PERSONALIZED MEDICINE
The tailoring of medical treatment to the individual characteristics of each patient. It does not literally mean the creation of drugs or medical devices that are unique to a patient, but rather the ability to classify individuals into subpopulations that differ in their susceptibility to a particular disease or their response to a specific treatment.¹

GENOMICS
The study of all of a person’s genes, including interactions of those genes with each other and the person’s environment.²

GENOME
A branch of biology that deals with the heredity and variation of organisms. Today, the term genetics is often used to refer to the study of single genes, one at a time, as snapshots.

Targeting Medicine for the Individual
New technologies are changing the way we think about and treat a wide range of diseases, including cancer. Scientists’ ability to map the human genome has dramatically improved our understanding of genes and how they vary from person to person.

With a patient’s unique genetic profile in hand, physicians will be better equipped to select drugs and treatment protocols that lead to more successful outcomes, with fewer harmful and debilitating side effects along the way. A genetic profile can also help physicians determine how susceptible a person is to a certain disease and establish a plan for monitoring the individual’s health and preventing the disease.

These exciting developments ultimately will end the one-size-fits-all approach to treating disease by allowing physicians to make clinical decisions on a patient-by-patient basis. Genomics and proteomics are two of the emerging fields that will help make this dramatic shift toward personalized medicine possible. Genomics provides information on genetic variations and the level of gene expression, while proteomics looks at proteins expressed by cells. Both fields rely on donated human tissue—including blood, urine, bone marrow, lymph nodes and fluid—to provide clinicians with the information they need to diagnose cancer and make appropriate treatment decisions.

The Potential of Proteomics
Many scientists believe that finding molecular indicators—or “biomarkers”—of cancer will yield the greatest promise for early detection and treatment. If you think of a gene as a cell’s recipe, the proteins encoded in that gene are the key ingredient that drives both health and disease. Proteomics seeks to understand everything about that key ingredient.

Researchers have learned that cancer tumors “leak” proteins into blood, urine and other bodily fluids. This discovery signals the possibility of diagnosing cancer at an early stage by testing these fluids for the presence of cancer-related biomarkers.

Certain blood proteins are already being used as cancer biomarkers. For example, elevated levels of prostate specific antigen (PSA) suggest the presence of prostate cancer, while elevated levels of cancer antigen 125 (CA-125) indicate recurrent ovarian cancer. Current research studies are looking at ways to measure multiple biomarkers simultaneously to identify patterns—known as protein signatures—that can lead to improved cancer diagnostics.

1 President’s Council of Advisors on Science and Technology, “Priorities for Personalized Medicine,” September 2008
2 National Human Genome Research Institute, National Institutes of Health
3 National Cancer Institute
4 NCI Clinical Proteomics Technologies for Cancer
Early diagnosis, of course, leads to better outcomes, regardless of the treatment approach. In the future, biomarkers found in bodily fluids might also be used to monitor how cancer responds during treatment or detect the recurrence of tumors at a later date.

**Genomics 101**

Genomics is the study of a person’s genes and how they interact with each other and the individual’s environment. This scientific study is applied to complex diseases such as cancer, heart disease, diabetes and asthma because they are caused by a combination of genetic and environmental factors. Researchers in this field seek to understand how multiple genes work in concert to perform a specific function.

Genomics can be likened to studying a whole bird to understand how it flies rather than focusing solely on its wing. Flying is the result of more than just a pair of wings flapping up and down; the complex act of flight requires many of the bird’s body parts to work together. We can learn a lot by studying a wing, but that alone cannot explain how the bird can fly. Similarly, we can learn a great deal by studying individual human genes, but we need to learn how multiple genes work together to understand how a human body functions.

National Institutes of Health statistics show that genomics plays a role in nine of the 10 leading causes of death in the United States, including heart disease, cancer and diabetes. According to the National Human Genome Research Institute, all human beings are 99.9% identical in their genetic makeup; the differences in the remaining 0.1% hold vital clues about what causes disease. The more we understand about the interactions between genes and the environment, the better equipped we will be to improve health and prevent disease.

Within the field of genomics there are a variety of specialties; one of particular importance to understanding and treating disease is called pharmacogenomics. This research area focuses on learning how variations in genes affect the ways people respond to drugs. (For example, studying the connection between genes and drug responses may help explain why some people experience severe side effects from a particular drug while others have no reaction at all.) The goal is to create drug therapy plans that identify the best type and dose of drugs based on an individual’s genetic makeup.

**The Cancer Genome Atlas (TCGA)**

TCGA is a joint effort of the National Cancer Institute and the National Human Genome Research Institute. The project is designed to identify all of the changes found in the DNA of cancer cells to better understand different forms of cancer and find ways to fight these diseases. It often takes several different DNA changes to result in cancer, so having a complete catalog—or “atlas”—will boost our understanding of how cancer manifests and how to treat its variations.

**Genomics vs. Genetics**

Genetics is the study of isolated genes and heredity; genomics is the study of an organism’s complete genetic material, including genes and their functions. The science of genomics would not exist without the science of genetics. To understand how genes function in relation to one another (the whole), we first needed to understand single genes (the parts). If we draw
an analogy between biology and music, a gene is a violin solo, and the genome is an entire symphony.

Geneticists study single genes and the roles they play in inheritance. Genes direct the activities of cells and how the body functions—they influence a person’s health and the development of disease. While genetics determines much of a person’s physical appearance and well being, environmental factors also play a role.

Genetic tests analyze human DNA, RNA or protein to detect variations in genes associated with a specific disease or condition. These tests are used to:
- Confirm a suspected diagnosis
- Predict the likelihood of future illness
- Detect whether a person is a carrier of a particular illness
- Predict response to therapy.

By contrast, genomicists study unique patterns of RNA from a tumor or tissue sample—often called a genomic profile—and compare them to those found in normal, healthy tissue. These profiles can also be used to study differences in tumors from patients with the same type of cancer. The goal of genomic research is to determine whether a genomic profile can be used to guide treatment decisions for individual patients based on their tumor’s unique pattern of gene expression.

**Genomics In Action**

Since nearly all human diseases and disorders have some basis in the genes—and many involve complex sets of genes—genomics is poised to have an enormous impact on all areas of medicine, particularly in biomedical research and the delivery of health care. In fact, the influence of genomics is already evident today.

**Understand disease.** Many of our most common and deadly diseases, including many types of cancer, high blood pressure, heart disease and diabetes, are now known to involve more than a single gene. Genomics is making it possible to investigate how sets of genes interact with one another and how they react to our behavior and environment to cause these diseases.

**Understand genetic differences.** Advances in genomics are helping scientists understand the genetic differences between people, which may lead to an understanding of why some people are more susceptible to certain diseases than others and why some respond better than others to different therapies.

**Develop drugs and biologic therapies.** Many current chemotherapies kill all cells—good ones as well as cancer cells—that grow and multiply rapidly. A patient’s body loses some important biological functions when some of those good cells die, and the result can lead to serious side effects. Genomics is helping us understand the basis for many diseases so that drugs and biologic therapies can be developed to target only diseased cells.

**Develop gene therapies.** Eventually, genomics may lead to the ability to fix or replace faulty genes directly—a process known as gene therapy. A great deal of research is currently underway in this area, although no gene therapies have yet been approved for use in the United States.

**Health Care Delivery**

Genomics will help us identify which disease we have, forecast the course of the disease, determine the best course of treatment and establish how much vigilance is needed to monitor the disease. This field of research will also make it possible to reduce our risk of disease and prevent us from...
gettingsick in the first place. Genomics is already affecting the practice of health care by helping us:

**Diagnose disease.** Genomic technologies are being used to help diagnose disease and subtypes of disease, and more tests are becoming available every year. Today, certain subtypes of cancers (e.g., colon and skin) can be diagnosed based on the results of genetic testing.

For example, genetic abnormalities can lead to a type of colon cancer that is called hereditary nonpolyposis colorectal cancer (HNPCC). About 90% of the patients with HNPCC have one of the two most common genetic abnormalities known to cause this syndrome. Individuals with these abnormalities have an 80% lifetime risk for developing colon cancer, and their children are also at significantly higher risk; however, fewer than 3% of colon cancer diagnoses are HNPCC-related. Advances in genomic technologies now allow us to distinguish HNPCC from other forms of colon cancer.

**Determine prognosis and treatment approach.** Doctors can already determine the aggressiveness of certain types of cancers based on genetic testing. As researchers continue to study large groups of people with slight differences in their genomes, they will be able to relate those differences to disease outcomes. Genetic assessments will then be developed to help predict the course an individual’s disease will take with treatment using a specific drug, based on that person’s genetic pattern.

One example of such a test already in use is Oncotype DX*—a test that examines a breast cancer patient’s tumor tissue at a molecular level, and gives information about her individual disease. This information can help tailor treatment for her breast cancer. Oncotype DX is the first and only gene expression test that has been accepted as demonstrating the ability to predict a patient’s benefit from chemotherapy as well as her risk of recurrence.

Other genetic assessments may show that two people who are diagnosed with lung cancer have very different prognoses—the disease may be more aggressive in one person than the other. As a result, they may receive different treatments. The genetic assessments may also predict that the two people will respond best to different drugs.

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*Oncotype DX is a product of Genomic Health, Inc.

**Reduce the risk of disease.** Genomics may help the practice of medicine shift from mostly reactive to more proactive. Rather than seeking treatment when something goes wrong with our health, genomics may help reduce our risk and prevent us from becoming sick. Genetic testing can determine whether we are at increased risk of certain diseases before we become ill. By knowing our risks in advance, we can change certain behaviors to help reduce that risk, such as increased cancer screening, preventive drugs or other therapy, and lifestyle modifications (e.g., quitting smoking, reducing sun exposure, eating a healthier diet).

**Treat disease more effectively.**

Through pharmacogenomics—the study of how genomics influences an individual’s response to drugs—we will be able to treat diseases by identifying their subtypes. Certain drugs may be highly effective in attacking some subtypes but less effective against others. Knowing which subtype a person has will help determine the best course of treatment. Genetic assessments will also help determine which drug and dose is best for an individual, based on the person’s ability to process the drug, which will likely reduce our chances of experiencing serious side effects.

**Monitor disease.** Genetic assessments may also be useful in the surveillance of cancer and other diseases. For example, individuals who possess a gene or genes that put them at increased risk for colorectal cancer can schedule more frequent colonoscopies. If polyps—which can develop into cancers—are detected, they can be removed to prevent cancer from developing. If a malignancy develops between colonoscopies, cancers found and removed early are highly curable.

In the future, after a successful treatment regimen, physicians may be able to use genetic assessments to determine whether a patient’s cancer-related genes have been turned on or off, which could indicate imminent recurrence of cancer or continued remission.

Genomics will help us diagnose diseases, predict the courses they will take, determine the most effective treatments, and monitor their status. This field of study will also help us determine our risk of disease so we can take preventive measures that can keep us healthy.
What You Can Do
Genomics and proteomics have already yielded some profound benefits, and their promise for advancing personalized medicine continues to grow. If you have cancer, you can help accelerate these benefits by donating your tissue.

Tissue is critical to the accurate diagnosis and staging of your cancer. It helps you and your doctor make sound treatment decisions that give you the best chance of surviving and preventing recurrence. But your tissue can also contribute to scientific discoveries that will prevent cancer and aid in the diagnosis and treatment for other cancer patients in the future.

For more information about the benefits of tissue donation and things you should consider before donating, go to [www.researchadvocacy.org](http://www.researchadvocacy.org) and enter tissue donation in the search box.

Additional Resources
Personalized Medicine Coalition – [www.personalizedmedicinecoalition.org](http://www.personalizedmedicinecoalition.org)
National Human Genome Research Institute – [www.genome.gov](http://www.genome.gov)

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Research Advocacy Network is committed to improving patient care through research. Our goals are to get results of research studies (new treatments) to patients more quickly, to give those touched by the disease an opportunity to give back and to help the medical community improve the design of its research to be more attractive to potential participants. Because research holds the hope for improvements in treatment, diagnostics and prevention, we are dedicated to patient focused research. We believe dissemination of research results to the medical community and patients can have a major impact on clinical practice.

The Research Advocacy Network (RAN) is a not for profit (501 c 3 tax exempt) organization that was formed in 2003 to bring together participants in the research process with the focus on educating, supporting, and connecting patient advocates with the medical research community. While there are many organizations addressing the needs of patients with specific diseases, political advocacy, cancer education and fundraising, no organization has focused on advancing research through advocacy.

RAN works with advocates and organizations to effectively integrate advocates into research activities. Please learn more about us at our Web site at [www.researchadvocacy.org](http://www.researchadvocacy.org) or contact us about our work by e-mailing us at info@researchadvocacy.org or by phone or FAX at 877-276-2187.

We look forward to hearing from you!