Lynch Syndrome

Precision Panel

Overview

Lynch Syndrome, also referred to as Hereditary Nonpolyposis Colorectal Cancer (HNPCC) is the most common form of hereditary colorectal cancer. It is inherited in an autosomal dominant pattern result of a defect in mismatch repair (MMR) proteins, which are in charge of repairing errors in cellular division, therefore leading to accumulated errors and thus the development of cells with abnormal growth. Colorectal cancer in patients with HNPCC present at an earlier age than in the general population characterized by an increase in other types of cancer, such as endometrial cancer, ovary, stomach and others.

Hereditary cancer syndromes are encountered in all medical specialties. Although they account for about 5% of all malignancies, it is of special importance to identify these patients because, unlike patients with sporadic cancers, they require special, long-term care as their predisposition can cause them to develop certain tumors at a relatively early age. These cancers can arise in the lungs, kidneys, liver, pancreas, skin, eyes, heart. Most hereditary cancers are associated with a “germline mutation” that will be present in every cell of the human body. Identification of patients at risk of inherited cancer susceptibility is dependent upon the ability to characterize genes and alterations associated with increased cancer risk as well as gathering a detailed personal and family history aiding in the identification of the mode of inheritance as well as other family members at risk of suffering from this susceptibility.

The Igenomix Lynch Syndrome Precision Panel can be as a screening and diagnostic tool ultimately leading to a better management and prognosis of the disease. It provides a comprehensive analysis of the genes involved in this disease using next-generation sequencing (NGS) to fully understand the spectrum of relevant genes involved.

Indications

The Igenomix Lynch Syndrome Precision Panel is indicated in those cases where there are:

- Family history of colorectal cancer (CRC) or suspected hereditary colorectal cancer syndrome, such as familial adenomatous polyposis (FAP) or Lynch syndrome (hereditary non-polyposis colon cancer or HNPCC)
- Family or personal history of Lynch syndrome or Lynch-like syndrome
- History of multiple colorectal adenomas
- Asymptomatic patient who wishes to know genetic risk for Lynch syndrome.

- History of colorectal and/or endometrial cancer in multiple relatives on the same side of a family.

Clinical Utility

The clinical utility of this panel is:

- The genetic and molecular diagnosis for an accurate clinical diagnosis of a patient with personal or family history suggestive of a hereditary cancer syndrome.
- Early initiation of treatment with a multidisciplinary team for appropriate total body screening, early surgical intervention or pharmacologic treatment.
- Risk assessment of asymptomatic family members according to the mode of inheritance
- Reduce the incidence of advanced adenomas at colonoscopy.
- Prevention of CRC.
- Reduce morbidity related to CRC, or morbidity secondary to complications of surveillance and treatment.
- Improved identification of hereditary CRC syndromes.
- Improved pathways from diagnosis to treatment in susceptible populations.

Genes & Diseases

GENE	OMIM DISEASES	INHERITANCE*	% GENE COVERAGE (20X)	HGMD**
EPCAM	Colorectal Cancer, Hereditary Nonpolyposis, Type 8, Lynch Syndrome	AR	99.94	52 of 70
MLH1	Colorectal Cancer, Hereditary Nonpolyposis, Type 2, Lynch Syndrome, Mismatch Repair Cancer Syndrome, Muir-Torre Syndrome	AD, AR	99.94	1079 of 1118
MSH2	Lynch Syndrome, Mismatch Repair Cancer Syndrome, Muir-Torre Syndrome	AD, AR	99.99	1032 of 1057
MSH6	Colorectal Cancer, Hereditary Nonpolyposis, Type 5, Lynch Syndrome, Mismatch Repair Cancer Syndrome, Muir-Torre Syndrome	AD, AR	99.28	613 of 641
PMS2	Colorectal Cancer, Hereditary Nonpolyposis, Type 4, Lynch Syndrome, Mismatch Repair Cancer Syndrome	AD, AR	97.17	264 of 285

*Inheritance: AD: Autosomal Dominant; AR: Autosomal Recessive; X: X linked; XLR: X linked Recessive; Mi: Mitochondrial; Mu: Multifactorial.
**Number of clinically relevant mutations according to HGMD

Methodology

Contact us

Call +34 963 905 310 or send an email to supportspain@igenomix.com for any of the following objectives:

- Get more information about the test.
- Request your kit.
- Request a pick up of the kit after collecting the sample.

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