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. These letters are called nucleotides.

In some parts of the instruction manual there are short strings of letters that repeat. These short strings of letters may include three (tri-), four (tetra-), five (penta-), six (hexa-), or more letters that repeat many times over. The number of repeats can vary between people even within the same family, but most people have a similar number of repeats. A repeat expansion is a genetic change where the number of repeats someone has increases in number. This can disrupt the instruction manual causing the gene to make either an abnormal protein, too much or too little protein, or no protein at all, which is what can cause some genetic conditions.

For example, a gene's letter sequence may have a three letter repeat (also called a trinucleotide repeat) that spells CAT over and over again. When someone has a typical amount of repeats, the gene produces a normal protein.

CAT CAT CAT = normal protein

Sometimes people can have extra repeats, but they don't affect how the instructions read, and thus don't create a problem.

CAT CAT CAT CAT CAT CAT = normal protein

When someone has too many repeats, that can change how the body reads that gene's instructions. This change impacts the protein that is produced, which is what can in turn cause some genetic conditions.

CAT CAT CAT CAT CAT CAT CAT CAT CAT CAT CAT = abnormal, too much/little, or no protein

Genetic conditions caused by this type of change are called repeat expansion disorders. For these types of conditions, we typically know how many repeats someone would need to have in order for that individual to be affected. For some (not all) of these disorders, we also know individuals can show different signs or symptoms, or be diagnosed at different ages based on the number of repeats they have.

Compared to other types of genetic changes that often are the same between family

members, repeat expansions can differ between family members. When there is a repeat expansion disorder in a family, family members may be affected differently based on the number of repeats they have. This is caused by what is called 'instability' of the repeat. It's also possible to see future generations be more severely affected than prior generations due to the repeat expansion growing as it's passed down in the family (called anticipation). For example, there may be a mother and daughter who are affected differently:

Mother = CAT CAT CAT CAT CAT CAT CAT CAT CAT CAT = mild symptoms

Daughter = CAT CAT CAT CAT CAT CAT CAT CAT CAT CAT CAT CAT CAT CAT CAT CAT CAT CAT CAT CAT CAT CAT = severe symptoms

The language that is used to describe the different categories of repeats outside of what is considered normal or typical varies depending on what genetic condition you're talking about. Possible descriptions may include:

- Normal/typical: repeat size is within the typical range; the gene is not impacted and works properly. These individuals are not affected.
- Mutable normal or intermediate: repeat size is on the higher side of the typical range; the gene is not impacted and works properly, but there is a higher risk the repeat could expand in future generations. These individuals are typically unaffected.
- Premutations or incomplete penetrance: repeat size is increased above what is typical, and the gene may be impacted. Some individuals may experience specific signs or symptoms, while others may not.
- Abnormal (also called full mutation or fully penetrant): repeat size is in a range where the gene is impacted and unable to work properly. All individuals with a full mutation will have signs or symptoms, but there can be variability in the severity (e.g. mild, classic, severe) and the age that someone is diagnosed.