February 20, 2007

This is an abbreviated version of Genomics and Health Weekly Update, published by the National Office of Public Health Genomics at CDC. View using the HTML option in Groupwise. The full the unabridged version, is available online at:

**Spotlight:** February is American Heart Month

Heart disease is the leading cause of death in the United States and a major cause of disability. Family medical history offers important information for identifying risk in individuals.

[Click here for more information.](#)

**Ashkenazi Jewish**

Selected genetic disorders affecting Ashkenazi Jewish families
Weinstein LB

**Blood Disorders**

“Mechanism driving iron overload in thalassemia identified”
(Feb 14) News-Medical.Net reports, “Led by researchers at Weill Cornell Medical College in New York City, an international group of scientists has pinpointed a specific genetic relationship as the cause of dangerous iron overload in persons with a form of the inherited blood disease, beta-thalassemia.”

**Bone Disorders**

“Second gene discovered for recessive form of brittle bone disease”
(Feb 8) EurekAlert! reports, “Researchers at the National Institutes of Health and other institutions have found a second genetic defect that accounts for previously unexplained forms of osteogenesis imperfecta (OI), a disorder that weakens bones, sometimes results in frequent fractures and is sometimes fatal.”

**Cancer**

“Variation in gene caspase-8 may help protect against breast cancer”
(Feb 13) News-Medical.Net reports, “A large-scale analysis of data on breast cancer risk has concluded that a common variation in the gene caspase-8 (CASP8) is associated with a somewhat lower risk of the disease. Variants are small changes that occur in a gene sequence.”

“Cold Spring Harbor Laboratory Scientists Discover New Gene That Prevents Multiple Types Of Cancer”
(Feb 9) Medical News Today reports, “A decades-old cancer mystery has been solved by researchers at Cold Spring Harbor Laboratory (CSHL).”

“International Study Points To New Breast Cancer-susceptibility Gene”
(Feb 9) Science Daily reports, “A gene whose existence was detected only a couple of years ago may increase women's risk of breast cancer when inherited in a mutated form, and may contribute to prostate cancer as well, researchers at Dana-Farber Cancer Institute and colleagues in Finland report in a new study.”

“Gene wrapper' discovery aids cancer fight”
(Feb 15) ABC News Online reports, “Cancer researchers in Sydney hope to prevent many early-onset cancer diagnoses with a new discovery.”

“Survey finds perceived risk of recurrence low in African-American breast cancer survivors”
(Feb 15) EurekAlert! reports, “A unique survey of African American breast cancer survivors at heightened risk for hereditary breast cancer has found the majority do not believe they have an increased chance of developing the cancer again.”

“Tumors' Genetic Profile Captured By Technology”
(Feb 14) Medical News Today reports, “A study led by researchers at Dana-Farber Cancer Institute and Broad Institute of the Massachusetts Institute of Technology and Harvard University provides the first demonstration of a practical method of screening tumors for cancer-related gene abnormalities that might be treated with 'targeted' drugs.”

“Simple 2-gene test sorts out similar gastrointestinal cancers”
(Feb 12) EurekAlert! reports, “A powerful two-gene test distinguishes between a pair of nearly identical gastrointestinal cancers that require radically different courses of treatment, researchers report this week in the online Early Edition of the Proceedings of the National Academy of Sciences.”

High-throughput oncogene mutation profiling in human cancer
Thomas RK, et al.
Nat Genet 2007 Feb

Cardiovascular, Diabetes, Hypertension, Stroke, and Related Conditions
“A unique twin study on the increased cardiometabolic risk in obesity”
(Feb 13) EurekAlert! reports, “Study finds that obesity, already in its early stages and independent of genetic influences, is associated with deleterious alterations in the lipid metabolism known to facilitate atherogenesis, inflammation and insulin resistance.”

Risk factors for coronary artery disease in patients with elevated high-density lipoprotein cholesterol
DeFaria Yeh D, et al.
Am J Cardiol 2007 Jan;99(1):1-4
“Brigham team devises model to better gauge heart risk”
(Feb 14) Boston.com reports, “It is a fact that has long frustrated doctors and their patients: Up to 20 percent of women who suffer heart attacks and other coronary problems had no obvious risk factors -- no high blood pressure or elevated cholesterol.”

“Type-2 diabetes genes identified”
(Feb 11) News-Medical.Net reports, “The most important genes associated with a risk of developing type-2 diabetes have been identified, scientists report today in a new study.”

Association of genetic variations with nonfatal venous thrombosis in postmenopausal women
Smith NL, et al.
JAMA 2007 Feb;297(5):489-98

Genetics of intracranial aneurysms
Nahed BV, et al.
Neurosurgery 2007 Feb;60(2):213-25; discussion 225-6

Ethical, Legal, and Social Issues
“People with genetic conditions twice as likely to report health insurance denial”
(Feb 12) eMaxHealth reports, “A new study published in the February 2007 issue of the American Journal of Medical Genetics reveals that individuals with genetic conditions are twice as likely to report having been denied health insurance than individuals with other chronic illnesses.”

“Patenting Life”
(Feb 13) The New York Times reports, “YOU, or someone you love, may die because of a gene patent that should never have been granted in the first place. Sound far-fetched?”

Access to health insurance: Experiences and attitudes of those with genetic versus non-genetic medical conditions
Kass NE, et al.
Am J Med Genet A 2007 Feb

Eye Disorders
“U-M team: Genetic testing sheds light on degenerative eye disease”
(Feb 13) EurekAlert! reports, “Genetic testing for eye disease is providing vital information about complex retinal diseases, especially when used to confirm a clinician’s diagnosis.”

Genomic Tools
“Biochip allows genes to express themselves”
(Feb 13) News-Medical.Net reports, “Biochip platforms that work as artificial cells are
attractive for medical diagnostics, interrogation of biological processes, and for the production of important biomolecules.”

“New patented technology for next generation of DNA and RNA microarrays” (Feb 13) News-Medical.Net reports, “A novel invention developed by a scientist from New York Institute of Technology (NYIT) could revolutionize biological and clinical research and may lead to treatments for cancer, AIDS, Alzheimer’s, diabetes, and genetic and infectious diseases.”

“Which genome variants matter?” (Feb 9) EurekAlert! reports, “Findings published today in Science will accelerate the search for genes involved in human disease.”

An evaluation of power and type I error of single-nucleotide polymorphism transmission/disequilibrium-based statistical methods under different family structures, missing parental data, and population stratification
Nicodemus KK, et al.

Statistical methods for haplotype-based matched case-control association studies
Zhang H, et al.
Genet Epidemiol 2007 Feb

Mental Health (including Addiction)
“Study identifies gene variation for mild form of Schizophrenia” (Feb 13) SAWF News reports, “University of Iowa researchers have learned more about a genetic variation that is a small risk factor for a mild form of schizophrenia, yet also is associated with improved overall survival.”

“Common Gene Version Optimizes Thinking — but With a Possible Downside” (Feb 8) NIH News reports, “Most people inherit a version of a gene that optimizes their brain’s thinking circuitry, yet also appears to increase risk for schizophrenia*, a severe mental illness marked by impaired thinking, scientists at the National Institutes of Health’s (NIH) National Institute of Mental Health (NIMH) have discovered.”

“Hooked By Genes: Studies Identify DNA Regions Linked To Nicotine Dependence” (Feb 12) Science Daily reports, “Americans are bombarded with antismoking messages, yet at least 65 million of us continue to light up.”

The genomic era and serious mental illness: a potential application for psychiatric genetic counseling
Austin JC & Honer WG
Psychiatr Serv 2007 Feb;58(2):254-61
Pharmacogenomics
Pharmacogenetics of antiparkinsonian drug treatment: a systematic review
Arbouw ME, et al.
Pharmacogenomics 2007 Feb;8(2):159-76

Various pharmacogenetic aspects of antiepileptic drug therapy: a review
Mann MW & Pons G
CNS Drugs 2007;21(2):143-64

Prenatal/Perinatal Screening and Issues
“New diagnostic technology for routine prenatal genetic testing”
(Feb 11) News-Medical.Net reports, “Research studies demonstrating the viability of an approach to routinely detect the presence of fetal DNA in a mother's blood to accurately diagnose or rule out genetic defects -- as early as the first trimester -- was presented today at the 27th Annual Meeting of the Society for Maternal-Fetal Medicine being held in San Francisco.”

“Genes may trigger repeated premature birth in blacks”
(Feb 8) STLtoday.com reports, “Black women may be genetically predisposed to giving birth prematurely, according to a new study from Washington University.”

“Baby's Breath: Newborns With Respiratory Distress Potentially Have Rare Genetic Disease”
(Feb 12) Science Daily reports, “Newborns with respiratory distress should be evaluated for primary ciliary dyskinesia, a rare genetic disease that has features similar to cystic fibrosis, says Thomas Ferkol, M.D., from Washington University School of Medicine in St. Louis.”

Let’s Go Surfing
The Minnesota Gene Pool Weblog: New postings on Gene Patenting, Women’s CVD Risk Assessment