OVASEQ:
NGS GENETIC TEST FOR OVARIAN, UTERINE AND BREAST CANCER

OvaSeq is an ultrasequencing panel (NGS), which analyzes simultaneously 24 genes and 89 SNPs associated with susceptibility to hereditary ovarian cancer, breast and uterus.

Ovarian cancer is the fifth most common cancer in women, it is estimated that 1 in 71 (1.4%) of women suffering from ovarian cancer over their lifetime. Meanwhile cervical cancer affects 1 in 38 women (2.61%). While breast cancer is the most common cancer in Western women. Affects approximately 1 in 8 women (12.29%) who suffering breast cancer over their lifetime.

Currently breast and ovarian cancer could be explained by inherited mutations in high penetrance genes BRCA1 and BRCA2 in 10-18% of cases. However, in recent years other genes have been identified by their relation to breast and ovarian cancer in families. Other genes associated with Lynch syndrome could significantly increase the risk of uterine cancer and ovarian cancer, while others such as PTEN could increase the risk of breast and uterine cancer. Although BRCA1 and BRCA2 are responsible for most cases of hereditary ovarian cancer, a significant proportion of cases of breast, ovary and uterus can be attributed to mutations in other genes. This genetic diversity makes the OvaSeq an ideal test to address the analysis of these patients.

Genes and regions covered in the OvaSeq 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
89 SNP	Bajo	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
MLH1	20-60%	Abdel-Rahman VW et al., 2006
MSH2	20-60%	
MSH6	20-60%	
PMS2	20-60%	Hedge MR y Roa BB, 2009

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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

OvaSeq is indicated to persons with personal or familial background according to following criteria:

• Breast and ovarian
• Two or more cases of ovarian cancer.
• Breast and uterine cancer
• Ovarian, uterine and/or colon cancer.
• Breast, ovary or uterus cancer diagnosed at early age (50 years)

An accurate molecular diagnosis can help estimate the risk of hereditary cancer, establish cancer prevention measures appropriate to each patient and evaluate surgical options. In some cases there are treatment recommendations, such as avoiding radiotherapy in patients with mutations in TP53 or the use of inhibitors of poly (ADP-ribose) polymerase in patients with BRCA gene mutations (Bayraktar and Glück, 2012)

Test description

OvaSeq Test is a panel of Target-directed sequencing or NGS that allows the detection of mutations in genes by sequencing 24 genes by their coding regions, with 25 nucleotides of the flanking introns. It also includes the detection of 89 SNP described in the literature.

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