Genetic test

BRCA1 & BRCA2

The test evaluates the predisposition of an individual to develop breast and ovarian cancer



BRCA 1 and BRCA 2 genes

Breast cancer is the most common cancer in women. It is estimated that approximately 7-8% of patients with breast cancer have a inherited genetic predisposition. Mutations of BRCA1 and BRCA2 genes are associated to hereditary breast cancer, these genes are also associated with an increased risk of developing ovarian cancer and other cancers.

The carriers of BRCA1 / BRCA2 mutations do not necessarily develop cancer but they have an increased risk compared to people who do not have mutations; they also have a 50% chance to transmit the mutation to their descendants.

Microgenomics test

Microgenomics genetic analysis is based on the new NGS technology - Next Generation Sequencing, through which you can perform complete and thorough analysis of your BRCA1 and BRCA2 genes.

The test does not require any complimentary analysis: it allows the identification of small DNA sequence variations as well as broaden gene rearrangements. This reduces the cost of the analysis and the reporting time; instead of the BRCA genes analysis carried out through two distinct tests, sequencing analysis and MLPA test. Microgenomics test is extremely accurate: the sensitivity and specificity are over 99%.

To perform the test you simply need a blood sample taken. The analysis and reporting of results are carried out in accordance with the recommendations of the Italian Society of Human Genetics (SIGU).

Technical specifications

The test analyses all the coding portions of BRCA 1 and BRCA 2 genes as well as intron/exon joints. Specifically, it is able to highlight both the sequence mutations and deletions/duplications of one or more exons of the BRCA genes.

SENSITIVITY: >99% [95% CI ≥ 99.02%] SPECIFICITY: >99% [95% CI ≥ 99.99%].

Why it's done?

The analysis of the BRCA1 and BRCA2 genes is suitable for women with a family history of breast or ovarian cancer, women with breast cancer in young age, women with bilateral breast cancer and some histological types of cancer (eg. Triple negative, multicentric).

Discovery of a BRCA1 or BRCA2 mutation helps establishing the proper prevention and surveillance lines, with a risk reduction through medical and/or surgical treatments. The identification of these gene alterations in young individuals leads to a reduction in mortality and also morbidity. Indeed these individuals are not normally included in population screening: if they are mutation carriers, they have a high risk of developing breast or ovarian cancer. Surveillance programs can significantly reduce mortality and it is possible to reduce morbidity through prophylactic surgery.

DID YOU KNOW THAT...

In male subjects BRCA2 mutations, and in rare cases BRCA1, are associated with an increased risk of breast (10%) and prostate cancer

Possible test results

PRESENCE OF PATHOGENETIC MUTATIONS

The test identified BRCA1 / BRCA2 mutations that confer a high risk of developing breast, ovarian or other cancers. Not all patients with pathogenic mutations develop a cancer, but they have an increased risk compared to the general population.



IDENTIFICATION OF MUTATIONS OF UNCERTAIN CLINICAL SIGNIFICANCE

Sometimes it may happen that the test detects mutations with an uncertain or unknown significance, that is, which is not possible to determine if they are benign variants or they confer an increased risk of developing cancer.

NO PATHOGENETIC MUTATIONS

The test did not identify BRCA1 and BRCA2 mutations that confer an increased risk of developing breast cancer or other cancers. These individuals still have a chance of developing cancer equal to the general population. In addition, a negative result does not exclude the presence of other risk factors for breast and ovarian cancer that cannot be identified with BRCA1 / BRCA2 testing.





MICROGENOMICS LABORATORY

Microgenomics is a highly specialized diagnostic centre, it provides molecular investigations.

The main goal is to offer high quality services by improving diagnostic methods in use, implementing innovative technologies, as well as monitoring the results for specific diseases. Microgenomics is an accredited laboratory to the Lombardy Region Health Service, it is certified ISO 9001:2008 and SIGUCERT – Human Genetics Italian Society Certification program.



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