WHITEPAPER

Deletion Detection with del-TECT Probes

BIOCARE MEDICAL

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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.[4] Deletion mutations are one type of mutation that can be detected via fluorescence in situ hybridization (FISH).

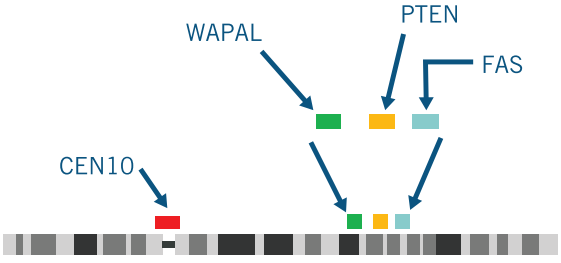
A deletion mutation occurs when a segment of DNA is not copied during replication and is therefore "deleted" from the genetic sequence.[5] The size of the deleted segment can be anywhere from a single nucleotide to an entire section of a chromosome.[5] Such mutations can be hemizygous (one copy) or homozygous (both copies) and have been implicated in a large number of diseases.[5] Homozygous deletion mutations in cancer often target tumor suppressor genes, such as PTEN, which can increase malignancy.[2]

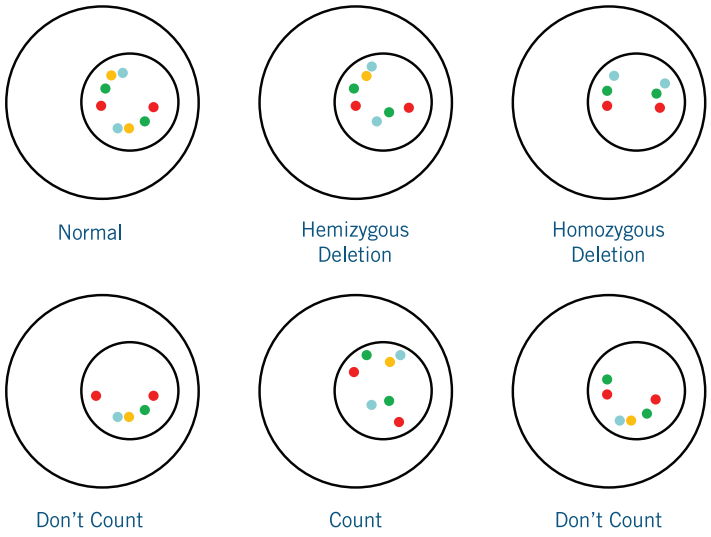
Since deletion mutations have been instrumental in identifying the causes of certain cancers, there is a great need for accurate detection and identification.[3] However, proving that a target sequence is truly absent presents a unique challenge. Cells do not sit neatly within the same plane, and so when a sample is cut into standard 4-6µm sections, a substantial number of cells will have their nuclei wholly or partially cut away.[6] This is referred to as cellular truncation.[6]

To address this issue, Biocare Medical provides a series of 4 color del-TECT FISH probes to significantly decrease the number of false positives caused by cell truncation. This is done by labeling probes on either side of the sequence of interest, which verify whether just the sequence of interest has been deleted or if the whole section of the chromosome has been cut away.

For example, the PTEN del-TECT (4 Color) FISH probe labels the PTEN Test probe orange. Then a second probe, Probe A (WAPAL/BMPR1A), which is located centromeric to the test probe, is labeled in green. A third probe, Probe B (FAS), is located telomeric to the test probe and is labeled in aqua. The combination of the three colored probes (green, orange, and aqua) in close proximity assists not only in eliminating the possibility of truncation of this region but also helps determine the actual size of the deletion. Finally, a centromere 10 probe labeled in red is included to help determine if chromosome 10 monosomies or polysomies are present.

Detection with del-TECH Probes





To learn more about Biocare's menu of 4 Color del-TECT probes, please visit us at biocare.net or call 1-800-799-9499.

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