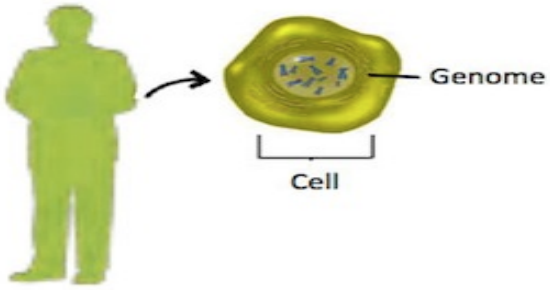
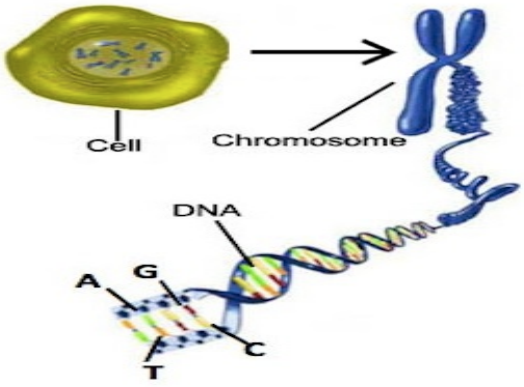
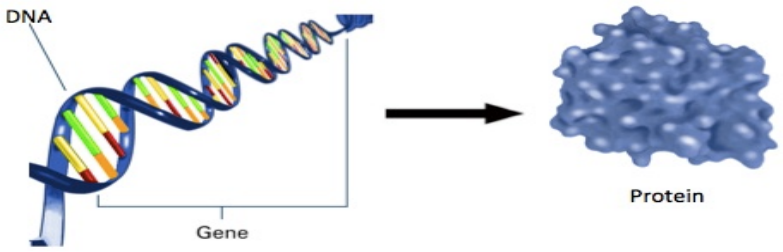
Genetics and Whole Genome Sequencing Handout

Information component	Information content
1. Genetics	
1a. What is a genome?	The human body is made up of trillions of cells. Cells are the building blocks of the body. The cells contain genetic material that guides the body's development and function. The **genome** is the full genetic information found in one cell.
1b. What are chromosomes and DNA?	Chromosomes contain our genetic material. In each cell of our body there are 23 pairs of chromosomes, 46 chromosomes in total. The chromosomes are passed from parents to children. 23 chromosomes come from our mother and 23 from our father. A chromosome is made up of 2 strands of DNA (the scientific word is deoxyribonucleic acid) that are bundled tightly. The DNA is built of a series of 4 chemicals called bases: **A**denine, **C**ytosine, **T**hymine, and **G**uanine. We use the first letter of each name – A, C, T, and G – to refer to these bases. The order, or sequence, of these bases gives the information that is necessary to take care of the body and its functions.
1c. What is a gene?	A **gene** is a length of DNA, usually a few thousand bases long. Human beings have over 20,000 genes. Genes tell our bodies how to make proteins. Proteins do most of the work in cells. They are necessary for proper structure, function, and regulation of the body's tissues and organs. Each chromosome is made up of many genes. Therefore, like chromosomes, genes come in pairs.

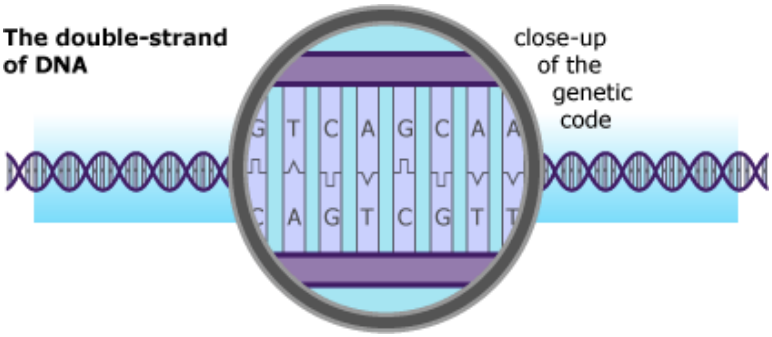






1d. What does 'DNA sequence' mean?

The DNA sequence is the exact order of the bases – adenine (A), cytosine (C), thymine (T), and guanine (G) – that are in the DNA of one cell. A part of a DNA sequence may read like this: ATTGCCGTATAGC. But, a full DNA sequence in the human genome is much longer. The human genome is made up of about three billion of these bases. More than 99% of these bases are the same in all people. This means that the genetic difference between individuals is less than 1%. Although this variation is small, it can influence risk for many different diseases. It can also account for different physical characteristics that make each of us unique.



2. What is whole genome sequencing?

Whole genome sequencing is a technology that reads the entire DNA or sequence of bases (A, C, T, and G) that make up a person's genome.

The goal of **whole genome sequencing** is to tell us the exact order, or sequence, of the bases in a person's genome. We can identify variations, or differences, in one person's DNA sequence if we compare them to a standard sequence. Finding variations in a genome may reveal a lot of personalized information.

3. What information can we learn from whole genome sequencing?

Whole genome sequencing can tell us about many different health conditions, including rare genetic disorders, common complex diseases, and how a person's body responds to medications. It can also give us information about physical traits and ancestry. In addition, we often find many DNA variants whose role we do not exactly understand. These are called variants of unknown significance.

More explanation of these types of information is given below:

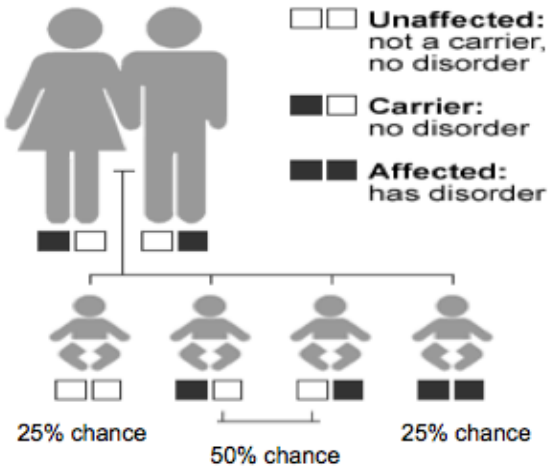
3a. Genetic disorders

Genetic disorders are diseases that are caused by abnormal variations, sometimes called mutations, in a person's DNA. A few examples of genetic disorders are: Down syndrome, sickle cell disease, and Huntington's disease.

3b. Genetic carrier

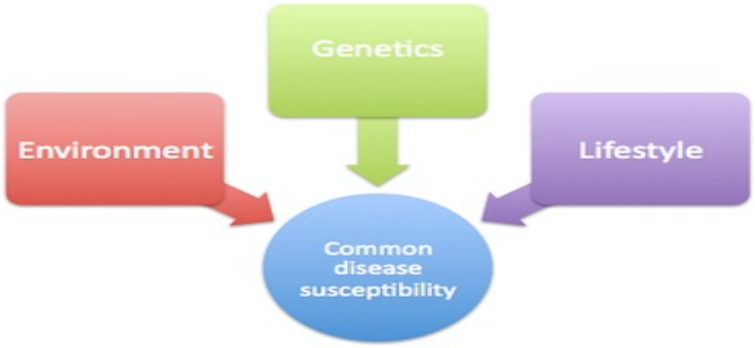
A genetic carrier is a healthy person who has a genetic mutation in a gene on one of their chromosomes and a normal gene on the other chromosome. In order for this kind of genetic mutation to cause a disorder, a person would need to have the mutation in both copies of a gene. These types of genetic disorders are called "autosomal recessive disorders." If both parents are carriers of a genetic mutation, they have a 25% chance of having a child with an autosomal recessive disorder. This is because children receive half of their chromosomes from their mother and half from their father. Cystic fibrosis is an example of an autosomal recessive genetic disorder.

The image below shows that a couple in which both parents are carriers has a 25% chance of having a child with a genetic disorder, a 50% chance of having a child who is a genetic carrier, and a 25% chance of having a child who does not have the mutation at all.



3c. Common disease susceptibility

Common diseases are caused by many different factors. Differences in a person's DNA sequence contribute to the development of common diseases. But genetic variations alone do not cause common diseases. Other important factors are the environment and lifestyle. A few examples of common diseases are: heart disease, type 2 diabetes, Alzheimer's disease, and some types of cancer. The exact combination of factors that causes a common disease varies among individuals.

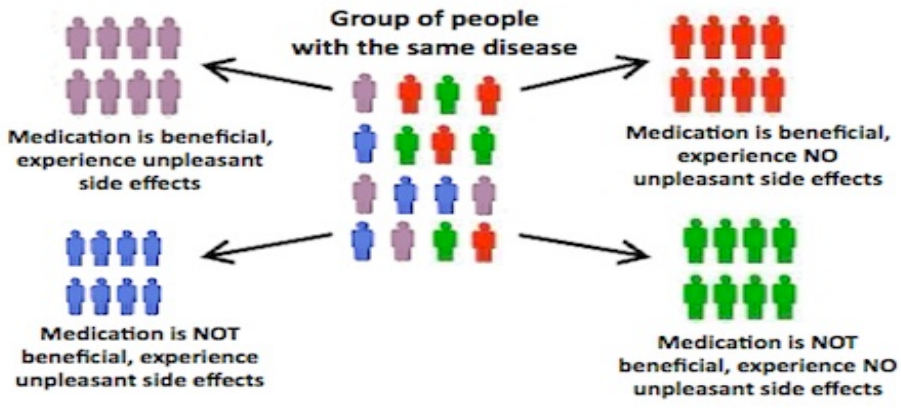


3d. Pharmacogenetics

Pharmacogenetics is the study of how a person's genetic makeup influences the way they respond to medications. For example, differences in a person's genome can influence whether or not a specific medication works for them, and whether or not they have unpleasant side effects from the medication.

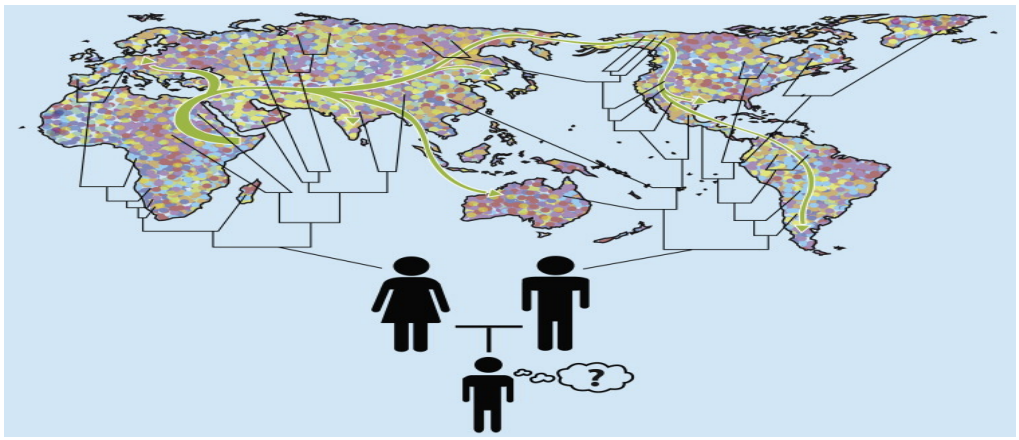
The image below shows how a group of people with the same disease can react differently to the

same medication.



3e. Physical traits Height, eye color, and hair color are examples of physical traits influenced by DNA.

3f. Ancestry A person's genome may give clues to their ancestry. These clues could identify relationships to close relatives, such as grandparents, or they could provide more general information such as identifying a continent of origin.



3g. DNA variant of A DNA variant of unknown significance is a variation in a person's DNA sequence that has an

unknown significance	unclear role. In the future, scientists hope to learn more about whether these variations really affect health or if they are just harmless.
4. What will whole genome sequencing mean to you?	It is possible that you have seen articles in the media about **whole genome sequencing**. Someday you might be invited to participate in a research study that uses **whole genome sequencing**. Or one day, it might even be possible to get **whole genome sequencing** done for your own personal use, either through your doctor or through a commercial company. We hope that these educational materials will help you to have a better understanding of whole genome sequencing in the future.