

## Facts of breast cancer

### Screening Guidelines for the Early Detection of Breast Cancer, American Cancer Society

Yearly mammograms are recommended starting at age 40.

A clinical breast exam should be part of a periodic health exam, about every three years for women in their 20s and 30s, and every year for women 40 and older.

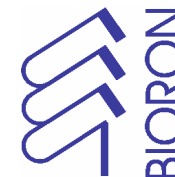
Women should know how their breasts normally feel and report any breast changes promptly to their health care providers. Breast self-exam is an option for women starting in their 20s.

Women at increased risk (e.g., family history, genetic tendency, past breast cancer) should talk with their doctors about the benefits and limitations of starting mammography screening earlier, having additional tests (i.e., breast ultrasound and MRI), or having more frequent exams.

### **E a r l y   D e t e c t i o n :**

Because of advancements in imaging and increased public awareness of the disease, almost two-thirds of all breast cancers are detected while the tumors are very small and limited to only the breast. In fact, the majority of women diagnosed with early stage breast cancer are candidates for treatment that saves the breast (breast conservation) and have very positive outcomes when treatment is completed.

***Every 13 minutes, a woman dies of breast cancer. Experts have said breast cancer self-exams should begin by the age of 20.***



Alternative Solutions

## Gene Pack DNA tubes for cancer-related mutations detection

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## Mutation detection

### Introduction

The most popular types of familial cancers are familial breast cancer and breast-ovarian cancer syndrome. Approximately 1% of all women are strongly genetically predisposed to the breast/ovarian cancer.

Women who are carrier of the mutation of BRCA1 or BRCA2 genes will get cancer with the probability more than 80% in the life span.

Analysis of mutations in these genes provides the possibility for early tumor detection in mutations carriers. The search for cancer mutation in the particular patient is complex due to the variety of mutations described and due to the involvement of multiple genes in predisposition formation.

Meanwhile, some mutations in BRCA1 and BRCA2 genes are recognized as the most frequent mutations in familial breast/ovarian cancers. Analysis of these mutations in cancer patients can easily reveal mutation carriers even without knowledge of familial history of the patient.

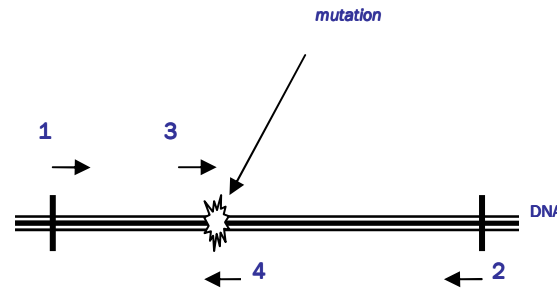
### Principle of mutation detection

Each kit is designed for the detection of one specific mutation.

The mutation detection is based on tetra primer allele-specific PCR.

Flanking primers provide amplification of a region around possible mutation in all cases. Internal primers can discriminate mutation allele and wild-type allele by the corresponding amplicon size (Fig 1):

## Storage at room-temperature



Primers 1 and 2 provide the synthesis of fragment A:



Primers 2 and 3 (mutation-specific) provide the synthesis of fragment B:



Primers 1 and 4 (wild-type specific) provide the synthesis of fragment C:



Figure 1. Schematic representation of tetra-primer allele-specific PCR; fragment B will be formed only in the case of mutation, fragment A will appear in all cases, fragment C will appear if wild-type sequence is present.

The kit is designed for the convenient and simple usage, storage and transportation. Each kit contains 100 tubes with lyophilised Master Mix with the following components:

- Taq DNA polymerase plus heat-sensitive Taq DNA inhibitors
- 4 primers for allele-specific PCR
- dNTP's
- special salts and stabilizers
- loading dye (bromphenol blue)
- Separate Tube of dilution buffer

The Master-Mix can be transported stored at ambient temperature at least for 1 Year without any change in performance of mutation detection.

To start PCR one have to add genomic DNA, dilution buffer and water. After PCR the product will be loaded on the Agarose gel (no need for loading dye).

The size of the fragments will indicate the presence or absence of the mutation in the sample studied. Fig. 2 shows an example of 5382insC mutation detection in BRCA1 gene:

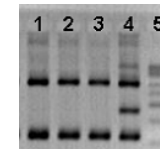


Figure 2. Analysis of DNA sample on the presence of 5382insC mutations in BRCA1 gene.

1,2,3 – no mutation detected

4 – mutation 5382insC (heterozygote)

5- Molecular Weight Marker

### List of mutations tests available in GenePack format:

BRCA1 – 6 mutations: 2963del10, 3819del5, 185delAG, 3875del4, 5382insC, 4154delA

BRCA2 – 3 mutations: S1099X, 9318dIAAAA, 1528deIAAAA

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