DNK s.r.o.
diagnostické centrum

Laboratory diagnosis of genetic predisposition to breast cancer and ovarian cancer

Diagnostic kit for detection of mutations in BRCA1 and BRCA2 genes using real-time PCR

Breast cancer occupies a leading position in the structure of oncological morbidity and mortality in women. Ovarian cancer ranks 7th in terms of its incidence in women worldwide. The most important risk factor for the development of breast cancer and ovarian cancer is hereditary predisposition. About 5-10% of breast cancer cases and 15-20% of ovarian cancer cases are hereditary. About 50–70% of hereditary breast and ovarian cancers are caused by mutations in the BRCA1 and BRCA2 genes.

BRCA1 and BRCA2 gene mutations – genetic risk factor for breast cancer/ovarian cancer

The BRCA1 and BRCA2 genes encode nuclear proteins that regulate DNA repair, act as tumor suppressors and ensure genome integrity. The protein product of the BRCA1 gene allows you to limit the excessive cell proliferation of estrogen-dependent organs.

The presence of only one mutant allele of BRCA genes can lead to malignant cell transformation. Defects of the BRCA gene are characterized by high penetration and wide geographical distribution. According to a meta-analysis (2007), the risk of developing breast cancer by the age of 70 is 55% for carriers of mutations in the BRCA1 gene and 47% for BRCA2. The risk of developing ovarian cancer is 39% for BRCA1 and 17% for BRCA2.

Mutations in the BRCA1 gene predominate in the population (about 80% of the total number of mutations in the BRCA genes). The most common mutation is 5382insC (68–90% of all BRCA1 mutations), followed by 4153delA (1–2%). The following BRCA1 mutations can also be detected in the population: 185delAG, T300G, 3875del4, 3819del5, 2080delA(insA).

Mutations in the BRCA2 gene are 1.5–2 times less common than in BRCA1, their hallmark is a higher incidence of breast cancer in men and a lower risk of ovarian cancer in women. One of the most common mutations found in breast cancer is 6174delT.

Indications for molecular genetic testing

- Family history: two or more cases of breast/ovarian cancer in relatives of degree I-II, early disease, bilateral breast cancer, male breast cancer, tumours of one localization, rare forms of cancer in two or more relatives
- Breast cancer at a young age, bilateral breast cancer
- Multiple primary tumors in different organs
- Atypical proliferative breast diseases
- Ethnicity (Ashkenazi Jews)

Diagnostic kits RealBest-Genetics BRCA

The principle of mutation detection is based on amplification of a selected area of human DNA and subsequent detection of melting curves of hybrid complexes of PCR products and specific fluorescent probes.

The kits are designed for analysis of 48 samples, including control samples, and are intended for use with CFX96 (Bio-Rad, USA) and DT-96 (DNA-Technology, Russia) devices.

Features and advantages of the diagnostic kit

- Ready Master Mix for PCR: lyophilised, simplifies the PCR analysis procedure and ensures high stability of test quality
- Analysis duration: 80-90 minutes, automatic interpretation of results with genotype identification
- Specimens: whole blood or buccal epithelium
- High kit stability: storage of all kit components at 2–8°C for 12 months; transport up to 26 °C for not more than 10 days

Cat. №	Kit name	Number of tests
3807	**RealBest-Genetics BRCA1 185delAG/3875del4**	48
3808	**RealBest-Genetics BRCA1 3819del5/T300G**	48
3809	**RealBest-Genetics BRCA1 2080delA(insA)/BRCA2 6174delT**	48
3841	**RealBest-Genetics BRCA1 4153delA/5382insC**	48

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