

The Office of
GENETICS
and Disease Prevention

Gene-Environment Interaction Fact Sheet

August 2000

Information from the Human Genome Project has caused scientists to re-examine the role of genetics and other risk factors involved in the development of disease. Understanding this complex interplay of genes and environment will lead us to new methods of disease detection and prevention.

Gene Environment Interaction



Virtually all-human diseases result from the interaction of genetic susceptibility factors and modifiable environmental factors, broadly defined to include infectious, chemical, physical, nutritional, and behavioral factors.

This is perhaps the most important fact in understanding the role of genetics and environment in the development of disease. Many people tend to classify the cause of disease as either genetic or environmental. Indeed, some rare diseases, such as Huntington or Tay Sachs disease, may be the result of a deficiency of a single gene product, but these diseases represent a very small proportion of all human disease. Common diseases, such as diabetes or cancer, are a result of the complex interplay of genetic and environmental factors.

Genetic Variations

Variations in genetic makeup are associated with almost all disease.

Even so-called single-gene disorders actually develop from the interaction of both genetic and environmental factors. For example, phenylketonuria (PKU) results from a genetic variant that leads to deficient metabolism of the amino acid phenylalanine; in the presence of normal protein intake, phenylalanine accumulates and is neurotoxic. PKU occurs only when both the genetic variant (phenylalanine hydroxylase deficiency) and the environmental exposure (dietary phenylalanine) are present.

Environmental Factors

Genetic variations do not cause disease but rather influence a person's susceptibility to environmental factors.

We do not inherit a disease state per se. Instead, we inherit a set of susceptibility factors to certain effects of environmental factors and therefore inherit a higher risk for certain diseases. This concept also explains why individuals are differently affected by the same environmental factors. For example, some health conscious individuals with "acceptable" cholesterol levels suffer myocardial infarction at age 40. Others individuals seem immune to heart disease in spite of smoking, poor diet, and obesity. Genetic variations account, at least in part, for this difference in response to the same environmental factors.

Intervention Strategies

Genetic information can be used to target interventions.

We all carry genetic variants that increase our susceptibility to some diseases. By identifying and characterizing gene-environment interactions, we have more opportunities to effectively target intervention strategies. Many of the genetic risk factors for diseases have not been identified, and the complex interaction of genes with other genes and genes with environmental factors is not yet understood. Clinical and epidemiological studies are necessary to further describe these factors and their interactions. However, as our understanding of genetic variations increases, so should our knowledge of environmental factors, so that ultimately, genetic information can be used to plan appropriate intervention strategies for high-risk individuals.

Contact

**Centers for Disease Control and Prevention
Office of Genetics and Disease Prevention**

1600 Clifton Road Mail Stop # K-28
Atlanta, Georgia 30333

770-488-3235

770 488-3236

<http://www.cdc.gov/genetics/>