Genetics & Genomic Science – FAQ

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What are genetics and genomics?

Genetics is a term that refers to the study of genes and their roles in inheritance - in other words, the way that certain traits or conditions are passed down from one generation to another. Genetics involves scientific studies of genes and their effects. Genes (units of heredity) carry the instructions for making proteins, which direct the activities of cells and functions of the body. Examples of genetic or inherited disorders include cystic fibrosis, Huntington's disease and phenylketonuria (PKU).

Genomics is a more recent term that describes the study of all of a person's genes (the genome), including interactions of those genes with each other and with the person's environment. Genomics includes the scientific study of complex diseases such as heart disease, asthma, diabetes, and cancer because these diseases are typically caused more by a combination of genetic and environmental factors than by individual genes. Genomics is offering new possibilities for therapies and treatments for some complex diseases, as well as new diagnostic methods.

Why are genetics and genomics important to my health?

Genetics and genomics both play roles in health and disease. Genetics helps individuals and families learn about how conditions such as sickle cell anemia and cystic fibrosis are inherited in families, what screening and testing options are available, and, for some genetic conditions, what treatments are available.

Genomics is helping researchers discover why some people get sick from certain infections, environmental factors, and behaviors, while others do not. For example, there are some people who exercise their whole lives, eat a healthy diet, have regular medical checkups, and die of a heart attack at age 40. There are also people who smoke, never exercise, eat unhealthy foods and live to be 100. Genomics may hold the key to understanding these differences.

Apart from accidents (such as falls, motor vehicle accidents or poisoning), genomic factors play a role in nine of the ten leading causes of death in the United States (for example, heart disease, cancer and diabetes. All human beings are 99.9 percent identical in their genetic makeup. Differences in the remaining 0.1 percent hold important clues about the causes of diseases. Gaining a better understanding of the interactions between genes and the environment by means of genomics is helping researchers find better ways to improve health and prevent disease, such as modifying

diet and exercise plans to prevent or delay the onset of type 2 diabetes in people who carry genetic predispositions to developing this disease.

Why are genetics and genomics important to my family's health?

Understanding more about diseases caused by a single gene (using genetics) and complex diseases caused by multiple genes and environmental factors (using genomics) can lead to earlier diagnoses, interventions, and targeted treatments. A person's health is influenced by his/her family history and shared environmental factors. This makes family history an important, personalized tool that can help identify many of the causative factors for conditions that also have a genetic component. The family history can serve as the cornerstone for learning about genetic and genomic conditions in a family, and for developing individualized approaches to disease prevention, intervention, and treatment.

What are some of the new genetic and genomic techniques and technologies?

Proteomics

The suffix "-ome" comes from the Greek for *all*, *every*, or *complete*. It was originally used in "genome," which refers to all the genes in a person or other organism. Due to the success of large-scale biology projects such as the sequencing of the human genome, the suffix "-ome" is now being used in other research contexts. Proteomics is an example. The DNA sequence of genes carries the instructions, or code, for building proteins. This DNA is transcribed into a related molecule, RNA, which is then translated into proteins. Proteomics, therefore, is a similar large-scale analysis of all the proteins in an organism, tissue type, or cell (called the proteome). Proteomics can be used to reveal specific, abnormal proteins that lead to diseases, such as certain forms of cancer.

Pharmacogenetics and Pharmacogenomics

The terms "pharmacogenetics" and "pharmacogenomics" are often used interchangeably in describing the intersection of pharmacology (the study of drugs, or pharmaceuticals) and genetic variability in determining an individual's response to particular drugs. The terms may be distinguished in the following way.

Pharmacogenetics is the field of study dealing with the variability of responses to medications due to variation in single genes. Pharmacogenetics takes into account a person's genetic information regarding specific drug receptors and how drugs are transported and metabolized by the body. The goal of pharmacogenetics is to create an individualized drug therapy that allows for the best choice and dose of drugs. One example is the breast cancer drug trastuzumab (Herceptin). This therapy works only for women whose tumors have a particular genetic profile that leads to overproduction of a protein called HER2.

Pharmacogenomics is similar to pharmacogenetics, except that it typically involves the search for variations in multiple genes that are associated with variability in drug response. Since pharmacogenomics is one of the large-scale "omic" technologies, it can examine the entirety of the genome, rather than just single genes. Pharmacogenomic studies may also examine genetic variation among large groups of people (populations), for example, in order to see how different drugs might affect different racial or ethnic groups.

Pharmacogenetic and pharmacogenomic studies are leading to drugs that can be tailor-made for individuals, and adapted to each person's particular genetic makeup. Although a person's environment, diet, age, lifestyle, and state of health can also influence that person's response to medicines, understanding an individual's genetic makeup is key to creating personalized drugs that work better and have fewer side effects than the one-size-fits-all drugs that are common today. For example, the U.S. Food and Drug Administration (FDA) recommends genetic testing before giving the chemotherapy drug mercaptopurine (Purinethol) to patients with acute lymphoblastic leukemia. Some people have a genetic variant that interferes with their ability to process this drug. This processing problem can cause severe side effects, unless the standard dose is adjusted according to the patient's genetic makeup.

Stem Cell Therapy

Stem cells have two important characteristics. First, stem cells are unspecialized cells that can develop into various specialized body cells. Second, stem cells are able to stay in their unspecialized state and make copies of themselves. Embryonic stem cells come from the embryo at a very early stage in development (the blastocyst staqe). The stem cells in the blastocyst go on to develop all of the cells in the complete organism. Adult stem cells come from more fully developed tissues, like umbilical cord blood in newborns, circulating blood, bone marrow or skin.

Medical researchers are investigating the use of stem cells to repair or replace damaged body tissues, similar to whole organ transplants. Embryonic stem cells from the blastocyst have the ability to develop into every type of tissue (skin, liver, kidney, blood, etc.) found in an adult human. Adult stem cells are more limited in their potential (for example, stem cells from liver may only develop into more liver cells). In organ transplants, when tissues from a donor are placed into the body of a patient, there is the possibility that the patient's immune system may react and reject the donated tissue as "foreign." However, by using stem cells, there may be less risk of this immune rejection, and the therapy may be more successful.

Stem cells have been used in experiments to form cells of the bone marrow, heart, blood vessels, and muscle. Since the 1990's, umbilical cord blood stem cells have been

used to treat heart and other physical problems in children who have rare metabolic conditions, or to treat children with certain anemias and leukemias.

There has been much debate nationally about the use of embryonic stem cells, especially about the creation of human embryos for use in experiments. In 1995, Congress enacted a ban on federal financing for research using human embryos. However, these restrictions have not stopped researchers in the United States and elsewhere from using private funding to create new embryonic cell lines and undertaking research with them. The embryos for such research are typically obtained from embryos that develop from eggs that have been fertilized *in vitro* - as in an *in vitro* fertilization clinic - and then donated for research purposes with informed consent of the donors. In 2009, some of the barriers to federal financing of responsible and scientifically worthy human stem cell research were lifted

Cloning

Cloning can refer to genes, cells, or whole organisms. In the case of a cell, a clone refers to any genetically identical cell in a population that comes from a single, common ancestor. For example, when a single bacterial cell copies its DNA and divides thousands of times, all of the cells that are formed will contain the same DNA and will be clones of the common ancestor bacterial cell. Gene cloning involves manipulations to make multiple identical copies of a single gene from the same ancestor gene. Cloning an organism means making a genetically identical copy of all of the cells, tissues, and organs that make up the organism. There are two major types of cloning that may relate to humans or other animals: therapeutic cloning and reproductive cloning.

Therapeutic cloning involves growing cloned cells or tissues from an individual, such as new liver tissue for a patient with a liver disease. Such cloning attempts typically involve the use of stem cells. The nucleus will be taken from a patient's body cell, such as a liver cell, and inserted into an egg that has had its nucleus removed. This will ultimately produce a blastocyst whose stem cells could then be used to create new tissue that is genetically identical to that of the patient.

Reproductive cloning is a related process used to generate an entire animal that has the same nuclear DNA as another currently or previously existing animal. The first cloned animals were frogs. Dolly, the famous sheep, is another example of cloning. The success rates of reproductive animal cloning, however, have been very low. In 2005, South Korean researchers claimed to have produced human embryonic stem cell lines by cloning genetic material from patients. However, this data was later reported to have been falsified.