Genetic Lifehacks

Learn. Experiment. Optimize.

Hi everyone,

Gregor Mendel, the 'Father of Genetics', was an Augustinian monk, mathematician, and biologist. You probably remember Mendel from high school biology... He experimented with pea plant breeding and meticulously recorded his findings. Mendel crossed tall peas and short peas, pink blooming peas and white blooming peas -- discovering the principles of inheritance before anyone knew about genes.

Mendelinan inheritance is a term applied to genetic diseases that indicates that a single mutation (or two copies of mutations) can cause a change.

Most of what I write about on Genetic Lifehacks involves subtle changes in genes that affect the risk of diseases when combined with lifestyle factors or other genetic variants. These common genetic changes make us all a little different.

Mendelian diseases, on the other hand, are serious hereditary disorders such as cystic fibrosis, phenylketonuria, and alpha 1 tryptase deficiency. The majority of these diseases are 'autosomal recessive,' which indicates that two copies of a mutation are required to cause a genetic disease. People who have one copy of the mutation are known as 'unaffected carriers.'

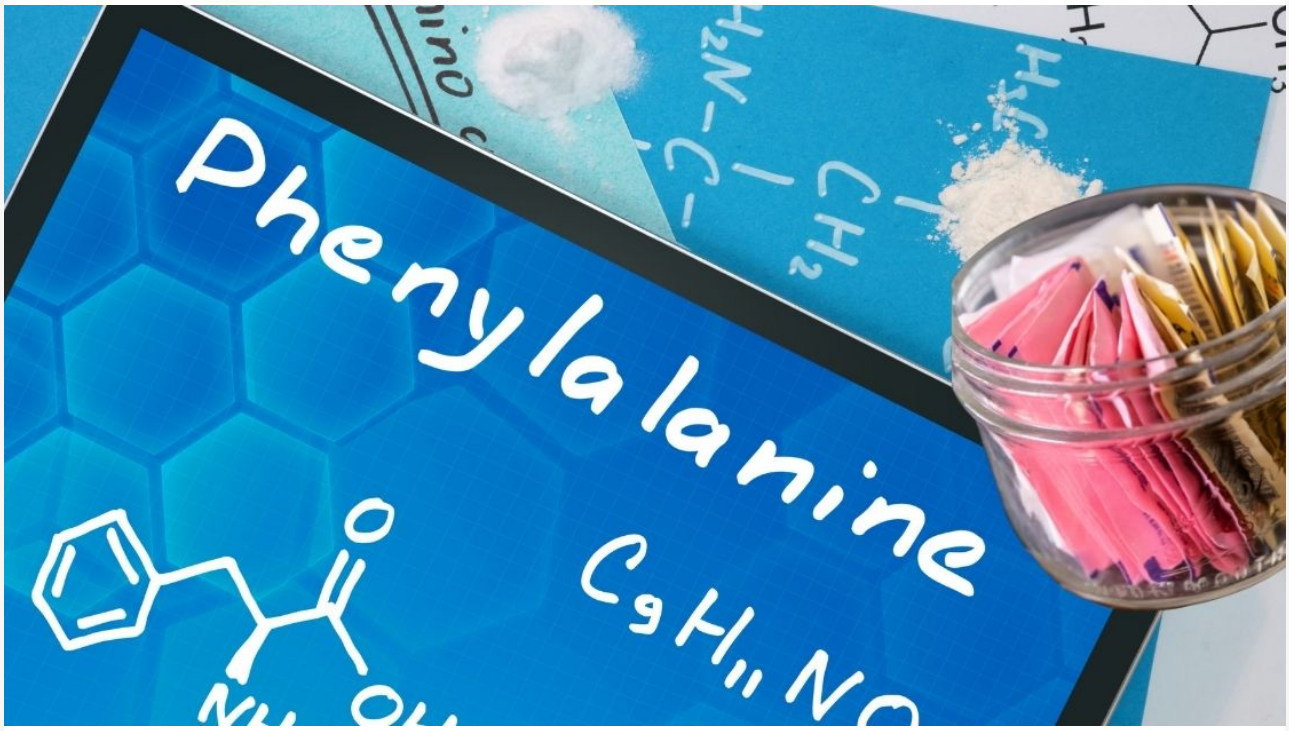
With the advent of wide-spread genetic testing, researchers now know that almost all of us carry multiple rare mutations. Interestingly, a study showed that a few people carry mutations that researchers think should cause significant genetic diseases, but they have no physical manifestations of the disease. In other cases, some may have physical symptoms from carrying just one copy of a mutation.[ref]

My latest article covers the Mendelian disease phenylketonuria (PKU). It occurs when someone has two mutations (one from mom, one from dad) in the PAH gene. In people with European ancestry, the carrier rate for PAH mutations can be ~1%. Interestingly, carriers of PAH mutations have specific neurological differences (e.g. lower verbal recall) that may be helped with dietary changes, such as cutting out diet sodas that contain asperame.

A quick caveat: Genetic data from 23andMe or AncestryDNA is pretty accurate. But 'pretty accurate' isn't good enough for diagnosing rare diseases; you should always verify any rare mutation with a clinical-grade test before making any medical decisions.

Gratefully yours,

~ Debbie Moon



Phenylketonuria: Mutations, Carrier Impact

Phenylketonuria, also called PKU, is a genetic metabolic disorder in which the amino acid phenylalanine is not properly metabolized. PKU can cause intellectual disabilities, seizures, behavioral issues, and psychiatric illnesses if left untreated. It is one of the genetic diseases that infants are tested for when they are born.

This article explores the physiological changes due to carrying a phenylketonuria mutation. We will dive into why the buildup of phenylalanine causes neurological issues, and how dietary changes can affect this. As always, I'll include the genetic mutations to check in your raw data.

Read the article, view your genes...

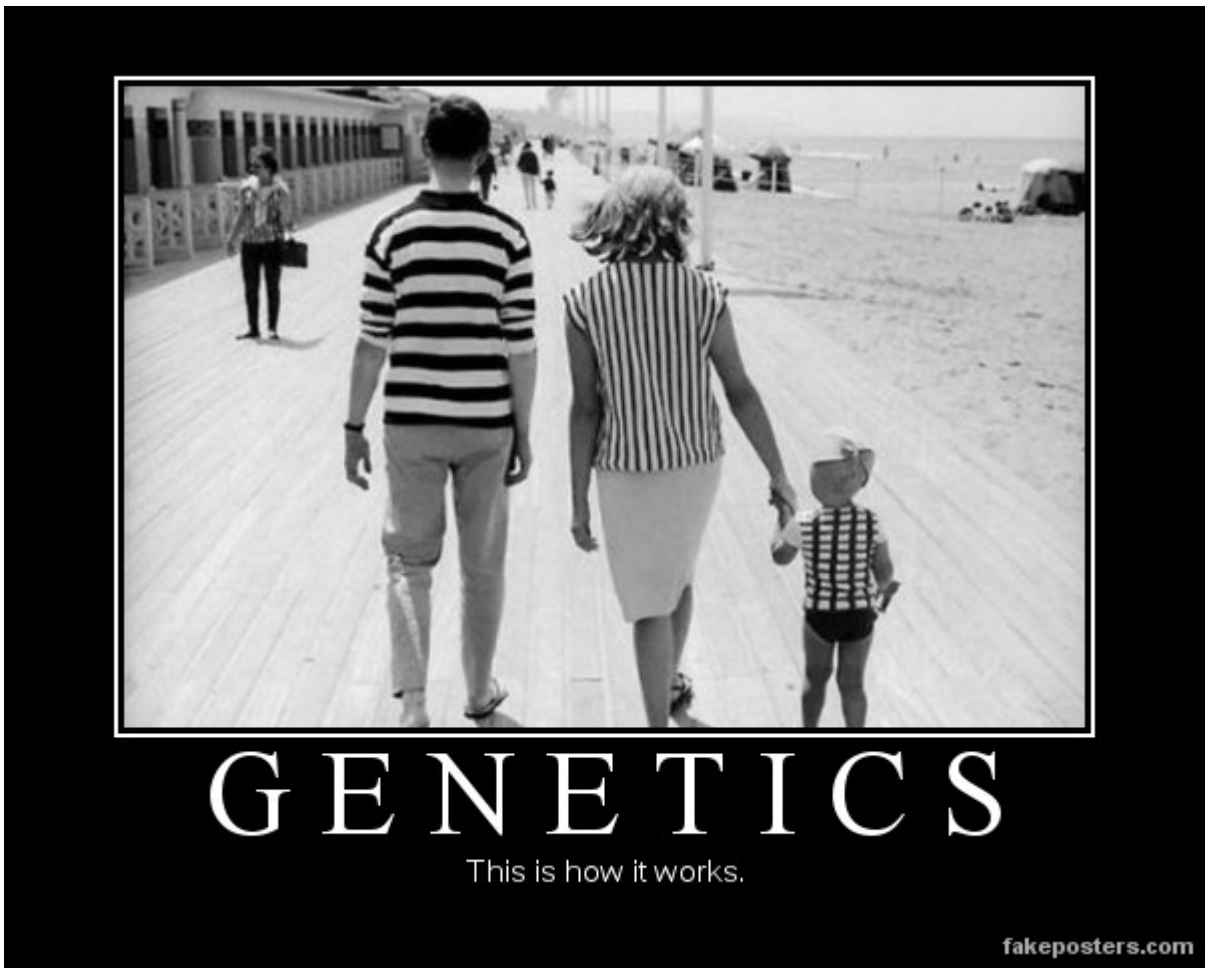
Related articles:

Cystic Fibrosis: Check to see if you are a carrier

Did you know that up to 3% of the population carries one copy of a mutation for cystic fibrosis? Being a carrier of a cystic fibrosis mutation increases the risk of several diseases including pneumonia from respiratory viruses, pancreatitis, and male infertility.

Alpha-1 Antitrypsin Deficiency

A genetic mutation in the SERPINA1 gene causes alpha-1 antitrypsin deficiency. This deficiency increases a person's susceptibility to respiratory issues, COPD (chronic obstructive pulmonary disease) and, in some cases, liver dysfunction.



Genetic Lifehacks

Summer will be here next week, MT