Hereditary Colorectal Cancer

Precision Panel

Overview

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. Most hereditary cancer syndromes follow an autosomal dominant inheritance, and the penetrance is high.

The Igenomix Hereditary Colorectal Cancer Precision Panel provides a comprehensive analysis of the most common hereditary Colorectal Cancer syndromes using next-generation sequencing (NGS) to fully understand the spectrum of relevant colorectal cancer predisposition genes.

Indications

The Igenomix Hereditary Colorectal Cancer Panel is indicated as a screening and diagnostic test in those cases where there is:

- 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 CRC.
- A personal history of getting radiation to the abdomen or pelvic area to treat prior cancer.
- Personal history of inflammatory bowel disease (Ulcerative Colitis or Crohn's disease).

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.

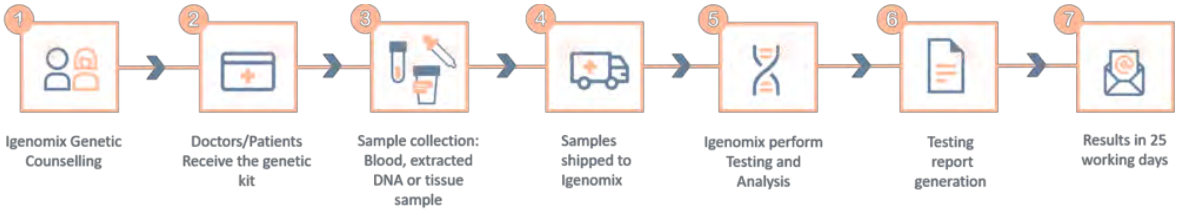
Genes & Diseases

GENE	OMIM DISEASES	INHERITANCE*	% GENE COVERAGE (20X)	HGMD**
AKT1	Cowden Syndrome 6	AD	100	6 of 6
APC	Familial Adenomatous Polyposis 1, Desmoid Disease, Gardner Syndrome	AD	98.92	1846 of 1882
AXIN2	AXIN2-Related Attenuated Familial Adenomatous Polyposis, Oligodontia-Colorectal Cancer Syndrome	AD	99.86	1608 of 1632
ATM	Ataxia-Telangiectasia, Breast Cancer, Mantle Cell Lymphoma	AD.AR	99.93	32 of 33
BLM	Bloom Syndrome	AR	97.19	133 of 141
BMPR1A	Familial Colorectal Cancer Type X, Juvenile Polyposis Syndrome Of Infancy, Polyposis Syndrome Juvenile Intestinal, Polyposis Syndrome Hereditary Mixed	AD	100	124 of 127
BUB1B	Mosaic Variegated Aneuploidy Syndrome	AD, AR	99.84	30 of 31
CDH1	Blepharo-Cheilo-Odontic Syndrome, Breast Cancer, Cleft Lip/Palate, Gastric Cancer, Prostate Cancer	AD	100	361 of 363
CHEK2	Li-Fraumeni Syndrome, Breast Cancer Susceptibility To, Prostate Cancer Susceptibility To, Colorectal Cancer Susceptibility To	AD	99.47	307 of 310
ENG	Familial Cerebral Saccular Aneurysm, Generalized Juvenile Polyposis/Juvenile Polyposis Coli, Hereditary Haemorrhagic Telangiectasia	AD	100	467 of 471
EPCAM	Colorectal Cancer, Hereditary Nonpolyposis, Type 8, Lynch Syndrome	AR	99.94	52 of 70
FLCN	Birt-Hogg-Dubé Syndrome, Colorectal Cancer, Renal Cell Carcinoma Nonpapillary	AD	100	200 of 205
GALNT12	Colorectal Cancer, Susceptibility To, 1		88.97	14 of 15
GREM1	Hereditary Mixed Polyposis Syndrome		99.89	5 of 5
MLH1	Colorectal Cancer, Hereditary Nonpolyposis, Type 2, Lynch Syndrome, Mismatch Repair Cancer Syndrome, Muir-Torre Syndrome	AD, AR	99.94	1079 of 1118
MLH3	Colorectal Cancer, Hereditary Nonpolyposis, Type 7, Endometrial Carcinoma	AD	99.98	32 of 32
MSH2	Lynch Syndrome, Mismatch Repair Cancer Syndrome, Muir-Torre Syndrome	AD, AR	99.99	1032 of 1057
MSH3	Familial Adenomatous Polyposis 4	AD, AR	99.42	23 of 24
MSH6	Colorectal Cancer, Hereditary Nonpolyposis, Type 5, Lynch Syndrome, Mismatch Repair Cancer Syndrome, Muir-Torre Syndrome	AD, AR	99.28	613 of 641
MUTYH	Familial Adenomatous Polyposis, 2	AR	100	183 of 183
NTHL1	Familial Adenomatous Polyposis 3	AR	100	13 of 13
PIK3CA	Cowden Syndrome 5	AD	99.58	54 of 58
PMS2	Colorectal Cancer, Hereditary Nonpolyposis, Type 4, Lynch Syndrome, Mismatch Repair Cancer Syndrome	AD, AR	97.17	264 of 285
POLD1	Colorectal Cancer, Susceptibility To, 10, Mandibular Hypoplasia, Deafness, Progeroid Features, And Lipodystrophy Syndrome, Polymerase Proofreading-Related Adenomatous Polyposis	AD	100	40 of 41
POLE	Colorectal Cancer, Susceptibility To, 12, FILS Syndrome, IMAGE-I Syndrome, Polymerase Proofreading-Related Adenomatous Polyposis	AD, AR	100	100 of 100
PTEN	Bannayan-Riley-Ruvalcaba Syndrome, Cowden Syndrome, Hereditary Breast And Ovarian Cancer Syndrome, Juvenile Polyposis Of Infancy, Lhermitte-Duclos Disease, Meningioma, Familial, Susceptibility To, Proteus Syndrome, Proteus-Like Syndrome, Segmental Outgrowth-Lipomatosis-Arteriovenous Malformation-Epidermal Nevus Syndrome	AD	99.97	609 of 629

RNF43	Sessile Serrated Polyposis Cancer Syndrome	AD	99.98	13 of 13
RPS20	Familial Colorectal Cancer		99.97	1 of 1
SMAD4	Juvenile Polyposis/Hereditary Haemorrhagic Telangiectasia Syndrome, Polyposis, Juvenile Intestinal, Myhre Dysplasia, Polyposis, Juvenile Intestinal	AD	99.56	136 of 136
STK11	Peutz-Jeghers Syndrome	AD	81.99	456 of 470
TP53	Adrenocortical Carcinoma, Paediatric, Basal Cell Carcinoma, Susceptibility To, 7, Bone Marrow Failure Syndrome 5, Breast Cancer, Colorectal Cancer, Familial Pancreatic Carcinoma, Glioma Susceptibility 1, Hereditary Breast And Ovarian Cancer Syndrome, Li-Fraumeni Syndrome, Osteosarcoma, Papilloma Of Choroid Plexus	AD, MU	98.92	557 of 563

*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.

References

1. Lorans M, Dow E, Macrae FA, Winship IM, Buchanan DD. Update on Hereditary Colorectal Cancer: Improving the Clinical Utility of Multigene Panel Testing. Clin Colorectal Cancer. 2018 Jun;17(2):e293-e305. doi: 10.1016/j.clcc.2018.01.001.
2. Guillén-Ponce C, Lastra E, Lorenzo-Lorenzo I, Martín Gómez T, Morales Chamorro R, Sánchez-Heras AB, Serrano R, Soriano Rodríguez MC, Soto JL, Robles L. SEOM clinical guideline on hereditary colorectal cancer (2019). Clin Transl Oncol. 2020 Feb;22(2):201-212. doi: 10.1007/s12094-019-02272-y.
3. National Comprehensive Cancer Network. (2021). Retrieved from https://www.nccn.org/professionals/physician_gls/default.aspx#detection
4. American Cancer Society https://www.cancer.org/cancer/colon-rectal-cancer.html
5. Brosens, L. A., Offerhaus, G. J., & Giardiello, F. M. (2015). Hereditary Colorectal Cancer: Genetics and Screening. The Surgical clinics of North America, 95(5), 1067–1080. https://doi.org/10.1016/j.suc.2015.05.004
6. Stoffel, E. M., Mangu, P. B., Gruber, S. B., Hamilton, S. R., Kalady, M. F., Lau, M. W., Lu, K. H., Roach, N., Limburg, P. J., American Society of Clinical Oncology, & European Society of Clinical Oncology (2015). Hereditary colorectal cancer syndromes: American Society of Clinical Oncology Clinical Practice Guideline endorsement of the familial risk-colorectal cancer: European Society for Medical Oncology Clinical Practice Guidelines. Journal of clinical oncology: official journal of the American Society of Clinical Oncology, 33(2), 209–217. https://doi.org/10.1200/JCO.2014.58.1322
7. Giardiello, F. M., Allen, J. I., Axilbund, J. E., Boland, C. R., Burke, C. A., Burt, R. W., Church, J. M., Dominitz, J. A., Johnson, D. A., Kaltenbach, T., Levin, T. R., Lieberman, D. A., Robertson, D. J., Syngal, S., Rex, D. K., & US Multi-Society Task Force on Colorectal Cancer (2014). Guidelines on genetic evaluation and management of Lynch syndrome: a consensus statement by the US Multi-Society Task Force on colorectal cancer. Gastroenterology, 147(2), 502–526. https://doi.org/10.1053/j.gastro.2014.04.001