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Abstract Student Research Conference
The DNA Repair Landscape of Women at Risk for Breast Cancer

Cancer treatments typically work by creating large amounts of DNA damage in the cells, which normally will result in cell death or apoptosis. Some cancer cells are extremely efficient at repairing DNA damage that has been induced by treatments such as radiation and chemotherapy, and are therefore resistant to the treatments. However, each individual person and cancer has a unique response. Different responses are attributed to the presence of variants in DNA repair genes in each individual. Analysis of the DNA of relatives, and specifically pairs of sisters, is particularly meaningful because it allows us to more easily identify the factors that are different for the sister that developed cancer compared to the sister that did not. This project aimed to identify pairs of female siblings with one sister who had had breast cancer and one sister who had not. One woman in each pair was a participant in the High Risk Breast Program of Vermont or a patient of Wendy McKinnon, MS, a genetic counselor at the Vermont Regional Genetics Center. All women who had had breast cancer were required to be negative for a BRCA1 or BRCA2 mutation. We successfully met our goal of enrolling six sister pairs to the Sister Study in summer 2012, and the next step in this study involves analysis of their genes by the CORE Facility at the University of Vermont. We will sequence the 5' and 3' UTRs, promoters and exons of 260 DNA repair genes using to identify significant single nucleotide polymorphisms. Interest in the Sister Study was so strong that we have obtained additional funding to expand the study to add 17 additional pairs.