

[Open Peer Review on Qeios](#)

# Mutation Analysis

National Cancer Institute (NCI)

## Source

*National Cancer Institute (NCI). Mutation analysis.*

A germline genetic testing method targeted to detect a specific variant or mutation (such as a deleterious MSH2 variant previously identified in a family), panel of variants (such as the 3 BRCA pathogenic variants comprising the founder mutation panel for individuals of Ashkenazi Jewish ancestry) or type of variant (such as large deletions or insertions in the BRCA1 gene). This type of testing is distinct from complete gene sequencing or variant scanning. The latter are designed to detect most variants in the region being tested. Current usage also applies this term to any genetic test.