Uncertain Genetic Test Results "Variants of Uncertain Significance"

What happens when we don't know if a change in a recipe (or gene) might change the final product?

Recipe 1

DNA Alphabet
Letters to write out the recipes

ATG
GCA
GGC

Chocolate

Gene = Recipe
Following the recipe, we will use chocolate.

Chocolate Cake
Following this recipe that used chocolate, we baked a chocolate cake.

Recipe 2

ATG
GGA
GGC

DNA Alphabet
Letters to write out the recipes

Chokalate

Gene = Recipe
The "chocolate" in the recipe has been changed to "chokalate." We don't know what this word means.

Final Product???
Following this recipe that used "chokalate," we don't know yet what the final product would be.

The change in the recipe from chocolate to "chokalate" may have changed the cake that was baked, but we do not know for certain. A variant of uncertain significance (VUS) acts the same way. When we see a change in our genes that has not been well studied, we cannot predict how this change might affect a person. Over time, a VUS may eventually be better understood so doctors know more about how the change affects people.

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New Genetic Changes ("De novo variants")

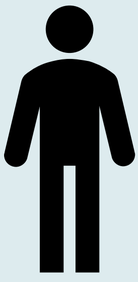
DNA Alphabet

Gene = Recipe

Chromosome = Cookbook

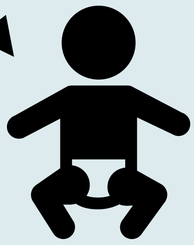
Genome = Cookbook Collection

The genome is a full set of 47 cookbooks:
23 from each parent
PLUS the extra mitochondrial DNA

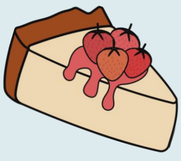


Chocolate

Chocolate



Cheese



New Changes/De novo variants

Sometimes a genetic change happens as a brand new change in a child and is not inherited from the parents. These new changes are called "de novo." In this example, both mom and dad have a recipe (gene) for chocolate cake, however, their child has a change in their recipe (gene) that made cheesecake instead. This child may be the first in the family to ever have this trait.

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