National Cancer Advisory Board

Genome-wide Association Studies: Introduction

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Categories of Cancer Causation

Environment

Genes

- Spontaneous

+ Hereditary syndromes
  Low-penetrant variants

- Microbes – 1960s
  Chemicals – 1970s
  Lifestyle – 1980s

+ Interactions
The Advent of GWAS at NCI

- A shift from candidate gene searches to systematic genome-wide scans (CGEMS)
- Large-scale association studies with staged replication strategies through intramural/extramural consortia with DCCPS
- Resequencing and fine-mapping of genomic loci to narrow the search for inherited genetic variants
- Downstream research with CCR: Functional studies, germline vs. somatic changes, pharmacogenomics, gene-environment interactions, risk prediction models, clinical applications
- Rapid dissemination of publicly available data (caBIG)
NCI Consortium of Cohorts

37 Population Cohorts with Nearly 4 Million Individuals (December 2008)
“There have been few, if any, similar bursts of discovery in the history of medical research...”