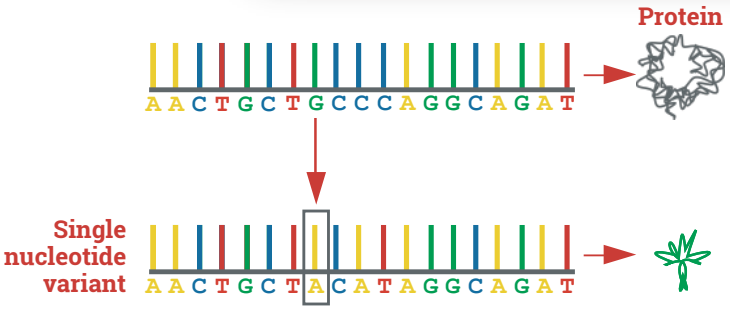
INTERNATIONAL ALLIANCE
OF ALS/MND ASSOCIATIONS

INTRODUCTION TO GENETICS

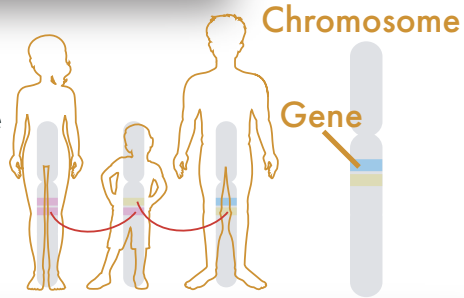
WHAT IS GENETIC VARIATION?



- Your DNA is made up of individual units. There are 4 types of **DNA units: A, T, C, and G.**
- Everyone has the same genes, but a gene can be spelled out slightly differently in different individuals, due to genetic variation.
- These are considered different "versions" of the genes, and give us our unique traits as humans.
- When your DNA is copied, mistakes can happen and this is a normal process.
- When a mistake in your DNA occurs, and results in a detrimental outcome such as a disorder, this is called a **mutation or pathogenic variant.**
- These detrimental mistakes can be caused by a change in your DNA units (such as a **single nucleotide variant)**, a portion of your DNA deleted or inserted, or a small region of DNA units repeated multiple times.

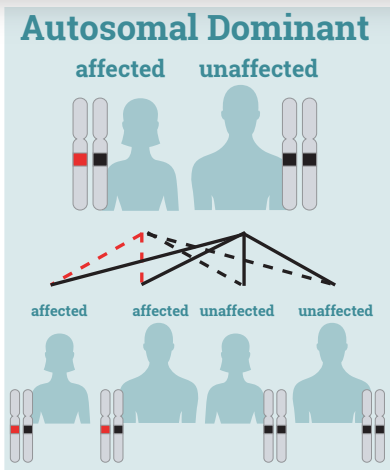
HOW ARE GENES INHERITED?

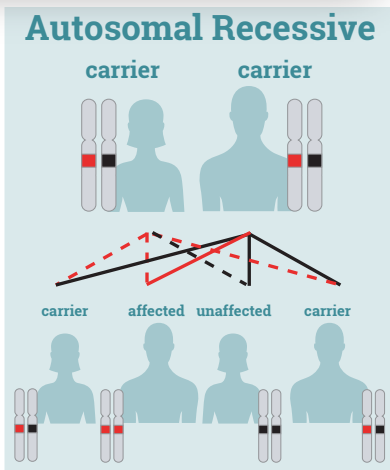
- You have 46 chromosomes in which your DNA is packaged.
- You inherited one set of 23 chromosomes from your mother, and the remaining set of 23 chromosomes from your father.
- Depending on which version of a gene you inherited from each parent, the gene will result in a different trait.



HOW DO WE INHERIT GENETIC DISORDERS?

- Some genetic disorders only need one copy of a mutant version of the gene to be present for an individual to be affected by the disorder. This is called **autosomal dominant inheritance.**
- Some genetic disorders need two copies of a mutant version of the gene to be present for an individual to be affected by the disorder. This is called **autosomal recessive inheritance.**
- Some people have a copy of a mutant version of the gene, but are unaffected by the disorder. The mutant version of the gene can be passed on, and such people are called **carriers**.





These resources have been developed by the International Alliance of ALS/MND Associations.
The material has been adapted by Kristiana Salmon and Helmut Bernhard from a presentation given by Dr. Kelly Williams, Macquarie University, Australia