genome Institute of Singapore
Exome Sequencing in Medical Genetics
Prof. Dr. Christian Gilissen
Radboud University Medical Center, Netherlands
Precision Medicine: What We Can Learn from Mendelian and Complex Genetic Disorders
dr. Gunadi, Ph.D., Sp.BA
Universitas Gadjah Mada & President of InaSHG
Sunday, Sept 11th, 2022
THE 1st ISGC CONFERENCE
KEYNOTE SPEAKER AND PLENARY LECTURE
Keynote Speaker:
The Support of Government and Indonesian Medical Association (IMA) for The Profession of Genetic Counselor in Genomic Era
dr. M. Adib Khumaidi, Sp.OT* - Indonesia Medical Association
Plenary Lecture:
Genetic Counseling Experience from India
Dr. Q. Annie Hasan, Ph.D, FNASc
President of Board Member Genetic Counseling India
Moderator
Prof. dr. Sofia Mubarika Haryana, M.Med.Sc, Ph.D*
Universitas Gadjah Mada
Genetic Counseling Education and Practice in Australasia
A/Prof. Alison McEwen - University of Technology Sydney & Human Genetic Society of Australasia
TOPICS OF SYMPOSIUM
1. Cancer Genomics
2. Genetic Counseling in Rare Disease
3. Genetic Diseases and Diagnosis
4. Genomic Counseling
Genomic Counseling: New Challenges and Opportunities in Clinical Medicine
Prof. MK Thong, MD, Ph.D, FHGSA
University of Malaya, Malaysia
Genetic Counseling in Reproductive Disorders
Prof. dr. Sultana MH Faradz, Ph.D
Universitas Diponegoro & President of ISGC
*) to be confirmed
REGISTER NOW
IMPORTANT DATE




## 22 OCTOBER 2022
### WORKSHOP: LONG READ SEQUENCING (NANOPORE TECHNOLOGY) FOR THAI REGISTRANT ONLY (30 PERSONS)

**Clinical applications of long read sequencing**

**Morning (08.30 - 12.30)**

Introduction to long read sequencing
long read sequencing workflow
long read sequencing and clinical research
long read sequencing and clinical application1 (Thailand experience)

**Lunch (12.30 - 13.30)**

**Afternoon (13.30 - 16.00)**

long read sequencing and clinical application2 (Thailand experience)
long read sequencing and clinical application3 (Thailand experience)
Facility tour of sequencing center in BKK (to be confirm, depends on availability and COVID situation)

| | | |
|---|---|---|
| **Host** | : | Asia Pacific Society of Human Genetics (APSHG) |
| **Co Hosts** | : | Medical Genetics and Genomic Association (Thailand)<br>Ministry of Public Health (MOPH)<br>The Royal College of Physicians of Thailand (RCPT)<br>Thailand Convention and Exhibition Bureau (TCEP) |
| **Venue** | : | Pullman Hotel Bangkok |
| **Chair** | : | Thanyachai Sura |
| **Co-chair** | : | Duangruedee Wattanasirichaikul<br>Brian Chung |

**Local organizing committee**

Duangruedee Wattanasirichaikul
Kanya Suphapeetiporn
Vorasak Chotilersak
Atchara Sathienkijkanchai
Nithiwat Vatanavicharn
Prasit Powthongkum
Tipwimon Tim-Aroon
Atchara Tunteeratum
Natinee Jinawat
Donnipat Dejsupong
Jakris Eu-ahsoonthornwattana
Chularak Kuptanon
Kitiwan Rojnueangnit
Khunton Wichajarn
Kitti Buranavuth
Boonchai Boonyawat
Surakameth Mahasirimongkol
Tim Phetthong

| | | |
|---|---|---|
| **Secretariat** | : | Atchara Tunteeratum<br>Prasit Powthongkum |
| **Scientific committee** | : | Prasit Powthongkum<br>Donniphat Dejsuphong<br>Kitti Buranavuth |
| **International advisery Board** | : | Poh San Lai (Singapore)<br>Brian Chung (Hong Kong)<br>Sanjeev Krishna (UK) |

ASIA PACIFIC SOCIETY OF HUMAN GENETICS

# 2022 SUMMER SCHOOL:
## GENETICS AND GENOMICS IN MEDICINE

## SCIENTIFIC PROGRAM : FRIDAY 21 OCTOBER 2022

### SESSION 1

**Chair: Duangruedee Wattanasirichaikul, Thailand**

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|---|---|---|
| 08.00 - 08.15 | **Opening Speech** | Thanyachai Sura<br>*President of Asia Pacific Society of Human Genetics* |

**Chair:Prasit Powthongkum, Thailand/ Yin-Hsiu Chien, Taipei**

| | | |
|---|---|---|
| 08.15 - 09.00 | **Translation Genomics Medicine: from research to practice** | Mark Williams<br>*President of ACMG. Professor and Director Emeritus Genomic Medicine Institute Geisinger USA* |
| 09.00 - 09.45 | **Genetics and Genomic testing technology** | Sanjeev Krishna<br>*Professor of Infectious Department St. George's Hospital University of London* |

### SESSION 2

**Chair:Catherine Lynn Silao, Philippines/Jakris Eau-ahsunthornwattana, Thailand**

| | | |
|---|---|---|
| 09.45 - 10.30 | **Genomic Medicine in Asia** | Poh San Lai<br>*Emeritus President of APSHG* |
| 10.30 - 11.15 | **Genetics and Genomic in Philippines** | Eva M. Cutiongco-de la Paz<br>*Immediate Past President of APSHG* |
| 11.15 - 12.00 | **Genomics and syndrome recognition** | Brian Chung<br>*President Elect APSHG* |
| 12.00 - 13.00 | **Lunch Break** | |

### SESSION 3

**Chair:Sultana Faradz , Indonesia/ Tipwimon Tim-Aroon, Thailand**

| | | |
|---|---|---|
| 13.00 - 13.45 | **Rare Diseases and Public Health Policy ( Where we are now?)** | Duangruedee Wattanasirichaikul<br>*Professor of Pediatrics Thai Rare Diseases Foundation.* |

**Exome Sequencing Nanopore Technology**

| | |
|---|---|
| 13.45 - 14.15 | **Long read Exome sequencing** |
| 14.15 - 14.45 | **Role of long read exome sequencing in clinical practice** |
| 14.45 - 15.15 | **Long read sequencing in Rare Diseases.** |

### SESSION 4

**Chair: B.R.Lakshmi ,India/Surakameth Mahasirimongkol, Thailand**

**Genetic counseling**

| | | |
|---|---|---|
| 15.30 - 16.00 | **Genetic Counseling in Genomic Era** | M.K. Thong.<br>*Professor of Paediatrics Department of Paediatrics Faculty of Medicine University of Malaya* |
| 15.30 - 16.15 | **Cancer Genetics and Genetic Counseling** | Mercy Laurino<br>*Director Cancer Genetics and Prevention at Seattle Cancer Care Alliance* |
| 16.15 - 17.00 | **Genetic Counseling in Cancer Genetics :experience in Thailand** | Atchara Tunteeratum<br>*Assistant Professor and clinical Geneticist Ramathibodi Hospital Bangkok, Thailand* |
| 17.00 - 17.10 | **Closing Remark** | Thanyachai Sura<br>*President of Asia Pacific Society of Human Genetics* |

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|---|---|---|
| **Plateform** | : | Hybrid |
| **Registration** | : | Online from 20 August 2022 |
| **Registration fee** | : | Free |
| **Number of Registrant** | : | onsite 50 (First come first serve persons)*<br>online 200 persons |
| **Abstract submission** | : | Online only |

**WRITING & SUBMITTING YOUR ABSTRAC**

Author's contact details (email, phone number, postal address)
Author's and co-Authors' details (Full first and family name(s), email)

| | | |
|---|---|---|
| **Affiliation details** | : | institution / company/ University,<br>city, state (if relevant), country |
| **Abstract title**<br>**Abstract text** | : | The abstract should not exceed 250 words |
| **Submission date** | : | 21 August 2022 - 30 September 2022 |
| **Abstract categories** | : | 1. Clinical genetics and genomics<br>2. Genetic counseling<br>3. Role of genomic technology to genetic diseases. |

**Best 4th APSHG Summer School abstracts:**

1st prize: 400 USD  2nd prize: 300 USD  3rd prize: 200USD