Types of Genetic Mutations

Germline Mutations: A hereditary mutation, passed directly from a parent to a child at the time of conception. Cancer caused by germline mutations is called inherited cancer and accounts for about 5% to 20% of all cancers.

Somatic Mutation: Mutations that can occur in any of the cells of the body but are not hereditary. These mutations may, in some cases, cause cancer or other diseases.

Breast Cancer Subtypes	Breast Cancer Genetic Mutations
▪ HER2-enriched breast cancer ▪ Luminal A breast cancer ▪ Luminal B breast cancer ▪ Luminal B-like breast cancer ▪ Triple-negative breast cancer	▪ **BRCA (BR**east **CA**ncer gene). **BRCA1** and **BRCA2** genes may increase one's risk of developing breast cancer and may impact the cancer's aggressiveness and treatment options. ▪ Other **genetic mutations** linked to breast cancer include: CHD1, CHEK2, PALB2, ATM, P53.

Glossary Terms

Biomarker Testing (molecular testing): Laboratory testing that identifies certain genes, proteins, or other molecules in a sample of tissue, blood, or other body fluid. In cancer, it may also be used to evaluate treatment or to make a prognosis.
Gene Mutation: A permanent change in the DNA sequence that makes up a gene. Mutations can be acquired or inherited and may impact treatment choices.
Genomic Testing (Molecular profiling): Laboratory testing that identifies certain gene mutations, proteins, chromosomal abnormalities, and/or other molecular changes that are unique to an individual's disease. In cancer, it may be used to evaluate treatment or to make a prognosis.
Immunotherapy: Type of therapy that harnesses one's own immune system to help the body fight cancer, infection, and other diseases.
PD-L1 Expression: PD-L1 is a receptor expressed on the surface of T cells. The presence of PD-L1 indicates that a patient may respond to immunotherapy.
PARP Inhibitors: A type of targeted treatment that inhibits the enzyme poly (ADP-ribose) polymerase.
Shared Decision-Making (SDM): Process of communication by which patients and clinicians collaborate to make healthcare decisions. The process encourages patients to take a more active role in their care and treatment.
Targeted Therapy: A type of personalized medicine that works by blocking specific mutations and by preventing cancer cells from growing and dividing, without affecting normal cells.
Triple-negative breast cancer: The cancer cells have tested negative for hormone epidermal growth factor receptor 2 (HER2), estrogen receptors (ER), and progesterone receptors (PR).

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