Molecular basis of inherited disease

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Thus, frameshift mutations typically exhibit complete loss of normal protein structure and function. Fragile X syndrome results from changes in the number of a CGG n repeat in the coding sequence of the FMR-1.

Molecular Basis of Inheritance notes

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Molecular Basis of Inheritance notes

It is known now that spontaneous mutations arise from a variety of sources, including errors in , spontaneous lesions, and transposable genetic elements. However, it is always true that mutations that reduce or eliminate gene function loss-of-function mutations are the most abundant class.

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