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MC-5293

Homologous Recombination Repair - HRR Panel

The Homologous Recombination Repair (HRR) pathway plays an important role in double strand break, which is the major cause of cancer development. HRR genes are involved in repair of damaged DNA and mutations in these genes can lead to deficiency in repair mechanism. It has been demonstrated that loss of function of HRR genes (e.g. BRCA1, BRCA2, PALB2) and homologous recombination deficiency (HRD) cause a higher risk of developing cancer. Breast and Ovarian cancers patients with HRR gene mutations show higher response to PARPi and platinum-containing therapies

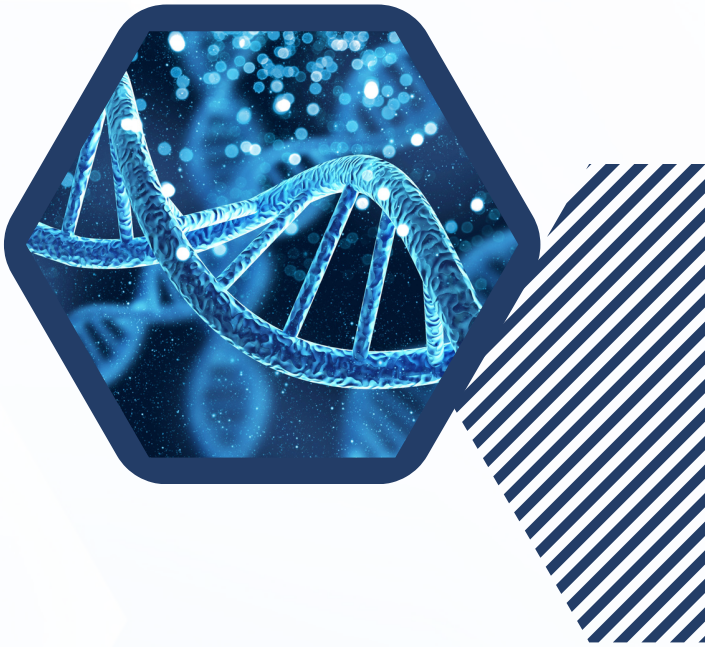
WHY TEST

- Hereditary Risk Assessment
- Targeted Therapy (guidance)
  - Prognostic and Predictive biomarker for platinum-based chemotherapy and PARP inhibitor therapy
  - Mutation in HRR genes (HRR defective tumors) in Triple Negative Breast Cancer is predictive of complete Pathological Response.
  - In relapsed / recurrent setting of ovarian cancers: Niraparib and Rucaparib have shown significant improvement in PFS

WHO SHOULD GET TESTED

- Family history of Breast/ Ovarian Cancer
- Breast/ Ovarian Cancer patient with negative BRCA1/ 2 test
- Ovarian cancer patient who may benefit from PARPi maintenance therapy
- Women with ovarian cancer resistant to platinum therapy
- Men with castration resistant prostate cancer who have progressed in prior treatment

METHOD : NGS



HRR NGS Panel is intended for qualitative detection of single nucleotide variants (SNVs) and insertions and deletions (Indels) variants in protein coding regions and intron/exon boundaries of 30 HRR genes and SNVs/Indels variants in hotspot regions of 4 driver genes (BRAF, ERBB2, KRAS and PIK3CA).

GENES

ATM ATR BARD1 BRCA1 BRCA2 BRIP1 CDK12 CHEK1 CHEK2 FANCA FANCC FANCD2 FANCE FANCF FANCG FANCL MLH1 MRE11A MSH2 MSH6 NBN PALB2 PMS2 RAD50 RAD51 RAD51B RAD51C RAD51D RAD54L TP53 BRAF, ERBB2, KRAS, and PIK3CA

SAMPLE

3-5 ml Blood in EDTA tubes, fresh-frozen tumor tissue or formalin-fixed paraffin embedded (FFPE) tumor tissue.
TAT: 3 weeks

*Quantitative or HRD (homologous repair deficiency) score assessment is not possible through this assay.

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