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. If there are any spelling mistakes in that instruction manual (called a variant), it can cause the gene to make either an abnormal protein or no protein at all, which is what can cause genetic conditions.

Sequencing is the process where the lab reads through the letters of a gene and looks for any spelling errors that may affect how the gene works.

A sequence variant is when there is some sort of spelling error in the gene. There may be an 'A' where there is normally supposed to be a 'C'. Some of these spelling errors happen in a part of the instructions that don't affect how the instructions read and don't create a problem (imagine an instruction that is supposed to say 'THE CAT RAN' but instead says 'THE CAT RUN'. The grammar isn't perfect, but the message is still pretty much the same). This is called a benign (non-harmful) variant.

Other spelling errors happen in a part of the instructions that change the whole meaning (an instruction that is supposed to say 'THE CAT RAN' but instead says 'THE CAB RAN'. That one letter difference changed the entire meaning of the sentence). This is called a pathogenic (harmful) variant. Pathogenic variants in genes can affect how the instructions make a protein. If the protein cannot work how it is supposed to in the body, it can cause a genetic condition.

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.

Related Articles

- Deletion/Duplication
  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 that…
- Next Generation Sequencing/Panel Testing
  Next generation sequencing (aka panel testing) is a relatively newer technology that allows the lab to look at many different genes in a faster and more cost-effective way than ever before. This type of testing can be particularly helpful in situations where there are many potential explanations for what we…
- Familial Pathogenic Variant
  Genetic testing for someone who may be at risk for an inherited disease is always easier if we know the specific genetic cause. Often times, the best way to find the genetic cause is to start by testing someone in the family who is known or strongly suspected to have…
- Traditional Chromosome Analysis
  A chromosome analysis, or karyotype, is the traditional testing that can been done on cells from chorionic villus sampling (CVS) and amniocentesis procedures, or from a blood sample. A karyotype involves the lab examining the cells and determining the number of chromosomes. How the lab determines the number of chromosomes…
- Chromosomal Microarray
  A chromosomal microarray (CMA) is a test that can be done to look for specific kinds of chromosome changes called microdeletions and microduplications. Much like a traditional chromosome analysis, CMA generally looks at all 23 pairs of chromosomes. While the chromosome analysis is looking for large changes (extra or missing…
- Genetic Testing Results
  With genetic testing, there are generally three different results that we can get: a positive, a negative, or a variant of uncertain significance (VUS). Positive A positive result means that a harmful change (called a pathogenic variant) was found in a gene that causes that gene to not work properly.…
- How to Decide
  Whether or not to pursue any genetic testing is a very personal decision. In some cases, moving forward with genetic testing may help to provide an answer to a health question that is running in someone's family, or may help to provide a name to a medical condition that someone…