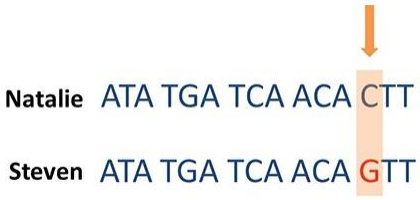
GENETIC VARIATION:

The Differences in characteristics within a population

If you take a look in a group of individuals you will notice that they are all distinctive. While everyone's DNA uses the same letters (A, T, C, and G), to compose the same genes, there still can be differences in our genes. In fact, about 99.9% of our DNA sequence is same. The remaining 0.1% makes a person unique. Each individual has its unique set of traits and even, offspring's of the same parents still differ a lot among themselves. This is called genetic variation.

Changes in the genetic sequence may affect many aspects of life. In the picture, the change from C to G in the genetic code gives Steven an increased risk for an early heart attack. This is not a diagnosis and clearly it is not Steven's total genetic risk, but it is a fraction of a bigger picture that gives him an increased risk for the disease because of his genetics.



So, except from genetic variants that affect appearance, some genetic variants, called risk variants, can increase your risk for developing a disease. Other genetic variants can decrease your risk for developing a certain health condition. These variants are called protective variants.

Only variation that arises in germ cells can be inherited from one individual to another and so affect population dynamics. Mutations and recombination are major sources of variation, but mechanisms such as sexual reproduction and genetic drift contribute to it as well.

Variation in the human genome can take several forms. One form is single nucleotide polymorphisms, (SNPs, pronounced ‘snips’), the most common type of genetic variation amongst people. Another form involves a larger-scale variation where you might have a stretch of DNA of hundreds, or even thousands, of base pairs that is different between people. Maybe I have three copies of that stretch and you have two. Or maybe it's a circumstance where I have the genes in the order ABC and you have them in the order of ACB because you have an inversion in that. Those don't have to be pathological. In fact, most of them won't be, but it's a different kind of variation that in some instances may be playing a role in disease risk.

The last few years have seen extensive efforts to catalogue human genetic variation and correlate it with phenotypic differences. Most common SNPs have now been assessed in genome-wide studies for statistical associations with many complex traits, including many important common diseases. Genetic variation can explain differences in disease susceptibility and how people react to drugs and reveal an insight into the role of human genetic variation behind differential susceptibility of human diseases, differential response to pharmacological agents and presence of varied phenotypes. This leads to the concept of personalized medicine, which is based on using an individual's genetic profile to make the best therapeutic choice by facilitating predictions about whether that person will benefit from a particular medicine or suffer serious side effects.

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