Panel de tumores solidos

Proporciona la secuenciación completa de 106 genes seleccionados asociados con el cáncer, así como el análisis de puntos críticos de regiones cancerosas relevantes en 43 genes. Detecta más de 5000 variantes oncogénicas validadas e incluye las últimas variantes basadas en evidencia asociadas con decisiones de tratamiento en tumores sólidos. El panel tiene más de 25 genes con terapias dirigidas aprobadas o que se están probando actualmente en ensayos clínicos. Además, se capturan las variantes somáticas con impacto en el pronóstico del tumor individual o en la eficacia de la terapia antitumoral estándar. Cubre más de 100 tipos diferentes de cánceres somáticos, incluidos el cáncer suprarrenal, de colon, hepático, de próstata, renal, de piel, testicular, tiroideo, glioma, esofágico, endometrial y de mama, entre otros.

Nº de genes: 149

Entrega: 10 días

Cobertura: >97% >200x

Línea: Somático

Resumen de genes y enfermedades asociadas (OMIM) incluidos en este panel

Genes	OMIM (Gen)	Enfermedades asociadas (OMIM)	Herencia
ABL1	189980	Congenital heart defects and skeletal malformations syndrome;Leukemia, Philadelphia chromosome-positive, resistant to imatinib	AD, SM
AKT1	164730	Breast cancer, somatic;Cowden syndrome 6;Colorectal cancer, somatic;Ovarian cancer, somatic;Proteus syndrome, somatic	
AKT2	164731	Diabetes mellitus, type II;Hypoinsulinemic hypoglycemia with hemihypertrophy	AD
AKT3	611223	Megalencephaly-polymicrogyria-polydactyly-hydrocephalus syndrome 2	AD

ALK	105590	Neuroblastoma, susceptibility to, 3	
APC	611731	Colorectal cancer, somatic;Gastric cancer, somatic;Gastric adenocarcinoma and proximal polyposis of the stomach;Desmoid disease, hereditary;Adenomatous polyposis coli;Gardner syndrome;Brain tumor-polyposis syndrome 2;Hepatoblastoma, somatic	AD
AR	313700	Androgen insensitivity;Androgen insensitivity, partial, with or without breast cancer;Prostate cancer, susceptibility to;Spinal and bulbar muscular atrophy of Kennedy;Hypospadias 1, XL	XLR, AD, SM
ARAF	311010		
ARID1A	603024	Coffin-Siris syndrome 2	AD
ASXL1	612990	Bohring-Opitz syndrome;Myelodysplastic syndrome, somatic	AD
ATM	607585	Breast cancer, susceptibility to;Ataxia-telangiectasia	AD, SM, AR
ATR	601215	?Cutaneous telangiectasia and cancer syndrome, familial;Seckel syndrome 1	AD, AR
ATRX	300032	Mental retardation-hypotonic facies syndrome, XL;Alpha-thalassemia/mental retardation syndrome;Alpha-thalassemia myelodysplasia syndrome, somatic	XLR, XLD
AXL	109135		
BAP1	603089	Tumor predisposition syndrome	AD
BRAF	164757	Cardiofaciocutaneous syndrome;Adenocarcinoma of lung, somatic;Noonan syndrome 7;Colorectal cancer, somatic;Melanoma, malignant, somatic,;LEOPARD syndrome 3	AD
BRCA1	113705	Breast-ovarian cancer, familial, 1;Pancreatic cancer, susceptibility to, 4;Fanconi anemia, complementation group S	AD, mi, AR

BRCA2	600185	Prostate cancer;Breast-ovarian cancer, familial, 2;Glioblastoma 3;Wilms tumor;Pancreatic cancer 2;Breast cancer, male, susceptibility to;Fanconi anemia, complementation group D1;Medulloblastoma	AD, SM, AD, AR, AD, AR, SM
BTK	300300	Agammaglobulinemia, XL 1;Isolated growth hormone deficiency, type III, with agammaglobulinemia	XLR
CBL	165360	Noonan syndrome-like disorder with or without juvenile myelomonocytic leukemia;Juvenile myelomonocytic leukemia	AD, AD, SM
CCND1	168461	Colorectal cancer, susceptibility to;von Hippel-Lindau syndrome, modifier of;Multiple myeloma, susceptibility to	AD, SM, AD, SM
CDH1	192090	Prostate cancer, susceptibility to;Breast cancer, lobular;Blepharocheilodontic syndrome 1;Ovarian cancer, somatic;Gastric cancer, hereditary diffuse, with or without cleft lip and/or palate;Endometrial carcinoma, somatic	AD, SM, AD
CDK12	615514		
CDK4	123829	Melanoma, cutaneous malignant, 3	AD
CDK6	603368	?Microcephaly 12, primary, AR	AR
CDKN1B	600778	Multiple endocrine neoplasia, type IV	AD
CDKN2A	600160	Melanoma and neural system tumor syndrome;Melanoma-pancreatic cancer syndrome;Melanoma, cutaneous malignant, 2	AD
CDKN2B	600431		
CHEK1	603078		
CHEK2	604373	Prostate cancer, familial, susceptibility to;Colorectal cancer, susceptibility to;Breast cancer, susceptibility to;Osteosarcoma, somatic;Li-Fraumeni syndrome	AD, SM

CREBBP	600140	Menke-Hennekam syndrome 1;Rubinstein-Taybi syndrome 1	AD
CSF1R	164770	Brain abnormalities, neurodegeneration, and dysosteosclerosis;Leukoencephalopathy, diffuse hereditary, with spheroids	AR, AD
CTNNB1	116806	Ovarian cancer, somatic;Colorectal cancer, somatic;Pilomatricoma, somatic;Neurodevelopmental disorder with spastic diplegia and visual defects;Exudative vitreoretinopathy 7;Medulloblastoma, somatic;Hepatocellular carcinoma, somatic	AD
DDR2	191311	Spondylometaepiphyseal dysplasia, short limb-hand type;Warburg-Cinotti syndrome	AR, AD
EGFR	131550	Nonsmall cell lung cancer, susceptibility to;?Inflammatory skin and bowel disease, neonatal, 2;Adenocarcinoma of lung, response to tyrosine kinase inhibitor in;Nonsmall cell lung cancer, response to tyrosine kinase inhibitor in	AD, SM, AR
ERBB2	164870	?Visceral neuropathy, familial, 2, AR;Ovarian cancer, somatic;Adenocarcinoma of lung, somatic;Glioblastoma, somatic;Gastric cancer, somatic	AR
ERBB3	190151	Visceral neuropathy, familial, 1, AR;?Lethal congenital contractural syndrome 2;?Erythroleukemia, familial, susceptibility to	AR, AD
ERBB4	600543	Amyotrophic lateral sclerosis 19	AD
ERCC2	126340	?Cerebrooculofacioskeletal syndrome 2;Xeroderma pigmentosum, group D;Trichothiodystrophy 1, photosensitive	AR

ESR1	133430	Breast cancer, somatic;Migraine, susceptibility to;Myocardial infarction, susceptibility to;Estrogen resistance	AD, AR
EZH2	601573	Weaver syndrome	AD
FANCA	607139	Fanconi anemia, complementation group A	AR
FANCD2	613984	Fanconi anemia, complementation group D2	AR
FANCI	611360	Fanconi anemia, complementation group I	AR
FBXW7	606278		
FGFR1	136350	Osteoglophonic dysplasia;Trigonocephaly 1;Pfeiffer syndrome;Encephalocraniocutaneous lipomatosis, somatic mosaic;Hypogonadotropic hypogonadism 2 with or without anosmia;Jackson-Weiss syndrome;Hartsfield syndrome	AD

FGFR2	176943	Crouzon syndrome;Saethre-Chotzen syndrome;Craniofacial-skeletal-dermatologic dysplasia;Antley-Bixler syndrome without genital anomalies or disordered steroidogenesis;Gastric cancer, somatic;LADD syndrome;Beare-Stevenson cutis gyrata syndrome;Scaphocephaly, maxillary retrusion, and mental retardation;Apert syndrome;Bent bone dysplasia syndrome;Pfeiffer syndrome;Jackson-Weiss syndrome	AD
FGFR3	134934	Achondroplasia;CATSHL syndrome;Thanatophoric dysplasia, type I;Bladder cancer, somatic;Hypochondroplasia;SADDAN;Colorectal cancer, somatic;Crouzon syndrome with acanthosis nigricans;Cervical cancer, somatic;Nevus, epidermal, somatic;Thanatophoric dysplasia, type II;Spermatocytic seminoma, somatic;Muenke syndrome;LADD syndrome	AD, AD, AR
FGFR4	134935		
FLT3	136351	Leukemia, acute lymphoblastic, somatic;Leukemia, acute myeloid, reduced survival in, somatic;Leukemia, acute myeloid, somatic	
FOXL2	605597	Blepharophimosis, epicanthus inversus, and ptosis, type 1;Blepharophimosis, epicanthus inversus, and ptosis, type 2;Premature ovarian failure 3	AD, AR, AD
GATA2	137295	Myelodysplastic syndrome, susceptibility to;Leukemia, acute myeloid, susceptibility to;Emberger syndrome;Immunodeficiency 21	AD, SM, AD
GNA11	139313	Hypocalciuric hypercalcemia, type II;Hypocalcemia, AD 2	AD

GNAQ	600998	Capillary malformations, congenital, 1, somatic, mosaic;Sturge-Weber syndrome, somatic, mosaic	
GNAS	139320	Pseudohypoparathyroidism Ic;Osseous heteroplasia, progressive;Pseudopseudohypoparathyroidism;Pseudohypoparathyroidism Ia;Pseudohypoparathyroidism Ib;Pituitary adenoma 3, multiple types, somatic;ACTH-independent macronodular adrenal hyperplasia;McCune-Albright syndrome, somatic, mosaic	AD, SM
H3-3A	601128	Bryant-Li-Bhoj neurodevelopmental syndrome 1	AD
H3C2	602819		
HNF1A	142410	Diabetes mellitus, insulin-dependent;Diabetes mellitus, insulin-dependent, 20;MODY, type III;Diabetes mellitus, noninsulin-dependent, 2;Hepatic adenoma, somatic;Renal cell carcinoma	AR, AD
HRAS	190020	Thyroid carcinoma, follicular, somatic;Spitz nevus or nevus spilus, somatic;Schimmelpenning-Feuerstein-Mims syndrome, somatic mosaic;Bladder cancer, somatic;Costello syndrome;Nevus sebaceous or woolly hair nevus, somatic;Congenital myopathy with excess of muscle spindles	AD
IDH1	147700	Glioma, susceptibility to, somatic	
IDH2	147650	D-2-hydroxyglutaric aciduria 2	
JAK1	147795	Autoinflammation, immune dysregulation, and eosinophilia	AD

JAK2	147796	Erythrocytosis, somatic;Polycythemia vera, somatic;Leukemia, acute myeloid, somatic;Thrombocythemia 3;Budd-Chiari syndrome, somatic;Myelofibrosis, somatic	AD, SM
JAK3	600173	SCID, AR, T-negative/B-positive type	AR
KDR	191306	Hemangioma, capillary infantile, susceptibility to;Hemangioma, capillary infantile, somatic	AD
KEAP1	606016		
KIT	164920	Gastrointestinal stromal tumor, familial;Germ cell tumors, somatic;Piebaldism;Leukemia, acute myeloid, somatic;Mastocytosis, systemic, somatic;Mastocytosis, cutaneous	AD, IC, AD
KMT2A	159555	Wiedemann-Steiner syndrome	AD
KMT2C	606833	Kleefstra syndrome 2	AD
KMT2D	602113	Kabuki syndrome 1	AD
KNSTRN	614718	?Roifman-Chitayat syndrome, digenic	DR
KRAS	190070	Arteriovenous malformation of the brain, somatic;Gastric cancer, somatic;Oculoectodermal syndrome, somatic;RAS-associated autoimmune leukoproliferative disorder;Pancreatic carcinoma, somatic;Lung cancer, somatic;Cardiofaciocutaneous syndrome 2;Bladder cancer, somatic;Leukemia, acute myeloid, somatic;Schimmelpenning-Feuerstein-Mims syndrome, somatic mosaic;Breast cancer, somatic;Noonan syndrome 3	AD
MAGOH	602603		

MAP2K1	176872	Cardiofaciocutaneous syndrome 3;Melorheostosis, isolated, somatic mosaic	AD
MAP2K2	601263	Cardiofaciocutaneous syndrome 4	AD
MAP2K4	601335		
MAPK1	176948	Noonan syndrome 13	AD
MAX	154950	Pheochromocytoma, susceptibility to	AD
MDM4	602704	?Bone marrow failure syndrome 6	AD
MED12	300188	Opitz-Kaveggia syndrome;Lujan-Fryns syndrome;Ohdo syndrome, XL;Hardikar syndrome	XLR, XLD
MEN1	613733	Multiple endocrine neoplasia 1	AD
MET	164860	Hepatocellular carcinoma, childhood type, somatic;?Deafness, AR 97;Osteofibrous dysplasia, susceptibility to;Renal cell carcinoma, papillary, 1, familial and somatic	AR, AD
MLH1	120436	Mismatch repair cancer syndrome 1;Muir-Torre syndrome;Colorectal cancer, hereditary nonpolyposis, type 2	AR, AD
MPL	159530	Myelofibrosis with myeloid metaplasia, somatic;Thrombocytopenia, congenital amegakaryocytic;Thrombocythemia 2	AR, AD, SM
MRE11	600814	Ataxia-telangiectasia-like disorder 1	AR
MSH2	609309	Muir-Torre syndrome;Colorectal cancer, hereditary nonpolyposis, type 1;Mismatch repair cancer syndrome 2	AD, AR

MSH6	600678	Colorectal cancer, hereditary nonpolyposis, type 5;Endometrial cancer, familial;Mismatch repair cancer syndrome 3	AD, AD, SM, AR
MTOR	601231	Focal cortical dysplasia, type II, somatic;Smith-Kingsmore syndrome	AD
MYC	190080	Burkitt lymphoma, somatic	
MYCN	164840	Feingold syndrome 1	AD
MYD88	602170	Macroglobulinemia, Waldenstrom, somatic;Immunodeficiency 68	AR
NBN	602667	Aplastic anemia;Leukemia, acute lymphoblastic;Nijmegen breakage syndrome	AR
NF1	613113	Watson syndrome;Leukemia, juvenile myelomonocytic;Neurofibromatosis, type 1;Neurofibromatosis, familial spinal;Neurofibromatosis-Noonan syndrome	AD, AD, SM
NF2	607379	Neurofibromatosis, type 2;Schwannomatosis, somatic;Meningioma, NF2-related, somatic	AD
NFE2L2	600492	Immunodeficiency, developmental delay, and hypohomocysteinemia	AD
NOTCH1	190198	Adams-Oliver syndrome 5;Aortic valve disease 1	AD
NOTCH2	600275	Alagille syndrome 2;Hajdu-Cheney syndrome	AD

NOTCH3	600276	?Myofibromatosis, infantile 2;Cerebral arteriopathy with subcortical infarcts and leukoencephalopathy 1;Lateral meningocele syndrome	AD
NRAS	164790	Schimmelpenning-Feuerstein-Mims syndrome, somatic mosaic;Colorectal cancer, somatic;Neurocutaneous melanosis, somatic;?RAS-associated autoimmune lymphoproliferative syndrome type IV, somatic;Noonan syndrome 6;Melanocytic nevus syndrome, congenital, somatic;Thyroid carcinoma, follicular, somatic;Epidermal nevus, somatic	AD
NTRK1	191315	Insensitivity to pain, congenital, with anhidrosis	AR
NTRK2	600456	Developmental and epileptic encephalopathy 58;Obesity, hyperphagia, and developmental delay	AD
NTRK3	191316		
PALB2	610355	Pancreatic cancer, susceptibility to, 3;Fanconi anemia, complementation group N;Breast cancer, susceptibility to	AD, SM
PDGFRA	173490	Hypereosinophilic syndrome, idiopathic, resistant to imatinib;Gastrointestinal stromal tumor/GIST-plus syndrome, somatic or familial	IC, SM

PDGFRB	173410	Myeloproliferative disorder with eosinophilia;Myofibromatosis, infantile, 1;Premature aging syndrome, Penttinen type;Basal ganglia calcification, idiopathic, 4;Kosaki overgrowth syndrome	AD
PIK3CA	171834	Keratosis, seborrheic, somatic;Ovarian cancer, somatic;CLAPO syndrome, somatic;Macrodactyly, somatic;CLOVE syndrome, somatic;Gastric cancer, somatic;Megalencephaly-capillary malformation-polymicrogyria syndrome, somatic;Colorectal cancer, somatic;Nevus, epidermal, somatic;Breast cancer, somatic;Cowden syndrome 5;Hepatocellular carcinoma, somatic;Nonsmall cell lung cancer, somatic	
PIK3CB	602925		
PIK3R1	171833	?Agammaglobulinemia 7, AR;Immunodeficiency 36;SHORT syndrome	AR, AD
PMS2	600259	Colorectal cancer, hereditary nonpolyposis, type 4;Mismatch repair cancer syndrome 4	AR
POLE	174762	Colorectal cancer, susceptibility to, 12;IMAGE-I syndrome;FILS syndrome	AD, AR
PPP2R1A	605983	Mental retardation, AD 36	AD
PTCH1	601309	Basal cell carcinoma, somatic;Holoprosencephaly 7;Basal cell nevus syndrome	AD

PTEN	601728	Cowden syndrome 1;Lhermitte-Duclos syndrome;Macrocephaly/autism syndrome;Glioma susceptibility 2;Meningioma;Prostate cancer, somatic	AD
PTPN11	176876	Leukemia, juvenile myelomonocytic, somatic;LEOPARD syndrome 1;Metachondromatosis;Noonan syndrome 1	AD
RAC1	602048	Mental retardation, AD 48	AD
RAD50	604040	Nijmegen breakage syndrome-like disorder	AR
RAD51	179617	Breast cancer, susceptibility to;Mirror movements 2;Fanconi anemia, complementation group R	AD, SM, AD
RAD51B	602948		
RAD51C	602774	Fanconi anemia, complementation group O;Breast-ovarian cancer, familial, susceptibility to, 3	AR
RAD51D	602954	Breast-ovarian cancer, familial, susceptibility to, 4	
RAF1	164760	Noonan syndrome 5;LEOPARD syndrome 2;Cardiomyopathy, dilated, 1NN	AD
RB1	614041	Bladder cancer, somatic;Retinoblastoma, trilateral;Small cell cancer of the lung, somatic;Osteosarcoma, somatic;Retinoblastoma	AD, SM
RBM10	300080	TARP syndrome	XLR

RET	164761	Hirschsprung disease, susceptibility to, 1;Multiple endocrine neoplasia IIA;Medullary thyroid carcinoma;Pheochromocytoma;Multiple endocrine neoplasia IIB;Hirschsprung disease, protection against;Central hypoventilation syndrome, congenital	AD
RHEB	601293		
RHOA	165390	Ectodermal dysplasia with facial dysmorphism and acral, ocular, and brain anomalies, somatic mosaic	
RIT1	609591	Noonan syndrome 8	AD
RNF43	612482	Sessile serrated polyposis cancer syndrome	AD
ROS1	165020		
SETD2	612778	Luscan-Lumish syndrome	AD
SF3B1	605590	Myelodysplastic syndrome, somatic	
SLX4	613278	Fanconi anemia, complementation group P	AR
SMAD4	600993	Juvenile polyposis/hereditary hemorrhagic telangiectasia syndrome;Pancreatic cancer, somatic;Myhre syndrome;Polyposis, juvenile intestinal	AD
SMARCA4	603254	Coffin-Siris syndrome 4;Rhabdoid tumor predisposition syndrome 2	AD
SMARCB1	601607	Coffin-Siris syndrome 3;Rhabdoid tumor predisposition syndrome 1;Schwannomatosis-1, susceptibility to;Rhabdoid tumors, somatic	AD
SMO	601500	Basal cell carcinoma, somatic;Curry-Jones syndrome, somatic mosaic;Pallister-Hall-like syndrome	AR

SPOP	602650	Nabais Sa-de Vries syndrome, type 2;Nabais Sa-de Vries syndrome, type 1	AD
SRC	190090	Colon cancer, advanced, somatic;?Thrombocytopenia 6	AD
STAT3	102582	Hyper-IgE recurrent infection syndrome;Autoimmune disease, multisystem, infantile-onset, 1	AD
STK11	602216	Peutz-Jeghers syndrome;Melanoma, malignant, somatic;Testicular tumor, somatic;Pancreatic cancer, somatic	AD
TERT	187270	Melanoma, cutaneous malignant, 9;Pulmonary fibrosis and/or bone marrow failure, telomere-related, 1;Dyskeratosis congenita, AD 2;Leukemia, acute myeloid;Dyskeratosis congenita, AR 4	AD, AD, AR, AD, SM
TOP1	126420		
TP53	191170	Hepatocellular carcinoma, somatic;Bone marrow failure syndrome 5;Basal cell carcinoma 7;Nasopharyngeal carcinoma, somatic;Choroid plexus papilloma;Glioma susceptibility 1;Pancreatic cancer, somatic;Breast cancer, somatic;Li-Fraumeni syndrome;Adrenocortical carcinoma, pediatric;Osteosarcoma;Colorectal cancer	AD, AD, SM, SM
TSC1	605284	Lymphangioleiomyomatosis;Focal cortical dysplasia, type II, somatic;Tuberous sclerosis-1	AD
TSC2	191092	?Focal cortical dysplasia, type II, somatic;Lymphangioleiomyomatosis, somatic;Tuberous sclerosis-2	AD
TSHR	603372	Hyperthyroidism, nonautoimmune;Hypothyroidism, congenital, nongoitrous, 1;Hyperthyroidism, familial gestational	AD, AR
U2AF1	191317		

VHL	608537	Pheochromocytoma;von Hippel-Lindau syndrome;Renal cell carcinoma, somatic;Erythrocytosis, familial, 2	AD, AR
XPO1	602559		