We have over 20,000 genes in our body, and each gene has a very specific job. To do that job, our genes make substances called proteins that make everything in the body work, grow, and function properly. A gene is made up of a long string of many letters (A, C, G, and T) that act as an instruction manual for how to make a specific protein.

Much like spelling errors (sequence variants) can affect how a gene makes a protein, so can extra and missing letters, which are called duplications and deletions. Adding or removing letters from an instruction manual affects how the instructions read, and can also cause the gene to make either an abnormal protein or no protein at all, which is what can cause genetic conditions.

A similar example is self-assemble furniture. Let's say you buy a kit at the store to build a jungle gym for your kids in the back yard. You lay all of the pieces out, start going through the instructions and putting things together. When you get done, the jungle gym doesn't look like it's supposed to and you have a bunch of extra pieces left over. You go back through the instructions and realize that there is a page that is missing. Without that missing page, you don't have proper instructions on how to put together your jungle gym. The instructions for how a gene builds a protein is very similar.

Some missing or extra pieces (called variants) do not cause a problem. If you're looking at your jungle gym instructions and it turns out you have a duplicate of one of the pages. It's not necessary, but it's also not hurting anything. Whether the missing or extra piece in a gene causes a problem depends on the gene, and where in the gene the extra or missing piece is at.

Variants in genes can be broken down into five different categories:

- Pathogenic variants are known to make the gene not work correctly, and can cause a genetic condition.
- Likely pathogenic variants probably make the gene not work correctly, but more information is needed to say for sure.
- A variant of uncertain significance means there is not enough information to say whether the variant is harmful or not, and more research on that variant is needed.
- Likely benign variants probably don't impact how the gene works, and thus likely does not cause a genetic condition.
- Benign variants are known to not impact how the gene works, and do not cause a genetic condition.