Genomics and Public Health Prior to the CDC Effort: The Linkage of Genetic Measurements to a Health Risk Assessment

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Background and objectives: Health Risk Assessments (HRA) or Health Hazard Appraisals, which they have also been called, have been used in the public health setting to identify and motivate behavioral change by providing users with estimates of their risk and/or relative risk of disease, injury or death from specific causes based on their characteristics, with recommendations for reducing the risk. In the late 1980's the investigators translated genomic discoveries into a public health program by developing an HRA which 1) accepted information about an individual's genetic risk status, 2) provided risk estimates based on published genetic risk information, and 3) made individual-specific recommendations including possible genetic testing. Because family history information is often necessary to stratify individuals with respect to the utility of including genetic measurements, it was necessary to develop and validate a family history collection instrument. Once the HRA was developed, it was necessary to evaluate the utility of the HRA to motivate behavioral compliance and the ability of clinicians to understand the output.

Our objectives of our program were: 1) To demonstrate that an HRA including input about genetic risk status is feasible; 2) To evaluate the validity of subject's knowledge of their family-history of specific diseases where family history and/or specific alleles have been demonstrated to add clinically significant information; and 3) To evaluate the effectiveness of the HRA in motivating behavioral change.

Methods:

1) For the evaluation of the validity of self-administered family history information, sensitivity and specificity of the subjects' family history information was compared to clinically - derived diagnoses with respect to coronary heart disease (CHD), hypertension (HT), diabetes (DM) and arthritis; and

2) For the evaluation of the effectiveness of the HRA, subjects who were told they were at increased risk because of their family history or allele presence were compared with those who were not provided this information

Results:

1. The sensitivity of detecting CHD, HT and DM in relatives (N=405) of cases with CHD was 79%, 70% and 89% respectively, and 67%, 58%, and 83% in relatives of unaffected controls (N=298). The specificity was 99%, 97% and 99% in relatives of cases, respectively and 98%, 95% and 99% in relatives of controls.

2. Those who were told they were at increased risk because of their family history or genetic status were significantly ($p<0.05$) more likely to comply with several preventive recommendations.

Discussion/Conclusion: The validity of family history of disease information depends of the disease being studied. It was very good for family history of CHD and DM and poor for hypertension and the arthritities. The provision of genetic risk information might lead to increased frequency of behavioral compliance in those who are at increased risk. The issues which surround the use of genomics information in the public heath setting today are similar to those we faced and still need further exploration.