Asia Pacific Society of Human Genetics

APSHG Newsletter

Volume 05 | Issue 01 | August 2025

Message from the President

June 23, 2025

Dear APSHG Community,

As we approach our biannual conference in Indonesia, I am thrilled to reflect on the remarkable achievements our society has accomplished this year and invite you to join us for what promises to be an exceptional gathering.

Our society continues to flourish through the dedication and innovation of our members. This summer, we celebrated the tremendous success of our summer school in India, expertly organized by Dr. Lakshmi. This educational initiative exemplified our commitment to building capacity and fostering the next generation of human geneticists across the Asia Pacific region.

We have also launched two groundbreaking initiatives that showcase our collaborative spirit. The Asia Pacific collaborative project on rapid genome sequencing for NICU, known as N-care and led by Dr. Nina Lee, represents a transformative approach to neonatal care that will benefit countless families across our region. Additionally, we are proud to have launched the APSHG HKGI Distinguished Lecture Series, which provides our community with access to cutting-edge research and thought leadership from distinguished experts in our field.

The upcoming conference in Indonesia offers an unparalleled opportunity to build upon these successes, share new discoveries, and strengthen the collaborative networks that make our society so vibrant. Under our theme " Equitable Future in Genomic Medicine: Innovation, Leapfrogging, and Advancing Technologies for Humanity" we will explore advances in precision medicine, prenatal genomics, bioinformatics, rare disease diagnostics, gene therapy, and the ethical, social, and legal dimensions of genomics.

I encourage each of you to participate in this vital gathering where we will shape the future of human genetics together. Your expertise, insights, and collaborative spirit are essential as we work toward making genomic medicine accessible and effective for all.

Dr. Brian Chung
President, Asia Pacific Society of Human Genetics

APSHG Committee Members

Board of Editors 2025

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Message from the Editor-in- Chief

Dear APSHG Members,

The year 2025 marks the fifth year of publication for the APSHG Newsletter. I would like to thank all APSHG Committee Members for their valuable article contributions to this edition.

As we enter the third quarter of 2025, we have successfully carried out numerous activities, many of which are highlighted in this issue.

In this edition, we feature a report on the APSHG–BGCI Summer School 2025, held from April 4–6, 2025, at IIT Madras, India. This newsletter also features important announcements about the Chen Travel Awards and the call for bids to host the APSHG 2027 Conference. These announcements highlight our commitment to fostering regional collaboration, supporting early-career researchers, and strengthening Asia's leadership in human genetics. We also cover major research initiatives in genomic medicine, including the N-Care Project led by Dr. Ni-Chung Lee from the National Taiwan University Hospital.

Additionally, this issue highlights a major research proposal currently under review for funding by the Wellcome Trust. This important collaborative effort focuses on rare diseases and involves multiple institutions across the Asia-Pacific region.

We are also pleased to announce that the 15th Asia-Pacific Conference of Human Genetics (APCHG) 2025 will be held in Yogyakarta, Indonesia. We encourage broad participation from around the world. The conference will feature a wide range of topics presented by distinguished experts in the field. For more information and to register, please visit the official conference website: https://tiketseminar.com/inashg/.

Thank you for your continued support and dedication to the APSHG community.

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

Published by
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APSHG–BGCI SUMMER SCHOOL 2025: EMPOWERING FUTURE LEADERS IN GENOMICS

|*Prepared by: B.R.Lakshmi, MD, India*

The APSHG–BGCI Summer School Workshop, hosted at IIT Madras from April 4–6, 2025, brought together 55 participants from across India and 2 international delegates, offering a unique blend of theory and hands-on training in genomic technologies.

Over three dynamic days, participants engaged in immersive sessions on cytogenetics, microarray, Sanger & next-generation sequencing (NGS), long-read platforms, and bioinformatics-based data interpretation. Global experts including Dr. Brian Chung and Dr. Q Annie Hasan delivered virtual keynotes, setting the tone for a forward-looking discourse on precision medicine.

The program featured rotational lab modules led by academic and industry collaborators such as Metasystems, LabIndia, and Omics Technology Pvt. Ltd., enabling real-time learning with practical exposure. A dedicated Genetic Counseling Workshop led by genetic counselors, offering role-play-based simulations for real world aplications.

A standout moment of the workshop was the classical Bharatanatyam performance of *Puranaooru* by Dr. Lakshmi Ramaswamy and team, weaving ancient Tamil literature with expressive art. The evening concluded with a Gala Dinner, offering space for networking and collaboration in an informal setting.

The final day featured a CME program attended by clinicians and faculty. It included:

- A panel discussion on cytogenomic technologies, exploring advancements, challenges, and clinical relevance
- A panel discussion on DNA sequencing, focusing on current applications, data complexities, and future directions

An industry session Talk by Dr. Laxman (ZEISS India) showcased state-of-the-art imaging solutions for cytogenetic diagnostics.

The workshop concluded with closing remarks and a vote of thanks from Dr. Janani Dakshinamoorthy and Dr. B R Lakshmi, highlighting the spirit of collaboration between BGCI, IIT Madras, academia, and industry.

Key Takeaways:

- Advanced skill-building in genomics and diagnostics
- Integrated perspective on technology, ethics, and patient care
- Enriched professional networks and cultural appreciation

TRANSFORMING RARE DISEASE DIAGNOSIS ACROSS ASIA-PACIFIC: THE N-CARE PROJECT

| *Prepared by: Ni-Chung Lee, M.D., Ph.D*

Toward the Advancing Genomics Through Long-Read Sequencing

Long read sequencing is revolutionizing the field of genomics through its cutting-edge, third-generation technology. Unlike traditional short-read platforms that sequence only a few hundred base pairs at a time, long-read sequencing enables real-time analysis of much longer DNA or RNA fragments—often spanning tens of thousands of bases—while preserving the native molecular structure. Among these technologies, Oxford Nanopore Technologies (ONT) has emerged as a leader, offering rapid detection of structural variants, accurate haplotype phasing, and deeper insights into complex genomes. Its clinical applications are particularly impactful in areas where time-sensitive and comprehensive genetic analysis is crucial, such as rare disease diagnostics and neonatal intensive care.

The utility of long-read sequencing is expanding rapidly, with ONT driving breakthroughs in diverse domains including rare disease diagnosis, cancer genomics, RNA sequencing, and epigenetic research.

The N-Care Project: Improving Diagnosis for Infants Across Asia-Pacific

To enhance pediatric care and accelerate diagnostics in critical care settings, the Asia-Pacific Society of Human Genetics (APSHG) launched the N-Care Project in February 2025. This multinational initiative leverages third-generation long-read genome sequencing to improve genetic diagnosis for critically ill infants and young children under 18 months of age. The project was initiated by APSHG President Brian H.Y. Chung and is guided by Professor Poh-San Lai. A collaborative network of leading experts from participating countries has been established, including Yin-Hsiu Chien (Taiwan), Prasit Phowthongkum and Thanyachai Sura (Thailand), Zilfalil Bin Alwi (Malaysia), Ai Ling Koh (Singapore), Catherine Lynn Silao (Philippines), Sultana Faradz (Indonesia), and B.R. Lakshmi (India). Two core sequencing laboratories are located in Taiwan (led by Ni-Chung Lee) and Thailand (led by Vorasuk Shotelersuk).

The N-Care Project is designed to provide real-time diagnostic results within 9 to 11 days, offering crucial, actionable information to medical teams caring for infants in intensive care units (ICUs). The program focuses on patients with severe or unexplained clinical conditions that suggest a genetic origin. Eligible participants include those presenting with syndromic features indicative of a genetic disorder or those identified as high-risk based on the criteria for a Brief Resolved Unexplained Event (BRUE). Informed consent for genome sequencing and data sharing is obtained from parents or legal guardians prior to participation.

By employing advanced long-read sequencing, N-Care enables the detection of a broad range of genetic variants including single nucleotide changes, indels, and complex structural variants that are often missed by short-read methods. In addition, the integration of genomic, clinical, and metabolic data supports a deeper understanding of genotype-phenotype relationships, facilitates precision treatment, and propels rare disease research throughout the Asia-Pacific region.

Together, Shaping the Future of Genomic Medicine

The N-Care Project represents more than a technological advance, it is a collaborative commitment to improving outcomes for some of the most vulnerable patients in our healthcare systems. By uniting experts across borders, we aim to accelerate diagnoses, advance rare disease knowledge, and bring new hope to critically ill children and their families. We invite clinicians, researchers, and partners across the Asia-Pacific region to join us in this mission advancing genomic medicine and building a healthier future, together.

UDN-APAC: ACHIEVING TIMELY ACCURATE RARE DISEASE DIAGNOSIS THROUGH INNOVATION AND COLLABORATION

| *Prepared by: Prof. Zilfalil bin Alwi, M.D., Ph.D*

The Asia Pacific Society of Human Genetics (APSHG) is taking a major step forward through its members' involvement in a high-impact international research grant application focused on rare diseases. The project, titled "UDN-APAC: Achieving Timely, Accurate Rare Disease Diagnosis through Innovation and Collaboration," brings together experts from eight countries across the Asia-Pacific region, Malaysia, Australia, New Zealand, Singapore, the Philippines, Indonesia, Cambodia, and Hong Kong. Representing APSHG, Prof. Zilfalil Alwi leads the initiative as Chief Investigator, supported by Brian Chung and five other Co-Investigators from across the region. The project also benefits from the expertise of Prof. John Christodoulou of Australia, who joins as Co–Chief Investigator, further strengthening the leadership and collaborative scope of this effort.

This project will establish an Undiagnosed Diseases Network across countries in the Asia Pacific region (UDN-APAC), harnessing nodes of clinical and scientific expertise, to implement robust, consistent, timely and scalable genomic sequencing to increase genetic diagnosis of individuals living with rare diseases. As part of this effort, the project also aims to strengthen human capital and infrastructure capacity in low-middle-income countries (LMICs) by fostering local expertise, investing in training, and supporting sustainable platforms for genomic diagnostics. This will help ensure that no country is left behind and that all rare disease patients including those in LMICs have access to timely and accurate diagnosis.

In partnership with the Asia Pacific Alliance of Rare Disease Organizations (APARDO) and the Malaysian Rare Disease Society (MRDS), the project will develop and refine governance processes to enable responsible and standardised sharing of expertise, data and diagnostic technology. We will systematically tackle cases through the use of state-of-the-art artificial intelligence driven platforms to analyse genomic sequencing data for those with rare disease. Additionally, the project will deploy frontier genomic technologies (including targeted RNA sequencing, proteomics and long-read sequencing) where needed. These technologies will be deployed equitably across the network, with targeted capacity-building in LMIC sites to enable local adoption, empower regional healthcare systems, and promote diagnostic equity across all participating countries.

We are hopeful that this grant application will be successful and serve as a powerful catalyst for advancing rare disease diagnosis and genomic medicine across the Asia-Pacific region. The UDN-APAC initiative stands to ignite collaboration, build capacity, and drive lasting change that ensures equitable access to cutting-edge diagnostics for all.

CHEN TRAVEL AWARDS

| Prepared by: Catherine Lynn Silao, MD, PhD, Philippines

The Chen Student Travel Award, funded with a generous donation from Professor Chen Yuan-Tsong through the Chenzyme Foundation, is given in recognition to students/young scientists with outstanding research work at the conference of the Asia Pacific Society of Human Genetics. A monetary award of USD500 will be given to a maximum of 10 award recipients to assist towards his/her travel expenses in attending the conference.

Awardees for the Chen Travel Awards will be selected by a Chen Travel Award Selection Committee comprising of international members from the APSHG Board. In order to make an application to be considered for an award, an applicant must:

1. Be registered for the 15th Asia-Pacific Conference of Human Genetics
2. Be an abstract submitter for oral and/or poster presentation (first author)
3. Inform the conference committee that he/she wishes to avail of the Chen Travel Award
4. Commit to attend the award ceremony of the conference

The Chen Travel Awardee(s) will be announced at the APSHG General Meeting Dinner of the 15th APCHG on November 8, 2025 (7 pm).

COUNTRY BID FOR THE ASIA PACIFIC CONFERENCE OF HUMAN GENETICS (APCHG) 2027

| Prepared by: Catherine Lynn Silao, MD, PhD, Philippines

To enable professionals in genetics and genomics to remain competitive in genomic sciences and to foster valuable collaborations, the Asia Pacific Society of Human Genetics organizes biennial conferences in member countries. Here, practitioners, researchers, students and industry partners engaged in human genetics and its applications convene to exchange the most recent advancements/ challenges and promote education in the field of genetics.

If you wish to propose a bid for the 2027 APCHG Meeting, it must be submitted to the board by October 3, 2025. Any submissions received after this deadline will not be taken into account. For further information about the Asia Pacific Society of Human Genetics (APSHG) Meeting Requirements and the proposal submission process, please contact us at apshg@apshg.info.

Asia Pacific Society of Human Genetics

APSHG MEMBERSHIP FEES

| *Prepared by: Catherine Lynn Silao, MD, PhD, Philippines*

The APSHG Board has approved the increase of its membership fees for the first time since 2006. Current membership fees are as follows:

- Lifetime membership: $250;
- Regular membership: $20;
- Junior membership: $10 and
- Platform fees: Credit card payments: 3.85 to 5+% ($1.20 - $10); Direct payments: 1 to 2.5% ($1 - $7).

Given that these are old rates since the society started, the board voted unanimously to increase the fees to the following -

- Lifetime membership: $300,
- Regular membership: $30,
- to keep the Junior membership at $10;
- and to add the platform fees.

The changes will start for new members signing up on or after November 8, 2025 and renewals occurring after that date. Annual fees have been one of few means to generate some income for the APSHG. The increase will allow the society to keep up with inflation and offset operational costs.

ANNUAL GENERAL MEETING (HYBRID): 8 NOVEMBER 2025

| *Prepared by: Catherine Lynn Silao, MD, PhD, Philippines*

5:30PM (UTC +5:30): India
7:00PM (UTC +07:00): Thailand/ Yogyakarta, Indonesia
8:00PM (UTC +08:00): Malaysia/Philippines/Singapore

Notice of Annual General Meeting
In accordance with the APSHG Constitution, notice is hereby given that the Annual General Meeting of the Society will be held at 7:00PM (UTC+07:00) on Saturday, November 8, 2025 at The Alana Hotel and Convention Center, Yogyakarta, Indonesia during the Asia Pacific Conference of Human Genetics 2025 conference.

AGM Agenda

- Welcome Address by Outgoing President Dr. Brian Chung
- Invited Speaker/s: Helene and Mikk Cederroth of the Wilhelm Foundation
- Chen Travel Award Prize Presentation
- Review of Accounts
- Induction of 2025-2027 Board of Directors
- Increase of APSHG Membership Fees
- Host Country for APCHG 2027
- Closing remarks of Incoming President Dr. Yin-Hsiu “Nancy” Chien

APSHG members will be able to attend the AGM live or via Zoom. The AGM is for APSHG members and invited guests only. Light refreshments will be served for on-site attendees. To facilitate planning, please

register for the AGM by 5 November 2025 by clicking on the button below.

RSVP for Annual General Meeting

[Reminder] Nominations for Leadership

Nominations are now open for 2025-2027 APSHG Board of Directors. We have several members of the Board who are at the end of their term. We are seeking nominations for the following roles:

- *President Elect** - this person will do a two-year term as President Elect, followed by two years as President and two years in the Past President role
- Three *Board Members*

Criteria for nominees are:

- Current APSHG member
- Attend Board of Director meetings (in-person or virtual)
- Align with the Society's objectives (for more info, go HERE)

- Interested in leadership experience for professional growth and development
- *Nominees for President Elect will be reviewed by the Board of Directors

Some Important Dates to Note

We also welcome feedback and suggestions on how APSHG can support you and our overall commitment. Please submit HERE.

Click the button below to access the nomination form.

Submit Nominations for APSHG Leadership

For more information, you may email apshg@apshg.info

Asia Pacific Society of Human Genetics

INTERNATIONAL

The 15th APCHG

Asia-Pacific Conference of Human Genetics 2025

"Equitable Future in Genomic Medicine: Innovation, Leapfrogging, and Advancing Technologies for Humanity"

Contact Person
dr. Siti Maisaroh
(+62 821-8640-4444)

dr. Petrus Gandi P
(+62 811-252-7777)

apchg2025@gmail.com

4th - 8th November 2025

The Alana Hotel & Convention Center Yogyakarta, Indonesia

in conjunction with:
The 5th Annual Meeting of Indonesian Society of Human Genetics (InaSHG)
The 2nd Conference of Indonesian Society of Genetic Counselor

Keynote Speakers

Prof. Sangkot Marzuki
Founder of APSHG

Mr. Budi Gunadi Sadikin*
Indonesian Minister of Health

Prof. Brian Hon-Yin Chung
President of APSHG

Plenary Speakers

Prof. Yin-Hsiu Chien
President-Elect APSHG

Prof. Dame Sue Hill
NHS England

Prof. Sultana MH Faradz
President ISGC

Prof. Herawati Sudoyo
MRIN

Prof. Gunadi
President InaSHG

Prof. Christian Gilissen
Radboud University

Dr. Bregje van Bon
Radboud University

Breana Cham
President PSGCA

Dr. Irene G. Romero
St. Vincent's Hospital
The University of Melbourne

Symposium Speakers

- **Prof. Damayanti Rusli Sjarif** (Universitas Indonesia)
- **Prof. Sofia Mubarika Haryana** (Universitas Gadjah Mada)
- **Prof. Tri Indah Winarni** (Universitas Diponegoro)
- **Prof. Erik Sistermans** (Amsterdam UMC)
- **Dr. Ivan Sini** (Universitas IPB)
- **Prof. Alison McEwen** (University of Technology Sydney)
- **Prof. Thong Meow Keong** (University of Malaya)
- **Dr. Tan Ee Shien** (KK Women's & Children Hospital)
- **Prof. Catherine Lynn T. Silao** (University of Philippines)
- **Prof. Eva Maria C.de la Paz** (University of Philippines)
- **Juliana Mei-Har Lee** (Genetic Couseling Asia)
- **Prof. Poh-San Lai** (National University of Singapore)
- **Prof. Vorasuk Shotelersuk** (Chulalongkorn University)
- **Dr. Syahrilnizam Abdullah** (MGVI, NIBM)
- **Prof. Mohammad Saifur Rohman** (Universitas Brawijaya)
- **Prof. Thanyachai Sura** (Mahidol University)
- **Prof. Ni-Chung Lee** (National Taiwan Univ. Hospital)
- **Dr. Prasit Phowthongkum** (Chulalongkorn University)
- **Dr. Koh Ai Ling** (KK Women's & Children Hospital)
- **Dr. Shih Wee Seow** (PRECISE Singapore)
- **Dr. B.R. Lakshmi** (MDCRC India)
- **Prof. Hiroyuki Awano** (Tottori University)
- **Dr. Elaine Lo** (National University Hospital)
- **Dr. Lim Weng Khong** (National University of Singapore)
- **Dr. Safarina Malik** (MRIN)
- **Dr. Tyas Hikmawan** (Universitas Gadjah Mada)
- **Dr. Rahadian Pratama** (MABBI)
- **Prof. David Aanansen** (Oxford University)
- **Prof. Muh. Nasrum Massi** (Universitas Hasanuddin)
- **Prof. Maria Inge Lucida** (Universitas Airlangga)
- **Prof. Zilfalil Alwi** (Universiti Sains Malaysia)
- **Prof. Kenjiro Kosaki** (Keio University, Japan)
- **Dr. Ahmad Utomo** (YARSI University)
- **Prof. Tiffany Boughtwood** (MCRI Australia)
- **Prof. Kazuto Kato** (Osaka University)
- **Dr. Helger Yntema** (Radboud University)
- **Dr. Irene Gallego Romero**
- **Prof. John Christodoulou** (University of Melbourne)
- **Dr. Ines Atmosukarto** (Indonesian Ministry of Health)
- **Indri Rooslamiati** (BB Binomika)

** to be confirmed*

Workshops

- **PSGCA Genetic Counselling Workshop**
  November 5th, 2025
- **Wellcome NGS Variant Calling & Interpretation Workshop**
  November 4-5th, 2025
- **Inborn Error of Metabolism: The Challenges of Newborn Screening to Diagnosis Confirmation & Prompt Treatment in Limited Resources Area**
  Course Director:
  Prof. Damayanti Rusli Sjarif
  November 5th, 2025

- **GA4GH Workshop ***
  November 5th, 2025

Symposiums

Day 1 *November 6th, 2025*
- Prenatal Genomics
- Expanded Newborn Screening in Inborn Error of Metabolism
- Rare Diseases
- Complex Genetic Disorder
- Gene Therapy

Day 2 *November 7th, 2025*
- Ethical, Social, and Legal Issues
- Omics Technologies
- Bioinformatics and AI in Genomics
- Metagenomics
- Cancer Genomics and Blood Disorders

Day 3 *November 8th, 2025*
- Precision Public Health
- Population Genomics
- Advancing Genomics in the Western Pacific Region: WHO's Vision and Initiatives
- Pharmaco and Nutrigenomics

Registration Fees

Workshops	Early Bird (USD)	Standard (USD)
PSCGA Genetic Counselling (5 Nov 2025)	100	150
Inborn Error of Metabolism from Newborn Screening to Genetic Therapy (5 Nov 2025)	100	150
NGS Variant Calling & Interpretation Workshop (4-5 Nov 2025)	Participants will be selected by Wellcome Committee (registration will be announced later)	
Global Alliance for Genomics and Health Workshop (GA4GH) (5 Nov 2025)	Participants will be selected by the Committee (registration will be announced later)	

Seminars	Early Bird (USD)	Standard (USD)
InaSHG and APSHG Member	200	300
General Non - Member	250	350
Trainee/Student (Proof by ID)	100	150

Important Dates

- Mar 27 Abstract Submission & Registration Opens
- Jun 1 : Abstract Deadline
- Jul 1 : Abstract Notification
- Jul 8 : Early Bird Deadline
- Oct 24: Late Registration Deadline

Nov 4th-8th APCHG 2025

Registration

UPDATE ON THE PROFESSIONAL SOCIETY OF GENETIC COUNSELORS IN ASIA (PSGCA) 2025

| *Prepared by: Breana Cham*

Dear colleagues, friends, and valued members of the PSGCA,

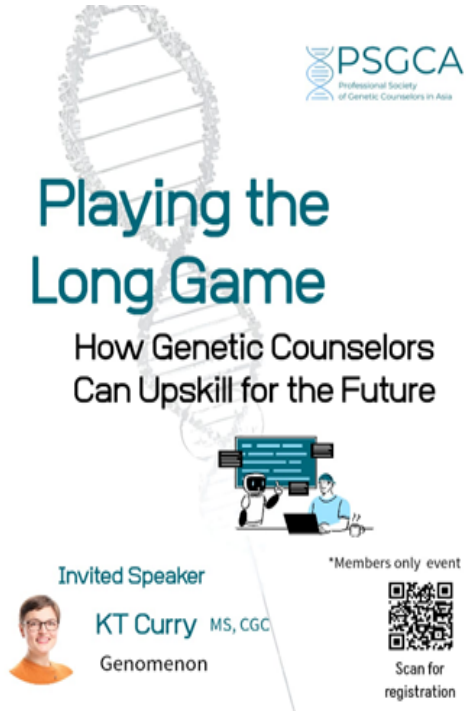
I hope this message finds you well as we move through the second half of 2025 and it is my pleasure to bring you 2 upcoming events in our calendar.

1. Upcoming PSGCA Webinar – 25 August 2025
Our next PSGCA webinar which will be held on 25 August 2025, 8pm (UTC+8). This session will focus on "Playing the long game: How genetic counsellors can upskill for the future", and will examine how genetic counsellors can stay relevant in a rapidly changing world. Please mark your calendars and register in advance—this is sure to be a thought-provoking session relevant to all practice settings.

2. Genetic Counseling Workshop – 5 November 2025, Yogyakarta, Indonesia
I am also thrilled to extend an invitation to our upcoming in-person Genetic Counseling Workshop to be held on 5 November 2025 in Yogyakarta, Indonesia. With the theme of "Pillars of Progress: Growing the Profession, Shaping the Professionals". The workshop will feature speakers and facilitators from across the region, offering insights on current and emerging career pathways for genetic counsellors, perspectives on multidisciplinary and interprofessional collaborations, and continued professional growth and development through supervision. Whether you are early in your career or an experienced genetic counsellor, this event promises meaningful takeaways for all.

In closing, I warmly encourage all current and lapsed members to renew their membership for the coming year. Your PSGCA membership not only connects you to a vibrant network of genetic counseling professionals across Asia, but your continued support ensures we remain a strong, unified voice in the region, and sustains the society's capacity to offer education, advocacy, and cross-border collaboration.

Yours Sincerely,
Breana Cham
President, Professional Society of Genetic Counselors in Asia



PSGCA Genetic Counselling Workshop 5 November 2025 - Pillars of Progress: Growing the Profession, Shaping the Professionals

- Keynote – Building Bridges, Not Walls: The Role of Technology in Expanding Genetic Counselling Services Worldwide
- Breakout sessions - Expanding Career Pathways
- Multidisciplinary teams - Collaboration in the Genomics Era
- Current Landscape of Genetic Counselling across Asia
- Genetic Counselling in Indonesia - Case Presentations
- Ongoing Professional Development - Supervision for Genetic Counsellors