



# Prostate Cancer Precision Panel



#### Overview

Prostate cancer is the second most common cancer in men after skin cancer and the second leading cause of cancer death in men after lung cancer. Risk factors for prostate cancer include increased age, family history and black race. In early stages, prostate cancer is asymptomatic and is typically detected by screening. Although many prostate cancers are sporadic, about 5-15% of prostate cancer diagnoses may be associated with a strong hereditary component. Genetic testing has found its place in routine testing for breast, ovarian and prostate cancers amongst others, providing actionable preventive measures to increase life expectancy and quality of life.

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. 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 Prostate Cancer Precision Panel provides a comprehensive analysis of the most common genes responsible for the development of a malignant growth in the prostate using next-generation sequencing (NGS) to fully understand the spectrum of relevant lung cancer predisposition genes.

#### **Indications**

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

- Family history of prostate cancer
- Blood in the urine
- Frequent urination
- Urge to urinate
- Pain or burning during urination
- Weak or interrupted urine flow
- Unintentional weight loss
- Fatigue





- Lower back pain
- Change in bowel habits

## 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 with predisposition to prostate cancer.
- Early initiation of treatment with a multidisciplinary team for appropriate total body screening, early surgical intervention, or pharmacologic treatment.
- Risk assessment and genetic counselling of asymptomatic family members according to the mode of inheritance.
- Reduce morbidity related to prostate cancer, or morbidity secondary to complications of surveillance and treatment.
- Categorization of genetic alterations into predictive levels of standard, investigational or hypothetic target therapies in the molecular pathology reports.
- Improved pathways from diagnosis to treatment in susceptible populations.

### Genes & Diseases

GENE	OMIM DISEASES	INHERITANCE*	% GENE COVERAGE	HGMD**
			(20X)	
APC	Colorectal Cancer, Gastric Cancer, Hepatocellular Carcinoma, Apc-Related Attenuated Familial Adenomatous Polyposis, Cenani-Lenz Syndrome	AD	98.92	1846 of 1882
AR	Androgen Insensitivity Syndrome, X-linked Hypospadias, Prostate Cancer, Reifenstein Syndrome, Kennedy Disease	AD,X,XR,G	97.96	NA of NA
BARD1	Breast Cancer, Hereditary Breast And Ovarian Cancer Syndrome	AD	99.86	195 of 195
BMPR1A	Hereditary Mixed Polyposis Syndrome, Familial Colorectal Cancer, Juvenile Polyposis Of Infancy	AD	100	124 of 127
BRCA1	Breast Cancer, Familial Breast-Ovarian Cancer, Fanconi Anemia Complementation Group, Familial Pancreatic Carcinoma, Primary Peritoneal Carcinoma	AD,AR,MU	98.97	2783 of 2894
BRCA2	Breast Cancer, Breast-Ovarian Cancer, Fanconi Anemia Complementation Group D1, Glioma Susceptibility, Medulloblastoma, Pancreatic Cancer, Prostate Cancer	AD,AR,MU	98.51	3343 of 3451
BRIP1	Breast Cancer, Fanconi Anemi, Complementation Group J, Hereditary Breast And Ovarian Cancer Syndrome	AD,AR	94.97	235 of 237
CDH1	Blepharocheilodontic Syndrome, Breast Cancer, Endometrial Carcinoma, Gastric Cancer, Prostate Cancer, Suppressor Of Tumorigenicity	AD	100	361 of 363
CDKN2A	Melanoma-Astrocytoma Syndrome, Melanoma-Pancreatic Cancer Syndrome, Li-Fraumeni Syndrome	AD	94.99	257 of 262
CHEK2	Breast Cancer, Li-Fraumeni Syndrome, Osteosarcoma, Prostate Cancer, Hereditary Breast And Ovarian Cancer Syndrome	AD	99.47	307 of 310
EPHB2	Prostate Cancer/Brain Cancer Susceptibility	AR	98.08	12 of 12
GREM1	Hereditary Mixed Polyposis Syndrome		99.89	5 of 5
KLF6	Gastric Cancer, Prostate Cancer	AD	100	1 of 1
MAD1L1	Prostate Cancer	AD	100	2 of 2
MDM2	Lessel-Kubisch Syndrome, Li-Fraumeni Syndrome, Well-differentiated Liposarcoma	AR	99.92	1 of 1
MLH1	Hereditary Nonpolyposis Colorectal Cancer, Mismatch Repair Cancer Syndrome, Muir-Torre Syndrome	AD,AR	99.94	1079 of 1118
MRE11	Ataxia-Telangiectasia-like Disorder, Hereditary Breast And Ovarian Cancer Syndrome	AR	99.95	NA of NA





MSH2	Lynch Syndrome, Mismatch Repair Cancer Syndrome, Muir-Torre Syndrome	AD,AR	99.99	1032 of 1057
MSH6	Hereditary Nonpolyposis Colorectal Cancer, Endometrial Carcinoma, Mismatch Repair Cancer Syndrome, Muir-Torre Syndrome	AD,AR	99.28	613 of 641
MXI1	Prostate Cancer	AD	94.55	NA of NA
NAB2	Solitary Fibrous Tumour/Hemangiopericytoma		99.43	NA of NA
NBN	Aplastic Anemia, Acute Lymphocytic Leukemia, Nijmegen Breakage Syndrome, Hereditary Breast And Ovarian Cancer Syndrome	AR,MU,P	100	200 of 200
PALB2	Breast Cancer, Fanconi Anemia Complementation Group N, Familial Pancreatic Carcinoma, Hereditary Breast And Ovarian Cancer Syndrome	AD,AR	98.78	601 of 617
PTEN	Cowden Disease, Meningioma, Prostate Cancer, Bannayan-Riley-Ruvalcaba Syndrome, Hereditary Breast And Ovarian Cancer Syndrome, Juvenile Polyposis Of Infancy, Lhermitte-Duclos Disease, Proteus Syndrome	AD	99.97	609 of 629
RAD50	Nijmegen Breakage Syndrome-like Disorder, Hereditary Breast And Ovarian Cancer Syndrome	AR	99.94	117 of 120
RAD51	Breast Cancer, Fanconi Anemia Complementation Group R, Hereditary Breast And Ovarian Cancer Syndrome	AD	99.98	16 of 16
RAD51C	Familial Breast-Ovarian Cancer, Fanconi Anemia Complementation Group O	AR	100	130 of 130
RAD51D	Hereditary Breast And Ovarian Cancer Syndrome		100	97 of 97
RNASEL	Hereditary Prostate Cancer	AD	99.83	7 of 7
RNF43	Sessile Serrated Polyposis Cancer Syndrome, Hyperplastic Polyposis Syndrome	AD	99.98	13 of 13
STAT6	Solitary Fibrous Tumour/Hemangiopericytoma		99.78	NA of NA
TP53	Basal Cell Carcinoma, Bone Marrow Failure Syndrome, Breast Cancer, Colorectal Cancer, Glioma Susceptibility, Hepatocellular Carcinoma, Li- Fraumeni Syndrome, Nasopharyngeal Carcinoma, Osteosarcoma, Pancreatic Cancer, Papilloma Of Choroid Plexus, Adrenocortical Carcinoma, Hereditary Breast And Ovarian Cancer Syndrome	AD,MU,P	98.92	557 of 563
ZFHX3	Prostate Cancer	AD	99.14	8 of 8

<sup>\*</sup>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





Call +34 963 905 310 or send an email to <a href="mailto:supportspain@igenomix.com">supportspain@igenomix.com</a> 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

National Comprehensive Cancer Network. (2021). Retrieved from https://www.nccn.org/professionals/physician\_gls/default.aspx#detection





- Zhen, J. T., Syed, J., Nguyen, K. A., Leapman, M. S., Agarwal, N., Brierley, K., Llor, X., Hofstatter, E., & Shuch, B. (2018). Genetic testing for hereditary prostate cancer: Current status and limitations. Cancer, 124(15), 3105–3117. <a href="https://doi.org/10.1002/cncr.31316">https://doi.org/10.1002/cncr.31316</a>
- Heidegger, I., Tsaur, I., Borgmann, H., Surcel, C., Kretschmer, A., Mathieu, R., Visschere, P., Valerio, M., van den Bergh, R., Ost, P., Tilki, D., Gandaglia, G., Ploussard, G., & EAU-YAU Prostate Cancer Working Party (2019). Hereditary prostate cancer - Primetime for genetic testing?. Cancer treatment reviews, 81, 101927. https://doi.org/10.1016/j.ctrv.2019.101927
- Pilarski R. (2019). The Role of BRCA Testing in Hereditary Pancreatic and Prostate Cancer Families. American Society of Clinical Oncology educational book. American Society of Clinical Oncology. Annual Meeting, 39, 79–86. https://doi.org/10.1200/EDBK\_238977
- 5. Tan, S. H., Petrovics, G., & Srivastava, S. (2018). Prostate Cancer Genomics: Recent Advances and the Prevailing Underrepresentation from Racial and Ethnic Minorities. *International journal of molecular sciences*, 19(4), 1255. https://doi.org/10.3390/ijms19041255
- Heise, M., & Haus, O. (2014). Dziedziczny rak gruczołu krokowego [Hereditary prostate cancer]. Postepy higieny i medycyny doswiadczalnej (Online), 68, 653–665. https://doi.org/10.5604/17322693.1104682
- 7. Pritzlaff, M., Tian, Y., Reineke, P., Stuenkel, A. J., Allen, K., Gutierrez, S., Jackson, M., Dolinsky, J. S., LaDuca, H., Xu, J., Black, M. H., & Helfand, B. T. (2020). Diagnosing hereditary cancer predisposition in men with prostate cancer. *Genetics in medicine : official journal of the American College of Medical Genetics*, 22(9), 1517–1523. https://doi.org/10.1038/s41436-020-0830-5