

**G E N E T I C**

**C O N N E C T I O N S**

**for Early Intervention Providers**



*A Workshop for Early Intervention Providers*



# **A Note from our Funders**

**This program is supported by the Health Resources and Services Administration (HRSA) of the U.S. Department of Health and Human Services (HHS) as part of an award totaling \$600,000.00 with 0 percent financed with non-governmental sources. The contents are those of the author(s) and do not necessarily represent the official views of, nor an endorsement, by HRSA, HHS, or the U.S. Government. For more information, please visit [HRSA.gov](https://www.hrsa.gov).**

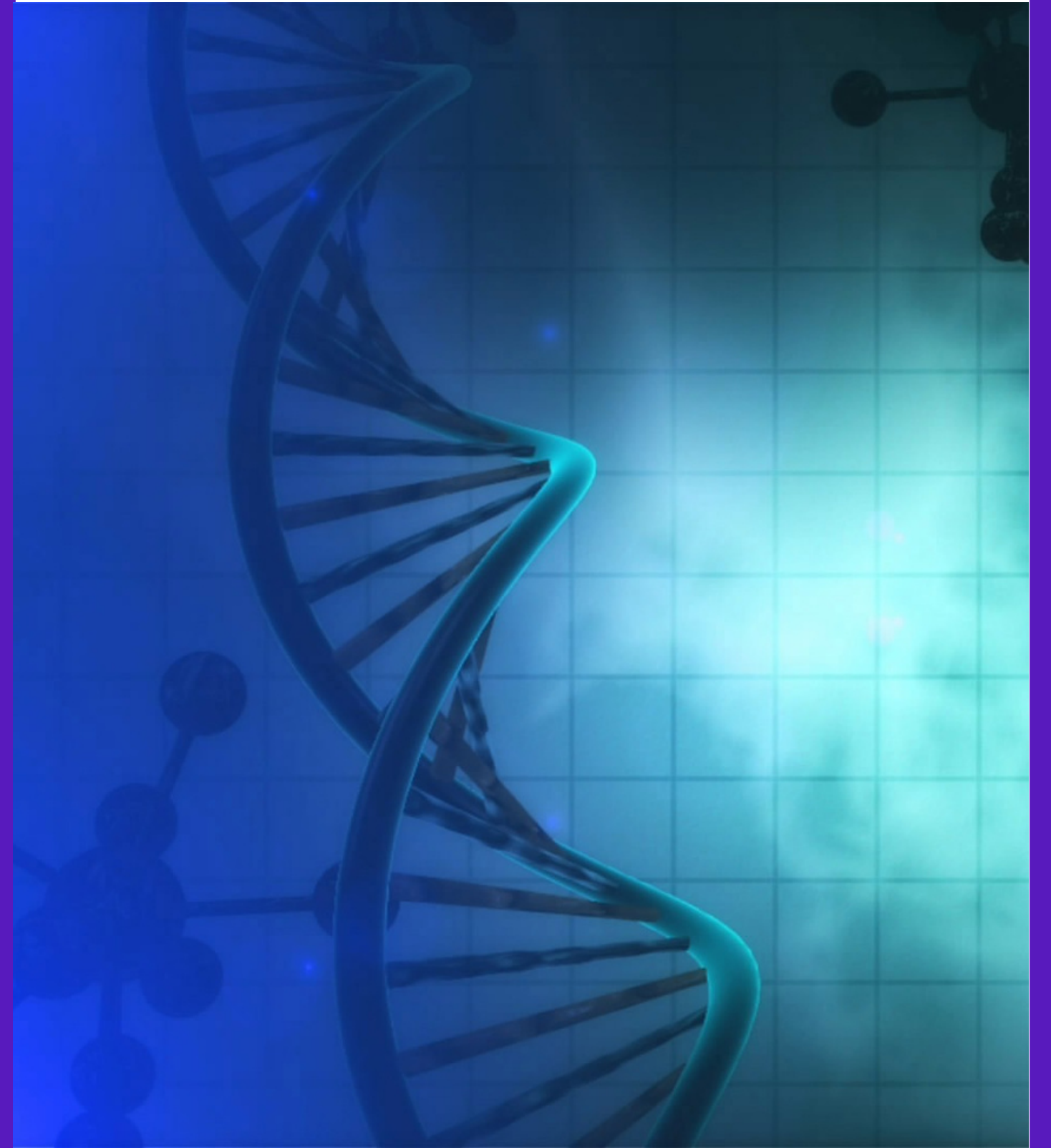
**M O D U L E 2**

**The Vocabulary of Genetics**

# Learning Objectives

The learner will be able to understand basic genetic terminology.

The learner will be able to utilize a simple analogy to illustrate a genetic concept to a family.



## Module 2: The Vocabulary of Genetics

# A G E N D A

### Topics Covered

What do you think of when you hear...

Vocabulary: DNA, Gene, Chromosome,  
Genome, Protein, Enzyme Genetic Change

Baking a Cake

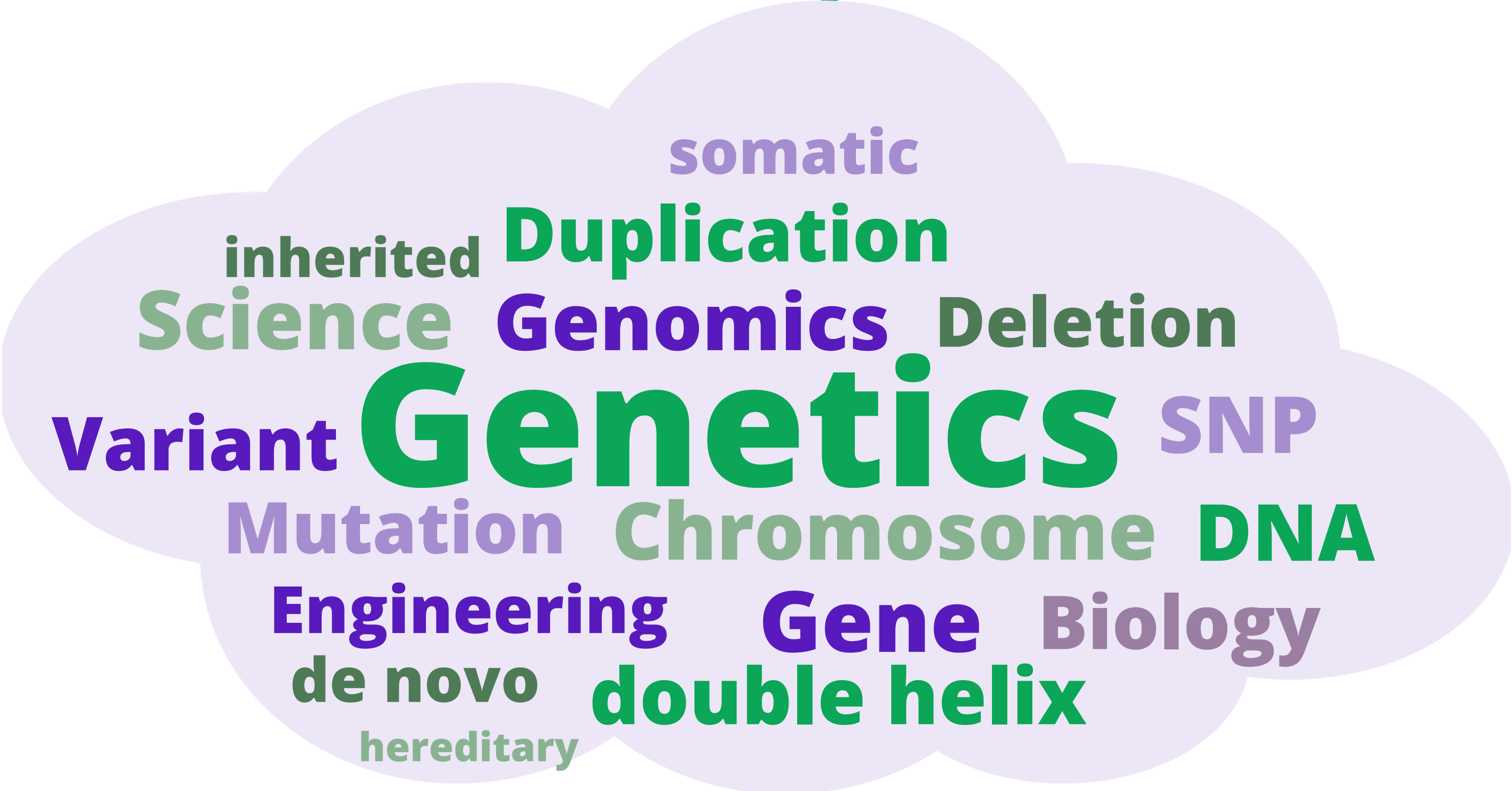
Genetic Changes & Red Flags

Resource: VUS Video

What do you think of when you hear the word...

**G E N E T I C S**

# Some words that may come to mind...



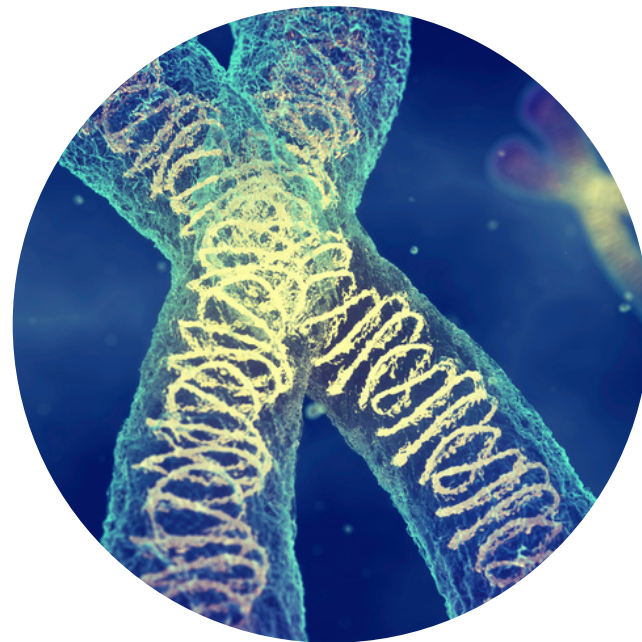
A word cloud of genetics-related terms inside a light purple cloud shape. The words are arranged in a roughly circular pattern, with 'Genetics' being the largest and most central word. Other prominent words include 'Duplication', 'Genomics', 'Deletion', 'SNP', 'DNA', 'Chromosome', 'Gene', 'Biology', 'double helix', 'Engineering', 'Mutation', 'Variant', 'Science', 'inherited', 'somatic', 'de novo', and 'hereditary'. The colors of the words vary, including shades of green, purple, and blue.

somatic  
inherited **Duplication**  
**Science** **Genomics** Deletion  
**Variant** **Genetics** SNP  
Mutation **Chromosome** **DNA**  
**Engineering** **Gene** **Biology**  
de novo **double helix**  
hereditary

# A few genetic words...



**DNA**



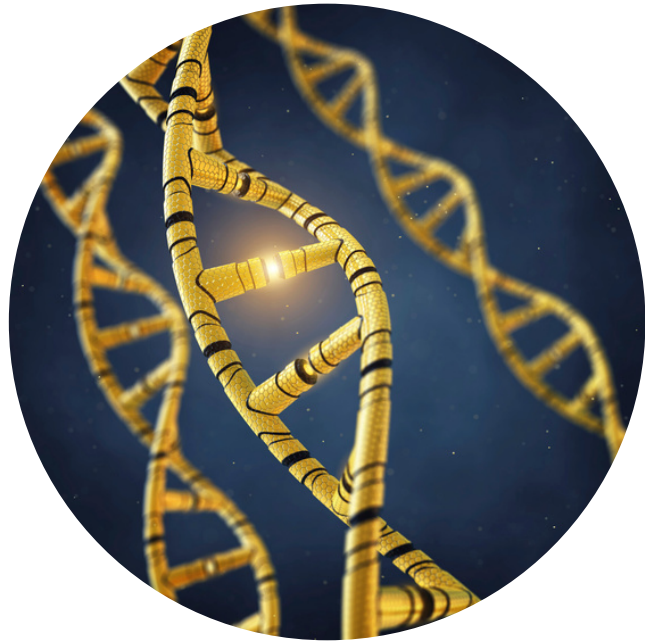
**Chromosome**



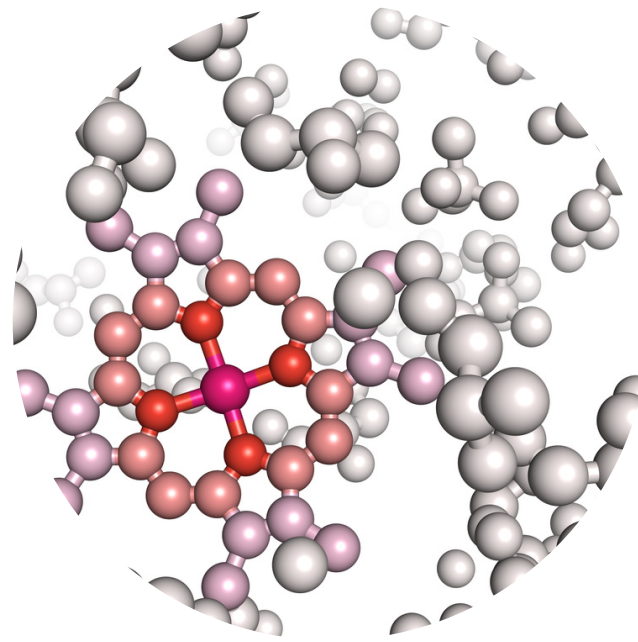
**Gene**



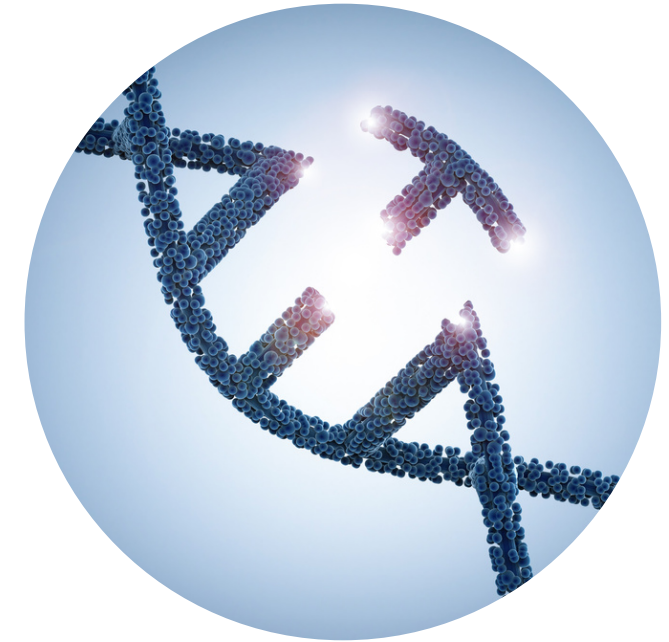
# A few more genetic words...



**Genome**



**Protein & Enzyme**



**Genetic Change**

# Let's Bake A Genetic Cake



# GENETIC ANALOGY: COOKING



Protein/Enzyme=  
Cake

DNA Alphabet =  
Letters used to write out a recipe

Gene = Recipe



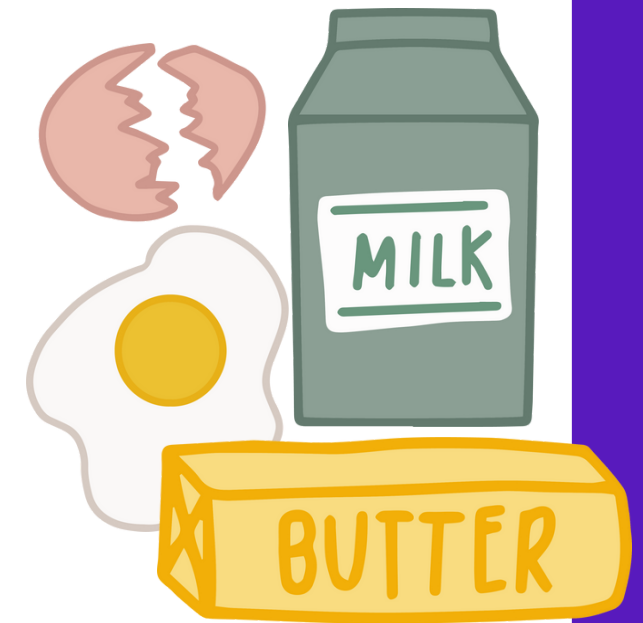
Chromosome = Recipe Book



Genome = Set of Recipe Books

AT

CG



# DNA

**DNA stands for Deoxyribonucleic Acid.**

**Simply stated, DNA is the alphabet used to write the instructions that tell our body how to grow and function.**

**DNA Alphabet =  
Letters used to write out a recipe**

**AT**

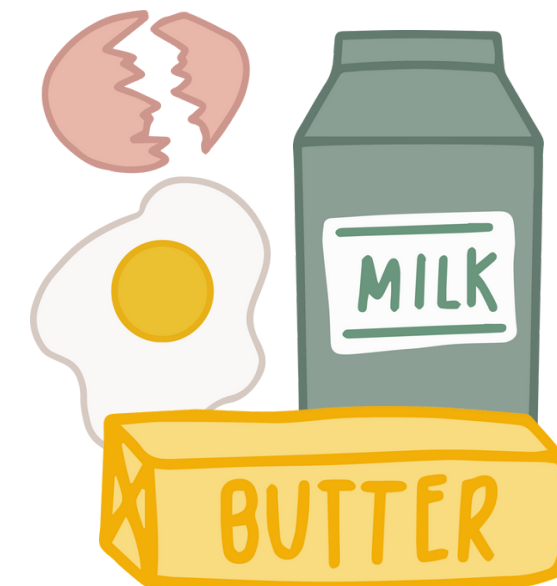
**CG**

# GENES

**Genes are sections of the DNA that contains instructions/recipe to make proteins, enzymes, and other products in the cells of the body.**

**One copy of each gene is inherited from each parent.**

**Gene = Recipe**



# CHROMOSOME

**DNA is packaged into chromosomes (recipe book).  
Each chromosome holds many genes.**

**Cells typically each have 46 chromosomes (23 pairs).  
These are numbered 1-22 and the 23rd pair is the sex  
chromosomes (X and Y).**



**Chromosome = Recipe Book**

# GENOME

**The genome is the entire set of genetic instructions found in a cell.**

**Genome = Set of Recipe Books**



# PROTEIN

**A protein is made up of various amino acids, the type and order of those amino acids is determined by the DNA sequence of the gene.**

**There are thousands of proteins that your body makes every single day that do important work in the body. In humans, there are approximately 20,000 genes that code for proteins.**



**Protein= Cake**



# ENZYME

**Enzymes are proteins that help speed up chemical reactions in our bodies. Enzymes come in all shapes and sizes depending on their function. Enzymes can help break down the foods we eat so the body can use them. Blood clotting is another example of enzymes at work. Too much or too little of a certain enzyme can cause health problems.**

**The enzyme is not destroyed during the chemical reaction and is used over and over. A cell contains thousands of different types of enzyme molecules, each specific to a particular chemical reaction.**



**Enzyme= Slice of  
Cake, Cup Cake,  
Whole Cake**

# GENETIC ANALOGY: COOKING



Protein/Enzyme=  
Cake

DNA Alphabet =  
Letters used to write out a recipe

Gene = Recipe



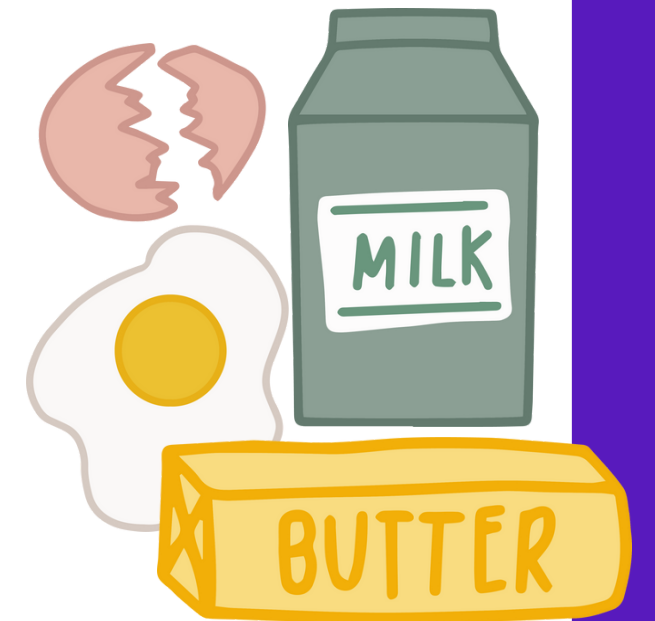
Chromosome = Recipe Book



Genome = Set of Recipe Books

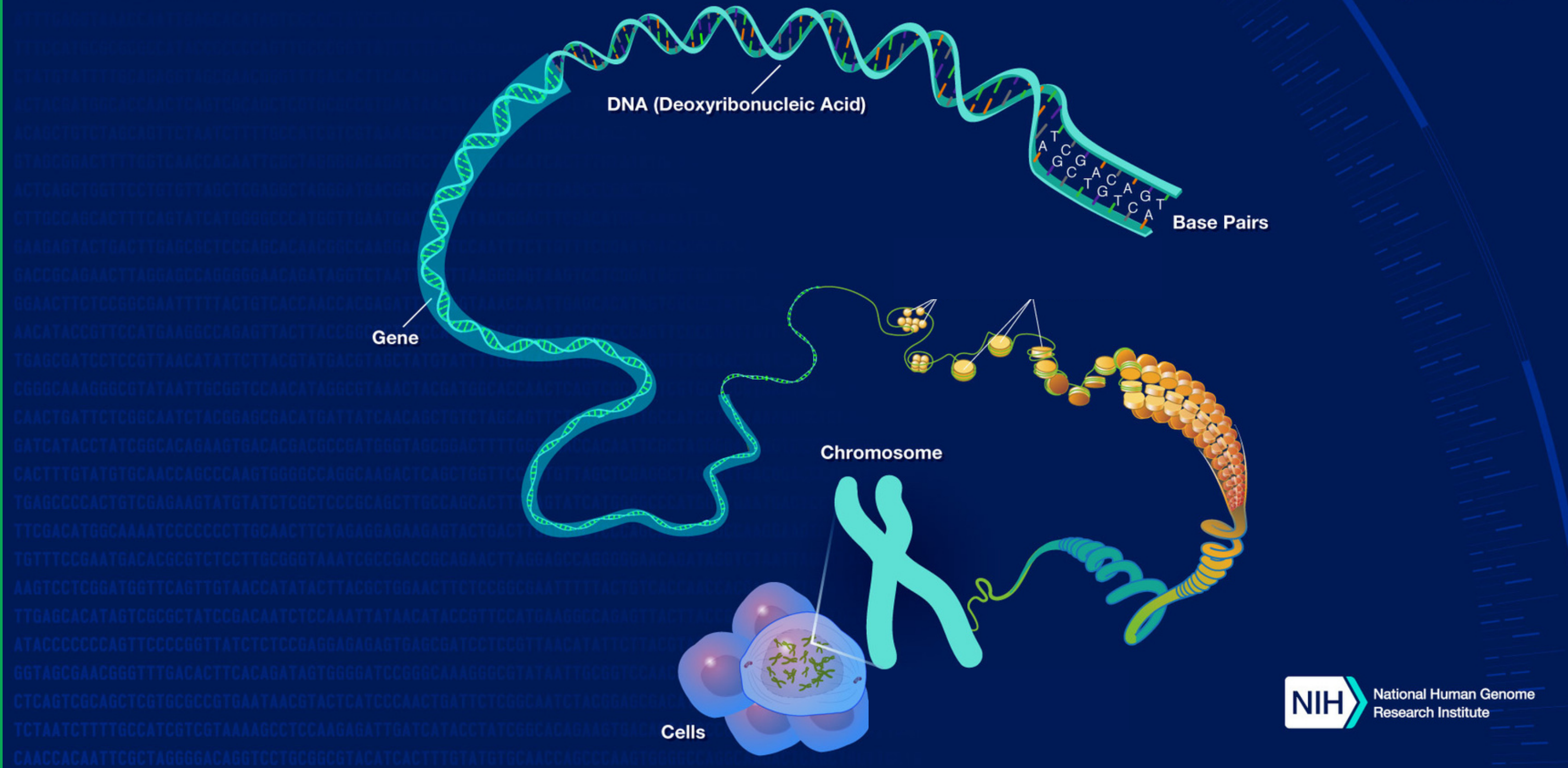
AT

CG



# A Brief Guide to Genomics

NHGRI FACT SHEETS  
genome.gov



source: [www.genome.gov/about-genomics/fact-sheets/A-Brief-Guide-to-Genomics](http://www.genome.gov/about-genomics/fact-sheets/A-Brief-Guide-to-Genomics)

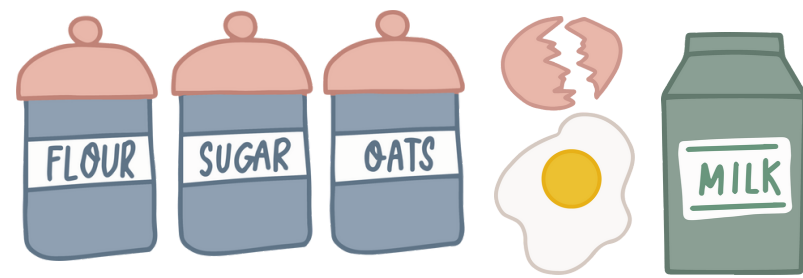
# How do genetic mutations and variants lead to health challenges and disability?

**It is important to note that genes themselves do not cause health challenges—genetic conditions are caused by DNA variants that alter or eliminate a gene's function.**

**For example, when people say that someone has “the Cystic Fibrosis Gene” they are usually referring to a version of the CFTR gene that contains a variant that causes the health condition. All people, including those without Cystic Fibrosis, have a version of the CFTR gene.**

<https://medlineplus.gov/genetics/understanding/mutationsanddisorders/mutationscausedisease/>

# GENETIC CHANGE: COOKING



DNA =

Letters used to  
write out a recipe



Chocolate



Gene = Recipe



Chocolate  
Cake



DNA =

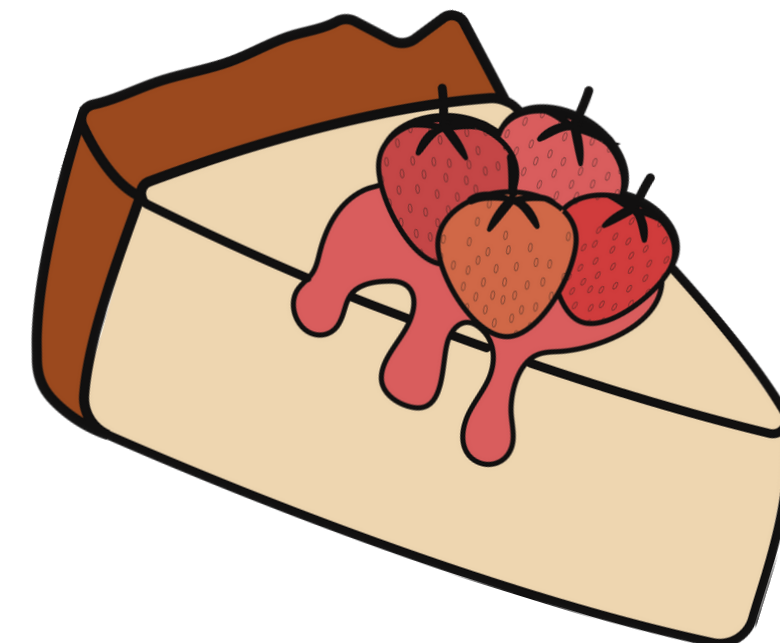
Letters used to  
write out a recipe



Cheese



Gene = Recipe



Cheese  
Cake

# GENETIC CHANGES

- **DNA can change, sometimes these changes result in a health challenge and sometimes they don't.**
- **These changes are referred to using various terms: "mutations" or "pathogenic variants" are changes in genes that can cause the protein to not form or function properly.**
- **When lots of DNA changes on a large scale and impacts sections of a chromosome, the changes are often referred to as: Chromosomal differences.**
- **Some types of chromosomal differences/ chromosomal rearrangements include:**
  - **Deletions**
  - **Insertion**
  - **Duplications**

# GENETIC CHANGES & RED FLAGS

**When a genetic change is happening inside the body and a protein/enzyme is taking a different shape or format, sometimes those differences will begin to "appear" to an outside observer in the form of a red flag.**

**The early intervention team is uniquely positioned to notice some of these red flags, signs, and symptoms due to your training in age 0-3 development.**

**The early intervention team may also be uniquely positioned to help a family advocate for genetic referral since the EI team is often in the family's home and working with the child on a more frequent basis than any other medical providers on the team.**

**A R E S O U R C E**

**A VIDEO:  
VARIANTS OF UNCERTAIN  
SIGNIFICANCE (VUS)**



V U S

Insert YouTube Link Here

**A VIDEO:  
VARIANTS OF UNCERTAIN  
SIGNIFICANCE (VUS)**



WHAT'S DNA HAVE TO DO WITH IT?

**DON'T NAVIGATE ALONE**

**DO NAVIGATE ALONGSIDE**

[www. MountainStatesGenetics.org](http://www.MountainStatesGenetics.org)

**T** H A N K  
Y O U

**Join us for Module 3:  
Four Families**