Summaries of UW ICTR-Marshfield Clinic Pilot Awards, 2015

1. **PI: Tonia Carter, PhD; Karen Hansen, MD, MS**
    Associate Professor, Medicine; Project Scientist, Personalized Medicine Project
    Title: *Do Polymorphisms in the ECE1 Isoform b Promoter Associate with Osteoporosis?*

    Summary
    Peak bone mineral density (BMD) is a critical factor determining one's lifetime risk of osteoporosis. Despite the significant influence of genetic variability on peak bone mass and subsequent risk of osteoporosis, researchers have only identified two genes, the vitamin D receptor gene and the estrogen receptor gene, as contributing to osteoporosis. If our proposed study confirms that the ECE1 genotype is associated with greater risk of osteoporosis in postmenopausal women, such knowledge can be used to identify patients and their kindred at high risk of osteoporosis, permitting early intervention.

2. **PI: Scott Hebbring, PhD; David C Page, PhD**
    Associate Research Scientist, Center for Human Genetics; Professor, Biostatistics and Medical Informatics
    Title: *Application of Pedigree Data in an Electronic Health Record for Precision Medicine*

    Summary
    Family history is one of the strongest predictors of a wide spectrum of diseases and may be the result of shared genetic and/or environmental factors. The goal is to identify clinically relevant diseases that can be predicted with EHR-linked familial data. This includes diseases that may have both strong genetic and/or environmental etiologies. We will apply standard logistic regression and machine learning techniques to predict disease risk; variables that will be considered in the predictive algorithms may include age, sex, diagnoses, cohabitation, and genetic relatedness.

3. **PI: Peggy L Peissig, PhD, MBA; Yirong Wu, PhD**
    Associate Research Scientist, Biomedical Informatics Research Center; Associate Scientist, Radiology
    Title: *Breast Cancer Phenotyping and Prediction using the Electronic Health Record*

    Summary
    We aim to develop an innovative **B**reast Cancer **M**achine-learned **A**dvice-based **P**henotyping (**BMAP**) model, which integrates physician expertise and machine-learned logic rules, for identifying subjects with the "most harmful" breast cancers from the EHR. The **BMAP** model will enable the use of the EHR data to develop risk prediction models for predicting the risk of the "most harmful" breast cancer. With accurate prediction of the "most harmful" breast cancers, we hope to increase the overall efficacy of breast cancer management by targeting more intense screening and preventive measures to those women at the highest risk.

UW ICTR Partners
School of Medicine and Public Health • School of Nursing • School of Pharmacy • School of Veterinary Medicine • College of Engineering • Marshfield Clinic

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