April 24th, 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: April is National Autism Awareness Month
Autism is one of a group of developmental disorders known as autism spectrum disorders (ASDs). Scientists think that both genes and the environment play a role in ASDs. Family studies have been helpful in understanding how genes contribute to autism. Read more.

Autism

“Autism research yields possibility of newborn diagnostic test”
(Apr 12) azcentral.com reports, “The lead researcher at Phoenix's non-profit Translational Genomics Research Institute (TGen) says a genetic test for autism is becoming more of a possibility.”

Cancer

“ATR Checkpoint-Activating DNA Structure”
(Apr 19) Medical News Today reports, “As published in Genes & Development, Dr. Karlene Cimprich and colleagues at Stanford University have determined the minimal DNA structure sufficient to activate the ATR-mediated DNA damage checkpoint.”

“World first test potential to identify breast cancer patients who will react badly to radiotherapy”
(Apr 19) EurekAlert! reports, “Medical scientists at the University of Leicester have announced a potentially unique advance in breast cancer research by identifying two genes associated with adverse reaction to cancer treatment.”

“Polymorphisms in the glutathione S-transferase genes and treatment outcomes in acute myeloid leukemia”
(Apr 18) News-Medical.Net reports, “Levels of an enzyme involved metabolizing chemotherapy may help patients with acute myeloid leukemia (AML) to be able to better tolerate chemotherapy, according to the results of research which will be presented by Kirsten Moysich, PhD, Department of Epidemiology, Roswell Park Cancer Institute (RPCI) at the 2007 centennial meeting of the American Association for Cancer Research (AACR), April 14-18, in Los Angeles, CA.”

“Expression Genetics, Inc. announces successful completion of phase I trial of gene-based IL-12 for treatment of ovarian cancer”
(Apr 17) Medical News Today reports, “Expression Genetics, Inc., announced today the
completion of a Phase I clinical study evaluating the Company's lead drug candidate, EGEN-001.”

“Gene therapy, cancer-killing viruses and new drugs highlight novel approaches to cancer treatment”
(Apr 17) EurekAlert! reports, “Studies presented at the 2007 meeting of the American Association for Cancer Research show how researchers are using the new, as well as the natural, to help design and test new drugs to treat cancer.”

“No solution to cancer: have our genes evolved to turn against us?”
(Apr 17) Science Daily reports, “Cancer is a natural consequence of human evolution.”

“Dysfunctional DNA repair pathways”
(Apr 15) News-Medical.Net reports, “Individuals who inherit two mutant copies of any one of about 12 genes that make the proteins of the Fanconi Anemia (FA) pathway develop FA, which is characterized by increased incidence of cancer and bone marrow failure, among other things.”

“Mailman School of Public Health researchers report blood DNA can be early predictor of liver cancer”
(Apr 15) EurekAlert! reports, “Researchers at Columbia University’s Mailman School of Public Health have discovered a means for early detection of liver cancer.”

“MicroRNAs as tumor suppressors”
(Apr 15) EurekAlert! reports, “In the May 1st issue of G&D, Drs. Yong Sun Lee and Anindya Dutta (UVA) reveal that microRNAs can function as tumor suppressors in vitro.”

“Study links breast cancer risk to epigenetic changes related to race, smoking and birth size”
(Apr 15) EurekAlert! reports, “Women can encounter environmental factors that increase their risk of breast cancer at various periods of their physical development, beginning before birth and extending until menopause.”

“Revolution in cancer treatment”
(Apr 12) Belfast Telegraph reports, “Cancer treatment could be on the brink of a revolution following a study showing that it may be possible significantly to improve the effectiveness of chemotherapy drugs without causing side effects.”

Rationale for, and approach to, studying modifiers of risk in persons with a genetic predisposition to colorectal cancer
Jenkins MA, et al.
Curr Oncol Rep 2007 May;9(3):202-7

Genotype-Phenotype Correlations in Autosomal Dominant and Autosomal Recessive Polycystic Kidney Disease
Rossetti S & Harris PC
J Am Soc Nephrol 2007 Apr

Adult weight gain and central obesity in women with and without a family history of breast cancer: a case control study
Harvie MN, et al.
Fam Cancer 2007 Apr

Cardiovascular, Diabetes, Hypertension, Stroke, and Related Conditions

“Twin studies reveal genetic components leading to cardiac and kidney disease” (Apr 17) EurekAlert! reports, “Daniel O’Connor, M.D., Professor of Medicine and Pharmacology at the University of California, San Diego (UCSD) School of Medicine has studied about 265 twin pairs over the past few years, which has led him to some surprising discoveries.”

“deCODE launches novel DNA-based reference laboratory test for assessing inherited risk of type 2 diabetes” (Apr 16) News-Medical.Net reports, “deCODE genetics has announced the launch of deCODE T2, a novel DNA-based reference laboratory test for the first common and well-validated genetic risk factor for type 2 diabetes (T2D).”

“Study shows that indigenous people are not genetically prone to diabetes” (Apr 16) EurekAlert! reports, “The high rate of diabetes among indigenous people is not due to their genetic heritage, according to a recently published study.”

“Study fails to verify gene variations as risk factors for certain cardiovascular problems” (Apr 15) Medical News Today reports, “New research has failed to confirm findings from smaller studies that 85 gene variations are associated with an increased risk for acute coronary syndromes (ACS), which includes heart attack and a type of angina, according to a study in the April 11 issue of JAMA.”

Atherosclerosis: the path from genomics to therapeutics
Miller DT, et al
J Am Coll Cardiol 2007 Apr;49(15):1589-9

Insights on Pathogenesis of Type 2 Diabetes from MODY Genetics
Weedon MN & Frayling TM
Curr Diab Rep 2007 Apr;7(2):131-8

“First gene test for diabetes risk now available” (Apr 16) Reuters Health reports, “The world's first genetic test to assess the risk of developing type 2 diabetes has been launched by Iceland's Decode Genetics Inc.”
Ethical, Legal, and Social Issues

“Prognosis -- predicting cancer risk in the long term” (Apr 16) EurekAlert! reports, “Every day, people make assumptions, educated and not, about their risk for developing cancer.”

“Health disparities -- Genetics, society and race play an important role in access to healthcare” (Apr 15) EurekAlert! reports, “Minority individuals are much more likely to develop and die from cancer than the general U.S. population.”

Genetic counseling utilization by families with offspring affected by birth defects, Hawaii, 1986-2003
Forrester MB & Merz RD
Am J Med Genet A 2007 Apr

Applications and implications of advances in human genetics: perspectives from a group of Black Americans
Sheldon JP, et al.
Community Genet 2007;10(2):82-92

“Understanding personal genetic risk for familial breast cancer eases anxieties” (Apr 17) EurekAlert! reports, “Services that help women understand the way that their inherited genetic make-up influences their risk of getting breast cancer ease distress and decrease their levels of cancer worry.”

The ethics of CYP2D6 testing for patients considering tamoxifen
Hartman AR & Helft P
Breast Cancer Res 2007 Apr;9(2):103

The essential role of genetic counseling in inherited thrombophilia
Lochhead P & Miedzybrodzka Z
Semin Hematol 2007 Apr;44(2):126-9

Genomic Tools

“Genetic study centre is launched” (Apr 17) BBC News reports, “A centre to undertake the largest study into the genetic and environmental causes of disease has opened its doors in Manchester.”

Bayesian multi-loci association method: allowing for higher-order interaction in association studies
Albrechtsen A, et al. 
Genetics 2007 Apr

New saliva DNA collection method compared to buccal cell collection techniques for epidemiological studies 
Rogers NL, et al. 

A simple and improved correction for population stratification in case-control studies 
Epstein MP, et al. 
Am J Hum Genet 2007 May;80(5):921-30

Improving Power in Contrasting Linkage-Disequilibrium Patterns between Cases and Controls 
Wang T, et al. 
Am J Hum Genet 2007 May;80(5):911-20

Immune System (including Autoimmunity)

“Astrocytes may contribute to Lou Gehrig's disease” ⚠️
(Apr 16) News-Medical.Net reports, “Two papers by Columbia and Harvard researchers report for the first time that astrocytes (the most abundant non-neuronal cells in the central nervous system), which carry a mutated gene known to cause some cases of amyotrophic lateral sclerosis (ALS/Lou Gehrig's disease), induce motor neuron death.”

“Research team identifies additional genetic risk factors for Crohn's disease” ⚠️
(Apr 15) EurekAlert! reports, “An international research team – including investigators from Massachusetts General Hospital (MGH) and the Broad Institute of Harvard University and Massachusetts Institute of Technology – has identified several novel genetic variations associated with the risk of Crohn's disease.”

Infectious Disease

“First successful attempt to determine the structure of RfaH transcription factor” ⚠️
(Apr 15) News-Medical.Net reports, “The results of a new study suggest that bacteria that cause diseases like bubonic plague and serious gastric illness can turn the genes that make them infectious on or off.”

“Gene that governs toxin production in deadly mold found” ⚠️
(Apr 12) EurekAlert! reports, “For the growing number of people with diminished immune systems - cancer patients, transplant recipients, those with HIV/AIDS - infection by a ubiquitous mold known as Aspergillus fumigatus can be a death sentence.”
Mental Health (including Addiction)

The Genetics of Anorexia Nervosa
Bulik CM, et al.
Annu Rev Nutr 2007 Apr

Nervous System (including Movement Disorders)

“Astrocytes may contribute to Lou Gehrig's disease”
(Apr 16) News-Medical.Net reports, “Two papers by Columbia and Harvard researchers report for the first time that astrocytes (the most abundant non-neuronal cells in the central nervous system), which carry a mutated gene known to cause some cases of amyotrophic lateral sclerosis (ALS/Lou Gehrig's disease), induce motor neuron death.”

“Novel gene therapy hints at improvement”
(Apr 16) The Denver Post reports, “The first dozen Parkinson's patients to have holes drilled in their skulls for a novel gene therapy attempt weren't harmed -- and hints at some improvement have researchers embarking on a larger study to see if the treatment really may work.”

“Researchers discover gene crucial for nerve cell insulation”
(Apr 16) NIH News reports, “Researchers funded by the National Institutes of Health have discovered how a defect in a single master gene disrupts the process by which several genes interact to create myelin, a fatty coating that covers nerve cells and increases the speed and reliability of their electrical signals.”

Invited Commentary: When Bad Genes Look Good--APOE*E4, Cognitive Decline, and Diagnostic Thresholds
Glymour MM
Am J Epidemiol 2007 Apr

Obesity

“Study identifies clearest link yet to obesity risk”
(Apr 12) News-Medical.Net reports, “Scientists have identified the most clear genetic link yet to obesity in the general population as part of a major study of diseases funded by the Wellcome Trust, the UK's largest medical research charity.”
“Mysterious, widespread obesity gene found through diabetes study.”
(Apr 13) Science reports, “The role that obesity plays in diabetes, cancer, and other diseases makes our expanding waistlines one of today's most pressing health problems.”

Pharmacogenomics

“Genes and biomarkers that allow doctors to choose the right therapy for the right patient.”
(Apr 17) EurekAlert! reports, “Genetic and epigenetic variations ensure that no two people are exactly alike, and the same holds true for any two cancers.”

Cost-effectiveness of pharmacogenetic testing to predict treatment response to angiotensin-converting enzyme inhibitor
Costa-Scharplatz M, et al.
Pharmacogenet Genomics 2007 May;17(5):359-68

Prenatal and Perinatal Health

“Genetic cause for preterm births found.”
(Apr 16) applesforhealth.com reports, “Researchers reported in Reno, Nev., Friday that they've linked preterm births in Hispanic women to a genetic cause.”

Overview of epidemiology, genetics, birth defects, and chromosome abnormalities associated with CDH
Pober BR
Am J Med Genet C Semin Med Genet 2007 Apr

Genetic factors in congenital diaphragmatic hernia
Holder AM, et al.
Am J Hum Genet 2007 May;80(5):825-4

Reproductive System

“Gene mutation leads to male infertility.”
(Apr 16) Science Daily reports, “French medical scientists have identified a mutation in a gene called AURKC (Aurora Kinase C) in 14 infertile men of North African descent.”

Respiratory System

Progress in the genetics of chronic obstructive pulmonary disease
Keicho N & Matsushita I
Nippon Rinsho 2007 Apr;65(4):611-4

Identification of individuals with alpha-1-antitrypsin deficiency by a targeted screening program
Bals R, et al.
Respir Med 2007 Apr

Let’s Go Surfing

The Minnesota Gene Pool Weblog