



# GENETICS BRIEF

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## Genes and the Environment

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Disease can be broadly defined as a complex trait that causes ill health and that involves the interaction of genes and the environment. Most diseases are thought to be the result of interactions between several low-penetrance<sup>1</sup> genes and environmental factors. Thus, most diseases are likely to be a combination of nature and nurture. There are a few diseases caused by genetic variation that will develop regardless of environmental factors. Even in these cases, environmental factors may affect the age of onset and severity of the disease.

Definitions of environmental and non-environmental factors vary. Generally, environmental factors can include other genes, infectious, chemical, and physical agents, and nutritional habits. In this brief, environmental factors refers to elements one may be exposed to, not behavioral factors such as tobacco use, alcohol consumption, and nutritional habits.

### Diseases, Genes and the Environment

According to the National Institute for Environmental Health Sciences' Environmental Genome Project (EGP), "environmentally associated diseases or dysfunctions" cover a wide spectrum of disease, including cancer, pulmonary diseases, neurodegenerative disorders, developmental disorders, birth defects, reproductive function, and autoimmune disease (EGP Web site, 2002). Certain genes are more likely than others to be "environmentally responsive." Thus far, the EGP has identified 554 potentially "environmentally responsive genes." For example, the GSTP1 gene is involved in the detoxification of carcinogens. Researchers are studying whether certain variations in the GSTP1 gene are associated with increased risk of prostate cancer, due to a reduced ability to detoxify environmental carcinogens. Variations in the gene encoding cytochrome P450, a metabolizing enzyme, are being studied in relation to various environmental exposures and diseases, including tobacco smoke exposure and risk of lung cancer.

### The Promise of Gene-Environment Research

Gene-environment research is still a relatively new field but promises to have many beneficial results. Genes that have evaluative and predictive potential for use in screening and diagnostics could be identified (Olden, et al., 2001). This research could be used to target public health strategies that help individuals avoid or reduce adverse exposures and to design better treatment regimes, including drug and gene therapy. According to Olden and colleagues, "the biotechnology revolution has opened new opportunities for addressing current inadequacies in decision making regarding environmental health (2001)." Regulation of environmental and occupational health standards currently relies on limited information regarding effects of various environmental exposures, usually based on research in animals and extrapolation to humans. Knowledge of the role of individual susceptibility and response to environmental exposures that may result in the onset of disease, may assist in refining these methods of regulation.

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<sup>1</sup> Penetrance: The degree to which having a genetic variation will result in disease, i.e., an individual with a low-penetrance disease gene has a lower chance of developing the disease based on that gene alone, whereas an individual with a high-penetrance disease gene has a higher probability of developing that disease.

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## Current Research Efforts

Most gene-environment research is currently taking place at the federal level. New York is an exception. The New York State Department of Health conducts the Cancer Surveillance Improvement Initiative, which includes a cancer registry that allows for the analysis of cancer incidence and its relationship to environmental and other factors. The Department also has an extensive gene-environment laboratory research component.

**National Institutes of Health, National Institute for Environmental Health Sciences, Environmental Genome Project.** Established in 1998, the EGP is a “long-term effort to characterize specific genetic variations or polymorphisms that contribute either to resistance or susceptibility to environmentally induced diseases (Environmental Health Perspectives, April 1999).” The goal of the EGP is to catalog information about human genetic variation and apply this information to understanding disease susceptibility and individual responses to environmental exposures. The goals are being carried out primarily through funding of extramural research, as well as investigator-initiated and intramural research. The types of research being conducted include discovery of genetic variations among humans; the study of the functions of these variations in producing disease; and population-based epidemiological studies, including, Parkinson’s disease, susceptibility to pesticides, and prostate cancer. The EGP also funds technology development to support this research. It is anticipated that the project will result in the development of improved risk assessments and regulations for environmental exposures, better protections for susceptible subgroups, and targeted disease screening for high-risk groups.

**National Institutes of Health, National Cancer Institute (NCI).** Research priorities of the NCI include understanding the interplay between inherited susceptibility to cancer and environmental risk factors. Research projects address a variety of issues, including identifying and characterizing gene variations; large population studies on gene-environment interactions; and family registries, including the Cooperative Family Registries for breast/ovarian and colorectal cancers. For example, the Cancer Family Registry collects data on cancer family history, demographics, environmental and lifestyle risk factors, as well as biological specimens for over 8,000 families at high-risk for breast and ovarian cancer, allowing for the study of genetic and environmental factors in the development of these cancers.

**Centers for Disease Control and Prevention, National Center for Environmental Health (NCEH), Office of Genomics and Disease Prevention (OGDP).** The mission of OGDP is to integrate advances in human genetics into public health research, policy, and programs, including clinical and epidemiological studies of gene-environment interactions in disease causation. For example, OGDP has established three Centers for Genomics and Public Health; each center will develop a regional hub of expertise to use information about gene-environment interactions associated with disease to develop new strategies for improving health. OGDP also funded a study of gene-environment interactions in cardiovascular disease to evaluate genetic polymorphisms related to lipid metabolism, obesity and blood pressure.

## Policy Issues

Genetic variation and individuals’ responses to environmental factors will present an opportunity and a challenge to policymakers. This research will provide insight into the role of individual susceptibility and the complexity of disease risk. It also will lead to questions concerning what “allowable” levels of exposure are, and how best to protect “sensitive” populations.

## Selected References and Resources

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