



**AACR Integrative Molecular Epidemiology Workshop**  
August 5-9, 2016  
Westin Copley Place Hotel • Boston, Massachusetts

**Director:**

Thomas A. Sellers, Moffitt Cancer Center and Research Institute, Tampa, Florida

**Co-Directors:**

Peter Kraft, Harvard School of Public Health, Boston, Massachusetts

Lorelei A. Mucci, Harvard School of Public Health, Boston, Massachusetts

**Faculty:**

1. Jiyoung Ahn, New York University School of Medicine, New York, New York
2. Paul L. Auer, University of Wisconsin-Milwaukee, Milwaukee, Wisconsin
3. Andrew H. Beck, Harvard Medical School and Beth Israel Deaconess Medical Center, Boston, Massachusetts
4. Kevin M. Brown, National Cancer Institute, Bethesda, Maryland
5. Maria Teresa Landi, National Cancer Institute, Bethesda, Maryland
6. Alvaro N. A. Monteiro, Moffitt Cancer Center and Research Institute, Tampa, Florida
7. Bogdan Pasaniuc, University of California, Los Angeles, California
8. Paul A. Scheet, The University of Texas MD Anderson Cancer Center, Houston, Texas
9. Shelley S. Tworoger, Harvard Medical School, Boston, Massachusetts

**Lecturers:**

1. Timothy R. Rebbeck, Dana Farber Cancer Institute, Boston, Massachusetts
2. Brian M. Wolpin, Dana-Farber Cancer Institute, Boston, Massachusetts

**Junior Faculty:**

1. Rebekah Baskin, St. Jude Children's Research Hospital, Memphis, Tennessee

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**DAY 1 - FRIDAY, AUGUST 5, 2016: INTRODUCTION, FUNDAMENTALS, AND GERMLINE VARIATION**

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12:00 p.m.-1:30 p.m.

**Registration**

1:30 p.m.-2:30 p.m.

**Welcome, Introductions, and Course Overview**

Thomas A. Sellers, Moffitt Cancer Center and Research Institute, Tampa, Florida

2:30 – 3:00 p.m.

**Workshop Pre-Test**

3:00 p.m.-3:45 p.m.

**Overview and Dimensions**

Thomas A. Sellers, Moffitt Cancer Center and Research Institute, Tampa, Florida

3:45 p.m. – 4:15 p.m.

**What Is Known About the Genetic Epidemiology of Cancer (and What Is Not)**

Peter Kraft, Harvard School of Public Health, Boston, Massachusetts

4:15 p.m.-4:30 p.m.	<b>Break</b>
4:30 p.m.-6:30 p.m.	<b>Publicly Available Online Genomics Resources</b> Paul A. Scheet, The University of Texas MD Anderson Cancer Center, Houston, Texas Paul L. Auer, University of Wisconsin-Milwaukee, Milwaukee, Wisconsin
6:30 p.m.-7:30 p.m.	<b>Discussion of Ioannidis and Rothman Articles</b> Peter Kraft, Harvard School of Public Health, Boston, Massachusetts
7:30 p.m.-9:00 p.m.	<b>Opening Reception and Group Buffet Dinner</b>

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## **DAY 2 - SATURDAY, AUGUST 6, 2016: INTEGRATING DATA ON GERMLINE VARIATION (PART 2)**

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7:00 a.m.-8:00 a.m.	<b>Continental Breakfast</b>
8:00 a.m.-9:00 a.m.	<b>Genome-Wide Association Studies and Imputation-Based Association Mapping</b> Paul L. Auer, University of Wisconsin-Milwaukee, Milwaukee, Wisconsin Paul A. Scheet, The University of Texas MD Anderson Cancer Center, Houston, Texas
9:00 a.m.-10:00 a.m.	<b>Mendelian Randomization</b> Paul L. Auer, University of Wisconsin-Milwaukee, Milwaukee, Wisconsin
10:00 a.m.-10:15 a.m.	<b>Break</b>
10:15 a.m.-11:45 a.m.	<b>Exome and Targeted Next-Generation Sequencing</b> Paul L. Auer, University of Wisconsin-Milwaukee, Milwaukee, Wisconsin Paul A. Scheet, The University of Texas MD Anderson Cancer Center, Houston, Texas
11:45 a.m.-12:45 p.m.	<b>Group Photo followed by Buffet Lunch with Career Development Conversations</b>
12:45 p.m.-1:30 p.m.	<b>Exome and Targeted Next-Generation Sequencing:</b> <b><i>“Designing your population sciences gene-mapping study on a budget!”</i></b> Paul L. Auer, University of Wisconsin-Milwaukee, Milwaukee, Wisconsin Paul A. Scheet, The University of Texas MD Anderson Cancer Center, Houston, Texas
1:30 p.m.-2:15 p.m.	<b>Categorical and Multivariate Outcomes</b> Peter Kraft, Harvard School of Public Health, Boston, Massachusetts
2:15 p.m.-2:30 p.m.	<b>Break</b>

- 2:30 p.m.-3:45 p.m.      **Application of NextGen Sequencing for Germline Analysis**  
 Maria Teresa Landi, National Cancer Institute, Bethesda, Maryland
- 3:45 p.m.-5:30 p.m.      **Microbiome**  
 Jiyoung Ahn, New York University School of Medicine, New York, New York
- 5:30 p.m.-5:45 p.m.      **Break**
- 5:45 p.m.-6:45 p.m.      **Integrative Molecular Epidemiology Applied Research Seminar #1**  
*The goal of these seminars will be to provide examples that highlight how integrative molecular epidemiology tools are utilized by top researchers. Speakers will present their latest research and highlight how integrative molecular epidemiology approaches can be applied as well as discuss challenges and practical limitations.*
- 6:45 p.m.-7:45 p.m.      **Multilevel Approaches To Address Cancer Disparities: From Nucleotide to Neighborhood**  
 Timothy R. Rebbeck, Dana Farber Cancer Institute, Boston, Massachusetts
- 6:45 p.m.-7:45 p.m.      **Group Buffet Dinner**

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**DAY 3 - SUNDAY, AUGUST 7, 2016: FUNCTIONAL FOLLOW-UP**

- 7:00 a.m.-8:00 a.m.      **Continental Breakfast**
- 8:00 a.m.-9:00 a.m.      **Establishing the *Functional* Chain of Causation**  
 Alvaro N. A. Monteiro, Moffitt Cancer Center and Research Institute, Tampa, Florida
- 9:00 a.m.-9:15 a.m.      **Research Vignettes: Mini-Brains and Organotypic Cultures**  
 Rebekah Baskin, St. Jude Children's Research Hospital, Memphis, Tennessee
- 9:15 a.m.-9:30 a.m.      **Research Vignettes: Identifying Allele-Specific Binding Proteins for Potential Gene-Regulatory Variants From GWAS**  
 Kevin M. Brown, National Cancer Institute, Bethesda, Maryland
- 9:30 a.m.-10:30 a.m.      **Overview of Epigenetics**  
 Alvaro N. A. Monteiro, Moffitt Cancer Center and Research Institute, Tampa, Florida
- 10:30 a.m.-11:00 a.m.      **Break**
- 11:00 a.m.-11:30 a.m.      **ENCODE and the Epigenome Roadmap in a Nutshell**  
 Alvaro N. A. Monteiro, Moffitt Cancer Center and Research Institute, Tampa, Florida  
 Kevin M. Brown, National Cancer Institute, Bethesda, Maryland  
 Rebekah Baskin, St. Jude Children's Research Hospital, Memphis, Tennessee

11:30 p.m.-12:30 p.m.	<b>Buffet Lunch with Career Development Conversations</b>
12:30 p.m.-1:00 p.m.	<b>Practical issues in Transcriptomics</b> Paul L. Auer, University of Wisconsin-Milwaukee, Milwaukee, Wisconsin
1:00 p.m. – 2:00 p.m.	<b>Overview of Functional Genomic Characterization of GWAS Loci</b> Kevin M. Brown, National Cancer Institute, Bethesda, Maryland
2:00 p.m.-2:30 p.m.	<b>Bioinformatics Toolkit for Functional Analysis</b> Rebekah Baskin, St. Jude Children's Research Hospital, Memphis, Tennessee
2:30 p.m. – 3:00 p.m.	<b>Break</b>
3:00 p.m.-5:30 p.m.	<b>Functional Analysis</b> Alvaro N. A. Monteiro, Moffitt Cancer Center and Research Institute, Tampa, Florida Kevin M. Brown, National Cancer Institute, Bethesda, Maryland Rebekah Baskin, St. Jude Children's Research Hospital, Memphis, Tennessee
5:30 p.m.-6:30p.m.	<b><u>Integrative Molecular Epidemiology Applied Research Seminar #2</u></b> <b>Genetic and Epigenetic Analysis in Tissue</b> Maria Teresa Landi, National Cancer Institute, Bethesda, Maryland
7:15 p.m.-	<b>Group Dinner and Social Event</b>

#### **DAY 4 - MONDAY, AUGUST 8, 2016: BIOMARKERS AND TUMOR PROFILING**

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7:00 a.m.-8:00 a.m.	<b>Continental Breakfast</b>
8:00 a.m.-9:30 a.m	<b>Biomarkers</b> Shelley S. Tworoger, Harvard Medical School, Boston, Massachusetts
9:30 a.m.-10:30 a.m.	<b>Overview of Tumor Profiling</b> Lorelei A. Mucci, Harvard School of Public Health, Boston, Massachusetts
10:30 a.m.-10:45 a.m.	<b>Break</b>
10:45 a.m.-11:45 a.m.	<b>Tumor Profiling Tools and Their Limitations</b> Andrew H. Beck, Harvard Medical School and Beth Israel Deaconess Medical Center, Boston, Massachusetts
11:45 a.m.-12:30 p.m.	<b>Buffet Lunch</b>
12:30 p.m.-2:30 p.m.	<b>Practical Study Design</b> Lorelei A. Mucci, Harvard School of Public Health, Boston, Massachusetts

## Offsite Laboratory Visits

2:45 p.m.-3:30 p.m.

Travel time to observational visits and walking to the labs

3:30 p.m.-5:00 p.m.

*Offsite Observational Laboratory Visits*

*Participants will attend one of the four tours below:*

- **Tour A: Beth Israel Deaconess Medical Center Pathology/Microarray Lab**
  - *Andrew H. Beck, Harvard Medical School and Beth Israel Deaconess Medical Center, Boston, Massachusetts*
  - *Benjamin Glass, Beth Israel Deaconess Medical Center, Boston, Massachusetts*

In this tour, the workshop participants will learn how histopathology specimens are received in the pathology laboratory and prepared for tissue-based molecular and morphological analyses. We will explain how the tissues are imaged, annotated, and sampled for tissue microarray construction. We will describe how tissue microarray sections are stained, imaged, and stored for analysis. The tour will then progress to a demonstration of how the slides are analyzed using various image analysis programs, some of which have been developed in the lab. Finally, we will have a presentation that describes how the quantitative measures obtained from these tissue-based analyses are incorporated into integrative molecular epidemiology studies.

- **Tour B: BWH/Harvard Cohorts Biorepository**
  - *Shelley S. Tworoger, Harvard Medical School, Boston, Massachusetts*
  - *Sherilyn Sawyer, Brigham & Women's Hospital, Boston, Massachusetts*

This tour of the Brigham and Women's Hospital/Harvard Cohorts Biorepository and the Dana Farber/Harvard Cancer Center High-Throughput Genotyping Core will provide students with an understanding of decision points in the collection and use of biological specimens in population-based studies. We will focus on practical issues underlying sample collection, processing, and storage, as well as provide an appreciation for the depth of quality control efforts needed to produce high-quality data from biospecimens.

- **Tour C: Broad Institute Metabolomics Platform**
  - *Clary B. Clish, Broad Institute of MIT and Harvard, Cambridge, Massachusetts*

The Broad Institute's Metabolomics Platform undertakes systematic studies of metabolites, the unique chemical fingerprints that result from both normal and pathological cellular processes. Because of their immediacy to cell physiology, metabolites are promising biomarkers for disease, as well as clues to genetic or drug alterations in the cell. The

Metabolomics Platform undertakes both discovery- and hypothesis-driven studies in collaboration with researchers across the Broad and external research communities. Current projects include the discovery of early metabolic markers of predisposition to type 2 diabetes, characterization of mitochondrial disorders, and elucidating metabolic networks in cellular studies of cancer metabolism, infectious disease, and metabolic disorders.

- **Tour D: Cancer Proteomics Core at Beth Israel Deaconess Medical Center**
  - *Towia A. Libermann, Beth Israel Deaconess Medical Center, Boston, Massachusetts*

The tour of the Cancer Proteomics Core at Beth Israel Deaconess Medical Center will provide students with an understanding of the tools of modern proteomics. The Cancer Proteomics Core (CPC) provides all DF/HCC members access to comprehensive interdisciplinary, translational, and clinically-oriented facilities for high-sensitivity, high-resolution, and high-throughput proteomics and metabolomics services, with particular emphasis on clinical sample analysis and in-depth scientific consultation for proteomics, metabolomics, lipidomics, and data analysis. The CPC also provides access to proteomic services for basic cancer research exploring signaling pathways and other aspects of cancer biology.

5:00 p.m.-6:00 p.m. Travel time back to hotel

6:00 p.m.- **Evening on own**

## **DAY 5 - TUESDAY, AUGUST 9, 2016: PUTTING IT ALL TOGETHER**

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7:00 a.m.-8:00 a.m. **Continental Breakfast**

8:00 a.m.-9:00 a.m. **Integrative Data Analysis Methods**  
 Peter Kraft, Harvard School of Public Health, Boston, Massachusetts  
 Bogdan Pasaniuc, University of California, Los Angeles

9:00 a.m.-10:00 a.m. **eQTL and mQTL**  
 Peter Kraft, Harvard School of Public Health, Boston, Massachusetts  
 Bogdan Pasaniuc, University of California, Los Angeles

10:00 a.m.-10:30 a.m. **Break**

10:30 a.m.-11:30 a.m. **Survival Skills and Ethics**  
 Thomas A. Sellers, Moffitt Cancer Center and Research Institute, Tampa, Florida  
 All faculty

11:30 a.m.-12:30 p.m.

**Integrative Molecular Epidemiology Applied Research Seminar #3**  
**Metabolomics and Cancer**

Brian M. Wolpin, Dana-Farber Cancer Institute, Boston, Massachusetts

12:30 p.m.-1:15 p.m.

**Buffet Lunch with Career Development Conversations**

1:15 p.m.-2:15 p.m.

**Integrative Molecular Epidemiology Applied Research Seminar #4**  
**Next generation computational pathology**

Andrew H. Beck, Harvard Medical School and Beth Israel Deaconess Medical Center, Boston, Massachusetts

2:15 p.m.-2:45 p.m.

**Closing Summary and Observations**

All faculty

2:45 p.m.-3:45 p.m.

**Post-Test and Evaluations**