

GENETIC CONNECTIONS

for Early Intervention Providers



A Workshop for Early Intervention Providers



A Note from our Funders

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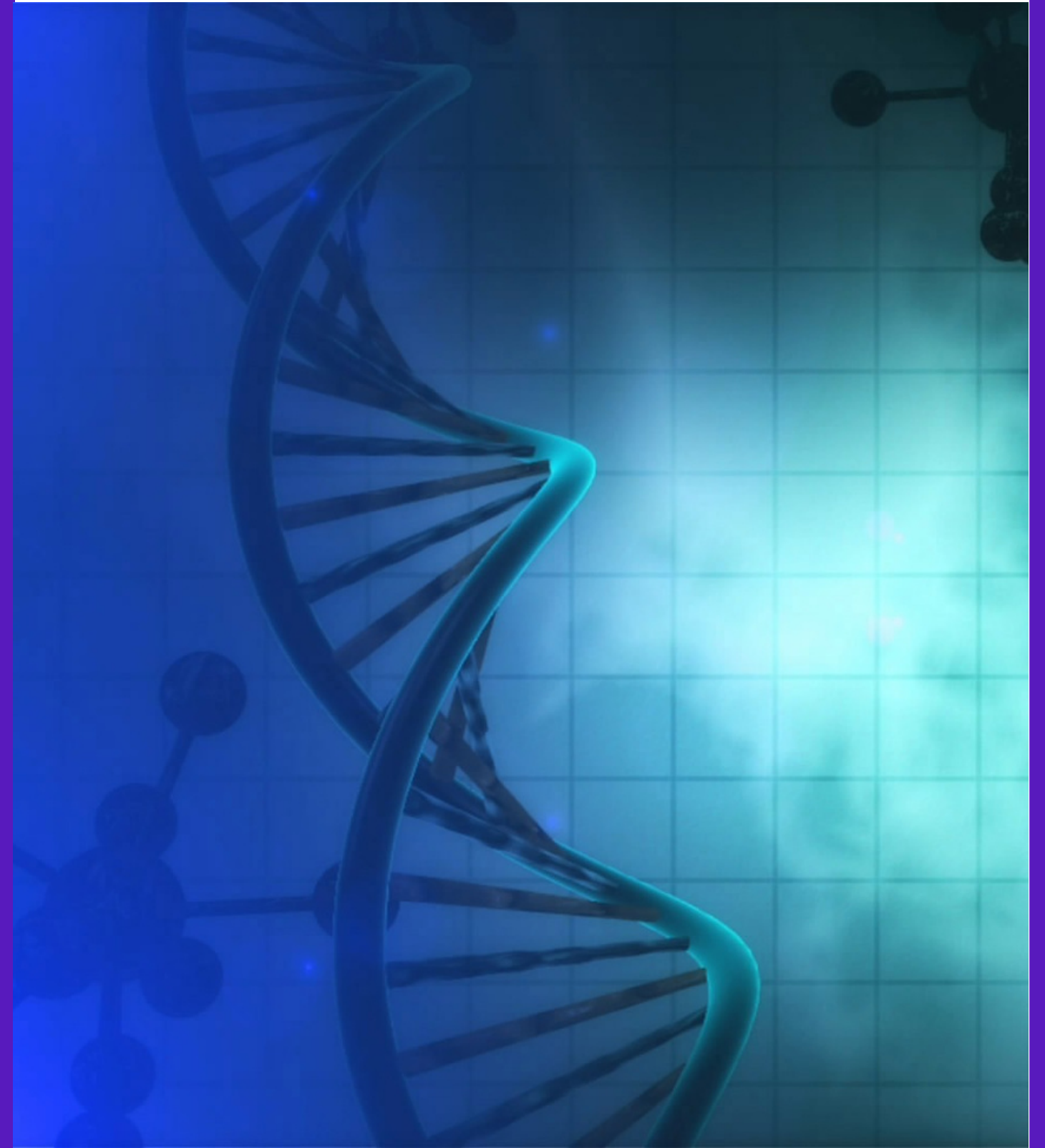
MODULE 2

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

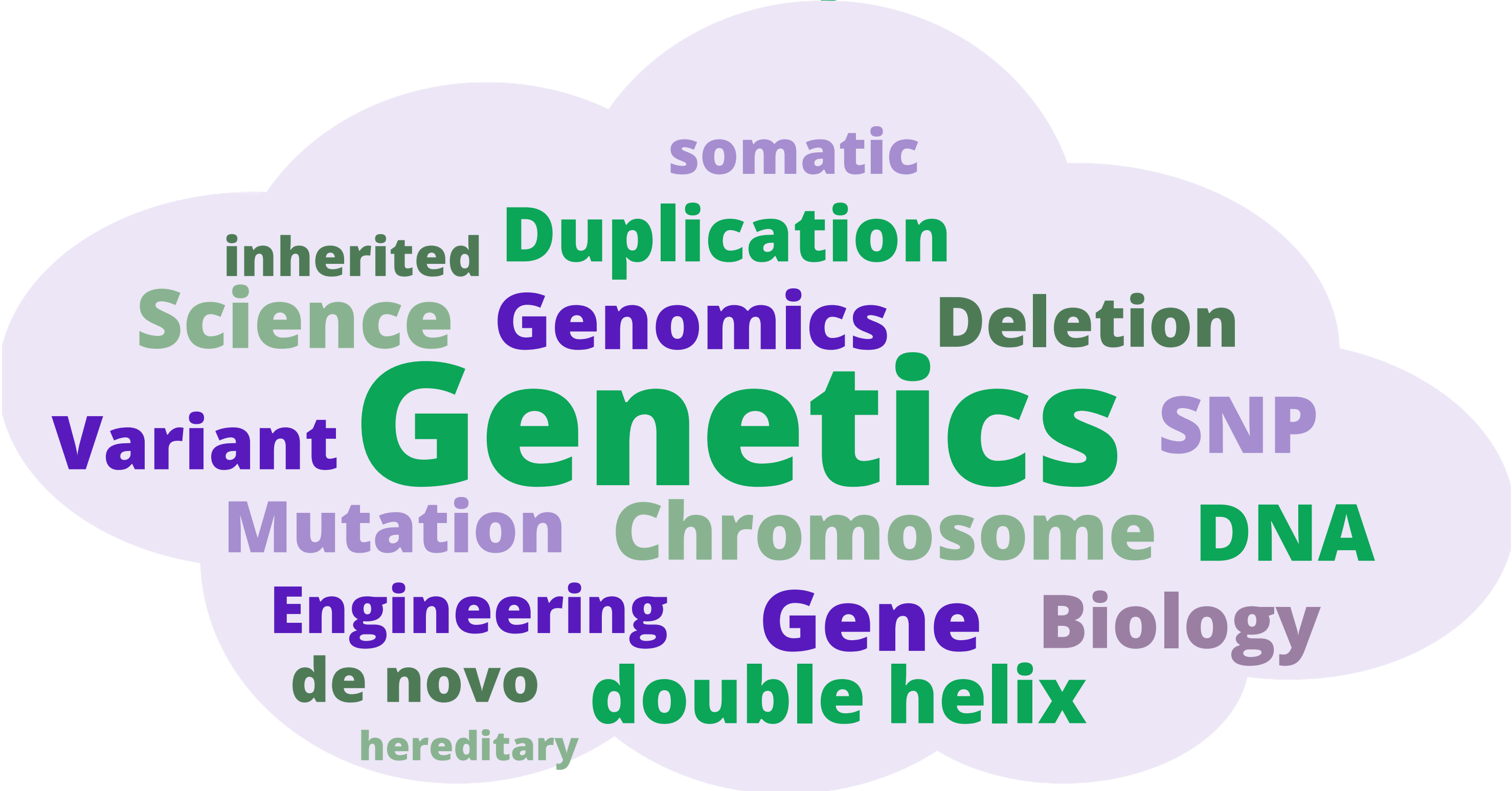
Genetic Changes & Red Flags

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...



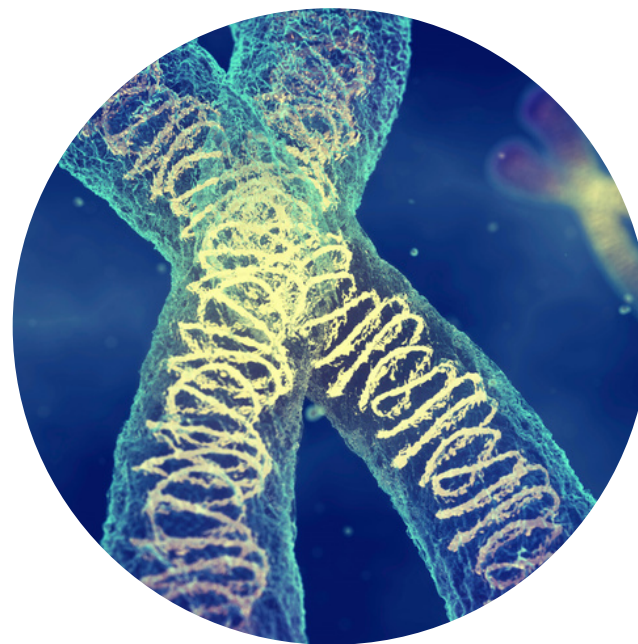
A word cloud centered around the word "Genetics". The word "Genetics" is the largest and most prominent, rendered in a bold green font. Surrounding it are various related terms in different sizes and colors (green, purple, and dark green). The words are arranged in a cloud-like shape with a light purple background. The words include: "somatic" (purple, top), "inherited" (green, top-left), "Duplication" (green, top-center), "Science" (green, left), "Genomics" (purple, center), "Deletion" (green, right), "Variant" (purple, left), "SNP" (purple, right), "Mutation" (purple, bottom-left), "Chromosome" (green, bottom-center), "DNA" (green, bottom-right), "Engineering" (purple, bottom-left), "Gene" (purple, bottom-center), "Biology" (purple, bottom-right), "de novo" (green, bottom-left), "double helix" (green, bottom-center), and "hereditary" (green, bottom-left).

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

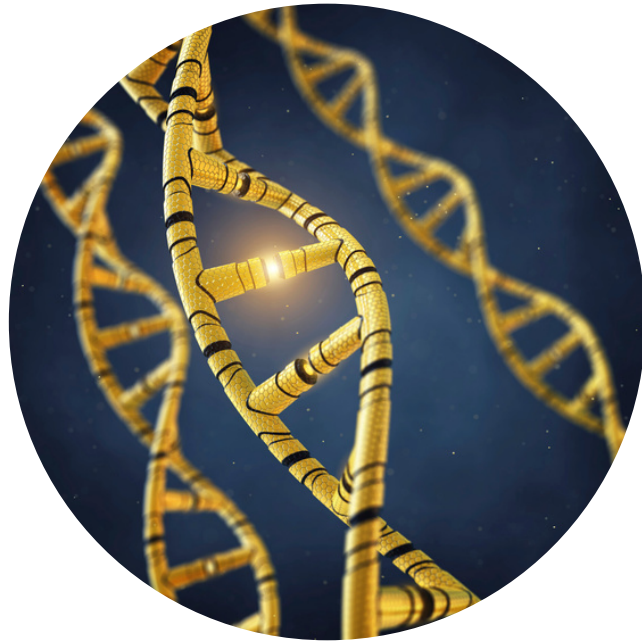


Chromosome

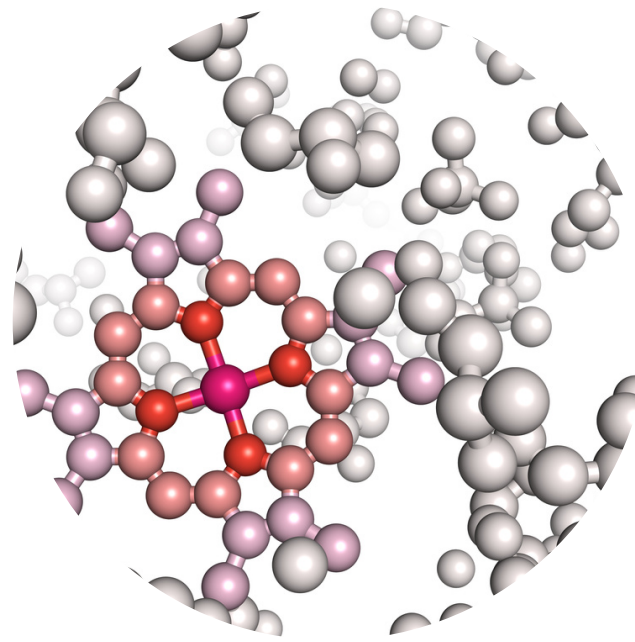


Gene

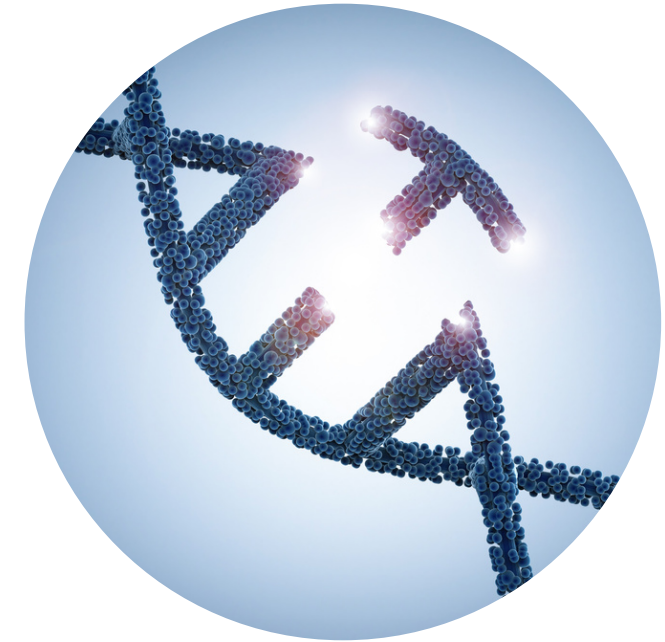
A few more genetic words...



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**

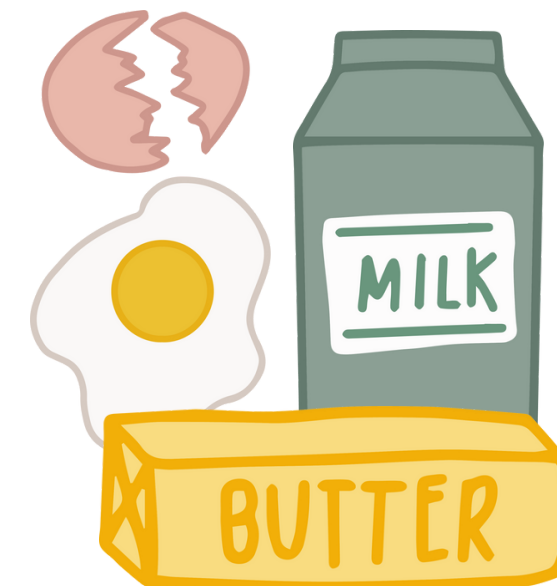
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GENES

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

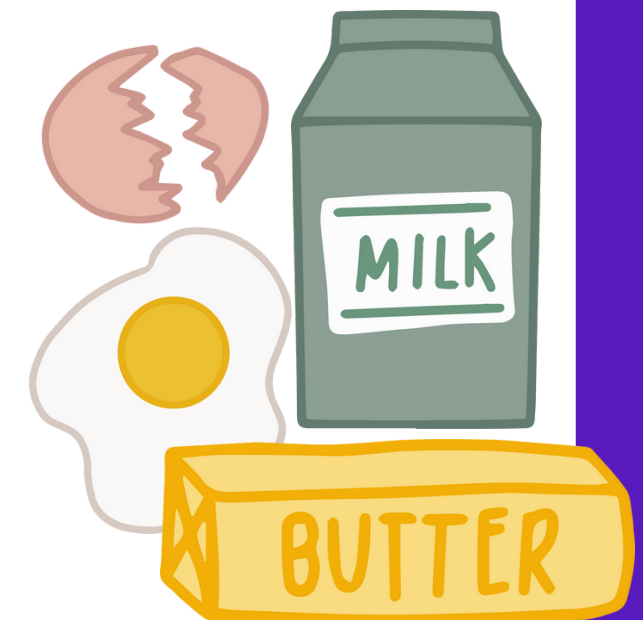
DNA Alphabet =
Letters used to write out a recipe

Gene = Recipe



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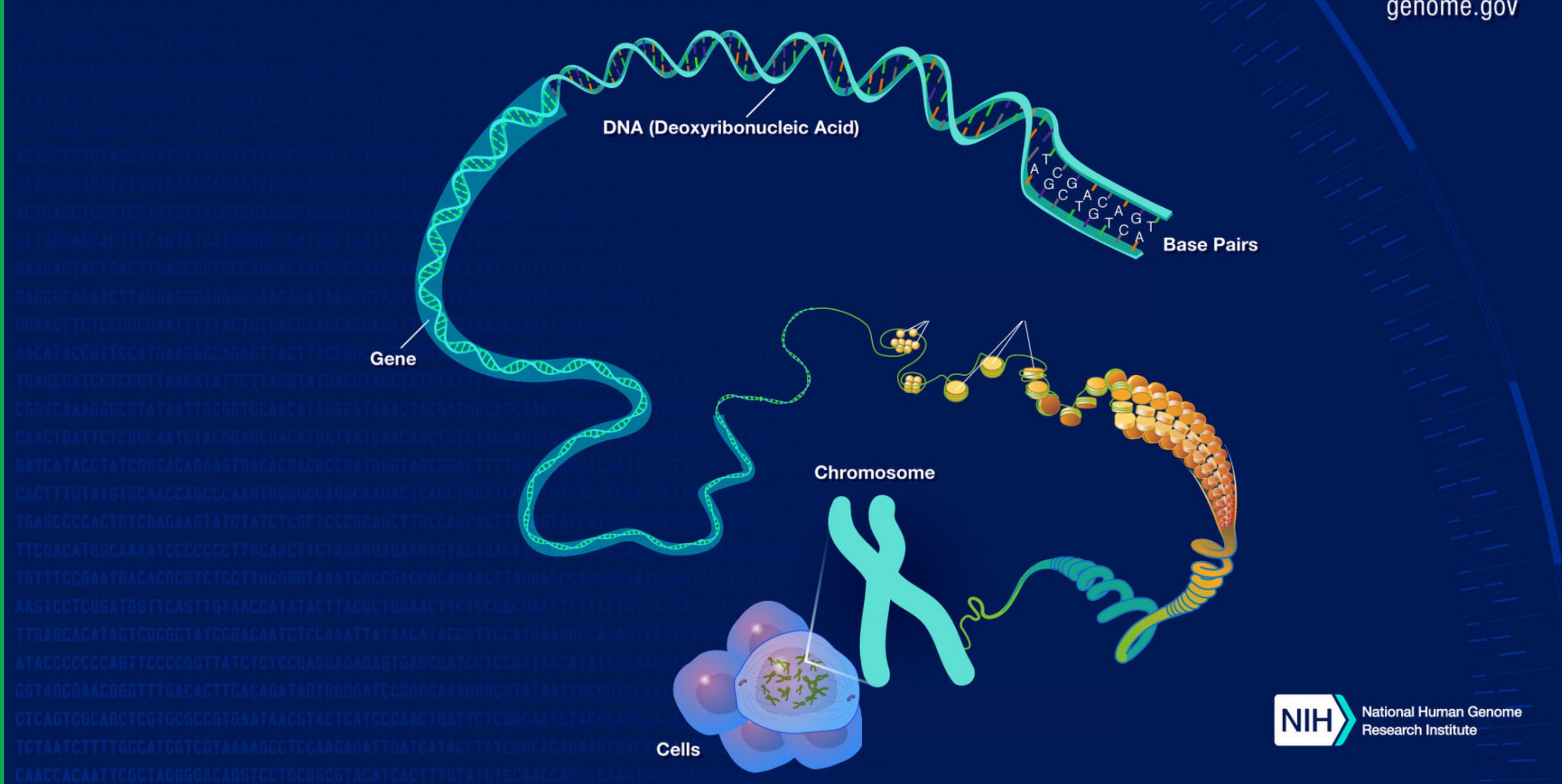
Chromosome = Recipe Book



Genome = Set of Recipe Books

A Brief Guide to Genomics

NHGRI FACT SHEETS
genome.gov



NIH National Human Genome
Research Institute

source: www.genome.gov/about-genomics/fact-sheets/A-Brief-Guide-to-Genomics

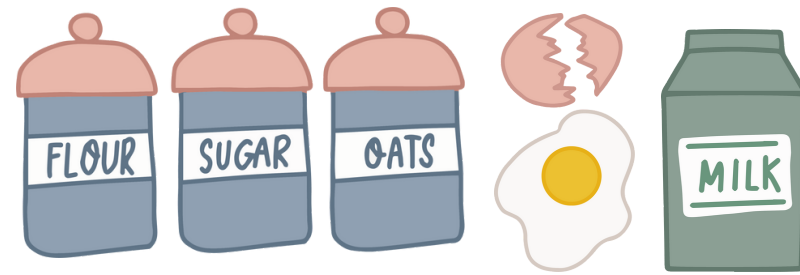
How do genetic mutations and variants lead to health challenges and disability?

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



Chocol**ate**



Gene = Recipe

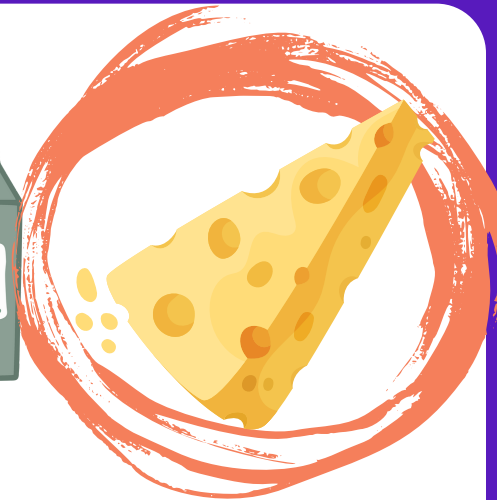


Chocolate
Cake



DNA =

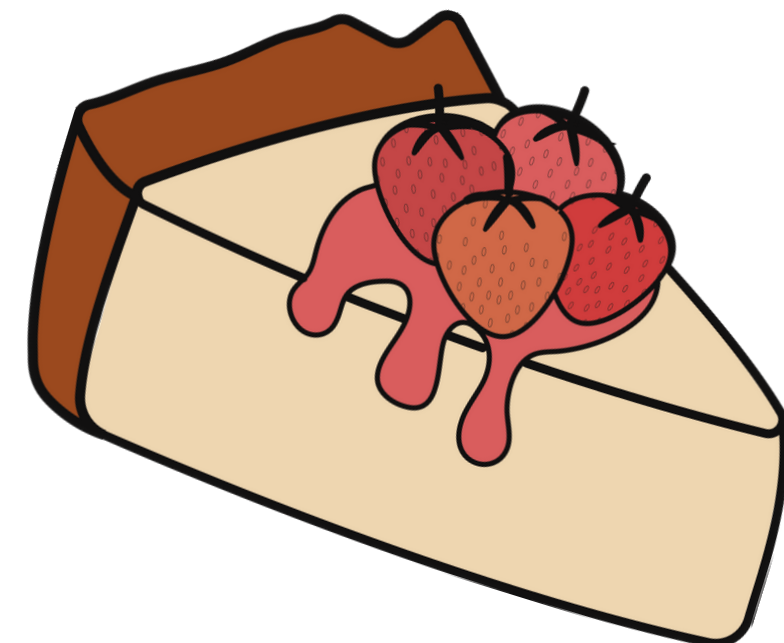
Letters used to
write out a recipe



Chees**e**



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 may not be produced in a high enough amount or may take a different shape or format. Sometimes these differences in quantity or function 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.

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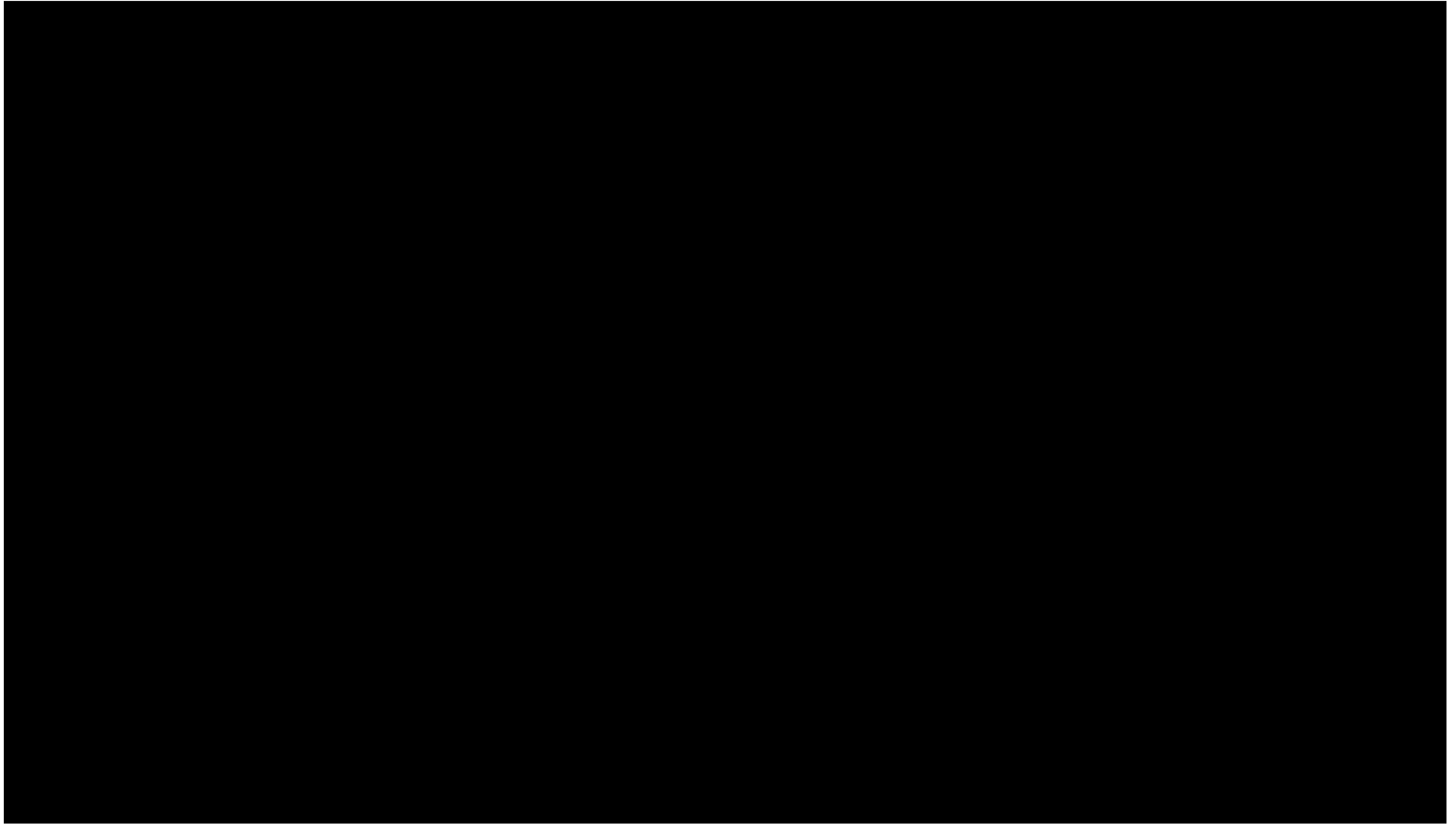
**A VIDEO:
VARIANTS OF UNCERTAIN
SIGNIFICANCE (VUS)**



You Tube Link:

<https://www.youtube.com/watch?v=QHrsia48fZA>

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



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**