HC-GEN TEST:
HEREDITARY CANCER GEN TEST FOR CLINICAL TRIALS AND RESERACH

AC-Gen Reading Life is a Spanish pioneer company in the application of Next-Generation Sequencing technology (NGS) for clinical use. It is an authorized centre for genetic analysis and diagnosis by the Spanish Health Authorities. The company focuses its efforts in offering advanced services for clinical uses by applying ultrasequencing technology or NGS (Next-Generation Sequencing)

The HC-Gen Test **is the ideal tool to identify genetic variations related to cancer in your patients**. It is a genetic test that allows the simultaneous detection of mutations in ALL the genes associated with hereditary cancer syndromes using ultrasequencing DNA techniques.

HC-Gen Test is a NGS Targeted panel, which is simultaneously looking at 37 genes, and 89 SNP linked to high risk of suffering a wide range of cancers as breast, ovary, uterus, colon and pancreas cancer. While mutations in each gene on this panel may be individually rare, they collectively account for a significant amount of hereditary cancer susceptibility. This panel may be appropriate in a number of scenarios, particularly if the family history shares features of several different hereditary cancer syndromes.

Genes and regions covered in the Hereditary Cancer Gene Test.

List of different genes with major hereditary cancer syndromes.

GENES	BreastSeq	OvaSeq	ColonSeq	PancSeq
BRCA1	✓	✓		✓
BRCA2	✓	✓		✓
ATM	✓	✓		✓
BARD1	✓	✓		
BRIP1	✓	✓		
FAM175A	✓	✓		
MRE11A	✓	✓		
NBN	✓	✓		
RAD50	✓	✓		
RAD51D	✓	✓		
RAD51C	✓	✓		
XRCC2	✓	✓		
89SNP	✓	✓		
PALB2	✓	✓		✓
STK11	✓	✓	✓	✓
CHEK2	✓	✓	✓	
PTEN	✓	✓	✓	
TP53	✓	✓	✓	✓
CDH1	✓	✓	✓	
MUTYH	✓	✓	✓	
MLH1		✓	✓	✓
MSH2		✓	✓	✓
MSH6		✓	✓	✓
EPCAM		✓	✓	✓
PMS2		✓	✓	✓
APC			✓	✓
BMPR1A			✓	
SMAD4			✓	
AXIN2			✓	
BLM			✓	
ENG			✓	
KIT			✓	
MLH3			✓	
MSH3			✓	
PDGFRA			✓	
PIK3CA			✓	
PMS1			✓	
SCG5			✓	

AC-Gen Reading Life makes all processes *in-house!*

From sample reception to bioinformatics, including genetic counselling

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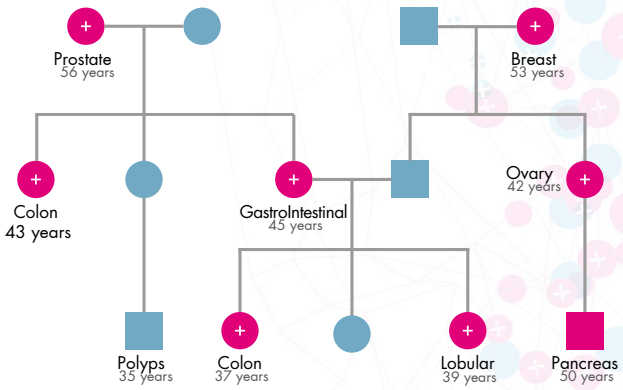
NGS Technology Advantages

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.

HC-Gen Genetic diagnosis Tests use NGS technologies for genes analysis, but also include validation of the detected mutations by Sanger sequencing. The resulting clinical report entails a thorough interpretation of the results through a detailed analysis of DNA variants detected and thus provides useful and relevant information for the diagnosis and prevention of hereditary cancer.

Detection of responsible mutations of diseases to analyze in a faster and more economical way the same DNA variant in other families.

Example of genealogic tree in highlighting multiple cancers in one family and which could fit a presumption of hereditary cancer syndrome. In this case it would be appropriate to perform the Hereditary Cancer Gen Test.



Common warning criteria for hereditary cancer

• Cancer diagnosed at early age (<50 years)
• Cancer diagnosed in different generations (dominant inheritance).
• Patients with multiple cancers. Bilateralism, ipsilaterality.

Test description

HC-Gen Test is a panel of Target-directed sequencing or NGS that allows the detection of mutations in 37 genes by massive sequencing of their coding regions, with 25 nucleotides of the flanking introns. This test includes the detection of 89 SNPs described in the literature in relation to breast and ovarian cancer in families. 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 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