A driver mutation, also known as an oncogenic or cancer driver mutation, is a specific genetic alteration in a cell's DNA that contributes to the initiation, development, or progression of cancer. These mutations provide a selective growth advantage to affected cells, promoting uncontrolled proliferation and survival. Driver mutations are key factors in the transformation of normal cells into cancerous cells.

Key characteristics of driver mutations include:

Functional Impact: Driver mutations result in changes to the structure or function of a protein, typically one involved in cell growth, division, or survival. These changes are often gain-of-function alterations that enhance the protein's activity, making it an oncogene.

Clonality: Driver mutations are usually present in a significant proportion of cancer cells, and they are considered clonal, meaning that they are found in the majority of tumor cells. Subclonal mutations, which occur in only a subset of cells, are less critical in driving cancer development.

Ubiquitous Presence: Driver mutations are present early in the disease's development and are found in most, if not all, tumor cells. They often represent the primary “driver” event behind cancer formation.

Selective Advantage: Cells with driver mutations have a growth or survival advantage over normal cells. This advantage allows them to proliferate more rapidly, ultimately leading to tumor formation.

Variability: Different types of cancer and even different patients with the same cancer type may harbor distinct driver mutations. Identifying the specific driver mutations in a particular cancer is crucial for developing targeted therapies.

Driver mutations can occur in various genes and signaling pathways. Some of the best-known examples include mutations in the RAS and BRAF genes in melanoma and colorectal cancer, the EGFR gene in lung cancer, and the BRCA genes in breast and ovarian cancer. Targeted therapies have been developed to block or inhibit the effects of driver mutations in these and other cancer types, making them essential for personalized cancer treatment.

In summary, driver mutations are genetic alterations that promote cancer development by providing growth and survival advantages to affected cells. Identifying these mutations and understanding their functional consequences is fundamental to cancer research, diagnosis, and the development of targeted therapies.