INSTITUTE FOR GENES AND ENVIRONMENT AT NORTHWESTERN MEDICINE

PERSONALIZED MEDICINE
INSTITUTE FOR GENES AND ENVIRONMENT AT NORTHWESTERN MEDICINE

The concept of Personalized Medicine has emerged from a growing appreciation of individual differences in our genes and environment. Through the new Institute for Genes and Environment at Northwestern Medicine, we are exploring and influencing this exciting and rapidly evolving frontier of study and practice.

Personalized Medicine holds the potential for healthcare that can be tailored to each individual based on his or her specific genes and environmental contacts. For each person, medical decisions, practices, and products can be customized based on a variety of observations and measurements. The goal is to improve and personalize care. At Northwestern, we are using the following 21st century observations and measurements to enhance our understanding of diseases and trailblaze a path to personalized medicine.

Deep Phenotyping

Most diseases are not due to a single cause—they are multifactorial and actually are syndromes. What is a syndrome? A syndrome is the association of specific recognizable features of diseases. These features can be signs that are observed by the physician and/or symptoms that are reported by the patient.

Diabetes is associated with high blood sugar and urine sugar content. Cancer is associated with tumor masses, but there may be many causes. The phenotype consists of observable traits associated with disease. Therefore, the healthcare provider can clinically and technically (laboratory tests) subdivide disease based upon observations and measurements. Such analysis can distinguish diseases and make them more easily studied and/or amenable to prevention, diagnosis, and/or treatment.

Environmental Measurements

We have known for some time that environmental factors impact or even cause disease. The associations of cigarette smoking with lung cancer and alcohol consumption with liver disease are obvious examples, but it is now clear that many environmental factors may contribute to a variety of diseases. Measuring these factors could impact our understanding of diseases such as asthma, autism, cancer, and heart disease.

DNA Sequencing

Sequencing an individual’s DNA can identify variations associated with or even directly causing disease. Inborn errors may be the direct result of a mutation in a person’s DNA such as in sickle cell anemia or cystic fibrosis. Other entities may be multifactorial where a combination of mutations or variations causes disease. Sequencing an individual’s DNA also contributes to our understanding of disease.

“We now know that environmental factors impact gene expression and cellular function. Diseases as diverse as cancer, heart disease, neurologic diseases, asthma, and diabetes result from an interplay between our inherited genes and environmentally-mediated changes in gene function.”

Andrea Dunaif, MD
Charles F. Kettering Professor of Endocrinology and Metabolism
Epigenetics
The expression of genes can be modified by environmental and other factors that impact how and when the genes produce their protein products. Measuring these marks on the DNA also contributes to our understanding of disease.

Gene Expression
Genes are transcribed into proteins, which are the building blocks of cells and life. New techniques are enabling the direct measurement of gene expression and the potential to associate such findings with disease occurrence and outcomes.

Protein Expression
Similar to gene expression, the expression of proteins is measurable and associated with disease.

Protein Modification
Proteins are further modified in a variety of ways that can be measured. Various metabolites, sugars, and lipids are measured and associated with disease. These new fields have been denoted “metabolomics, glycomics, and lipidomics.”

Big Data Interpretation
All of this information has to be collated and correlated in ways much too complex for the human brain. Computer systems and new information technologies permit complex computation and an association of the variety of observations and measurements outlined above.

Integration (systems biology, pharmacogenetics/genomics)
The data can then be integrated to understand disease causes and associations. Systems biology involves the complex computations of such interactions. In the specific areas of drug development and pharmacology, experts can measure the impact of gene sequence and expression on how an individual will respond to a particular therapy. These fields have been denoted pharmacogenetics (gene sequence) and pharmacogenomics (gene expression). Common drugs, such as blood thinners, work very differently in various individuals based upon these individual variations.

Diagnosis, Treatment, Drug Discovery, Screening, Prevention
Lastly, all of this information is analyzed in the context of human disease with a goal to develop new approaches to diagnosis, treatment, drug discovery, screening procedures, and prevention of disease.

LOOKING TO THE FUTURE
At Northwestern, our ultimate goal is to bundle all of this rich clinical and technical information and then endeavor to understand the specifics of how our individual genes, groups of genes, and environmental factors work together to determine the human condition. This ground-breaking work requires a thorough understanding of how the aforementioned information relates and interacts as a complex system. To ultimately develop personalized medicine—individualized diagnostics, treatments, and preventive approaches to human health and disease—we will use mathematical models and computer science tools in concert with sophisticated clinical and medical insights. This all will be accomplished through our breakthrough Institute for Genes and Environment.
THROUGH NORTHWESTERN MEDICINE, WE INTEND TO CREATE A NATIONAL EPICENTER FOR HEALTHCARE, EDUCATION, RESEARCH, COMMUNITY SERVICE, AND ADVOCACY.

NORTHWESTERN MEDICINE

Northwestern Memorial HealthCare and Northwestern University Feinberg School of Medicine are seeking to impact the health of humankind through Northwestern Medicine. We aspire to be the destinations of choice for people seeking quality healthcare; for those who provide, support, and advance that care through leading-edge treatments and breakthrough discoveries; and for people who share our passion for educating future physicians and scientists. Our commitment to transform healthcare and to be among the nation’s top academic medical centers will be accomplished through innovation and excellence.

Through our world-class Institute for Genes and Environment at Northwestern Medicine, we are seizing the opportunity to pursue Personalized Medicine and fully explore its potential to improve and save lives. As physicians and scientists, we now know that differences in gene sequence, gene expression, and environmental factors impact whether a person develops an illness or not. The genes each one of us expresses may determine vulnerabilities, susceptibilities, or our resistance to disease. As important, the environment in which we live impacts gene expression and function and, hence, our ultimate health and/or disease.

Through our experts at the Institute for Genes and Environment, we will boldly move forward and use these revolutionary insights about our genes and environment to improve the care experience for each patient.