Genetics Overview

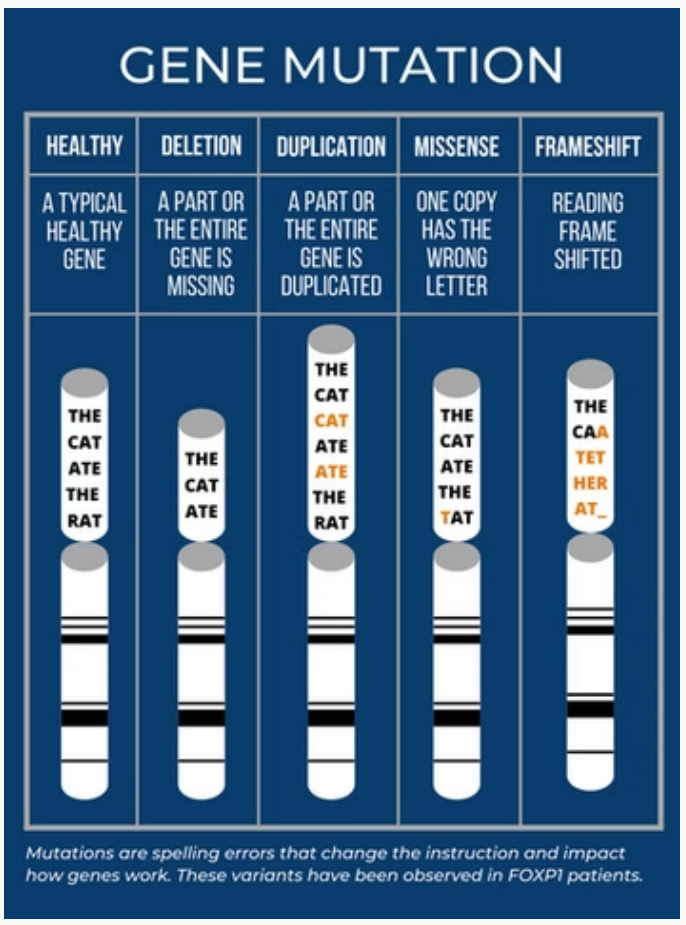
Genes are the body's instructions for building proteins, much like a recipe guides you in making a cake. FOXP1 is a gene on chromosome 3 that directs the production of a protein crucial for development. If this gene is mutated, the "recipe" is altered, which can lead to various developmental and medical issues.

Genetic testing, like whole exome sequencing, can identify changes in a person's entire genetic code, while panel tests focus on specific genes linked to certain conditions (e.g., autism, intellectual disability, epilepsy). These tests have become more accessible, helping doctors diagnose conditions like FOXP1 syndrome earlier.

Most FOXP1 mutations are "de novo," meaning they occur spontaneously during early development and are not inherited from either parent. Genetic testing can confirm this, and if the mutation is de novo, the chance of having another child with the same mutation is very low. However, the affected child could pass the mutation on to their own children later in life.

Gene Mutation

In the alphabet, different letters can be combined to produce useful and meaningful words – but only if the letters are put together in the correct way. The same works for DNA. Certain nucleotides (the base unit of DNA) whether it is adenine (A), thymine (T), cytosine (C), or guanine (G) can be arranged in a specific manner to form the FOXP1 gene, which in turn encodes a specific protein. A missense mutation is when there is a single DNA nucleotide switch in the gene. Sometimes such a change is silent and has no effect on the protein encoded by the DNA. Other changes result in an amino acid change in the protein the gene codes for, which sometimes can profoundly change the way the protein functions.



Mutations are spelling errors that change the instruction and impact how genes work. These variants have been observed in FOXP1 patients.

Nonsense and Frameshift Mutations

Like a missense mutation, a nonsense mutation also involves a single alteration in the DNA. However, in the case of a nonsense mutation, this single change results in the production of a stop codon, (stop codons signal the end of the protein coding sequencing), there by terminating protein synthesis (process in which cells make proteins) prematurely. The protein may not be made at all, or a shortened version of the protein may be made. Similarly, frameshift mutations arise when one or a small number of nuclootides are deleted or inserted, causing the 3-letter "words" of the gene to come out of alignment; this kind of mutation can introduce a premature stop codon, or can cause all the "downstream" words to be misread. Either outcome can result in major disruption to the way the protein functions.

Missense Mutation

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