

ESR1

Gene mutation testing Next Generation Sequencing (NGS) based liquid biopsy test

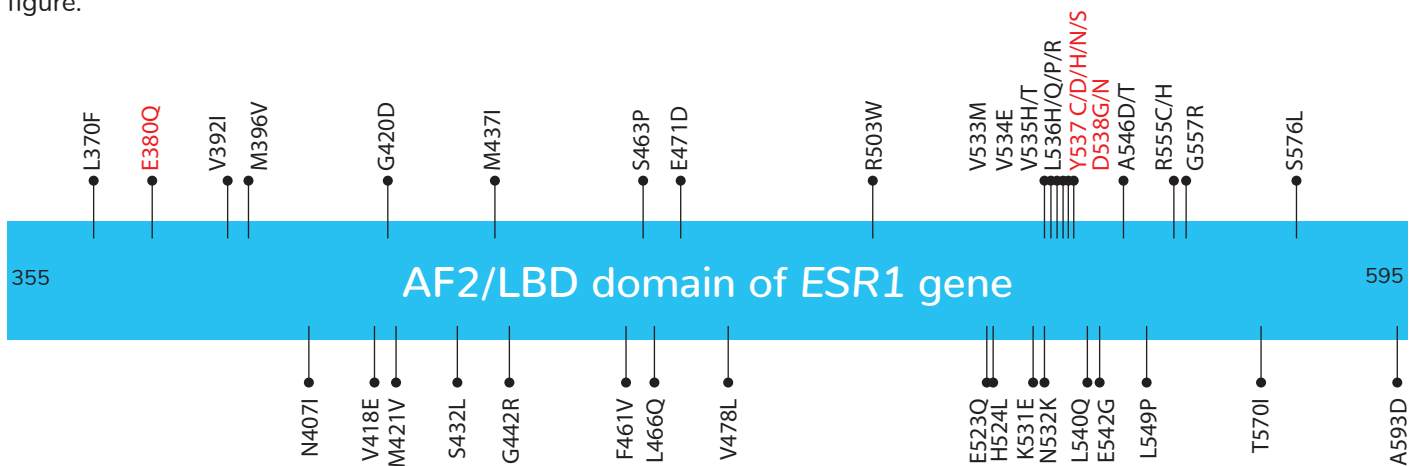


Minimally invasive assay performed on the blood (plasma cfDNA) to detect all reported hotspot mutations in ESR1 gene

- Helps in the detection for acquired resistance in advanced hormone-positive breast cancer patients who have progressed on endocrine treatment
- Aids in selection of patients where alternate combination therapy options can be used
- Detects the mutations in plasma cfDNA using blood samples; Serial monitoring is possible without rebiopsy

Medgenome's ESR1 gene test by NGS- Liquid Biopsy

Hotspot mutations reported in Ligand Binding domain (LBD) of ESR1 gene are covered in this assay as shown in the figure.

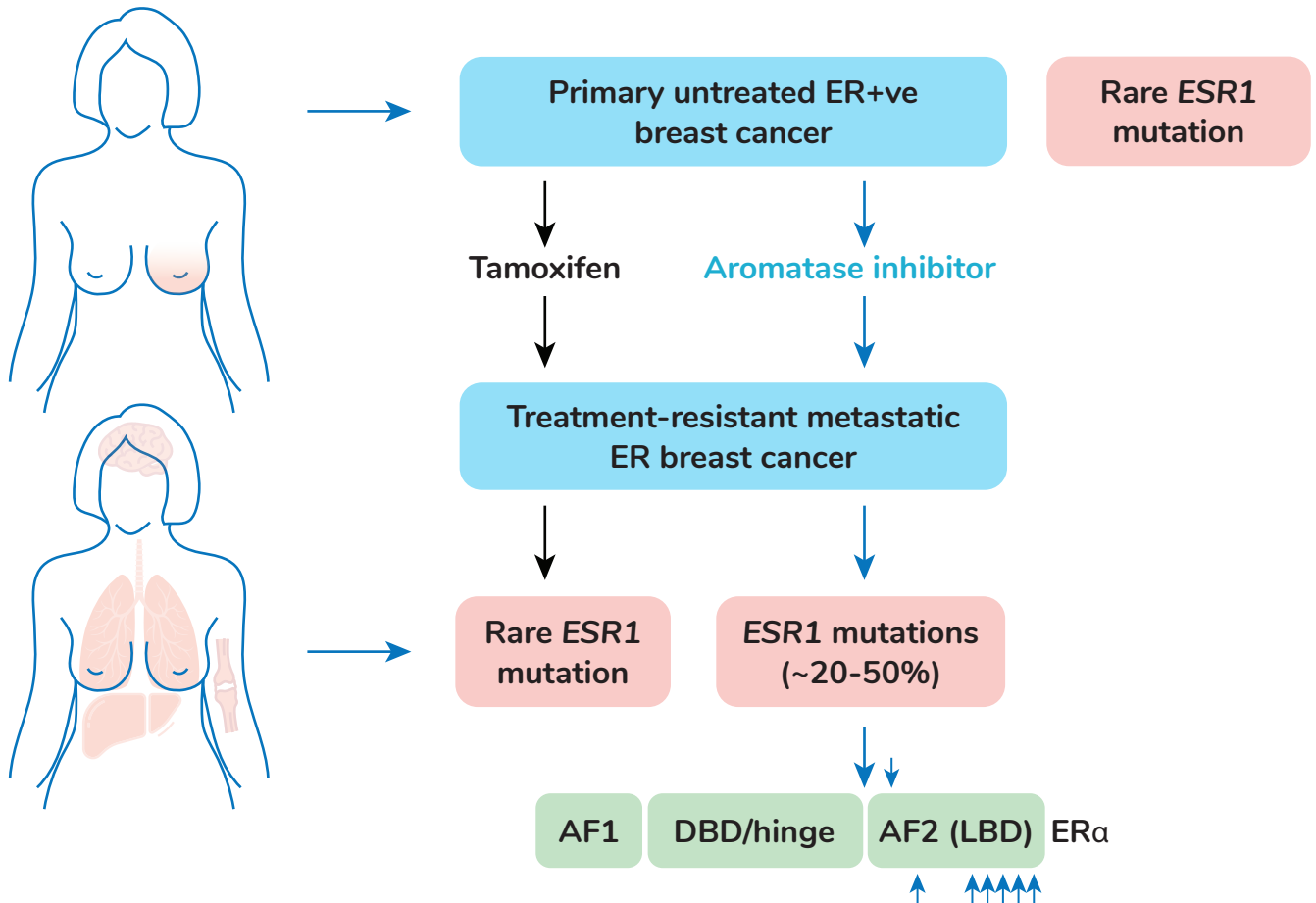


Test Details

Name of the test	Test code	Sample type	Shipping Conditions	TAT
ESR1 gene testing by NGS -Liquid Biopsy (Hot Spot Mutations)	MGM2732	Peripheral Blood in Streck Tube (10ml X 2)	Ship same or next day at room temperature. Do not freeze or refrigerate	14 Working days from sample receipt at the laboratory to result

Importance of ESR1 gene testing

- 70% of breast cancers are hormone positive/ luminal subtypes which are treated with Endocrine therapy (ET).
- **ESR1** mutations play an important role in resistance to endocrine therapy, leading to advanced breast cancer.
- **ESR1** mutations present rarely in primary treatment-naïve ER+ breast cancer, whereas they are highly prevalent in metastatic tumors, suggesting that these mutations may potentially arise from rare clones of primary tumors through clonal selection against ET.



Oesterreich S, Davidson NE. The search for *ESR1* mutations in breast cancer. *Nature Genetics*. 2013 Dec;45(12):1415-1416. DOI: 10.1038/ng.2831. PMID: 24270445; PMCID: PMC4934882

Assay specifications:

Cancer type	Advanced Breast cancer
Assay type	Next generation Sequencing
Sequencing Platform	Illumina platforms
Limit of detection	0.25% VAF for SNV and InDels
Average depth of sequencing	>25000X ; >2000X post UMI correction
Analytical sensitivity	100%
Analytical specificity	100%