**Speaker:** Bernard Futscher, PhD  
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**Presentation:** 3:45 – 4:10 PM  
**Q&A:** 4:10 – 4:15 PM

**Title: Written in Blood: Ultra-Sensitive and -Specific Cancer Detection Using DNA Methylation Biomarkers**

Earlier detection of cancer and its recurrence offers an opportunity to improve the treatment and management of the disease. Therefore, techniques for minimally invasive and cost-effective cancer diagnosis and monitoring are needed. Analysis of cell-free DNA (cfDNA) from cancer patient’s plasma represents one approach being developed for a wide variety of unmet clinical needs in cancer detection and monitoring.

5-methylcytosine is an obligate epigenetic modification that is temporally and spatially controlled and acts to regulate cell identity in healthy cells. In contrast, all cancer cells have an altered epigenome with global changes in their patterns and levels of DNA methylation. Detection of aberrant, cancer-specific DNA methylation in cfDNA samples provides an approach for minimally invasive cancer diagnostics and monitoring. Since multiple DNA regions are aberrantly methylated in tumors, DNA methylation could be superior to DNA mutations as a cancer-specific marker, as very few specific gene mutations are present in a large fraction of tumors. Therefore, detection of DNA methylation of several cancer-specific methylated regions may provide higher sensitivity over detecting a single or few mutation markers.

Applying artificial intelligence and machine learning tools to analyze all data in TCGA as our DNA methylation biomarker discovery dataset and hundreds of GEO datasets to validate our DNA methylation biomarkers, we discovered ultra-sensitive and -specific suites of DNA methylation markers that could be used to detect all common human cancers. We further show that our DNA methylation biomarkers also detect these cancers in their premalignant state and may predict progression to frank malignancy. Finally, we translated these DNA methylation biomarkers to clinical studies in multiple cancers at the UACC, which have validated their clinical performance.