

### NEW GENOMICS COURSE FOR PATHOLOGY TRAINEES

# **Jan 6 - 10, 2014**

## NEW GENOMICS COURSE FOR PATHOLOGY RESIDENTS

#### JAN 6-10, 2014

We have developed a highly integrative, practical and focused approach to genomics teaching for residents and trainees in the Department of Pathology and Laboratory Medicine to be offered as a 1 week intensive program in Jan 2014. Simple genetic tests with a limited detection threshold are gradually being replaced by multiplex, deeply interrogative assays which coincide with the recent revolutions in massively parallel sequencing and advanced molecular profiling technology. This is further propelled by simultaneous ongoing discoveries of novel cancer pathway aberrations that drive the development and clinical application of novel targeted therapeutics. More questions are being asked of the pathologist and biopsied tissues are getting smaller. This course provides the residents a review and refresher of relevant topics in genomics and epigenomics followed by a survey of available clinically significant molecular diagnostic assays as well as bioinformatic tools.

Genomic medicine and lab medicine, especially pathology, are natural and symbiotic partners in the future of patient care. In many cases, molecular diagnostic assays complement rather than supplement "glass-based" pathology. For example, correct identification of tumor cells and estimation of tumor fraction facilitates downstream analysis. In an era of personalized medicine, understanding of the genomic and epigenomic makeup of a disease lesion, via the appropriate application of advanced molecular tests and bioinformatic analytic tools, represents a logical progression from special stains to epitopespecific antibodies to genetic tests, for making an accurate and informative diagnosis. We will draw from the community of local experts at UBC, the Michael Smith Genome Sciences Centre, the Centre for Disease Control and the BC Cancer Agency in the fields of molecular pathology, molecular biology, microbiology, bioinformatics and genomics to offer an immersive introductory program on current state of the art and emergent technologies and their applications.





Sohrab Shah

Stephen Yip

The core competencies students will obtain and the course outline are listed below:

#### Core competencies:

- Solid understanding of the concepts of human molecular biology including the human genome, resources to navigate it, the nature and extent of human genetic variation as it applied to pathology and genetic disorders
- Available and emerging molecular assays for diagnosis
  - understanding the nature of the test, its limitations and assumptions for interpretation
  - appreciate that specific molecular variants can lead to unique histopathology, biochemical findings, and biological behaviour with direct impact on clinical management of patients
  - understand that careful evaluation of the tissue (for extraction of genetic material) and proper pathologic diagnosis are necessary prerequisites for molecular assays
- Understand qualitative differences between human genomes and prokaryotic genomes and highlight the role of genomics in the field of microbiology and infectious diseases
- Understanding the role of computation and bioinformatics in emergent assays

## Syllabus, lead instructors: Dr. Stephen Yip and Dr. Sohrab Shah

Day 1: Concepts in molecular biology
Day 2: Diagnostic tests in 'omics'
Day 3: Whole genome/transcriptome approaches + systems biology
Day 4: Pathogen genomics

Day 5: Future trends near and long term

## UBC Pathology Genomics/Bioinformatics Core 2013 – TOR

- Provide clinical trainees and graduate students in the Department of Pathology & Laboratory Medicine with an inclusive yet focused and practical course on molecular diagnostic assays relevant to anatomical pathology, hematopathology, medical biochemistry, medical microbiology, and neuropathology.
- Acknowledge that this is a rapidly- evolving field with constantly changing technology as well as expectations and demands from the end-users. The course will include the most up-to-date topics and will also briefly touch on issues such as ethical/medico-legal concerns (including DTC genomics such as 23andMe). However, given the limited time frame of the course these topics may be discussed in detail in subsequent lectures (or PAL series).
- The course will review basic concepts of molecular genetics and how aberrations in the genome/ epigenome can lead to phenotypic changes from congenital malformations to cancers, liquid and solid. This is followed by survey of the available molecular diagnostic assays used to identify these changes and review of tests "on the horizon".
- The course will devote significant amount of time and resources into survey of open source bioinformatic tools and resources and how they can be utilized in laboratory medicine in clinical practice or as part of research project.
- Survey of relevant topics on pathogen genomics (eg. rapid identification of pathogens in an outbreak), microbiome (?), and liquid- based diagnostics (biochemistry) will be included.
- There will be a discussion on "careers" in molecular diagnostics with active participation by current fellows (Hector Chang and Tomo).
- Highlight the local expertise and resources in advanced translational genomics to trainees and clinical staff to foster collaborations.
- Guest speaker should be experienced in advanced molecular diagnostics and highlight his/her contribution(s) to the field. Also, there should be interactions with the trainees to facilitate career development.

## A Glimpse of Some of Our Outstanding 2014 Speaker Line-up:

#### **Michael S. Anglesio, PhD**

Research Associate, Department of Pathology and Laboratory Medicine BC Cancer Research Centre, Cancer



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#### Michael M. Burgess, PhD

Chair in Biomedical Ethics at the W. Maurice Young Centre for Applied Ethics and the Department of Medical Genetics at the University of British Columbia <u>mburgess@ethics.ubc.ca</u>



#### Bruce Carleton, B.Pharm, Pharm.D.

Senior Clinician Scientist, CFRI Professor, Department of Pediatrics, University of British Columbia Director, Pharmaceutical Outcomes Programme, BC Children's Hospital <u>bcarleton@popi.ubc.ca</u>



#### Hector Chang - To get it from Drs. Yip/Shah

#### Martin Hirst, PhD

Head of Epigenomics, Michael Smith Genome Sciences Centre, BC Cancer Agency Assistant Professor, Dept. of Microbiology and Immunology, Centre for High-Throughput Biology, University of British Columbia <u>hirst@chibi.ubc.ca</u>



Anthony John Lafrate, MD Associate Professor in Pathology Harvard Medical School <u>aiafrate@partners.org</u>

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#### Janessa Laskin, MD Medical Oncologist

BC Cancer Agency jlaskin@bccancer.bc.ca



#### Ryan D. Morin, MSc, PhD

Assistant Professor, Simon Fraser University Affiliations: Department of Molecular Biology and Biochemistry and School of Computing Science (Associate Member), Simon Fraser University Scientist, Genome Sciences Centre, BC Cancer Agency <u>rdmorin@sfu.ca</u>



#### Torsten Nielsen, PhD, MD

Professor, Departments of Pathology, University of British Columbia Associate Professor, Orthopaedics and the Department of Urologic Sciences, University of British Columbia <u>torsten@mail.ubc.ca</u>



#### Kasmintan (Intan) A. Schrader, MBBS UBC Medical Genetics & BCCA



#### Sohrab Shah, MSc, PhD (UBC)

Scientist, Department of Molecular Oncology, BC Cancer Agency; and Assistant Professor, Department of Pathology, UBC <u>sshah@bccrc.ca</u>



#### Stephen Yip, MD, PhD, FRCPC

Assistant Professor, Department of Pathology and Laboratory Medicine Clinician-Scientist and Consultant Neuropathologist, BC Cancer Agency <u>syip@bccancer.bc.ca</u>



#### MONDAY, JANUARY 6, 2014

TIMETABLE

#### THEME - DAY 1: CONCEPTS IN MOLECULAR BIOLOGY (REVIEW) // LOCATION: VGH, TAYLOR FIDLER AUDITORIUM

TIME	TITLE	LECTURER
0830-0900	REGISTRATION/HOUSEKEEPING (iClicker assignment ?)	
0900-1010	Introduction to course/pretest/central dogma and beyond	Stephen Yip & Sohrab Shah
1010-1025	DICER aberrations in human cancers - clinical discovery and functional consequences	Michael S. Anglesio
1030-1200	Somatic mutation and cancer and congenital abnormalities and de novo mutations- what is known?	Stephen Yip
1200-1300	LUNCH	
1300-1430	Sequencing technology - from sanger to illumina	Martin Hirst
1430-1600	Impact of advanced diagnostics on society	Michael M. Burgess

#### TUESDAY, JANUARY 7, 2014

#### THEME - DAY 2: DIAGNOSTIC TESTS IN ,OMICS' // LOCATION: VGH, TAYLOR FIDLER AUDITORIUM

TIME	TITLE	LECTURER
0850-0900	Feedback of 'day 1' talks	
0900-0950	Survey of practical germline genetic testing	Intan Schrader
1000-1050	Survey of practical somatic mutation testing and pathology considerations (FFPE/ Mol Fx)	Hector Chang
1100-1150	ALK translocation in NSCLC	Anthony John Lafrate
	BCCA ONCOLOGY GRAND ROUNDS	
1200-1250	Implementation of clinical somatic mutation testing - The MGH experience	Anthony John Lafrate
1330-1420	Fusion events in malignant gliomas - functional and clinical implications	Anthony John Lafrate
1430-1520	Other molecular diagnostic assays in current pathology use - Oncotype Dx/PAM50/Mammoprint	Torsten Nielsen
1530-1630	Personalized Oncogenomics - clinical translation of genomic discoveries	Janessa Laskin

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#### WEDNESDAY, JANUARY 8, 2014

#### THEME - DAY 3: WHOLE GENOME/TRANCRIPTOME APPROACHES/LAB DAY // LOCATION: THE LIFE SCIENCES INSTITUTE

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TIME	TITLE	LECTURER
0850-0900	Feedback of 'Day 2' talks	
0900-0950	Systems biology apporaches to molecular research in cancer - TCGA	Sohrab Shah & Ryan Morin
1000-1050	Resources for human genome browsing (UCSC genome browser)	Ryan Morin
1100-1150	Basic approaches to determining human genome mutation data	Sohrab Shah & Ryan Morin
1200-1250	LUNCH	
1300-1400	Pharmacogenomics	Bruce Carleton & Ryan Morin
1400-1500	Analytical workflow in germline testing	Intan Schrader & Ryan Morin
1500-1600	Data resources for cancer genomics (TCGA portal, etc)	Sohrab Shah & Ryan Morin
1600-1700	Functional validation (CIITA)	Sohrab Shah & Ryan Morin

#### THURSDAY, JANUARY 9, 2014

#### THEME - DAY 4: MICROBIAL GENOMICS AND METAGENOMICS // LOCATION: VGH, TAYLOR FIDLER AUDITORIUM

TIME	TITLE	LECTURER
0850-0900	Feedback of 'Day 3' talks	
0930-1020	Genomics in Medical Microbiology (introduction to concepts, applications and lab methods)	PT??
1030-1120	Bioinformatics analysis of microbial WGS data - seqeunce analysis	WH??
1130-1220	Bioinformatics analysis of microbial WGS data - comparative genomics	WH??
1230-1320	LUNCH	
1330-1420	Outbreak investigations using genomics data	PT??
1430-1520	Metagenomics in Medical Microbiology (microbiome and pathogen disovery)	PT??
1630-1650	Future Directions for Genomics in Microbiology	PT/WH??

#### FRIDAY, JANUARY 10, 2014

#### THEME - DAY 5: MICROBIAL GENOMICS AND METAGENOMICS // LOCATION: VGH, TAYLOR FIDLER AUDITORIUM

TIME	TITLE	LECTURER
0850-0900	Feedback of 'Day 4' talks	
0900-1030	Implementation of advacned molecular diagnostics into everyday pathology practice	Ryan Morin
1030-1200	Panel discussion - faculties (SA, DH, MM, SS, SY, TON)	DH??
1230-1330	PALS - TBC - patient advocate	Patient advocate
1330-1400	WRAP UP	DH??