ACTIVE RESEARCH PROTOCOLS

You, your family or your patient may be eligible for one or more of these protocols. To speak with one of our study coordinators contact them by E-mail, Phone: (614) 293-6694, Toll-free: 888-329-1654.

PROTOCOLS-SHORT EXPLANATIONS AND COORDINATOR

BRCA Modifiers study (07014) **(Leigha Senter)**
An international consortium (CIMBA: The Consortium of Investigators of Modifiers of BrcA1/2) in which 22 centers pool resources and expertise to have sufficient power to detect low-penetrance mutations and new breast and ovarian cancer susceptibility genes.

BRCA Variants study (05082) **(Leigha Senter)**
OSU is a contributing site for this NCI-funded Mayo-based study which seeks to functionally, genetically, statistically and phyllogenetically decipher the pathogenicity of missense variants within the *BRCA1/2* genes. The local PI is Amanda Toland, PhD, and she is specifically recruiting individuals with breast or ovarian cancer who have a deleterious mutation or variant of uncertain significance in *BRCA1* or *BRCA2*.

Breast Cancer Tissue Bank 2007C0066 **(Rob Pilarski)**
An ongoing protocol to collect specimens and clinical, treatment and outcomes data on women diagnosed with breast adenocarcinoma. Specimens and data are banked and made available to researchers with IRB-approved protocols. Tissue microarrays have been made on a subset of these patients.

Cancer DNA Banking protocol (OSU9952/1999C0245) **(Ilene Lattimer)**
This is a general "umbrella" protocol that allows for storage of DNA and tissues for future research.

Cardiovascular Genetics Collaborations with Mohler Lab **(Amy Sturm)**
Novel variants of uncertain significance in genes underlying inherited arrhythmia syndromes will undergo functional studies in the Mohler laboratory. One specific focus is the *ANK2* gene.

Cardiovascular Genetics Data Repository **(Amy Sturm)**
The aim of this study is to create a data repository that will contain clinical information on patients who have been diagnosed with, or are at increased risk for, cardiovascular genetic conditions.

CLL (chronic lymphocytic leukemia) study (07087/2007C0074) **(Leigha Senter)**
Individuals diagnosed with CLL and their relatives are accrued to this protocol to store genetic material and to collect personal and family history to identify predisposing factors to CLL.

Columbus Foundation Down Syndrome **(Dawn Allain)**
The project has two arms: The first is a survey of caregivers for adults with Down syndrome aimed at assessing health status, access to healthcare and medical screening/care. The second component includes a targeted physical assessment and labs.

Controls for Genetics Research study (2005H0249) **(Elizabeth Solinger)**
The primary objective of this protocol is to create a bank of control samples for use as a general resource for researchers in the field of human genetics. This will be done by collecting and storing clinical and family history information gained from a questionnaire, and by collecting a blood sample.

Dilated Cardiomyopathy (DCM) Research Project (Protocol #: 2012H0185) **(Ana Morales)**
Study aimed at identifying the genetic basis of DCM. Currently recruiting individuals with DCM and their family members. Protocol involves pedigree and cardiovascular history intake, medical record analysis, recruitment and cardiovascular screening of family members, a blood sample for exome sequencing, and availability of results notification for confirmatory testing.

Endocrine Neoplasia Repository study (06032) **(Rebecca Nagy)**
A clinicopathologic data and biological samples repository for individuals with various endocrine neoplasia, with a focus on differentiated thyroid cancer and benign thyroid disease. This project corresponds to Core A of the P01 program project entitled "Genetic and Signaling Pathways in Epithelial Thyroid Cancer"; PI, Matthew Ringel, MD, Funding period 3/2008-3/2013.

Familial Papillary Thyroid Cancer (PTC) study (9812) **(Rebecca Nagy)**
A gene-hunting study looking for genes that predispose to papillary thyroid cancer

Family Health Link (FHL) **(Amy Sturm/Kevin Sweet)**
Family HealthLink is a online automated family history risk assessment triage tool for the two most common diseases, cancer and coronary heart disease, with tailored risk assessments and health messages for the user; applicable for use in the clinical setting and the general public. User information is collected, stored and available for query in a data repository through the OSUMC Information Warehouse (IW).

Georgetown ADAPT/ASSURE Projects **(Leigha Senter)**
Multicenter study to test the use of decision making tools in individuals undergoing testing for BRCA1/2 gene mutations.

Genomic counseling
Through interviews of patients with chronic disease and community participants on the OSU-Coriell Personalized Medicine Collaborative, we will develop and evaluate a novel genomic counseling delivery model for multiplexed genomic and pharmacogenomic results.

Jane Engelberg Memorial Foundation Study (JEMF) (60028055) **(Dawn Allain/Kate Shane)**
A study to determine the value of genetic counselors in genetic testing for breast-ovarian cancer.

Low Penetrance Genes study (05102) **(Heather Hampel)**
This study aims to identify multiple human low penetrance genes that control genetic susceptibility and resistance to cancer by testing genes that map to regions that have been identified in mice as housing such genes. Multiple cancer types, primarily from archived tissue, will be studied. We are particularly interested in identifying combinations of genetic variants that interact to affect cancer risk.

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Metastatic Squamous Cell Carcinoma (MSCC) **(Dawn Allain)**
The goal of this study is to determine the feasibility of using microRNA profiling and exome sequencing to identify changes which occur in SCCs that become metastatic.

MOPD (microcephalic osteodysplastic primordial dwarfism) study **(Rebecca Nagy)**
A study of genotype/phenotype correlations in children with microcephalic osteodysplastic primordial dwarfism type I (MOPD I), caused by mutations in U4atac, a small nuclear RNA.

Non-cancer DNA Banking protocol (0590/2004H0136) **(Amy Sturm)**
This is a study to store the biological material of participants for the discovery and characterization of genes involved in hereditary disease predisposition.

Ohio Colorectal Cancer Prevention Initiative (OCCPI) **(Rachel Pearlman/Heather Hampel)**
OCCPI is a demonstration study to show that universal screening for Lynch syndrome (LS) can be accomplished on a large-scale basis. The study includes around 40 Ohio hospitals and enrolls all newly diagnosed colorectal cancer patients. In addition to being screened for LS, patients and their FDRs will be contacted by Dr. Paskett's team for a colonoscopy screening adherence study. Leftover samples form a biorepository.

OSU Heart Center Biobank (2008H0113) **(Amy Sturm)**
The aim of this study is to collect and store DNA to be used in the investigation of the genomic basis for cardiovascular diseases and genomic determinants of disease outcome and response to therapy. This Repository will allow researchers access to DNA from a broad sample of patients with a variety of co-morbidities which is essential for the further understanding of the genomic basis of cardiovascular diseases.

Pharmacogenomics (PGRN TPP) **(Kevin Sweet/Amy Sturm)**
Project to start using pharmacogenomic/biomarker information in clinical settings for both research and clinical use. The use of novel EMR methods will make this information available to clinicians and help with clinical decision making support

PMS2 study (2006C0005) **(Leigha Senter)**
A study to characterize mutations in the PMS2 gene in individuals with HNPCC-associated cancers.

Prospective Cohort BRCA study (0107) **(Leigha Senter)**
A prospective questionnaire-based study offered to all female patients who test positive for *BRCA* mutations to identify hormonal, reproductive and lifestyle factors that are associated with the risk of developing breast and ovarian cancer.

***PTEN* Immune Dysregulation Study** **(Rob Pilarski)**
An investigation of the immune effecter cells of patients with PTEN mutations to determine how immune system dysregulation may contribute to their heightened risk for cancer.

Squamous Cell Carcinoma (SCC) study (05078) **(Dawn Allain)**
A collaborative study with UCSF to determine the genetic etiology of SCC of the skin. Multiple molecular methods, especially array comparative genomic hybridization, will be used to analyze both normal and tumor DNA.

The OSUWMC MD Anderson thyroid cancer SPORE(multiple studies within)
Multi-institutional SPORE focusing on thyroid cancer pre-disposition, progression and treatment.

Triple Negative Modifiers **(Rob Pilarski)**
A study done in collaboration with Dr. Fergus Couch using a GWAS approach to identify genetic factors contributing to the development of triple-negative breast cancers.

Uveal Melanoma study (06036) **(Rob Pilarski)**
A study of the molecular genetics of uveal melanomas including analyzing prognostic molecular genetic markers for detection of aggressive disease, and germline mutations predisposing to early onset of the disease and/or high frequency of other cancers in uveal melanomas patients.

Villalona FancD2 nuclear foci (NCI 8472/OSU09100) **(Rob Pilarski)**
A study of the effectiveness of a tissue assay in determining use of PARP inhibitors and presence of germline mutations in BRCA1/2.