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BRCA1 Gene Mutation

National Cancer Institute

Source

National Cancer Institute. *BRCA1 Gene Mutation*. NCI Thesaurus. Code C19635.

A mutation that is typically a heritable, permanent change in the nucleotide sequence of the BRCA1 gene. Single nucleotide substitutions and small deletions or insertions (1-20 bases) account for the majority of mutations in the BRCA1 gene. Approximately 75% of these alterations result in a truncated form of the breast cancer type 1 susceptibility protein. Mutations in the BRCA1 gene predispose individuals to breast and ovarian cancers.