BREASTSEQ:
NGS GENETIC TEST FOR BREAST CANCER

BreastSeq is an ultrasequencing panel (NGS) which analyzes simultaneously 19 genes and 76 SNPs associated with susceptibility to hereditary breast cancer.

Breast cancer is the most common tumor in Western women. Affects approximately 1 in 8 women (12.29%) suffering from breast cancer over their lifetime. Breast cancer is a complex and multifactorial disease caused by genetic and environmental factors. Several studies have shown that between 5 and 10% of breast tumors cases have a specific hereditary component.

Today hereditary breast cancer can be explained by mutations in BRCA1 and BRCA2 genes in 15% of cases. However, in recent years other genes have been identified by their relation to hereditary breast cancer. Current knowledge suggests that mutations in the genes analyzed in the panel BreastSeq may confer a risk between 25-80% of breast cancer throughout life.

Some of these genes are also associated with an increased risk of other cancers such as: PALB2 with pancreatic cancer, RAD50 with ovarian cancer or TP53 with different sarcomas. In addition, we have included the detection of 76 SNPs (Single Nucleotide Polymorphism) associated with breast cancer in recent studies of GWAS (Genome-wide association study).

Genes and regions covered in the BreastSeq panel.

Estimation of risk associated with mutations in these genes derived from the literature.

GENES	RISK	REFERENCE
BRCA1	40-80%	Miki et al., 1994
BRCA2	20-85%	Wooster et al., 1995
ATM	15-20%	Renwick et al., 2006
BARD1	Variable	Ghimenti et al., 2002
BRIP1	Variable	Seal et al., 2006
FAM175A	Variable	Solyom et al., 2012
MRE11A	Variable	Bartkova et al., 2008
NBN	Variable	Seemanová et al., 2007
RAD50	Variable	Heikkinen et al., 2006
RAD51D	Variable	Osher et al., 2012
RAD51C	Variable	Somyajit et al., 2010
XRCC2	Variable	Park et al., 2012
76 SNP	Low	Bahcall, 2013
PALB2	20-40%	Erkko et al., 2007
STK11	57-81%	Hearle et al., 2006
CHEK2	25-37%	Walsh et al., 2007
PTEN	25-50%	Tan et al., 2012
TP53	56-90%	Walsh et al., 2006
CDH1	60%	Pharoah PD el at., 2001
MUTYH	Variable	Rennert et al., 2012

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From sample reception to bioinformatics

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NGS Technology Advantages

1. NGS technology allows addressing the study of multiple genes in a time and cost similar to that used in studying one or two genes with other methodologies.
2. Massive sequencing exceeds microarray approach because it´s not being limited to known mutations of a particular population.
3. NGS technology offers the best cost / benefit in the diagnosis of genetic - hereditary diseases.

Indication

In relation to international recommendations **BreastSeq** is suitable for people with a personal or family background according to the following criteria:

• Breast cancer at a young age (<50 years) or bilateral cancer
• Two primary breast cancers and ovarian and breast cancer.
• Male breast cancer.
• Risk ethnic populations.

A precise molecular diagnosis can help to estimate the risk to personal and family cancer, establishing cancer prevention measures appropriate to each patient and evaluate surgical options

Test description

BreastSeq Test is a panel of Target-directed sequencing or NGS that allows detection of mutations in 19 genes by mass sequencing of the coding regions of these genes, along with 25 nucleotides of the flanking introns. Also included 76 SNPs detection described in the literature in relation to hereditary breast cancer.

The genomic DNA of patients is extracted using standard procedures. The enrichment of the selected regions for the analysis is performed by digestion and amplification with primers overlapping to each gene of interest. It then proceeds to perform the massive sequencing of interesting regions. The extracted information is processed through a comprehensive bioinformatic analysis. Detected DNA variants with clinical interest are additionally verified by Sanger sequencing. The sensitivity of the analysis method is 96-99% for the mutations described.

Test application

You can find our study application form on our website:
http://www.ac-gen.com/apply-for-test.html

Sample shipping requirements:

- Peripheral blood: 5-10 ml of peripheral blood. Delivery Recommended Temp 4-8 °C.
- Saliva: pickup with a Self Collection kit supplied by our laboratory.
- 10µg of genomic DNA, preferably diluted to 200ng/ul. OB DNA 260-280 ratio (1.8-1.9). Delivery Recommended Temp 4-8 °C.

Delivering address:
AC-Gen Reading Life S.L. (Att Laboratory)
Parque Científico UVa
Edif. CTTA 2ª planta
Paseo de Belén nº9
47011 - Valladolid - Spain

Call us and we will handle the shipping of samples

More information: info@acgen.es www.ac-gen.com