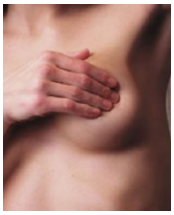
BRCA 1 and 2 Test

Genetic susceptibility to hereditary breast and ovarian cancer



Breast and ovarian cancer are associated with a hereditary component in 5-10% of the cases. This type of cancer is characterized by its incidence at early ages, even before 40 years of age. The origin of this susceptibility resides frequently in mutations of the BRCA 1 and 2 genes. Both are tumor suppressor genes, when their functionality is altered an accumulation of other genetic alterations is produced that finally cause the tumor process.

The BRCA Test makes possible the analysis of the BRCA 1 and BRCA 2 genes in search of mutations in them. The identification of a pathological mutation in the germ line confirms the hereditary etiology of the cancer and has obvious clinical implications for the patient as well as for her family.

The detection of mutations in the BRCA genes in the patient allows detecting rapidly and economically this same mutation in the rest of the family members.

The presence of alterations in the BRCA 1 and 2 genes is sufficient for the carriers to present throughout their life a higher accumulated risk of developing breast and ovarian cancer. These mutations can predispose to other types of cancers such as prostate or pancreatic cancer.

BRCA 1 and 2 Test

The test is analyzed trough **Next Generation Sequencing (NGS)**. This technique allows to detect point mutations and small insertions or deletions distributed along BRCA 1 and BRCA2 genes. If any pathogenic mutation is detected, these will be confirmed by a second sequencing analysis.

The carriers of a mutation in the germ line in the BRCA genes have a risk of breast cancer during their lives of 45-80%. The probability of developing the disease increases with age, from 20% at 40 years of age, 37% at 50, 55% at 60 and over 70% at 70 years of age.

Around 15% of patient with hereditary breast cancer have large deletions/duplications in BRCA 1 and BRCA2 genes, which cannot be detected through sequencing. If no pathogenic mutation has been identified it is recommended to perform large deletions/duplications study in both genes through **MLPA technique (Multiplex Ligation-dependent Probe Amplification)**.

Advantages of BRCA 1 and 2 Test

The identification of mutations in the BRCA 1 and BRCA 2 genes makes it possible to implement monitoring measures in the patients diagnosed with breast and/or ovarian cancers and preventive measures in their family members who are carriers that have not developed the disease. The preventive measures can be applied effectively at early ages. Among these measures can be highlighted early diagnosis, prophylactic surgery or chemoprevention.

The complete sequencing of the BRCA 1 and BRCA 2 genes allows obtaining a risk assessment of developing hereditary breast and ovarian cancer and makes possible the realization of appropriate personalized monitoring of the patient and of her family members. This sequencing process usually means a very high cost due to the processing of the sample and to the amount of genetic information integrated.

Indications

The sequencing of the BRCA genes is indicated for patients with:

- Men or women with a family history of breast or ovarian cancer.
- Women with breast or ovarian cancer.
- Women who wish to know their genetic risk associated to this type of pathology.

Requirements

Fasting is not necessary.

Sample: 2 tubes of 3 mL of total blood EDTA.

Documentation: Specific Test requisition form and Informed consent, together with clinical report if possible.

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