

RCPA, RCPA Faculty of Science and HGSA

Short Course in Medical Genetics and Genetic Pathology

Saturday 15 – Tuesday 18 June 2013

Intercontinental Sanctuary Cove Resort, Gold Coast, Queensland, Australia



Co-organised by the RCPA and the HGSA, this short course in genetic pathology focuses on contemporary and emerging training/ CPD requirements for pathologists and medical scientists involved in medical genetic/ genomic (constitutional or cancer-related) laboratory service provision.

The course, which will be delivered at an advanced rather than introductory level, will particularly serve the needs of genetic pathology trainees and scientists preparing for Fellowship examinations and senior practitioners delivering genetic diagnostic laboratory services.

Saturday June 15 th	Sunday June 16 th	Monday June 17 th	Tuesday June 18 th
Genome structure in health and disease (9.30-11.00am) Chromosome Architecture, Genome Organisation and Cell Division – an update (Damien Hudson, Murdoch Childrens Research Institute, Melbourne) Human Variome: What does it mean to be 'Normal'? (John Pearson, Institute for Molecular Bioscience, University of Queensland, Brisbane) Conserved regulatory elements (Karen Lower, Flinders Medical Centre, Adelaide)	Massively parallel sequencing (9.30-11.00am) Whole genome and exome sequencing - overview (Graham Taylor, Department of Pathology, University of Melbourne) Diagnosis of Mendelian disorders (Tony Roscioli, Prince of Wales Hospital, Sydney) Epigenetic biomarkers (Jeffrey Craig, Murdoch Childrens Research Institute, Melbourne)	Epigenetic mechanisms & clinical relevance (9.00-10.30am) Epigenetic Mechanisms (Richard Saffery, Murdoch Childrens Research Institute, Melbourne) Gene Imprinting and regulation (Elizabeth Algar, Monash Medical Centre, Melbourne) Cancer genome assembly through plasma DNA analysis (Rossa Chiu – Prince of Wales Hospital Hong Kong)	Clinical evaluation of genomic data (9.00-10.30am) Rare and novel CNC genetic variants (Chiyen Lau, Pathology Queensland) Massively Parallel Sequencing-based diagnostic screening in practice (Michael Buckley, South East Area Laboratory Service, Sydney) Assessing the potential clinical significance of rare missense variants (Peter Taylor, South East Area Laboratory Service, Sydney)
Cellular function in health & disease (cont.) (11.30-1.00 pm) What is Systems Biology? (Mark Ragan, Institute for Molecular Bioscience, University of Queensland, Brisbane) What is Systems Genetics? (Joseph Powell, Diamantina Institute, University of Queensland, Brisbane) An overview of computational requirements for systems biology (Leslie Burnett, RNSH, The University of Sydney)	Massively parallel sequencing in clinical practice (11.30-1pm) Cell-Free Circulating DNA: Biology and clinical applications (Damien Bruno, Victorian Clinical Genetics Services, Melbourne) Non-Invasive Prenatal Testing Beyond Down Syndrome (Rossa Chiu – Prince of Wales Hospital Hong Kong) Applications in cancer (Sean Grimmond, Institute for Molecular Bioscience, University of Queensland, Brisbane)	Epigenetics (continued) (11.00-12.30pm) Interpreting ENCODE from a clinical perspective (Edwin Kirk, Prince of Wales Hospital, Sydney) Current and emerging technologies for DNA methylation analysis (Alex Dobrovic, Peter MacCallum Cancer Centre, Melbourne) Characterising histone modifications (Graham Kay, Queensland Institute of Medical Research, Brisbane)	Cancer Genetic Diagnostics (11.00-12.30pm) Personalised Medicine (Paul Waring, Department of Pathology, University of Melbourne) Clinical validation of cancer biomarkers (Sebastian Lunke, Department of Pathology, University of Melbourne) Colorectal cancer genetics (Vicki Whitehall, Queensland Institute of Medical Research, Brisbane)
Gene Expression in health & disease (2.00-3.30pm) Transcriptional Profiling Using High Throughput Sequencing (Sean Grimmond, Institute for Molecular Bioscience, University of Queensland, Brisbane) Towards implementing an integrated "omics" approach to medicine (Mervyn Thomas, Emphron Informatics, Brisbane) Biochemical and functional assays for characterising VOUS (John Beilby, PathWest, Perth)	Microarrays (2.00-3.30pm) Clinical Manifestations of Genetic Mosaicism (Mac Gardner – Genetic Health Service, NZ) Susceptibility CNVs (Howard Slater, Victorian Clinical Genetics Services, Melbourne) Homozygosity from a clinical perspective (David Ravine, PathWest, Perth)	Medical genomics and the clinic (1.30-3.00pm) Pharmacogenomics (Les Sheffield, GenesFX Health & Victorian Clinical Genetics Services, Melbourne) Clinical utility of CNV positional information (James Harraway, Sullivan & Nicolaides Pathology, Brisbane) The place of follow-on testing in relatives (David Amor, Victorian Clinical Genetics Services, Melbourne)	Clinical case presentations (1.30-3.00pm) MPS-based diagnostic screening (Michael Buckley, South East Area Laboratory Service, Sydney) Microarray-based diagnostic screening (Damien Bruno – Victorian Clinical Genetics Services, Melbourne) Prenatal diagnosis (David Ravine, PathWest, University of Western Australia) Epimutation (e.g. unmethylated FRAX full mutation) (Val Hyland, Pathology Queensland, Royal Brisbane Hospital)
Haematological Genetics (4.00 – 5.00pm) DNA sequence-based blood grouping (Robert Flower, Australian Red Cross Blood Service, Brisbane) Haemophilia genetic testing (John Rowell, Pathology Queensland, Royal Brisbane Hospital) Clinical utility of thrombophilia testing (Rob Bird, Princess Alexandra Hospital, Brisbane)	Microarrays (4.00 – 5.00pm) Place of microarrays in prenatal diagnosis (George McGillivray, Victorian Clinical Genetics Services, Melbourne) Utility of microarray for cancer diagnostics (Val Hyland, Pathology Queensland, Royal Brisbane Hospital) Incidental findings (Chris Barnett, Women's and Children's Hospital, Adelaide)	Biochemical Genetics (3.30-4.30pm) Clinical and Biochemical markers of mitochondrial dysfunction (Jim McGill, Royal Children's Hospital, Brisbane) Genetics of mitochondrial dysfunction (David Thorburn, Murdoch Childrens Research Institute, Melbourne)	
Societal/ Ethical issues Evening Panel Discussion Direct to consumer testing (Panel: Mac Gardner, Ron Trent, David Weisbrot, Les Sheffield)	Professional issues Evening Panel Discussion CPD and upskilling (Panel: Graham Taylor, Kevin Carpenter, Paul Waring, Graeme Suthers, Leslie Burnett, Leah Bloomfield)	Professional issues Evening Panel Discussion Medical genomic test reports (Panel: Tony Roscioli, Margaret Otlowski, David Ravine, Honey Heussler, Andrew Fellowes)	

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TAX INVOICE PLEASE KEEP A COPY OF THIS FORM FOR YOUR RECORDS

DELEGATE REGISTRATION FORM

Details		RCPA ID (if applicable)	
Prefix	First	Last	
Organisation			
Address			
City	State	Post Code	
Email		Phone	

Registration Fees (in Australian dollars and including GST)	Full registration	1 Day registration
RCPA Fellow	<input type="checkbox"/> AUD \$1100.00	<input type="checkbox"/> AUD \$385.00
RCPA Trainee and Scientist	<input type="checkbox"/> AUD \$660.00	<input type="checkbox"/> AUD \$220.00
Day Registration please tick which day/s you will be attending		
<input type="checkbox"/> Saturday 15 June <input type="checkbox"/> Sunday 16 June <input type="checkbox"/> Monday 17 June <input type="checkbox"/> Tuesday 18 June		

Flights/Transfers & Accommodation

Special accommodation rates are available at the Intercontinental Sanctuary Cove. Please fill out the accompanying Travel reservation form and return directly to **Concierge Travel**. Airport transfers have also been arranged at a reduced rate and we recommend you book your flights according to these.

Payment		
Mastercard <input type="checkbox"/>	Visa <input type="checkbox"/>	American Express <input type="checkbox"/>
Card Number <input type="text"/> <input type="text"/> <input type="text"/> <input type="text"/> / <input type="text"/> <input type="text"/> <input type="text"/> <input type="text"/> / <input type="text"/> <input type="text"/> <input type="text"/> <input type="text"/> / <input type="text"/> <input type="text"/> <input type="text"/> <input type="text"/>		
Expirers <input type="text"/> <input type="text"/> / <input type="text"/> <input type="text"/>		Signature
Cheques		
Payable to RCPA Send to Royal College of Pathologists Durham Hall 207 Albion Street Surry Hills NSW 2010		
Registration and Enquiries		
Susan Lohan Phone: 02 8356 5824 Fax: 02 8356 5828 Email: susanl@rcpa.edu.au		

Registration Closes on Tuesday 4 June 2013. No refunds will be available after 17 May 2013

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TRAVEL RESERVATION FORM

Please complete this form and fax or email to Concierge Business Travel by **THURSDAY 16 MAY 2013**

Bookings made after this date cannot be guaranteed. We strongly recommend booking prior to this date.

Key Contact: Allysa Abalos **Tel:** +61 2 8270 4888 **Fax:** +61 2 9290 1306 **Email:** aabalos@conciergebt.com.au

STEP 1: DELEGATE DETAILS

(Please print clearly)

NAME AS PER ID: TITLE: FIRST NAME: SURNAME:

ORGANISATION:

ADDRESS:

SUBURB/TOWN: STATE: POSTCODE:

TELEPHONE: FAX:

MOBILE: EMAIL:

STEP 2: BOOKING REQUESTS

ACCOMMODATION: 14-18 June 2013

Single \$205.00 per night with one full breakfast / Double \$223.00 per night with two full breakfasts

HOTEL	ROOM TYPE	ARRIVAL DATE	DEPARTURE DATE	NO. OF NIGHTS	TOTAL AUD COST	SPECIAL REQUEST
Intercontinental Sanctuary Cove						

AIR: A \$22 booking fee (including GST) will apply to all Air/Transfer bookings processed by Concierge Business Travel

AIRLINE PREFERENCE	DATE OF TRAVEL	FROM	DEPARTURE TIME	TO	CLASS OF TRAVEL	FLEXIBLE or BEST FARE please stipulate	AIRLINE MEMBERSHIP NO

TRANSFERS:

Coach transfers are available in conjunction with flight bookings made via Concierge Business Travel

One Way transfer cost per person: Brisbane Airport-\$25.00 / Coolangatta Airport (Gold Coast)-\$20.00

14 June 2013 : Departs Brisbane Domestic - Intercontinental: 6.30pm or 8.30pm. Departs Coolangatta Airport -Intercontinental 6.30pm or 7.30pm

18 June 2013: Departs Intercontinental - Brisbane Domestic 3.30pm. Departs from Intercontinental - Coolangatta Airport 4.00pm

DATE OF TRAVEL	FROM	TO	PREFERRED TIME	DATE OF TRAVEL	FROM	TO	TIME
	Brisbane Airport	Intercontinental			Intercontinental	Brisbane Airport	
	Coolangatta Airport	Intercontinental			Intercontinental	Coolangatta Airport	

PLEASE NOTE: For private transfers Hughes Limousines can assist attendees by booking directly online

Refer to the Hughes Limousines website from 1 January 2013. Quote the conference code in the free format area. Payable direct.

Website: www.hugheslimousines.com.au **Conference code:** RCPA14JUNCBT

STEP 3: PAYMENT DETAILS

Please note: Merchant fees are applicable to credit card transactions and will be charged at time of final payment directly by the supplier

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CARD NUMBER:

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EXPIRY DATE:

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CCV/SECURITY CODE:

CARD HOLDERS NAME:

SIGNATURE:

DATE:

CCV - Visa/ Mastercard verification is the 3-digit number printed on the reverse side of your credit card. With Amex cards the Security Code is the 4-digit code number above your credit card number. Your signature gives permission to charge your credit card.

Cancellation and Change fees will vary dependent on; the service of the providers terms and conditions and ; of the date of written notification. Penalties can be up to 100% non refundable.