

HOW TO TEST GERMLINE VS SOMATIC TESTING

Germline INHERITED MUTATIONS



CARRIED IN ALL
CELLS OF THE BODY



DETECTED IN A
BLOOD OR SALIVA
SAMPLE

Germline mutations, such as inherited *BRCA* mutations, can be passed on to the next generation.

Germline mutations can be detected in a blood sample, cells in a cheek swab, or saliva. The sample is sent to a genetic laboratory for analysis and results are available in as early as 7 to 10 days.²⁻⁴

Somatic ACQUIRED MUTATIONS



USUALLY ONLY
PRESENT IN THE
TUMOR



DETECTED WITH A
TUMOR BIOPSY

A biopsy of tumor tissue must be analyzed to detect somatic mutations.

BRCA mutations detected in DNA extracted from a sample of tumor tissue can either be germline or somatic. A subsequent blood test can determine what type of mutation it is. The entire testing process may take several weeks.^{2,5,6}

References: **2.** Lynch JA, Venne V, Berse B. Genetic tests to identify risk for breast cancer. *Semin Oncol Nurs.* 2015;31(2):100-107. **3.** Cook-Deegan R, Niehaus A. After Myriad: genetic testing in the wake of recent Supreme Court decisions about gene patents. *Curr Genet Med Rep.* 2014;2:223-241. doi:10.1007/s40142-014-0055-5. **4.** Myriad. HBOC FAQs: BRCA1 or BRCA2 mutation and BRCAAnalysis. Myriad Web site. <https://myriadgenetics.eu/healthcare-professional-treating-diseases/hereditary-cancer-testing/hereditary-breast-and-ovarian-cancer-hboc-syndrom/hboc-faqs/>. Accessed June 15, 2018. **5.** Vergote I, Banerjee S, Gerdes A-M, et al. Current perspectives on recommendations for *BRCA* genetic testing in ovarian cancer patients. *Eur J Cancer.* 2016;69:127-134. **6.** FoundationOne. Genomic testing. <https://www.foundationmedicine.com/genomic-testing/foundation-one>. Accessed June 15, 2018.