



HKUSPACE

香港大學專業進修學院
HKU School of Professional and Continuing Education

Advanced Workshop on Genomic Testing and Genetic Counselling for Hereditary Cancers

8, 15, 22 and 29 May 2021



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HKU School of Professional and Continuing Education

Week 1 Sessions (8th May 2021)

Chairperson: **Dr Stephen Tak Sum LAM**

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|----------------------|---|
| 14:00 – 15:00 | Lecture 1 Introduction to hereditary cancer diseases Dr Ivan Fai Man LO |
| 15:00 – 15:10 | Q&A |
| 15:10 – 15:40 | Tea break |
| 15:40 – 16:40 | Lecture 2 Overview of hereditary cancer genetic testing Dr Jeanette MCCARTHY |
| 16:40 – 16:50 | Q&A |
| 16:50 – 17:50 | Lecture 3 Understanding genetic test reports Dr Jeanette MCCARTHY |
| 17:50 – 18:00 | Q&A |

Lecture 1 Introduction to hereditary cancer diseases

(8 May 2021, 14:00 – 15:00)



Dr Ivan Fai Man LO

MBChB, FHKCPaed, FHKAM(Paed)

Honorary Clinical Associate Professor, Departments of Paediatrics and Obstetrics & Gynaecology, The Chinese University of Hong Kong

Dr Ivan FM Lo is Head of the Clinical Genetic Service of the Department of Health, a government funded tertiary referral centre of clinical genetic service in Hong Kong for over 30 years. Dr Lo was trained in genetics and genomics in the Clinical Genetic Service since 1991, and received training in biochemical and molecular genetics at the University of British Columbia, Canada, in 1996. He also received Paediatric training in Hong Kong and became a Fellow of the Hong Kong College of Paediatricians in 1998. He is in-charge of the overall supervision of genetic counselling service, laboratory genetic service and newborn screening programme since 2015. He is also Honorary Clinical Associate Professor of the Department of Paediatrics & Adolescent Medicine of both medical schools in Hong Kong, and Chairman of the Genetics & Genomics (Paediatrics) subspecialty board under the Hong Kong College of Paediatricians. Dr Lo's special interests include dysmorphology, neurogenetics, molecular genetics and application of next generation sequencing technology. He has more than 100 publications in international peer reviewed medical/genetic/genomic journals.

Lecture Highlights

- ❖ Introduction to human genetics
- ❖ Overview of hereditary cancer diseases
- ❖ Common hereditary cancer patients' characteristics
- ❖ Variability in clinical symptoms expression
- ❖ Hereditary cancer genetics, inheritance mode, predisposition and risk factors
- ❖ Review of cancer susceptibility genes, somatic and germline alterations
- ❖ Clinical symptoms & family history suitable for referral to genomic testing
- ❖ Informed consent in tests

Lecture 2 Overview of hereditary cancer genetic testing (8 May 2021, 15:40 – 16:40)



Dr Jeanette MCCARTHY

MPH, PhD

Founder, Precision Medicine Advisors

Adjunct Associate Professor, Community and Family Medicine, Duke University

Adjunct Associate Professor, Department of Medicine, University of California, San Francisco

Dr Jeanette McCarthy is a UC Berkeley trained genetic epidemiologist and spent the early part of her career in industry at Millennium Pharmaceuticals before transitioning to academia. She currently holds adjunct faculty positions at Duke University and UCSF. Her previous research had focused on the genetic underpinnings of complex diseases, both infectious and chronic. More recently, she has become a leading educator in the field of genomic and precision medicine involved in demystifying genomics for non-technical audiences, including health care providers, patients and other stakeholders. In 2014 she helped launch the first consumer-facing magazine in this field, *Genome*, where she served as editor-in-chief until 2016. She teaches genomic and precision medicine through UCSF and UC Berkeley Extension and online through the Precision Medicine Academy (precisionmedicineacademy.org). She also designs and delivers custom workshops and courses to international audiences and advises companies on strategic and technical aspects of precision medicine.

Lecture Highlights

- ❖ Pedigree analysis and hereditary risk assessment
- ❖ Factors to consider when selecting hereditary cancer genetic tests
- ❖ Variety of hereditary cancer genetic tests
- ❖ Genetic counselling prior to testing
- ❖ Genomic test results analysis and disclosure
- ❖ Family members hereditary risk evaluation

Lecture 3 Understanding genetic test reports (8 May 2021, 16:50 – 17:50)



Dr Jeanette MCCARTHY

MPH, PhD

Founder, Precision Medicine Advisors

Adjunct Associate Professor, Community and Family Medicine, Duke University

Adjunct Associate Professor, Department of Medicine, University of California, San Francisco

Dr Jeanette McCarthy is a UC Berkeley trained genetic epidemiologist and spent the early part of her career in industry at Millennium Pharmaceuticals before transitioning to academia. She currently holds adjunct faculty positions at Duke University and UCSF. Her previous research had focused on the genetic underpinnings of complex diseases, both infectious and chronic. More recently, she has become a leading educator in the field of genomic and precision medicine involved in demystifying genomics for non-technical audiences, including health care providers, patients and other stakeholders. In 2014 she helped launch the first consumer-facing magazine in this field, *Genome*, where she served as editor-in-chief until 2016. She teaches genomic and precision medicine through UCSF and UC Berkeley Extension and online through the Precision Medicine Academy (precisionmedicineacademy.org). She also designs and delivers custom workshops and courses to international audiences and advises companies on strategic and technical aspects of precision medicine.

Lecture Highlights

- ❖ Lab selection, pathogenicity of variants and test report interpretation
- ❖ Case studies for hereditary cancer genetic testing
- ❖ Examples on hereditary breast, ovarian, colorectal, prostate and pancreatic cancers
- ❖ Possible problems that may encounter in practice



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Week 2 Sessions (15th May 2021)

Chairperson: **Professor Ava KWONG**

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|---------------|--|
| 14:00 – 14:40 | Lecture 4 Hereditary breast cancer & genetic counselling Professor D. Gareth EVANS |
| 14:40 – 14:50 | Q&A |
| 14:50 – 15:30 | Lecture 5 Clinical management of hereditary breast cancer from a surgeon's view Professor Ava KWONG |
| 15:30 – 15:40 | Q&A |
| 15:40 – 16:20 | Lecture 6 Precision oncology medicines in hereditary breast cancer Professor Roger Kai Cheong NGAN |
| 16:20 – 16:30 | Q&A |
| 16:30 – 16:40 | Tea Break |

Chairperson: **Professor Roger Kai Cheong NGAN**

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|---------------|---|
| 16:40 – 17:20 | Lecture 7 Hereditary ovarian cancer & genetic counselling Professor D. Gareth EVANS |
| 17:20 – 17:30 | Q&A |
| 17:30 – 18:10 | Lecture 8 Clinical management of hereditary ovarian cancer Professor Hextan Yuen Sheung NGAN |
| 18:10 – 18:20 | Q&A |

Lecture 4 Hereditary breast cancer & genetic counselling (15 May 2021, 14:00 – 14:40)



Professor D. Gareth EVANS

MB, BS, MD, FRCP, FLSW, FRCOG ad eundem

Professor of Medical Genetics and Cancer Epidemiology, The University of Manchester

Professor Evans is professor of medical genetics and cancer epidemiology at university of Manchester UK. He has established a national and international reputation in clinical and research aspects of cancer genetics, particularly in neurofibromatosis and breast/colorectal cancer. He has published 864 peer reviewed research publications; 305 as first or senior author (total publications 1015). He has an ISI web of knowledge H-index of 116 and google scholar of 152 having only published his first article in 1990. In the last 8 years he has raised over £50 million in grants for multicentre and local studies – approximately £42 million to Manchester. He is Chief Investigator on two NIHR program grants (2009-2014-£1.59 million) (2017-2020-£1million) on breast cancer risk prediction and also has an NIHR RfPB grant as CI (2011). He has supervised 14 successful doctoral theses and is currently supervising five. He has led a successful bid for a Nationally funded NF2 service (£7.5 million pa) that started in 2010 and is involved in the national complex NF1 service. He is overall cancer lead (3 themes) and Cancer Prevention Early detection theme lead on the successful all Manchester NIHR Biomedical research centre bid (2017-2022-£28.5million). He is lead clinician on the NICE familial breast cancer guideline group and until recently a trustee of Breast Cancer Now and the Neuro Foundation. He is on the editorial board of JNCI and several other journals. He is the recipient of a number of awards including the Theodor Schwann, von Recklinghausen and Sir Patrick Forrest prize twice.

Lecture Highlights

- ❖ Signs and symptoms of hereditary breast cancer patients
- ❖ Common mutations in hereditary breast cancer (e.g. BRCA1/2, PTEN, p53, HRD genes)
- ❖ Individual risk assessment and referral criteria for hereditary breast cancer testing
- ❖ Different genomic tests available for hereditary breast cancer
- ❖ Counselling advice for genomic testing in hereditary breast cancer
- ❖ Genetic counselling for hereditary breast cancer patients

Lecture 5 Clinical management of hereditary breast cancer from a surgeon's view

(15 May 2021, 14:50 – 15:30)



Professor Ava KWONG

MBBS, BSc, PhD, FRCS, FRCSEd, FCSHK, FHKAM(Surgery)

Daniel C K Yu Professor in Breast Cancer Research

Clinical Professor and Chief of Division of Breast Surgery, The University of Hong Kong

Professor Kwong is Daniel C K Yu Professor in Breast Cancer Research and Clinical Professor at The University of Hong Kong, Chief of Division of Breast Surgery at Queen Mary Hospital, Tung Wah Hospital and The University of Hong Kong-Shenzhen Hospital. In 2007, she founded and is Chairman of the Hong Kong Hereditary Breast Cancer Family Registry. In 2012, she was appointed as the co-leader of the Cancer Work Group in the development of Cancer Services planning of the Hong Kong West Cluster. In 2013, she was elected to be the Deputy Chief and Committee Member of the Shenzhen Breast and Endocrine Cancer Society, China. She has been appointed to be a member of the Cancer Coordinating Committee of the Food and Health Bureau, the Government of the HKSAR since 2014. She is also currently a member of several committees including Consortium on Harmonization of Institutional Requirements for Clinical Research (CHAIR) and Central Committee on Cancer Service of Hospital Authority, Expert Advisory Panel (Cancer) of Health and Medical Research Fund (HMRF) of Food and Health Bureau, the Government of the HKSAR. Professor Kwong's research interests focus on breast and ovarian cancer genetics, advanced surgical technologies, epidemiology, psychosocial and clinical trials in oncological treatment. She has gained multiple awards in various local and international meetings including the "Uccio Querci della Rovere Award" of The Royal College of Surgeons of England in 2019, "Young Investigator Awards" of Hong Kong international Cancer Congress in 2006 and 2008.

Lecture Highlights

- ❖ Hereditary breast cancer prevalence in Hong Kong and South East Asia
- ❖ Management and recommendations for mutation carriers:
 - ❖ Breast self-examination
 - ❖ Annual screening
 - ❖ Chemoprevention
 - ❖ Prophylactic measures
 - ❖ Risk reducing mastectomy
 - ❖ Psychosocial support
 - ❖ Testing in carrier's children
- ❖ Conventional clinical management for breast cancer patients

Lecture 6 Precision oncology medicines in hereditary breast cancer

(15 May 2021, 15:40 – 16:20)



Professor Roger Kai Cheong NGAN

MBBS (HK), LRCP (Lond), MRCS (Eng), DMRT (UK), FFRRCS(I), FRCR, FHKCR, FHKAM (Radiology)

Clinical Professor, Department of Clinical Oncology, The University of Hong Kong

Professor Roger Ngan is currently Clinical Professor in the Department of Clinical Oncology of the University of Hong Kong, and is the Chief of Specialty in Oncology at Gleneagles Hospital Hong Kong. He is also an honorary consultant in the Departments of Clinical Oncology of both Queen Mary Hospital and Queen Elizabeth Hospital, and in the Department of Surgery of Kwong Wah Hospital of Hong Kong. Prof Ngan is a specialist in clinical oncology practicing in Hong Kong who has a special interest in the clinical research of multimodality management of breast cancers, head and neck cancers, gynecological cancers, soft tissue sarcomas and brachytherapy. He has published more than 130 articles in peer-reviewed journals and book chapters. Prof Ngan has worked as the Director of the Hong Kong Cancer Registry from 2012 to 2018, and the Chief of Service of Department of Clinical Oncology, Queen Elizabeth Hospital from 2013 to 2018. He is the immediate past President of the Hong Kong Breast Oncology Group, and deputy Chief Editor of the Hong Kong Journal of Radiology.

Lecture Highlights

- ❖ Somatic genomic test interpretation
- ❖ Effect of genomic test results on breast cancer treatment
- ❖ Targeted therapeutic drugs
 - ❖ HER2 antibodies
 - ❖ Tyrosine kinase inhibitors
 - ❖ PIK3K inhibitor
 - ❖ CDK inhibitors
 - ❖ mTOR inhibitors
- ❖ Emerging new therapeutic measures in breast cancer patients

Lecture 7 Hereditary ovarian cancer & genetic counselling

(15 May 2021, 16:40 – 17:20)



Professor D. Gareth EVANS

MB, BS, MD, FRCP, FLSW, FRCOG ad eundem

Professor of Medical Genetics and Cancer Epidemiology, The University of Manchester

Professor Evans is professor of medical genetics and cancer epidemiology at university of Manchester UK. He has established a national and international reputation in clinical and research aspects of cancer genetics, particularly in neurofibromatosis and breast/colorectal cancer. He has published 864 peer reviewed research publications; 305 as first or senior author (total publications 1015). He has an ISI web of knowledge H-index of 116 and google scholar of 152 having only published his first article in 1990. In the last 8 years he has raised over £50 million in grants for multicentre and local studies – approximately £42 million to Manchester. He is Chief Investigator on two NIHR program grants (2009-2014-£1.59 million) (2017-2020-£1million) on breast cancer risk prediction and also has an NIHR RfPB grant as CI (2011). He has supervised 14 successful doctoral theses and is currently supervising five. He has led a successful bid for a Nationally funded NF2 service (£7.5 million pa) that started in 2010 and is involved in the national complex NF1 service. He is overall cancer lead (3 themes) and Cancer Prevention Early detection theme lead on the successful all Manchester NIHR Biomedical research centre bid (2017-2022-£28.5million). He is lead clinician on the NICE familial breast cancer guideline group and until recently a trustee of Breast Cancer Now and the Neuro Foundation. He is on the editorial board of JNCI and several other journals. He is the recipient of a number of awards including the Theodor Schwann, von Recklinghausen and Sir Patrick Forrest prize twice.

Lecture Highlights

- ❖ Signs and symptoms of hereditary ovarian cancer patients
- ❖ Common mutations in hereditary ovarian cancer
 - ❖ BRCA1/2, LGRs
 - ❖ MLH1, MSH2, MSH6
 - ❖ PMS2, STK11
 - ❖ Selected HRD genes
- ❖ Shared risks associated with hereditary breast cancer
- ❖ Referral criteria for genomic testing in hereditary ovarian cancer
- ❖ Counselling advice for genomic testing in hereditary ovarian cancer
- ❖ Genetic counselling for hereditary ovarian cancer patients

Lecture 8 Clinical management of hereditary ovarian cancer

(15 May 2021, 17:30 – 18:10)



Professor Hextan Yuen Sheung NGAN

MBBS, MD(HK), FRCOG, FHKCOG, FHKAM (O&G); Cert RCOG (Gynae Onc); Accredited gynaecological oncologist (UK and HK); Accredited colposcopist (HKCOG/HKSCCP, HK)

Tsao Yin-Kai Professor in Obstetrics & Gynaecology

Professor, Chair of Obstetrics & Gynaecology, The University of Hong Kong

Head of Department of Obstetrics & Gynaecology, The University of Hong Kong

Professor Hextan Ngan graduated in the University of Hong Kong in 1978. She obtained MRCOG in 1983 and MD in 1995. She has been a gynaecological oncologist accredited by the Royal College of Obstetricians and Gynaecologists (RCOG) since 1993, and has vast experience in open, laparoscopic and robot-assisted cancer surgery. She has got more than 300 publications in peer-reviewed journals. Her research interest is in biomolecular studies, cancer screening, clinical trials and psychosocial studies in gynaecological oncology. Prof Ngan is the past president of the Hong Kong College of Obstetricians and Gynaecologists (HKCOG), past president of the Asia Oceania Research Organization of Genital Infections & Neoplasia (AOGIN), past Chair of the FIGO Gynecology Oncology Committee, and currently the President of the Family Planning Association of Hong Kong. She is now the Head of Department of Obstetrics & Gynaecology, the University of Hong Kong, and has been endowed with Tsao Yin-Kai Professorship in Obstetrics and Gynaecology in 2009. Prof Ngan is also the head of the gynaecological oncology team and laboratory of the University of Hong Kong, the Chief of Service in Obstetrics & Gynaecology of the University of Hong Kong-Shenzhen Hospital, and the Chair of Specialty ("COS") for Obstetrics & Gynaecology, Gleneagles Hong Kong Hospital Limited.

Lecture Highlights

- ❖ Management and recommendations for carriers to reduce risks:
 - ❖ Clinical abdominal exams
 - ❖ Oophorectomy or fallopian tube removal
 - ❖ Oral contraceptive use
 - ❖ Transvaginal ultrasound or endometrial biopsy
 - ❖ CA-125 blood levels
 - ❖ Hysterectomy
- ❖ Other associated risks that can be modified
- ❖ Conventional clinical management of ovarian cancer patients
- ❖ Emerging new therapeutic measures in ovarian cancer patients



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Week 3 Sessions (22nd May 2021)

Chairperson: **Dr Ka On LAM**

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| 14:00 – 14:45 | Lecture 9 Hereditary colorectal cancer & genetic counselling Professor Siu Tsan YUEN |
| 14:45 – 14:55 | Q&A |
| 14:55 – 15:40 | Lecture 10 Clinical management of hereditary colorectal cancer Dr Dominic Chi Chung FOO |
| 15:40 – 15:50 | Q&A |
| 15:50 – 16:10 | Tea Break |

Chairperson: **Dr Chi Leung CHIANG**

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| 16:10 – 16:55 | Lecture 11 Pancreatic cancer – clinical management and hereditary susceptibility Professor David WHITCOMB |
| 16:55 – 17:05 | Q&A |

Chairperson: **Professor Anthony Chi Fai NG**

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|---------------|--|
| 17:05 – 17:50 | Lecture 12 Prostate cancer: The genetics of disease risk and management Professor Leonard GOMELLA |
| 17:50 – 18:00 | Q&A |

Lecture 9 Hereditary colorectal cancer & genetic counselling

(22 May 2021, 14:00 – 14:45)



Professor Siu Tsan YUEN

MBBS(HK), MD(HK), FCSHK, FRCSEd, FRCPath(UK), FHKCPath, MIAC, MRCPPath(UK), FHKAM(Pathology)

Co-Director & Honorary Clinical Professor, Hereditary Gastrointestinal Cancer Genetic diagnosis Laboratory, Department of Pathology, The University of Hong Kong

Professor Siu Tsan Yuen is the Co-Director and Honorary Clinical Professor, Hereditary Gastrointestinal Cancer Genetic diagnosis Laboratory, Department of Pathology, The University of Hong Kong. He is the Founding Fellow, Faculty of Science, The Royal College of Pathologists of Australasia, and the First Fellow in Genetic and Genomic Pathology, Hong Kong College of Pathologist. Professor Yuen became a fellow of the Royal College of Surgeons of Edinburgh in 1989, a fellow of the College of Surgeons of Hong Kong in 1991, and a fellow of the Hong Kong Academy of Medicine (Pathology) in 1995. He is also the Honorary Professor of Beijing Army General Hospital in China and the Medical Advisor of The Hong Kong Cancer Fund. He was appointed as the Consultant Pathologist in the Department of Pathology of St. Paul's Hospital in 2004 and the Deputy Medical Superintendent of St. Paul's Hospital in 2011. His main research interest is hereditary gastrointestinal (gastric, colorectal) cancers and genetic diagnosis. He has more than 150 publications in peer-reviewed international journals including Nature and Nature Genetics.

Lecture Highlights

- ❖ Clinical presentation and genetic cause
- ❖ Hereditary colorectal cancer subtypes & characteristics
- ❖ Autosomal dominant inheritance
- ❖ Family history
- ❖ Referral criteria for genomic tests
 - ❖ Amsterdam criteria
 - ❖ Bethesda criteria
 - ❖ MSI & IHC screening
- ❖ Counselling advice for genomic testing in hereditary colorectal cancer
- ❖ Genetic counselling for hereditary colorectal cancer

Lecture 10 Clinical management of hereditary colorectal cancer

(22 May 2021, 14:55 – 15:40)



Dr Dominic Chi Chung FOO

MBBS, FRCSEd, FCSHK, FHKAM(Surgery)

Clinical Assistant Professor, Department of Surgery, The University of Hong Kong

Dr Dominic Chi Chung Foo is Clinical Assistant Professor in the Division of Colorectal Surgery of The University of Hong Kong. He graduated from The University of Hong Kong with a Bachelor of Medicine and Bachelor of Surgery degree in 2002. He undertook his surgical training in Queen Mary Hospital and joined the Division of Colorectal Surgery in 2009. He became a specialist in general surgery, a fellow of the Hong Kong Academy of Medicine (Surgery) and a fellow of the College of Surgeons of Hong Kong in 2011 and a fellow of the Royal College of Surgeons of Edinburgh in 2012. His main interest is in the field of minimally invasive colorectal surgery, in particular, rectal surgery. He has personally performed over 100 robotic rectal surgeries. His publications, including book chapters and peer-reviewed articles, focus on robotic rectal surgery, transanal surgery and single-incision colorectal surgery. Dr Foo's active involvement in various professional societies includes being a council member of the International Society for Digestive Surgery – Asian Pacific, a member of the Simulation Working Group, Research Committee, Examination Committee and Education Committee in the College of Surgeons of Hong Kong.

Lecture Highlights

- ❖ Actionable lifestyle changes to reduce colorectal cancer risk
- ❖ Mutation carriers' management
 - ❖ Colonoscopy
 - ❖ Endoscopic ultrasound
 - ❖ Urinalysis
 - ❖ Physical examination
 - ❖ Prophylactic colectomy
 - ❖ NSAIDs use in reducing CRC risk
 - ❖ Associated cancer screening (e.g. endometrial & ovarian)
- ❖ Conventional clinical management
- ❖ Emerging new therapeutic measures

Lecture 11 Pancreatic cancer – clinical management and hereditary susceptibility

(22 May 2021, 16:10 – 16:55)



Professor David WHITCOMB

MD, PhD

Giant Eagle Foundation Professor of Cancer Genetics, The University of Pittsburgh School of Medicine

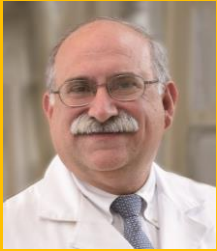
Professor David C. Whitcomb is a pioneer in the use of mathematics, genetics, neurosciences, immunology, epidemiology and clinical sciences to study complex inflammation disorders and cancer risks of the digestive system, and he is among the most-cited authors in the field of pancreatic diseases with multiple landmark papers. He co-founded the Center for Genomic Sciences at the University of Pittsburgh (now part of the Genomics and Proteomics Core Laboratory), and served as Director of the Nutrition Support Service and Director of Gastroenterology for the Pittsburgh VA Healthcare System. He served as Chief, Division of Gastroenterology, Hepatology and Nutrition at the University of Pittsburgh from 1999 to 2016, building it into a top-tier Gastroenterology program. With over 25 years of continuous NIH funding, he served as principal investigator on multiple National Institutes of Health-sponsored studies, building one of the top human genetics programs for complex digestive diseases. He has published over 400 papers, reviews and book chapters with nearly 30,000 citations, and h-index of ~ 85 and i10index of ~280. His research, education and organizational innovations in precision medicine continue to be studied and emulated by other leading programs throughout the world.

Lecture Highlights

- ❖ Signs, symptoms and clinical presentation of hereditary pancreatic cancer patients
- ❖ Hereditary inheritance
 - ❖ PRSS1/TRYP1, STK11/LKB1 and P16INK4a in pancreatic cancer
- ❖ Risk association with other cancers
- ❖ Other associated disease: hereditary pancreatitis
- ❖ Conventional clinical management
- ❖ Emerging new therapeutic measures

Lecture 12 Prostate cancer: The genetics of disease risk and management

(22 May 2021, 17:05 – 17:50)



Professor Leonard GOMELLA

MD, FACS

*The Bernard W. Godwin, Jr. Professor of Prostate Cancer, Chairman,
Department of Urology, Thomas Jefferson University*

Professor Leonard Gomella is the Bernard W. Godwin, Jr. Professor of Prostate Cancer and Chairman of the Department of Urology at the Sidney Kimmel Medical College. Originally from New York, Professor Gomella completed medical school, general surgery and urology training at the University of Kentucky in Lexington. After a Urologic Oncology Fellowship in the Surgery Branch of the NCI in Bethesda, Maryland, he joined the Jefferson faculty in 1988 and was appointed Chair in 2002. He serves as Senior Director for Clinical Affairs for the Sidney Kimmel Cancer at Jefferson. From 2008 until 2019 he was Clinical Director of the Sidney Kimmel Cancer Center Network and from 1999-2020 Urology Chair for RTOG (now NRG). In 2019 Dr. Gomella was named Urology Service Line Director for the entire Jefferson Health hospital system. Professor Gomella is involved in translational basic science and clinical research developing new diagnostic tests and treatments for prostate, bladder and kidney cancer through the Sidney Kimmel Cancer Center where he serves as the Co-Leader of the Biology of Prostate Cancer Program. His team was first to use molecular techniques (RT-PCR) to detect circulating prostate cancer micrometastasis (now called "circulating tumor cells"), a discovery that led to a new field of investigation in this disease. He is also recognized as an early contributor to urologic laparoscopy, starting Jefferson's program in 1990. He led the Urology effort in the 2017 and 2019 Philadelphia Prostate Cancer Consensus that provided the first multidisciplinary guidance on the genetic testing for prostate cancer. He has given over 600 presentations nationally and internationally and written over 400 papers, chapters and monographs in the field of Urology.

Lecture Highlights

- ❖ Signs and symptoms of hereditary prostate cancer
- ❖ Common mutations in hereditary prostate cancer
 - ❖ BRCA1/2, ATM, RAD51, BRIP1, PABL2, CHEK2 & HOXB13 mutations
- ❖ Risk assessment and referral criteria for genomic testing
- ❖ Risk factors
- ❖ Factors that can be modified to reduce risks
- ❖ Surveillance in high-risk patients with guideline recommendations
- ❖ Conventional treatments
- ❖ Emerging new treatments



Week 4 Sessions (29th May 2021)

Chairperson: **Dr Joseph Siu Kie AU**

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| 14:00 – 15:00 | Lecture 13 Clinical genomic tests for detecting cancer susceptibility genes Dr Shu Jen CHEN |
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| 15:00 – 15:10 | Q&A |
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| 15:10 – 15:20 | Tea Break |
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Chairperson: **Dr Shu Jen CHEN**

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| 15:20 – 16:10 | Lecture 14 Laboratory workflow in clinical genomic testing by NGS Dr Alfa BAI |
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| 16:10 – 16:20 | Q&A |
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| 16:20 – 16:30 | Intermission |
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| 16:30 – 17:50 | Lecture 15 Interactive session in real life case sharing Dr Stephen Tak Sum LAM, Mr Daniel C. SIU and Ms Maggie LAW |
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| 17:50 – 18:00 | Q&A |
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Course completed

Lecture 13 Clinical genomic tests for detecting cancer susceptibility genes

(29 May 2021, 14:00 – 15:00)



Dr Shu Jen CHEN

PhD

Chief Scientific Officer, ACT Genomics

Dr Shu-Jen Chen obtained her Ph.D. degree in Biochemistry from Virginia Commonwealth University, USA. She received her postdoctoral training at Baylor College of Medicine and became a Research Assistant Professor at SUNY Buffalo. She joined National Health Research Institute (NHRI) as an Assistant Investigator and established the high throughput screening program for the institute. As one of the founding scientists, Dr. Chen joined TaiGen Biotechnology in 2001 to lead the in vitro pharmacology group. In 2006, she moved to Chang Gung University as an Associate Professor in the Department of Biomedical Sciences. As a co-founder of ACT Genomics, Dr. Chen has served as the Chief Scientific Officer since 2014. Dr. Chen specializes in automated drug screening system, genomics and transcriptomics technologies, omics data analysis and biological database integration. She is also familiar with cancer biology, system integration and database design. She currently leads the sequencing group and the bioinformatics group at ACT Genomics to implement cancer genome sequencing for research and clinical applications.

Lecture Highlights

- ❖ Principle & practical aspects in NGS tests
- ❖ Methodologies in hereditary cancer risk tests
- ❖ Analyses of SNV, Indel, CNV, LGR and MLPA in somatic and germline testing
- ❖ Bioinformative and pharma-bioinformative analysis
- ❖ Testing for BRCA1/2, HRD genes and genomic scars
- ❖ FDA, NCCN, AMP and ACMG guidelines in cancer risk tests
- ❖ Clinical interpretation of test results
 - ❖ Different emphasis between somatic and germline tests

Lecture 14 Laboratory workflow in clinical genomic testing by NGS

(29 May 2021, 15:20 – 16:10)



Dr Alfa BAI

BSc, MPhil, Dr.rer.nat., MLT Part1
Laboratory Director, ACT Genomics

Dr Alfa Bai Hsing Chen is a registered medical laboratory technologist in Hong Kong. He finished the professional training and obtained a Bachelor of Science degree in Medical Technology from Chang Gung University, Taiwan in 1999. Dr Bai started his academic research career, which focused in cancer genetics and epigenetics, in 2000; and he obtained a Master of Philosophy degree in Medical Sciences from The Chinese University of Hong Kong and Doctor of Science degree (Doctor rerum naturalium) from Ruprecht-Karls-Universität Heidelberg, Germany in 2004 and 2011 respectively. Dr Bai published twenty research papers in international peer-review journals namely, Nature Medicine and Gut, in his research career.

Lecture Highlights

- ❖ NGS workflow in Ion torrent and Illumina platforms
- ❖ Quality assurance tests
- ❖ Requirement differences in somatic and germline testing
- ❖ Sample preparation for blood, saliva, buccal swab & tumour tissues
- ❖ The importance of tissue macro-dissection in somatic tests
- ❖ Library preparation and the importance in preventing cross-contamination in NGS lab
- ❖ NGS test reporting

Lecture 15 Interactive session in real life case sharing (29 May 2021, 16:30 – 17:50)



Dr Stephen Tak Sum LAM

MBBS (HK), DCH (Lond), MRCP (Irel), MRCP (UK), FRCP(Edin), MHA (New South Wales), MD (HK), FHKCPaed, FHKAM (Paediatrics)

Director, Clinical Genetics Service, Honorary Consultant in Clinical Genetics, Hong Kong Sanatorium & Hospital

Dr Stephen Lam is a Fellow of Hong Kong College of Paediatricians, Fellow of Royal College of Physicians (Edinburgh), and Fellow of Hong Kong Academy of Medicine. He was the Consultant Clinical Geneticist, and Head of Clinical Genetic Service, Department of Health, Hong Kong (1990-2015). He is an Honorary Professor of the Faculty of Medicine in the Chinese University of Hong Kong since 2012. He was the Past President of the Asia Pacific Society of Human Genetics (2011-12) and the International Federation of Human Genetic Societies (2012-14). Since July 2016, he is the Director of Clinical Genetics Service and Honorary Consultant in Clinical Genetics in the Hong Kong Sanatorium and Hospital.

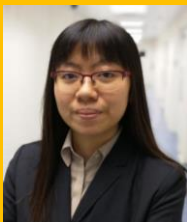


Mr Daniel C. SIU

MS, MBA

CEO and Founder, Rainbow Genomics

Mr Daniel C. Siu received his B.S. in molecular biology from the University of Texas, Austin, his M.S. in bioinorganic chemistry from the University of Minnesota, Minneapolis, and his MBA from San Francisco State University. In 2011, Mr Siu co-founded US-based Axiq Technologies with the chairman of MacroGen. He co-founded Nextcode Health in 2013, a US and Iceland-based clinical bioinformatics and "big data" company. Mr Siu founded Rainbow Genomics in 2016, which provides clinical whole genome and whole exome sequencing tests with unique methodologies for paediatric / adult rare disease and germline cancer diagnosis.



Ms Maggie LAW

BSc, MMS, MGC

Associate genetic counsellor, International Genetic Counselling Centre

Ms Law received both of her Bachelor of Science degree in Biotechnology, and her Master of Medical Sciences in Pathology, from the University of Hong Kong. She also received her Master of Genetic Counselling degree from the University of Technology Sydney. Ms Law received her clinical training at Hong Kong Sanatorium & Hospital, Canberra Hospital and Prince of Wales Hospital in Australia.

Lecture Highlights

- ❖ Case sharing of examples in hereditary cancer patients

Credits

- CME Point Allocation by the following colleges through Hong Kong Academy of Medicine

| College | Maximum point for attending all 4 sessions | Session point on 8 May 2021 (14:00 - 18:00) | Session point on 15 May 2021 (14:00 - 18:00) | Session point on 22 May 2021 (14:00 - 18:00) | Session point on 29 May 2021 (14:00 - 18:00) |
|---|--|---|--|--|--|
| Hong Kong College of Radiologists | 15.5 | 3.5 | 4.0 | 4.0 | 4.0 |
| The Hong Kong College of Obstetricians and Gynaecologists | 10.0 | 3.0 | 4.0 | 3.0 | 3.0 |
| Hong Kong College of Physicians | 14.5 | 3.5 | 4.0 | 3.5 | 3.5 |
| The College of Surgeons of Hong Kong | 14.5 | 3.5 | 4.0 | 3.5 | 3.5 |
| The Hong Kong College of Pathologists | 15.5 | 3.5 | 4.0 | 4.0 | 4.0 |
| Hong Kong College of Paediatricians* | 10.0 | N/A | N/A | N/A | N/A |
| Hong Kong College of Community Medicine | 10.0 | 3.0 | 4.0 | 3.0 | 3.0 |
| The Hong Kong College of Family Physicians | 10.0 | 3.0 | 3.0 | 3.0 | 3.0 |

* CME point is only given upon completion of the whole function (no session point given); attendance of 80% is required in order to obtain 10 CME points.

- 14.5 CNE points from The Nursing Council of Hong Kong
- 12 CPD points from MLT Board (Activity Code: 05200611)

Note: No CME/CNE/CPD point will be given for watching the lecture video recordings.