WHITEPAPER

Biocare Basics: Deletion Mutations

BIOCARE MEDICAL

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Immunohistochemistry (IHC) is designed to locate and label cellular structures in order to detect disease states. However, in the case of diseases such as cancer, genetic mutations may be the underlying cause, and so the specific type and location of these mutations are of diagnostic and prognostic interest. In situ hybridization (ISH) procedures may be utilized to detect specific mutations and yield diagnostic and prognostic information through the application of molecular probes. One type of mutation that ISH probes may be able to detect is deletion mutations.

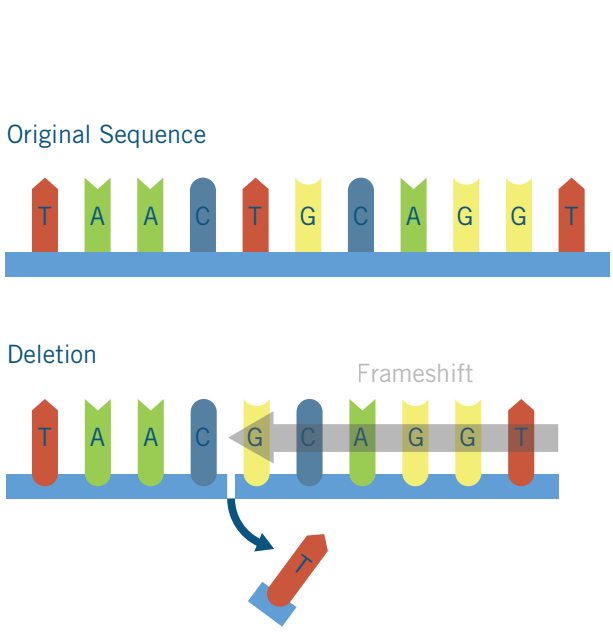
A deletion mutation is a type of mutation where a segment of DNA is not copied during replication and is therefore deleted from the genetic sequence.[4] The size of the deleted segment can be anywhere from a single nucleotide to an entire section of a chromosome.[4] Such mutations can be hemizygous (one copy) or homozygous (both copies) and have been implicated in a large number of diseases, including the majority of cystic fibrosis cases.[4]

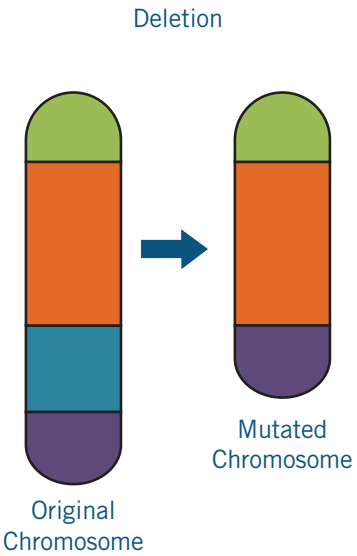
Frameshift mutations can also be a devastating result of deletion mutations. These mutations arise when the genetic deletions do not occur in multiples of three.[4] This is because the genetic code is read by the cell in groups of three bases at a time, known as "triplet codons."[4] If a deletion mutation disrupts this reading frame by throwing off the codon count, then the entire genome sequence will be read incorrectly.[4]

Research has shown that human cancer tends to result from an accumulation of genetic errors or mutations that eventually transform normal cells into cancerous tumor cells.[1] This may take the form of cancer-promoting mutations combined with the loss of tumor suppressor genes.

For example, p16 INK4a is a tumor suppressor protein that has been found to be involved in the pathogenesis of a variety of malignancies.[3] Analyses of the p16 INK4a gene have revealed the presence of homozygous deletions or frameshift mutations in several human cancers.[2,3] Likewise, mutations or deletions in the gene for transcription factor and tumor suppressor SMAD4 have also been implicated in multiple human cancers, including cholangiocarcinoma, colorectal cancer, head and neck cancer, and pancreatic cancer.[6]

In addition to IHC antibodies that can detect the presence of proteins coded by genes of interest, Biocare Medical offers a line of ISH probes designed to detect the presence or absence of genes of interest directly.





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