International Workshop and Symposium on
Integrating Genetics in the Medical Curriculum
Waters Edge, Colombo, Sri Lanka
12-14 June 2013

organized in association with the
Human Genetics Unit, Faculty of Medicine, University of Colombo
Sri Lanka Medical Association
and the National Science Foundation, Sri Lanka
International Workshop and Symposium on
Integrating Genetics in the Medical Curriculum

Waters Edge, Colombo, Sri Lanka
12-14 June 2013
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MESSAGE ON BEHALF OF THE INTERNATIONAL GENETICS EDUCATION NETWORK

The International Genetics Education Network (IGEN) is a network of genetics education providers and those interested in genetics education for health professionals. IGEN was established in 2006 and its overall aim is to facilitate human genetics education by promoting and sharing expertise and resources.

This workshop in Colombo, Sri Lanka is the third international forum facilitated by IGEN. We are delighted to partner with the Human Genetics Unit, Faculty of Medicine, University of Colombo, Sri Lanka Medical Association and the National Science Foundation of Sri Lanka and we especially thank Professor Vajira H. W. Dissanayake for his enthusiasm and invitation to hold this workshop in his beautiful country.

We are also grateful to our commercial sponsors Illumina Singapore Pte Ltd, Alliance Global Group, Dubai, UAE, Invitrogen Bioservices India Pvt. Ltd (Life Technologies, India), Abbott Molecular and Dowell International, as well as the many providers of educational resources.

It is our pleasure to meet and interact with all of the delegates from the region. We hope that the topics and activities that are covered in the next couple of days will emphasize the importance of genomic medicine today and foster your enthusiasm for genetics education within the medical curriculum and in a doctor’s professional life beyond.

Welcome and enjoy.

The IGEN Organizing Committee:

Judith Allanson MD - Canada
Joseph D. McInerney MA, MS - USA
Sylvia Metcalfe PhD - Australia
Kunal Sanghavi MBBS, MS - USA
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MESSAGE ON BEHALF OF THE SRI LANKAN HOSTS

It gives us great pleasure to welcome all of you to Colombo for this meeting. We are particularly pleased that the International Genetic Education Network (IGEN) has chosen Sri Lankan to hold their first South Asian workshop and symposium.

Formal teaching in Medical Genetics was introduced to Sri Lanka with the establishment of the Human Genetics Unit (HGU) of the Faculty of Medicine, University of Colombo in 1983. So this landmark IGEN South Asian workshop and symposium is being held when we are celebrating the 30 years of Medical Genetics Education in Colombo.

The HGU spanned its wings outside the shores of Sri Lanka, when in 2010 we took on the task of training Clinical Geneticists from Nepal. Today Nepal is developing their Medical Genetics infrastructure around the first Clinical Geneticist in the country trained in Colombo. There is more to be done in countries in the region through such regional collaboration. We are pleased that we were able to facilitate the participation of delegates from affluent centers in the region as well as from those who are aspiring to introduce Medical Genetics to their country at this meeting.

We hope that the network that is created as a result of this meeting in Colombo would become a strong regional network in the years to come. That process can be catalyzed only through collaboration with the best in the world. IGEN has brought a galaxy of Medical Geneticists and Medical Genetics Educators from North America and Australia to this meeting; the very best in the world. We are indeed grateful to all of them for the efforts that they put into bringing this meeting to the region, and then planning for over a year and now executing it. We hope that they will be with us in the long journey of improving Medical Genetics Education in the region in the years to come.

Today Industry is emerging as a strong partner in Medical Genetics Education. We are also particularly pleased that the 'big giants' in genomics are here in Colombo at this meeting. We hope that they would continue to support us in the years to come.

Once again, let me warmly welcome all of you to Colombo and wish you a productive meeting.

Prof. Rohan W Jayasekara MBBS, PhD, C.Biol., MSB(Lond)
Founder Director, Human Genetics Unit
Dean, Faculty of Medicine
University of Colombo
Sri Lanka
# MISEQ PERSONAL SEQUENCER

<table>
<thead>
<tr>
<th>Feature</th>
<th>Description</th>
</tr>
</thead>
<tbody>
<tr>
<td>Sequencing Technology</td>
<td>Sequencing by Synthesis (SBS) chemistry</td>
</tr>
<tr>
<td>Output</td>
<td>MiSeq routinely exceeds 10Gb with a roadmap to achieve 15Gb</td>
</tr>
<tr>
<td>Cluster generation to data analysis</td>
<td>Built-in Automated workflow</td>
</tr>
</tbody>
</table>
| Workflow                  | • Single integrated instrument  
• Entire sequencing process occurs on the MiSeq  
• Does not require emulsion PCR                                                                                                                     |
| Data analysis             | Fully automated on instrument                                                                                                                                                                            |
| Accessory equipments      | No need                                                                                                                                                                                                 |
| Hands-on time             | As little as 30-45 minutes total hands-on time from genomic DNA to analyzed data                                                                                                                          |
| Read length               | up to 2 x 250 bp (Pair End Read)                                                                                                                                                                           |
| Data quality              | MiSeq has the highest quality data  
• >80% Q30 at 2x150  
• >75% Q30 at 2x250                                                                                                                                     |
| Turnaround time           | Samples to analyzed data in as little as eight hours                                                                                                                                                  |
| Instrument control computer | Integrated in the sequencer                                                                                                                                                                             |
| Light emitting diode(LED) | 530 nm , 660 nm                                                                                                                                                                                      |
| Homopolymer repeats       | As the MiSeq uses a competitive sequencing strategy where during base incorporation all bases are added simultaneously, the sequencing data is highly accurate with no issues in sequencing runs of the same base. |
| Informatics               | MiSeq comes with BaseSpace integration(cloud computing platform)                                                                                                                                       |
| Mappable Data             | MiSeq achieves >90% mappable reads                                                                                                                                                                        |
| Applications              | Single-end Sequencing  
Paired-end sequencing  
Capillary electrophoresis (CE) sequencing applications  
Amplicon Sequencing  
Small genome sequencing  
Small RNA sequencing  
RNA sequencing protocol  
De novo sequencing  
ChIP sequencing  
Metagenomics (16S rRNA-sequencing)  
Tag-based gene expression  
Ribosome profiling  
DNA imprinting and allele specific expression  
Disease specific panels |
BACKGROUND

At the 2006 International Congress of Human Genetics (ICHG) in Brisbane, a group of people with an interest in genetics education took part in a short workshop to discuss the prospect of developing an International Genetics Education Network. The purpose of the network (now called IGEN) that grew out of those discussions is to expand human genetics education at an international level by promoting and sharing expertise and resources, with its activities drawing on evidence-based educational principles.

IGEN's first global activity was a 1.5-day satellite workshop at the 2011 ICHG in Montreal. The workshop, titled “Genetics, Primary Care, and Developing Countries”, brought together teams of two (a primary-care provider and a geneticist) from developing countries with an expert faculty to consider the integration of genetic medicine into mainstream clinical practice. Thirty-five participants from 16 different countries attended the workshop. The countries represented included China, Egypt, India, Jordan, Sri Lanka, Thailand, Vietnam as well as Brazil, Canada, Chile, Cuba, Saudi Arabia, Turkey, and USA along with ten faculty members (organizers, speakers, and facilitators) from Australia, Canada, Switzerland (originally from Iraq), the UK and the USA.

The workshop commenced with a reception and a brief presentation by each participant. The following day comprised a mix of didactic and interactive sessions on public health genomics, role of advocacy, consanguinity, genetics education, and program evaluation. Participants received a pack of resources, which added to their enthusiasm for the workshop. They expressed a desire to continue the dialogue and to expand their networks. This workshop relied heavily on sponsorship from a number of sources, and delegates from developing countries received travel support to attend.

We implemented follow-up and evaluation measures in the form of a post-workshop survey and the sharing of presentations and video recordings of the sessions through an online networking group.

Discussions with the participants and feedback from their experiences, revealed a universal concern: a dearth of genetics education and tools to integrate genetics education into training programs for health professionals. Developed nations also face this challenge and have tried to address it at various levels by involving specialty trainers in the development and delivery of genetics education (Burke et al., 2005).

IGEN’s latest workshop, in Colombo, Sri Lanka in June 2013, will engage medical educators in South Asia in strategies for the integration of genetics into medical education in different settings, including formal medical education and continuing professional education. We ascertained the extent and modes of current genetics education and services in the participant countries with the help of a needs assessment survey. We are using the survey results to help guide the structure of the workshop as we identify potential measures to address any existing challenges through group discussions among the participants.
The objectives of the workshop are to:

1. Create awareness about the need for incorporating genetics into education curricula at all levels of physician education among medical educators in South Asia
2. Demonstrate strategies for medical education through the life course of the physician
3. Review the status of genetic medicine in South Asia
4. Discuss the future prospects for genetic medicine worldwide
5. Formulate strategies for implementing genetics education programs
6. Design a preliminary roadmap for international collaboration on genetics education for physicians in South Asia

We expect to adopt ask-tell-ask approach that promotes tailored conversations and development of value-based strategies to provide appropriate resources and establish a new model of collaborating with different genetics education settings (Gaster B et al., 2010). A post-workshop survey will serve as an evaluation tool, which may help us assess the impact of the workshop and provide an evidence base for implementing relevant resources and programs (Metcalf et al., 2008). We also intend to promote genetic counseling through this workshop, as genetic counselors, are well suited to address many of the challenges associated with increasing awareness about genetics education (McInerney 2008).

References:


PROGRAMME

Inauguration & Welcome Reception
Wednesday, 12 June 2013

18:00 Welcome Address on behalf of the local hosts
Prof. Rohan W Jayasekara, Dean, Faculty of Medicine

18:10 Address on behalf of IGEN
Prof. Sylvia Metcalfe, Member, Organising Committee, IGEN

18:20 Country Presentations
Bhutan
India
Malaysia
Nepal
Pakistan
Sri Lanka

19:20 Vote of Thanks
Prof. Vajira H. W. Dissanayake, Local Organiser

19.30 Welcome Reception
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Workshop: Genetics through the Life Course of the Doctor
Thursday, 13 June 2013

08.30 Registration

09:00 – 9:30 Genetics and genomics in South Asian Medical Schools
- **Speaker:** Vajira H W Dissanayake (Sri Lanka)
- **Session highlight:** Current trends, experiences, and needs assessment in introducing genetics education
- **Q/A and discussion with the audience**

9:30 – 09:45 Review of educational resources for medical educators and clinicians
- **Speaker:** Judith Allanson (Canada)

09:45 – 11:00 Problem Based Learning (PBL) for medical student/trainees
- **Speaker:** Sylvia Metcalfe (Australia)
- **Session highlight:** Examples and experiences from medical school teaching in Australia, what do medical students need to know and learn about genetics and why? Approaches to teaching genetics in medical school

11:00 – 11:15 Tea/Coffee Break

11:15 – 12:30 Interactive Group Session 1: Integrating PBL in genetics education
- Experiences and interaction with medical students/residents in your region
- Identify common themes/issues that challenge integrating genetics into medical education
- Discuss strategies for addressing these issues and facilitate this integration

12:30 – 13:15 Lunch

13:15 – 14:30 A Case Based Learning (CBL) approach to genetics education for hospital staff
- **Speaker:** Kate Reed & Vinayak Kottoor (USA)
- **Session highlight:** Successful model in promoting genetics education for hospital staff, challenges in introducing genetics education, how to best address these challenges, how to apply in other clinical settings, importance of family history in genetics and relevance of consanguinity
14:30 – 15:45 **Interactive Group Session 2: Integrating CBL into professional development for physicians**
   - Discuss strategies to encourage this integration
   - Does this apply to your setting?
     - If yes, reflect how you can apply this concept in your region
     - If no, how would you like to promote genetics education for healthcare providers in your region/institution

15:45 – 16:00 **Tea/Coffee Break**

16:00 – 16:30 **Summary from Interactive Group Sessions 1 and 2**
   - Representative from each group will present the summary

16:30 – 17:00 **Next Steps**
   - **Expert opinion:** Joseph Mcinerney (USA)
   - Lessons learned from the interactive group sessions and a perspective on the next steps – How these findings help us in facilitating genetics education

17:00 – 17:30 **Concluding Remarks**
PARTICIPANTS AT THE WORKSHOP

**Bhutan**
1. Dr. Purushotam Bhandari, Paediatrician, Central Regional Referral Hospital, Gelephug, Bhutan. purub@druknet.bt
2. Dr. Purbh Dorji, Obstetrician and Gynaecologist, National Referral Hospital, Thimpu, Bhutan. phurbd@yahoo.com

**India**
1. Dr. Madulika Kakabra, Assistant Professor, Genetics Unit, Department of Paediatrics, All India Institute of Medical Sciences, New Delhi, India. madhulikakabra@hotmail.com
2. Dr. Anurag Agrawal, Additional Director, National Board of Examinations, New Delhi, India. anurag@natboard.edu.in
3. Dr. Ashwin Dalal, Head, Diagnostics Division, Center for DNA Finger Printing and Diagnostics, Hyderabad, India. ashwinda@gmail.com
4. Dr. Partha Majumder, Director, National Institute of Biomedical Genomics, Kalyani, India. ppm1@nibmg.ac.in
5. Dr. Ratna Puri, Clinical Geneticist, Center of Medical Genetics, Sir Ganga Ram Hospital, New Delhi, New Delhi, India. ratnápuri@yahoo.com
6. Prof. Pragna Rao, Professor, Director, Postgraduate Studies, Department of Biochemistry, Kasturba Medical College, Manipal University, Karnataka, India. pragna.rao@manipal.edu
7. Prof. Nutan Kamath, Professor, Department of Paediatrics, Kasturba Medical College, Manipal University, Mangalore, India. nutankamath@yahoo.com

**Pakistan**
1. Dr. Saqib Mahmood, Assistant Professor, Department of Human Genetics & Molecular Biology, University of Health Sciences, Lahor, Pakistan. medgen@uhs.edu.pk

**Nepal**
1. Dr. Nilam Thakur, Clinical Geneticist, National Academy of Medical Sciences, Kathmandu, Nepal. nilu_thakur1212@yahoo.com
2. Prof. Ranga Bahadur Basnet, Professor of Pathology, National Academy of Medical Sciences, Kathmandu, Nepal. rangbasnet111@hotmail.com
3. Prof. Damodar Prasad Pokhrel, Vice Chancellor, National Academy of Medical Sciences, Kathmandu, Nepal. rangbasnet111@hotmail.com

**Malaysia**
1. Dr. Mohammad Zahirul Hoque, Associate Professor, Department Of Pathobiology & Medical Diagnostics, School Of Medicine, University Malaysia Sabah, Kota Kinabalu, Sabah, Malaysia. drzahir@ums.edu.my
2. Prof. Zainal Ariffin Mustapha, Deputy Dean, Department Of Pathobiology & Medical Diagnostics, School Of Medicine, University Malaysia Sabah, Kota Kinabalu, Sabah, Malaysia. drzahir@ums.edu.my
Sri Lanka

University of Colombo

1. Prof. Rohan W. Jayasekara, Dean, Faculty of Medicine and Director, Human Genetics Unit, University of Colombo. rohanwj@hotmail.com
2. Dr. Hemali Goonasekera, Senior Lecturer, Department of Anatomy and the Human Genetics Unit, Faculty of Medicine, University of Colombo. hemaliww_g@hotmail.com
3. Dr. N.D. Sirisena, Lecturer and Clinical Geneticist, Department of Anatomy and the Human Genetics Unit, Faculty of Medicine, University of Colombo. nirmalasirisena@yahoo.com
4. Dr. Dulika Sumathipala, Lecturer and Clinical Geneticist, Department of Anatomy and the Human Genetics Unit, Faculty of Medicine, University of Colombo. dulikasanjeewani@gmail.com
5. Dr. Indika Karunatilake, Director, Medical Education and Research Center, Faculty of Medicine, University of Colombo. karunatilake@hotmail.com
6. Dr. Asela Olupeliyawa, Senior Lecturer, Medical Education and Research Center, Faculty of Medicine, University of Colombo. asela_o@yahoo.com
7. Dr. Ashwini Abrew, Lecturer, Medical Education and Research Center, Faculty of Medicine, University of Colombo. ashwinie@yahoo.com
8. Dr. Nilakshi Samaranayake, Senior Lecturer, Department of Parasitology, Faculty of Medicine, University of Colombo. nilakshis35@gmail.com

University of Peradeniya

9. Prof. M. Chandrasekara, Professor of Anatomy, Faculty of Medicine, University of Peradeniya. mmsriyani@hotmail.com.
10. Dr. J. Dissanayake, Senior Lecturer, Department of Anatomy, Faculty of Medicine, University of Peradeniya. jayamkd@yahoo.com

University of Ruhuna

11. Prof. Isurani Ilayperuma, Professor in Anatomy, Department of Anatomy, Faculty of Medicine, University of Ruhuna. lisurani@yahoo.com
12. Dr. Lahiru Prabodha, Senior Lecturer in Anatomy, Department of Anatomy, University Of Ruhuna. lahiruprabodha@gmail.com

University of Jaffna

13. Dr. Shivananthini Udhayakumar, Senior Lecturer, Department of Anatomy, Faculty of Medicine, University of Jaffna. sivananthini3@yahoo.com
14. Dr. T. Chenthuran, Lecturer, Department of Anatomy, Faculty of Medicine, University of Jaffna. tchenthuran@yahoo.com
University of Kelaniya

15. Prof. Suji Salgado, Professor of Anatomy, Faculty of Medicine, University of Kelaniya. sujeesal@yahoo.com

University of Sri Jayewardenepura

16. Prof. S.G. Yasawardene, Professor of Anatomy, Department of Anatomy, Faculty of Medical Sciences, University of Sri Jayewardenapura. surangiy@hotmail.com
17. Dr. Varuni. Thennakoon, Senior Lecturer, Department of Anatomy, Faculty of Medical Science, University Of Sri Jayewardenapura. varuni16email@yahoo.com

Rajarata University of Sri Lanka

18. Dr. Asantha Jayawardena, Lecturer and Postgraduate Trainee in Clinical Genetics, Department of Anatomy, Rajarata University of Sri Lanka, Anuradhapura. smajayawardana@gmail.com
19. Dr. Sampath Pathiginige, Lecturer, Department of Anatomy, Faculty of Medicine and Allied Health Sciences, Rajarata University of Sri Lanka, Anuradhapura. paththinige@yahoo.com

Ministry of Health

20. Dr. Padmapani Padeniya, Clinical Geneticist, De Soysa Hospital for Women, Colombo. padmapanip@gmail.com
21. Dr. Subashi Karunarathne, Clinical Geneticist, Lady Ridgeway Hospital for Children, Colombo. subhashi@subhashi.com
22. Dr. Thilina Wanigasekara, Postgraduate Trainee in Clinical Genetics, Human Genetics Unit, Faculty of Medicine, University of Colombo. thilinaw71@yahoo.com
23. Dr. Samuditha Senaratne, Postgraduate Trainee in Clinical Genetics, Human Genetics Unit, Faculty of Medicine, University of Colombo. ssamudita@gmail.com
24. Dr. Niluka Dissanayake, Postgraduate Trainee in Clinical Genetics, Human Genetics Unit, Faculty of Medicine, University of Colombo. home_niluka@yahoo.co.uk
25. Dr. Nuwanthi Weerapperuma, Postgraduate Trainee in Clinical Genetics, Human Genetics Unit, Faculty of Medicine, University of Colombo. kusharanuwanthi@yahoo.com
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A Promise for Life
Symposium: An Update on Genomic Medicine
Friday, 14 June 2013

08.00  Registration

08:30 – 09:00 Genetic Medicine: What Is Possible in South Asia, and Where?
Vajira H.W. Dissanayake (Sri Lanka)

09:00 – 09:45 Panel Discussion and Audience Participation: Integration of genetic medicine into every-day practice

09:45 – 11:15 Symposium – 1: Future prospects in genetic medicine

09.45  Companion Diagnostic: Age of Personalized Medicine - Jee Hian Lim (Singapore)

10.15  Utility of FISH Panels in Classification and Diagnosis in Breast Cancer and Haematology Malignancies - Yan Chin Tai (Singapore)

10.45 – 11.15 Tea/Coffee Break

11:15 – 13:00 Symposium-2: Future prospects in genetic medicine

11.15  New dimensions of next generation sequencing - Sailesh Gochhait (India)

11.45  Illumina NGS Technology and Its Clinical Applications - Arjuna Kumarasuriyar (Singapore)

12.15  Application of targeted resequencing on cancer samples in a translational lab - Ana Carrera (Singapore)

13:00 – 14:00 Lunch

14:00 – 15:15 The Human Variome Project - Timothy Smith (Australia)

15:15 – 15:30 Coffee/Tea Break

15:30 – 16.00 Panel Discussion and audience participation: planning for the future of genetic medicine in education and practice

16:00 – 16:30 Concluding Remarks
**INTERNATIONAL FACULTY**

**Judith Allanson MD, FRCP, FRCP(C), FCCMG, DABMG**

Judith Allanson is a Professor of Pediatrics at the University of Ottawa and retired Clinical Geneticist at Children's Hospital of Eastern Ontario, Canada. She received her MB ChB degree from Liverpool Medical School and trained in Internal Medicine in Britain. Her Fellowship in Medical Genetics was completed at the University of British Columbia, Canada, under the mentorship of Dr Judith Hall. Her clinical and research interests have focused on evaluation of facial dysmorphisms and methods of syndrome identification, evaluation of genetic services, psychosocial impact of genetics services, models of genetics service delivery; and the understanding, perception and application of genetic knowledge by primary care providers. She is co-editor of a book on management of common genetic syndromes and the co-Director of GEC-KO, the Genetics Education Centre – Knowledge for Ontario.

allanson@cheo.on.ca

**Ana J Carrera MS**

Ana has worked at Illumina for the past 3 years in regional marketing and as technical trainer for APAC. Prior to Illumina, Ana worked for over 6 years as a Senior Field Application Scientist for Applied Biosystems in Washington D.C and New York, as a Senior Research Associate at the School of Computational Sciences at George Mason University in Virginia, and as a Bioinformatics Analyst at The Institute for Genomic Research (TIGR) in Maryland. She holds a Bachelor in Molecular Biology from U.C Berkeley and a Masters in Bioinformatics from the School of Computational Sciences at George Mason University.

acarrera@illumina.com

**Vajira H. W. Dissanayake MBBS, PhD**

Vajira Dissanayake is a Professor in Anatomy and Medical Geneticist at the Department of Anatomy and the Human Genetics Unit at the Faculty of Medicine of the University of Colombo, Sri Lanka. He graduated from the University of Colombo and read for his PhD at the University of Nottingham in the UK. Since returning back to Sri Lanka in 2004 he has been instrumental in developing clinical genetic and genetic diagnostic services as well as research in the field of Medical Genetics both in the public and private sectors. He established several MSc courses in the University of Colombo. They include three MSc courses in Clinical Genetics, Genetic Diagnostics, and Biomedical Informatics in collaboration with the University of Oslo, Norway and an MSc course in Regenerative Medicine in collaboration with Manipal University, India. He also founded the Asiri Center for Genomic and Regenerative Medicine of the Asiri Group of Hospitals – the largest private
hospital chain in Sri Lanka in 2006 and co-founded Credence Genomics – the first Next Generation Genomics Company in Sri Lanka in 2012. He has been helping Nepal develop their manpower in Medical Genetics by training doctors and scientists from Nepal in Clinical Genetics and Genetic Diagnostics in Colombo. He was elected a fellow of the National Academy of Sciences of Sri Lanka in 2013.

vajirahwd@hotmail.com

**Vinayak Kottoor MD**

Vinayak Kottoor is an Instructor in General Internal Medicine at Johns Hopkins University in Baltimore, Maryland. He completed his medical school training in Bangalore, India. Thereafter he completed an Internal Medicine and Pediatrics residency in Saint Louis, Missouri and a Clinical Genetics Fellowship at Johns Hopkins University. Dr. Kottoor’s areas of focus include primary care in the outpatient setting with special interests in care transition from pediatric to adult medical homes and in the integration of genomic principles as well as the potential for newer technologies in the provision of care. He is also very involved in medical student and resident education.

vkottoo1@jhmi.edu

**Arjuna Kumarasuriyar PhD**

Arjuna has a Bachelor of Biotechnology, first class honors from the University of Queensland, and a PhD from the same University in Molecular Biology conducted at the Institute of Molecular and Cell Biology, Singapore. He joined Illumina in October 2007 as the Regional Account Manager responsible for sales in South East Asia. In July 2009, he moved to the marketing team as Product Marketing Manager, South Asia Pacific where he assisted the sales team and managed the marketing of Illumina’s entire product range in the Indian Subcontinent, South East Asia and Australia/New Zealand. In 2011 he transitioned to his current role as Illumina’s District Sales Manager, South Asia, responsible for managing the Illumina sales team covering the Indian subcontinent, South East Asia and Singapore. He also has previous experience working in R&D (Peplin Biotech, Australia), Quality Assurance (Progen Industries, Australia), Government Regulation (Qld government Biotechnology Regulation Unit, Australia) and Technology Transfer (UniQuest Pty Ltd, Australia and Innovation and Technology Transfer Office, Nanyang Technological University, Singapore).

arjuna@illumina.com
Jee-Hian Lim BSc, MSc

Jee-Hian Lim works as Asia Pacific Marketing Manager in Abbott Molecular. He received his undergraduate degree in Biomedical Sciences from University Putra of Malaysia and completed his Master in Molecular Biology in 2003. His focus is providing Fluorescence in situ Hybridization (FISH) probes with high quality, reliable, cutting-edge tools for molecular diagnostics, personalized molecular medicine and companion diagnostics.

jeehian.lim@abbott.com

Sylvia Metcalfe BSc, PhD

Sylvia Metcalfe is Professor in Medical Genetics in the Department of Paediatrics at the University of Melbourne and Group Leader of Genetics Education and Health Research in the Murdoch Children's Research Institute, based at the Royal Children's Hospital in Melbourne, Australia. She has a BSc (Hons) and a PhD in Biochemistry from the UK, with a broad background of biomedical laboratory based research from London, New York and Melbourne. Recently her research interests include the understanding of genetics by health professionals and the community with respect to the implications of genetic technologies, genetic screening/testing and the impact of genetic diagnosis. She teaches human genetics at The University of Melbourne, as well as providing continuing professional development nationally and internationally, and has produced a number of educational resources.

sylvia.metcalfe@mcri.edu.au

Joseph D McInerney MA, MS

Joseph D. McInerney is Executive Vice President of the 8000-member American Society of Human Genetics. Previously, he was Executive Director of the National Coalition for Health Professional Education in Genetics (NCHPEG) and a former director of the Colorado-based Biological Sciences Curriculum Study (BSCS). His focus has been the development of textbooks and other educational programs in human genetics, genetic medicine, and evolution for audiences ranging from K-12 students and teachers to physicians and other health professionals.

McInerney, who lives in Baltimore, Maryland, received his MS in Human Genetics and Genetic Counseling from the State University of New York at Stony Brook. He is a Fellow of the American Association for the Advancement of Science, a former member of the editorial board of the Quarterly Review of Biology, and a recipient of the Award for Excellence in Human Genetics Education, from the American Society of Human Genetics.

jmcinerney@ashg.org
Sailesh Gochhait PhD

Prior to joining Life Technologies Sailesh completed his PhD from National Centre of Applied Human Genetics, School of Life Sciences, Jawaharlal Nehru University, India under the guidance of Prof. R.N.K. Bamezai (2008). He also undertook collaborative projects on Genetic susceptibility to Leprosy with Prof. Adrian Hill [WTCHG, Oxford in 2006] and on Epigenetic Inheritance with Dr. Peter de Boer [Radboud University Nijmegen Medical Centre].

Sailesh joined Life Technologies in June 2008 as an Application Scientist for Applied Biosystems Instruments (PCR, Real Time PCR and Capillary Electrophoresis) and products being used extensively in both basic as well as applied fields (Molecular Diagnostics and Forensic Sciences). Since 2010 he has been supporting Next Generation Sequencing platforms (SOLiD and Ion Torrent).

Sailesh is currently the FAS Lead for South Asia for Next Generation Sequencing at Life Technologies.

sailesh.gochhait@lifetech.com

Kate Reed ScM, MPH, CGC

Kate Reed is a project director at the National Coalition for Health Professional Education in Genetics (NCHPEG) and also a genetic counselor at Johns Hopkins University in Baltimore, MD. Currently at NCHPEG, Kate directs educational projects including a 10-month CME course on genomics for physicians at a community hospital, a web-based educational program on hereditary breast and ovarian cancer, a resource on chromosomal microarray for pediatricians, and point-of-care tools focusing on the management of genetic conditions. She is a Board-certified genetic counselor who earned her ScM from Johns Hopkins University/National Institutes of Health and her MPH in public health genomics from the University of Washington.

kreed@nchpeg.org

Kunal Sanghavi MBBS, MS

Kunal Sanghavi is a genetics program specialist at the McKusick Nathans Institute of Genetics Medicine at the Johns Hopkins University School of Medicine, Baltimore, USA and the Patient & Family project coordinator at New York Mid-Atlantic Consortium for Genetics and Newborn Screening Services (NYMAC), USA. He completed his medical education in Mumbai, India and pursued a program in genetic counseling at the Boston University School of Medicine, USA. He shares special interest in genetics education, cultural competency, and translational research. He develops and manages different programs related to genetic counseling, community partnerships, public health genomics, family history, and short-term internships. At NYMAC, he is pioneering the formation of a Consumer Interest Group (CIG)
aimed at helping families with special needs to access appropriate information and resources. Kunal is also involved in developing cross-border collaborations for genetics education, counseling, and services.

smileheals@gmail.com

**Timothy Smith BSc (Hons), PhD**

Timothy D. Smith is the Communications Officer for Human Variome Project International Ltd., the Coordinating Office for the Human Variome Project. He studied biochemistry and molecular biology at the University of Melbourne before starting his PhD which looks at how the process of genetic variation database curation is conducted and investigates novel methods for supporting that process through the application of different technologies. He is the author of the VariVis software program for genetic data visualisation.

Timothy is also the Laboratory Liaison Officer for the Human Variome Project Australian Node, a project—based within the Department of Pathology at the University of Melbourne— to provide a mechanism for diagnostic labs and clinicians to share information on genetic variations discovered in Australian patients.

tim@variome.com

**Yan Chin Tai MSc**

Yan Chin graduated from University of Malaya, Malaysia with a Masters in Medical Science in 2004. Her study on the molecular genetics of lymphomas had generated extensive publications in regional and international peer-reviewed journals.

Yan Chin then joined Republic Polytechnic as Academic Staff, and was appointed as Head of Laboratories in School of Applied Sciences. She helped to establish the laboratory processes and safety measures, in compliance with the Integrated Quality, Environmental, Health and Safety (QEHS) Management System, to accommodate the needs of five diplomas (Diplomas of Biomedical Sciences, Biotechnology, Pharmaceutical Science, Material Science and Environmental Science).

Yan Chin has been a Vysis user since 1999 while she was working in the Department of Pathology in University of Malaya, and also for her research project in the molecular genetics of lymphomas. She joined Abbott Molecular in 2008 as Molecular Application Specialist for Oncology for Asia Pacific region. She is responsible for managing and supporting the integration of FISH diagnostic tests by providing the necessary pre- and post-sales support and training. She is also extensively involved in regional events in promoting the use of FISH technique. She is currently Senior Molecular Application Specialist, Asia Pacific at Abbott Molecular, Singapore.

yanchin.tai@abbott.com
Ann P. Walker MA, LCGC

Ann Walker is Professor Emerita in the University of California, Irvine School of Medicine’s Department of Pediatrics—Division of Genetics and Metabolism. She is a licensed and certified genetic counselor who received her MA from UC Irvine’s Genetic Counseling Program in 1977, returning to the faculty there in 1978. She directed this graduate program from 1986 until her retirement in 2008. Ann continues to teach cancer genetics to medical and genetic counseling students, and neonatology and maternal-fetal medicine fellows. She is a member of the UCI Speakers bureau and provides lectures to community hospitals on a range of genetics topics. Ann’s recent clinical practice focused primarily on hereditary cancers and late-onset neurogenetic diseases. She has held leadership positions in the American Society of Human Genetics, National Society of Genetic Counselors, and American Board of Genetic Counseling. Her research has included educational, cultural and ethical issues in genetic counseling and hemochromatosis.

awalker@uci.edu
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