

TEST ID: ESR1

ESTROGEN RECEPTOR 1 (*ESR1*) MUTATION ANALYSIS, TUMOR

USEFUL FOR

- ▶ Assisting in the clinical management of patients with metastatic breast cancer by assessing gene targets in the *ESR1* gene
- ▶ Stratifying prognosis of metastatic breast cancer

TESTING ALGORITHM

When this test is ordered, slide review will always be performed at an additional charge.

CLINICAL INFORMATION

The *ESR1* gene encodes an estrogen receptor that regulates cell growth through activation of downstream signaling pathways upon binding of estrogen. Tumors demonstrating estrogen receptor expression (ER-positive) are candidates for endocrine therapy such as selective estrogen receptor modulators (SERM) and aromatase inhibitors. *ESR1* mutations are rarely observed in primary tumors; however, mutations in the ligand-binding domain of *ESR1* have been reported at a higher frequency in ER-positive metastatic breast tumors. Preclinical data suggests that *ESR1* mutations mitigate resistance to aromatase inhibitors and decrease sensitivity to SERMs and estrogen-receptor downregulators. Studies also suggest that *ESR1* mutations are an independent indicator of poor prognosis.

This test assesses for somatic mutations in the ligand-binding domain of the *ESR1* gene associated with acquired resistance to endocrine therapy (ie, aromatase inhibitors) in patients with ER-positive metastatic breast cancer.

INTERPRETATION

An interpretive report will be provided.

REFERENCE VALUES

An interpretative report will be provided.

ANALYTIC TIME

12 days

SPECIMEN REQUIRED

Detailed on back side of this sheet.

ESTROGEN RECEPTOR 1 (ESR1) MUTATION ANALYSIS, TUMOR**SPECIMEN REQUIRED****PREFERRED****Specimen Type**

Tissue block

Collection Instructions

Submit a formalin-fixed, paraffin-embedded tissue block

ACCEPTABLE**Slides**

1 stained and 10 unstained slides

Collection Instructions

Submit 1 slide stained with hematoxylin and eosin and 10 unstained slides (nonbaked, charged slides preferred) with 5-micron thick sections of the tumor tissue.

Specimen Type

Cytology slide (Direct smears or ThinPrep)

Slides

1-2 slides

Collection Instructions

Submit 1-2 slides stained and coverslipped with at least 5,000 total nucleated cells

Additional Information

Cytology slides will not be returned.

SUPPORTIVE DATA

This next-generation sequencing assay detects somatic mutations that can be used to assist in the clinical management of metastatic breast cancer patients.

This assay has been shown to be very reproducible, having a 100% concordance for intra- and interassay reproducibility experiments. All somatic mutations that had been previously identified by various other molecular methods were detected by this assay during accuracy studies. No pathogenic variants were detected in known mutation negative samples.



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