COLONSEQ:
NGS GENETIC TEST FOR COLORECTAL CANCER

ColonSeq is an ultrasequencing panel (NGS), which analyzes simultaneously 24 genes associated with susceptibility to different types of hereditary colorectal cancer.

Colorectal cancer is the second leading cause of death from tumor in the Western world. It is estimated that 1 in 20 (5.2%) people will have colorectal cancer throughout his life.

Most colorectal cancers are sporadic, though is estimated that at least 25 % are familial. A subset of the same shows strong genetic etiology. As in most forms of cancer prevention and early detection is critical. Have characterized different syndromes associated with the development of colon cancer.

ColonSeq is an analysis tool to analyze the main genes associated with colon cancer syndromes such as Lynch syndrome, familial adenomatous polyposis, MUTYH -associated polyposis, disorders related to PTEN, Li-Fraumeni syndrome, juvenile polyposis, cancer or diffuse gastric Peutz - Jeghers syndrome.

Genes included in the panel and associated syndromes in ColonSeq.

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

GENES	SYNDROME	RISK	REFERENCE
PTEN	Cowden syndrome	10%	Orloff y Eng et al., 2008
TP53	Li-Fraumeni syndrome	Variable	Olivier M et al., 2003
STK11	Peutz-Jeghers syndrome	39%	Hearle et al., 2006
CDH1	Diffuse gastric cancer	Variable	Guilford P et al., 2010
MSH3	Nonpolyposis colorectal cancer (HNPCC)	Variable	Hedge MR y Roa BB, 2009
MLH1	Lynch syndrome, HNPCC	40-80%	Abdel-Rahman WV et al., 2006
MSH2	Lynch syndrome, HNPCC	40-80%	
MSH6	Lynch syndrome, HNPCC	20-44%	
EPCAM	Lynch syndrome, HNPCC	40-80%	
PMS2	Lynch syndrome, HNPCC	15-20%	
PMS1	Lynch syndrome, HNPCC	Variable	
BLM	Multiple	Variable	Russell y Groden, 2009
CHEK2	Multiple	24-37%	Cybulski et al., 2004
PIK3CA	Multiple	Variable	Miyaki et al., 2007
MLH3	Multiple	Variable	Wu et al., 2001
AXIN2	Oligodontia – Colorrectal cancer	Variable	Lammi et al., 2004
MUTYH	Associated polyposis to MUTHY	35-63%	Rennert et al., 2012
BMPR1A	Juvenile polyposis	17-68%	Van Hattem et al., 2008
SMAD4	Juvenile polyposis		Van Hattem et al., 2008
ENG	Juvenile polyposis		Sweet et al., 2005
APC	Familial adenomatous polyposis (FAP)	90%	Pedace L et al., 2008
SCG5	Hereditary mixed polyposis syndrome	Variable	Jaeger et al., 2012
PDGFRA	Gastrointestinal stromal tumour (GIST)	Variable	Joensuu et al., 2013
KIT	Gastrointestinal stromal tumour (GIST)		

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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

Certain patterns of colon cancer as colon polyps or tumor characteristics shared in a concrete family, can direct the diagnosis to a certain type of colon cancer. However, the ColonSeq test can be critical in cases of uncertainty as to the preventive level. Knowledge about genetic susceptibility to colon cancer can help the clinical management of patients, suggesting more frequent periodic reviews more frequent as colonoscopies or consider prophylactic colectomy patients with pathogenic mutations in genes such as APC or MLH1.

Test description

ColonSeq Test is a panel of Target-directed sequencing or NGS that allows detection of mutations by sequencing 24 genes by their massive sequencing coding regions, with 25 nucleotides of the flanking introns.

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