Lung Cancer

Precision Panel

Overview

Lung cancer is the leading cause of cancer death worldwide, with 90% of cases being attributable to smoking. It is the second most common cancer, and it is a malignancy that affects either the lung tissue or the airways. Risk factors for lung cancer include cigarette smoking, asbestos, radon and family history of lung cancer. Lung cancer can be sporadic or associated to a hereditary cancer syndrome.

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

Indications

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

- Cough
- Wheezing
- Unintentional weight loss
- Hemoptysis
- Chest pain
- Dyspnea
- Horaseness
- Family history of lung cancer

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 lung 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 lung 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

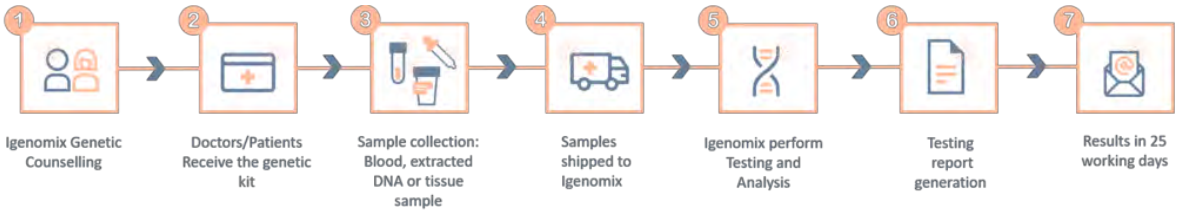
GENE	OMIM DISEASES	INHERITANCE*	% GENE COVERAGE (20X)	HGMD**
ABL1	Congenital Heart Defects And Skeletal Malformations Syndrome, Chronic Myeloid Leukemia	AD	99.93	8 of 8
AKT1	Breast Cancer , Colorectal Cancer , Cowden Syndrome, Proteus Syndrome, Suppressor Of Tumorigenicity	AD	100	6 of 6
ALK	Neuroblastoma		99.84	16 of 16
APC	Colorectal Cancer , Desmoid Disease, Hereditary , Familial Adenomatous Polyposis, Gastric Cancer, Hepatocellular Carcinoma, APC-Related Attenuated Familial Adenomatous Polyposis, Cenani-Lenz Syndrome	AD	98.92	1846 of 1882
AR	Androgen Insensitivity Syndrome, Prostate Cancer, Reifenstein Syndrome, Kennedy Disease	AD,X,XR,G	97.96	-
ARID1A	Coffin-Siris Syndrome	AD	95.32	40 of 42
ASXL1	Bohring-Opitz Syndrome, Myelodysplastic Syndrome, Systemic Mastocytosis With Associated Hematologic Neoplasm	AD	99.96	41 of 41
ATM	Ataxia-Telangiectasia, Breast Cancer	AD,AR	99.93	1608 of 1632
AXL	Hypogonadotropic Hypogonadism Without Anosmia	AR	100	10 of 10
BRAF	Cardiofaciocutaneous Syndrome, Leopard Syndrome, Lung Cancer	AD	100	80 of 80
CASP8	Breast Cancer, Caspase 8 Deficiency, Hepatocellular Carcinoma, Lung Cancer	AD,AR	100	8 of 8
CDH1	Blepharocheilodontic Syndrome, Breast Cancer, Endometrial Carcinoma, Gastric Cancer, Prostate Cancer, Suppressor Of Tumorigenicity	AD	100	361 of 363
CDK4	Familial Melanoma, Well-Differentiated Liposarcoma	AD	100	22 of 22
CDKN2A	Melanoma-Astrocytoma Syndrome , Melanoma-Pancreatic Cancer Syndrome, Li-Fraumeni Syndrome	AD	94.99	257 of 262
CTNNB1	Colorectal Cancer, Hepatocellular Carcinoma, Suppressor Of Tumorigenicity, Desmoid Tumor	AD,AR	100	63 of 63
CYP2A6	Lung Cancer	AD	100	-
DDR2	Spondylometaepiphyseal Dysplasia, Short Limb-hand Type, Warburg-Cinotti Syndrome	AD,AR	100	13 of 13
EGFR	Lung Cancer	AD,AR	100	27 of 27
ERBB2	Gastric Cancer, Glioma Susceptibility, Lung Cancer	AD	96.97	10 of 10
ERCC6	Cerebrooculofacioskeletal Syndrome, Cockayne Syndrome, De Sanctis-Cacchione Syndrome, Lung Cancer	AD,AR	99.98	127 of 128
FASLG	Autoimmune Lymphoproliferative Syndrome, Lung Cancer	AD	99.98	8 of 9
FGFR1	Encephalocraniocutaneous Lipomatosis, Hartsfield Syndrome, Jackson-Weiss Syndrome	AD	100	279 of 280

FGFR2	Antley-Bixler Syndrome Without Genital Anomalies Or Disordered Steroidogenesis, Apert Syndrome, Crouzon Syndrome, Gastric Cancer, Jackson-Weiss Syndrome, Pfeiffer Syndrome, Saethre-Chotzen Syndrome	AD	98	140 of 143
FGFR3	Bladder Cancer, Cervical Cancer, Colorectal Cancer, Crouzon Syndrome With Acanthosis Nigricans, Muenke Syndrome, Testicular Tumor	AD,AR	99.89	77 of 78
FGFR4	Prostate Cancer, Neuroendocrine Carcinoma	-	98.59	2 of 2
GNA11	Hypocalciuric Hypercalcemia, Familial, Uveal Melanoma	AD	92.08	11 of 12
GNAQ	Sturge-Weber Syndrome, Uveal Melanoma	AD	99.97	3 of 3
GNAS	McCune-Albright Syndrome	AD	99.95	263 of 273
HRAS	Bladder Cancer, Costello Syndrome, Schimmelpenning-Feuerstein-Mims Syndrome	AD	100	34 of 34
IDH1	Glioma Susceptibility, Maffucci Syndrome, Ollier Disease	AD	100	4 of 4
IDH2	D-2-Hydroxyglutaric Aciduria 2, Maffucci Syndrome, Ollier Disease	AD	99.99	4 of 4
IRF1	Gastric Cancer, Lung Cancer	AD	100	-
JAK2	Myelofibrosis With Myeloid Metaplasia, Budd-Chiari Syndrome	AD,AR	99.63	25 of 27
JAK3	Severe Combined Immunodeficiency	AR	99.98	86 of 88
KDM6A	Kabuki Syndrome	AD,X,XD,G	99.98	-
KDR	Capillary Infantile Hemangioma	AD	100	26 of 26
KIT	Gastrointestinal Stromal Tumor, Testicular Tumor, Bullous Diffuse Cutaneous Mastocytosis, Systemic Mastocytosis With Associated Hematologic Neoplasm	AD	100	112 of 112
KMT2A	Cornelia De Lange Syndrome, Wiedemann-Steiner Syndrome	AD	98.14	144 of 149
KMT2C	Kleefstra Syndrome	AD	98.76	55 of 59
KMT2D	Kabuki Syndrome	AD	99.71	839 of 847
KRAS	Bladder Cancer, Breast Cancer, Gastric Cancer, Acute Myeloid Leukemia, Lung Cancer, Pancreatic Cancer, Ras-Associated Autoimmune Lymphoproliferative Syndrome, Schimmelpenning-Feuerstein-Mims Syndrome, Cardiofaciocutaneous Syndrome, Encephalocraniocutaneous Lipomatosis, Familial Pancreatic Carcinoma, Linear Nevus Sebaceus Syndrome, Lynch Syndrome, Toriello-Lacassie-Droste Syndrome	AD	100	38 of 38
MAP2K1	Cardiofaciocutaneous Syndrome, Isolated Melorheostosis	AD	100	31 of 31
MAP3K8	Lung Cancer	AD	99.91	1 of 1
MET	Hepatocellular Carcinoma, Osteofibrous Dysplasia, Renal Cell Carcinoma	AD,AR	99.8	41 of 41
MLH1	Mismatch Repair Cancer Syndrome, Muir-Torre Syndrome, Lynch Syndrome	AD,AR	99.94	1079 of 1118
MTOR	Focal Cortical Dysplasia Of Taylor, Smith-Kingsmore Syndrome, Macrocephaly-Intellectual Disability-Neurodevelopmental Disorder-Small Thorax Syndrome	AD	99.98	39 of 39
NF1	Juvenile Myelomonocytic Leukemia, Neurofibromatosis-Noonan Syndrome, Watson Syndrome	AD	97.97	3082 of 3166
NOTCH1	Adams-Oliver Syndrome	AD	99.83	178 of 179
NRAS	Colorectal Cancer, Neurocutaneous Melanosis, Ras-Associated Autoimmune Lymphoproliferative Syndrome, Schimmelpenning-Feuerstein-Mims Syndrome	AD	100	15 of 15
NTRK3	Fibrosarcoma, Congenital Mesoblastic Nephroma	-	94.93	7 of 7
PDGFRA	Gastrointestinal Stromal Tumor/Gist-Plus Syndrome, Hypereosinophilic Syndrome	AD	100	24 of 24
PDGFRB	Basal Ganglia Calcification, Kosaki Overgrowth Syndrome, Myeloproliferative Disorder, Chronic, With Eosinophilia, Infantile Myofibromatosis	AD	99.64	28 of 28
PIK3CA	Breast Cancer, Colorectal Cancer, Congenital Lipomatous Overgrowth, Vascular Malformations, And Epidermal Nevi, Cowden Syndrome, Gastric Cancer, Hepatocellular Carcinoma, Lung Cancer	AD	99.58	54 of 58
PIK3R1	Agammaglobulinemia, Immunodeficiency, Short Syndrome	AD,AR	99.89	29 of 29
PPP2R1B	Lung Cancer	AD	100	2 of 2
PRKN	Lung Cancer, Suppressor Of Tumorigenicity	AD,AR	100	-
PTCH1	Basal Cell Carcinoma, Basal Cell Nevus Syndrome, Gorlin Syndrome	AD	98.89	498 of 502
PTEN	Cowden Disease, Prostate Cancer, Bannayan-Riley-Ruvalcaba Syndrome, Lhermitte-Duclos Disease, Proteus Syndrome	AD	99.97	609 of 629
PTPN11	Juvenile Myelomonocytic Leukemia, Leopard Syndrome, Metachondromatosis	AD	100	150 of 151

RB1	Bladder Cancer, Osteosarcoma, Retinoblastoma, Small Cell Cancer Of The Lung	AD	99.41	941 of 995
RET	Multiple Endocrine Neoplasia, Pheochromocytoma, Thyroid Carcinoma, Haddad Syndrome	AD	100	453 of 454
ROS1	Lung Cancer, Lung Larce Cell Carcinoma, Adenocarcinoma	-	99.86	5 of 5
SLC22A18	Breast Cancer, Lung Cancer, Rhabdomyosarcoma	AD,AR	99.98	1 of 1
SMAD4	Myhre Syndrome, Pancreatic Cancer, Hereditary Hemorrhagic Telangiectasia	AD	99.56	136 of 136
SMARCA4	Coffin-Siris Syndrome, Rhabdoid Tumor Predisposition Syndrome	AD	100	68 of 69
SMARCB1	Coffin-Siris Syndrome, Rhabdoid Tumor Predisposition Syndrome, Schwannomatosis, Coffin-Siris Syndrome	AD	100	97 of 99
SMO	Basal Cell Carcinoma, Congenital Hypothalamic Hamartoma Syndrome, Curry-Jones Syndrome	AR	94.03	10 of 10
STK11	Pancreatic Cancer, Peutz-Jeghers Syndrome, Testicular Tumor	AD	81.99	456 of 470
TP53	Adrenocortical Carcinoma, Basal Cell Carcinoma, Breast Cancer, Colorectal Cancer, Glioma, Hepatocellular Carcinoma, Li-Fraumeni Syndrome	AD,MU,P	98.92	557 of 563
TSC1	Lymphangioleiomyomatosis, Tuberous Sclerosis Complex	AD	99.86	390 of 406
TSHR	Familial Gestational Hyperthyroidism, Nonautoimmune Hyperthyroidism	AD,AR	99.94	160 of 160
VHL	Renal Cell Carcinoma, Von Hippel-Lindau Syndrome	AD,AR	100	511 of 544

*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. Parikh A. R. (2019). Lung Cancer Genomics. Acta medica academica, 48(1), 78–83. https://doi.org/10.5644/ama2006-124.244
2. National Comprehensive Cancer Network. (2021). Retrieved from https://www.nccn.org/professionals/physician_gls/default.aspx#detection
3. Lara-Guerra, H., & Roth, J. A. (2016). Gene Therapy for Lung Cancer. *Critical reviews in oncogenesis*, *21*(1-2), 115–124. https://doi.org/10.1615/CritRevOncog.2016016084
4. Tímár, J., Méhes, G., & Vass, L. (2020). A tüdőrák molekuláris diagnosztikájának modern szemlélete és klinikai jelentősége [Molecular diagnostics of lung cancer and its clinical relevance]. *Magyar onkologia*, *64*(3), 183–189.

5. Nishimura, T., Nakamura, H., Végvári, Á., Marko-Varga, G., Furuya, N., & Saji, H. (2019). Current status of clinical proteogenomics in lung cancer. *Expert review of proteomics*, *16*(9), 761–772. https://doi.org/10.1080/14789450.2019.1654861
6. Swanton, C., & Govindan, R. (2016). Clinical Implications of Genomic Discoveries in Lung Cancer. *The New England journal of medicine*, *374*(19), 1864–1873. https://doi.org/10.1056/NEJMra1504688
7. Gazdar, A., Robinson, L., Oliver, D., Xing, C., Travis, W. D., Soh, J., Toyooka, S., Watumull, L., Xie, Y., Kernstine, K., & Schiller, J. H. (2014). Hereditary lung cancer syndrome targets never smokers with germline EGFR gene T790M mutations. *Journal of thoracic oncology : official publication of the International Association for the Study of Lung Cancer*, *9*(4), 456–463. https://doi.org/10.1097/JTO.0000000000000130
8. Fehringer, G., Kraft, P., Pharoah, P. D., Eeles, R. A., Chatterjee, N., Schumacher, F. R., Schildkraut, J. M., Lindström, S., Brennan, P., Bickeböller, H., Houlston, R. S., Landi, M. T., Caporaso, N., Risch, A., Amin Al Olama, A., Berndt, S. I., Giovannucci, E. L., Grönberg, H., Kote-Jarai, Z., Ma, J., … African American Breast Cancer Consortium (AABC) and African Ancestry Prostate Cancer Consortium (AAPC) (2016). Cross-Cancer Genome-Wide Analysis of Lung, Ovary, Breast, Prostate, and Colorectal Cancer Reveals Novel Pleiotropic Associations. *Cancer research*, *76*(17), 5103–5114. https://doi.org/10.1158/0008-5472.CAN-15-2980