2025/06/29 01:30 1/1 p.V600E mutation

p.V600E mutation

The p.V600E mutation is a specific genetic mutation that occurs in the BRAF gene. This mutation results in the substitution of valine (V) with glutamic acid (E) at position 600 of the BRAF protein.

BRAF is a protein that plays a critical role in cell signaling pathways that regulate cell growth and division. The p.V600E mutation in BRAF results in a constitutively active protein, meaning that it is constantly "switched on" and signals cells to grow and divide uncontrollably.

The p.V600E mutation is particularly important in cancer biology, as it has been found in a variety of human cancers, including melanoma, colorectal cancer, papillary thyroid cancer, and non-small cell lung cancer. In fact, it is one of the most common mutations found in melanoma, a type of skin cancer.

Because the p.V600E mutation results in a constitutively active protein, it can be targeted by drugs that specifically inhibit the activity of mutated BRAF proteins. These drugs are known as BRAF inhibitors and have been shown to be effective in treating some cancers that harbor the p.V600E mutation. However, some tumors may develop resistance to these drugs, highlighting the need for further research and development of new therapies targeting this mutation.

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