C-B3-02:

Association of FTO, INSIG2, MC4R, and PCSK1 Obesity SNPs With Binge Eating in Morbidly Obese Patients

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Background/Aims: Obesity has a strong genetic component. Recent genome-wide association studies have identified single nucleotide polymorphisms (SNPs) in or near over a dozen genes that are related to body mass index (BMI). Despite the association of these SNPs with BMI, the mechanism by which they influence the determination of body weight is not yet known. Recently, the fat-mass and obesity-associated (FTO) obesity SNP was related to energy intake and preference for foods of high caloric density in children. FTO genotype was not associated with resting energy expenditure. We have extended this type of analysis to eating behaviors in the morbidly obese. **Methods:** DNA was obtained from approximately 900 morbidly obese (BMI > 40 kg/m²) patients and used to genotype obesity SNPs in or near the FTO, INSIG2, MC4R, and PCSK1 genes. Binge eating status (normal, episodic overeating, or any binge eating) was determined using the validated Questionnaire on Eating and Weight Patterns (QEWP). Binge eating status was correlated with each individual genotype, the combined obesity allele burden, and the combined homozygous obesity gene burden. **Results:** Binge eating data was obtained from 640 patients who had completed the QEWP. Of these 640, 116 (18%) were classified as manifesting binge eating behavior. No association was present between heterozygous or homozygous FTO ($P=0.59$), MC4R ($P=0.30$), or PSK1 ($P=0.77$) obesity SNPs. However, 29% of those who were homozygous for the INSIG2 obesity SNP were classified as binge eaters, versus 17% of heterozygous or homozygous normal patients ($P=0.006$). Association was also found with binge eating status and the presence of 2 or more homozygous obesity genotypes (28% versus 17%, $P=0.041$), likely due to the INSIG2 gene. Cumulative obesity allele burden (0–8 alleles for the 4 genes) was not associated with binge eating status ($P=0.42$). **Conclusions:** The INSIG2 obesity SNP appears to influence binge eating behavior in morbidly obese adults. The FTO obesity SNP appears to influence eating behavior in children suggesting that different genes may influence obesity at different ages. For both genes, excess caloric intake appears to be the major mechanism influencing BMI. How other obesity genes influence body weight regulation has not yet been determined.

C-B3-03:

Association of Epidemiologic and Genetic Factors With Abdominal Aortic Aneurysm (AAA)

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Background: Abdominal Aortic Aneurysm (AAA) rupture is the thirteenth leading cause of death in the U.S. and a major cause of death in the elderly. Since most AAAs are asymptomatic, mortality could be greatly reduced by determining which patients are at risk, then screening those patients prior to a rupture. Our aim was to determine which clinical and genetic risk factors are associated with AAA. **Methods:** This was a case-control study of patients from the Geisinger Vascular Clinic and the Geisinger MyCode Project (a biobanking project of Geisinger Clinic patients who volunteered to participate in genetic research projects). Clinical and environmental risk factor data was obtained from patient electronic medical records (EMR) and analyzed by multivariate logistic regression. In a subset of participants, we also analyzed three promising genetic polymorphisms, two from a prior genome-wide association study in this population (rs12039875, 1q41; rs7635818, 3p12.3) and one from the recent literature (rs10757278, 9p21). The genetic data was combined with the clinical data using multivariate logistic regression. **Results:** In the clinical analysis the number of Vascular Clinic and MyCode AAA cases totaled 722, with 11,761 controls. Adjusted

OR showed a significant AAA risk for age, gender, smoking, intermittent claudication and peripheral artery disease, while body mass index (BMI) and diabetes were surprisingly protective. The genetic analysis consisted of 502 AAA cases and 295 controls from the Vascular Clinic. The GC genotype of rs7635818 showed an increased risk, but the rs12039875 AA genotype showed a significant protective effect when controlling for the clinical variables. This SNP is located in KCNK2, a gene in the potassium channel protein family. Although this gene has not previously been linked with AAA, our laboratory showed that this gene is expressed in vascular tissue. **Conclusions:** Our study indicates a significant elevated risk of AAA for individuals who are older, male, ever smoked, have peripheral artery disease and a GC genotype of rs7635818. Higher BMI, diabetes and the AA genotype of rs12039875 in the KCNK2 gene significantly lower AAA risk. This is one of the first studies to utilize the MyCode population in a research study and establish this population source as a valuable and convenient asset of Geisinger Health System for clinical research.

C-B3-04:

Media Analysis of Genetic Testing Information for Common Disease Presented to the Public

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Background/Aims: With the advent of personalized medicine, consumers are routinely exposed to information and direct-to-consumer (DTC) advertising of various genetic tests. Specifically, 2007–2008 saw a return of the DTC advertisement for BRACAnalysis® in the eastern US, and the advent of three new companies offering DTC genetic screening for under \$1000. This abstract presents the results of a media analysis for information presented to the general public on genetic testing for common diseases, such as cancer or heart disease, or for full genome scans for disease risk in general. **Methods:** Lexis Nexis search was conducted using the broad term genetic testing. The search was limited to all major world publications, news wire services, TV and radio transcripts, blogs, and web publications between the dates of September 1, 2007 and September 30, 2008. The purpose was to assess information presented to the lay public; therefore, scientific literature was not searched. These media reports were examined for number of reports, report type (print, television, online), target audience, polarity of information (positive for genetic testing or negative against genetic testing), and test type. **Results:** One-thousand articles were retrieved from the initial search. Immediately excluded were articles about ancestry testing, diagnostic testing, psychiatric testing, food safety, forensic testing, cloning, and pre-marital genetic testing due to ethnicity or consanguinity. Additional exclusions were made for all areas surrounding prenatal genetic testing, including pre-implantation diagnosis and paternity testing. Stock announcements, hearing testimonies, and reports on non-U.S. companies or from non-U.S. sources were also excluded. This resulted in a total of 173 media reports over the one year time period to which the lay public was exposed. These 173 reports were analyzed further for specific information presented. **Conclusions:** Over a one year period, consumers are routinely exposed to information about genetics through the media. These reports span all methods of media delivery and vary in polarity.

PS2-08:

Development and Implementation of a Genetic Fingerprinting Assay for the Personalized Medicine Research Project

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Background: For any biorepository, it is necessary to develop measures that determine sample quality and ensure each sample can be correctly identified. One method of devising this type of quality control measure is to rely on DNA polymorphism panels developed for forensic applications. Although valid for identification, these panels are not useful for other purposes, such as medical research. **Aims:** We developed an identification panel for the Personalized Medicine Research Project (PMRP) that uses medically relevant polymorphisms. This panel not only uniquely identifies samples and tests for sample quality but can also be used by investigators for candidate gene studies. **Methods:** Polymorphism candidates were taken from investigator