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Número de genes: 97

Gen	OMIM (Gen)	Enfermedades asociadas (OMIM)	Herencia
APC	611731	Colorectal cancer, somatic	-
		Gastric cancer, somatic	-
		Gastric adenocarcinoma and proximal polyposis of the stomach	AD
		Desmoid disease, hereditary	AD
		Adenomatous polyposis coli	AD
		Gardner syndrome	AD
		Brain tumor-polyposis syndrome 2	AD
		Hepatoblastoma, somatic	-
ATM	607585	Breast cancer, susceptibility to	AD, SM
		Ataxia-telangiectasia	AR
AXIN2	604025	Oligodontia-colorectal cancer syndrome	AD
		Colorectal cancer, somatic	-
BAP1	603089	Tumor predisposition syndrome	AD
		Kury-Isidor syndrome	AD
		Uveal melanoma, susceptibility to, 2	-
BARD1	601593	Breast cancer, susceptibility to	AD, SM
BLM	604610	Bloom syndrome	AR
BMPR1A	601299	Polyposis, juvenile intestinal	AD
		Juvenile polyposis syndrome, infantile form	AD
		Polyposis syndrome, hereditary mixed, 2	-
BRCA1	113705	Breast-ovarian cancer, familial, 1	AD, MF
		Pancreatic cancer, susceptibility to, 4	-
		Fanconi anemia, complementation group S	AR
BRCA2	600185	Prostate cancer	AD, SM
		Breast-ovarian cancer, familial, 2	AD
		Glioblastoma 3	AR
		Wilms tumor	AD, SM
		Pancreatic cancer 2	-
		Breast cancer, male, susceptibility to	AD, SM
		Fanconi anemia, complementation group D1	AR
		Medulloblastoma	AD, AR, SM
BRIP1	605882	Fanconi anemia, complementation group J	-
		Breast cancer, early-onset, susceptibility to	AD, SM
CDH1	192090	Prostate cancer, susceptibility to	AD, SM
		Breast cancer, lobular	AD, SM
		Blepharocheilodontic syndrome 1	AD
		Ovarian cancer, somatic	-

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		Gastric cancer, hereditary diffuse, with or without cleft lip and/or palate	AD
		Endometrial carcinoma, somatic	-
CDK4	123829	Melanoma, cutaneous malignant, 3	AD
CDKN2A	600160	Melanoma and neural system tumor syndrome	AD
		Melanoma-pancreatic cancer syndrome	AD
		Melanoma, cutaneous malignant, 2	AD
CHEK2	604373	Prostate cancer, familial, susceptibility to	AD, SM
		Colorectal cancer, susceptibility to	AD, SM
		Breast cancer, susceptibility to	AD, SM
		Osteosarcoma, somatic	-
		Li-Fraumeni syndrome	-
DICER1	606241	Pleuropulmonary blastoma	AD
		Goiter, multinodular 1, with or without Sertoli-Leydig cell tumors	AD
		GLOW syndrome, somatic mosaic	-
		Rhabdomyosarcoma, embryonal, 2	-
DIS3L2	614184	Perlman syndrome	AR
EPCAM	185535	Colorectal cancer, hereditary nonpolyposis, type 8	AD
		Diarrhea 5, with tufting enteropathy, congenital	AR
FANCC	613899	Fanconi anemia, complementation group C	AR
FH	136850	Fumarase deficiency	AR
		Leiomyomatosis and renal cell cancer	AD
FLCN	607273	Birt-Hogg-Dube syndrome	AD
		Pneumothorax, primary spontaneous	AD
		Renal carcinoma, chromophobe, somatic	-
		Colorectal cancer, somatic	-
GALNT12	610290	Colorectal cancer, susceptibility to, 1	-
HOXB13	604607	Prostate cancer, hereditary, 9	-
KIT	164920	Gastrointestinal stromal tumor, familial	AD, IC
		Germ cell tumors, somatic	-
		Piebaldism	AD
		Leukemia, acute myeloid, somatic	-
		Mastocytosis, systemic, somatic	-
		Mastocytosis, cutaneous	AD
		[Skin/hair/eye pigmentation 2, blond hair/fair skin]	AR
		[Skin/hair/eye pigmentation 2, red hair/fair skin]	AR
		Melanoma, cutaneous malignant, 5	-

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MC1R	155555	UV-induced skin damage	AR
		[Analgesia from kappa-opioid receptor agonist, female-specific]	-
		Albinism, oculocutaneous, type II, modifier of	AR
MEN1	613733	Multiple endocrine neoplasia 1	AD
MET	164860	Hepatocellular carcinoma, childhood type, somatic	-
		?Deafness, autosomal recessive 97	AR
		Osteofibrous dysplasia, susceptibility to	AD
		Renal cell carcinoma, papillary, 1, familial and somatic	-
		?Arthrogryposis, distal, type 11	-
MITF	156845	Melanoma, cutaneous malignant, susceptibility to, 8	-
		Waardenburg syndrome, type 2A	AD
		Waardenburg syndrome/ocular albinism, digenic	-
		Tietz albinism-deafness syndrome	AD
		COMMAD syndrome	AR
MLH1	120436	Mismatch repair cancer syndrome 1	AR
		Muir-Torre syndrome	AD
		Colorectal cancer, hereditary nonpolyposis, type 2	-
MLH3	604395	Colorectal cancer, somatic	-
		Endometrial cancer, susceptibility to	AD, SM
		Colorectal cancer, hereditary nonpolyposis, type 7	-
MRE11	600814	Ataxia-telangiectasia-like disorder 1	AR
MSH2	609309	Muir-Torre syndrome	AD
		Colorectal cancer, hereditary nonpolyposis, type 1	AD
		Mismatch repair cancer syndrome 2	AR
MSH3	600887	Endometrial carcinoma, somatic	-
		Familial adenomatous polyposis 4	AR
MSH6	600678	Colorectal cancer, hereditary nonpolyposis, type 5	AD
		Endometrial cancer, familial	AD, SM
		Mismatch repair cancer syndrome 3	AR
MUTYH	604933	Adenomas, multiple colorectal	AR
		Gastric cancer, somatic	-
NBN	602667	Aplastic anemia	-
		Leukemia, acute lymphoblastic	-
		Nijmegen breakage syndrome	AR
NF1	613113	Watson syndrome	AD
		Leukemia, juvenile myelomonocytic	AD, SM
		Neurofibromatosis, type 1	AD

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		Neurofibromatosis, familial spinal	AD
		Neurofibromatosis-Noonan syndrome	AD
NTHL1	602656	Familial adenomatous polyposis 3	AR
PALB2	610355	Pancreatic cancer, susceptibility to, 3	-
		Fanconi anemia, complementation group N	-
		Breast cancer, susceptibility to	AD, SM
PMS2	600259	Colorectal cancer, hereditary nonpolyposis, type 4	-
		Mismatch repair cancer syndrome 4	AR
POLD1	174761	Colorectal cancer, susceptibility to, 10	AD
		Mandibular hypoplasia, deafness, progeroid features, and lipodystrophy syndrome	AD
POLE	174762	Colorectal cancer, susceptibility to, 12	AD
		IMAGE-I syndrome	AR
		FILS syndrome	AR
POT1	606478	Melanoma, cutaneous malignant, susceptibility to, 10	AD
		Glioma susceptibility 9	AD
PRSS1	276000	Pancreatitis, hereditary	AD
PTCH1	601309	Basal cell carcinoma, somatic	-
		Holoprosencephaly 7	AD
		Basal cell nevus syndrome	AD
PTEN	601728	Cowden syndrome 1	AD
		Lhermitte-Duclos syndrome	AD
		Macrocephaly/autism syndrome	AD
		Glioma susceptibility 2	-
		Meningioma	AD
		Prostate cancer, somatic	-
RAD50	604040	Nijmegen breakage syndrome-like disorder	AR
RAD51C	602774	Fanconi anemia, complementation group O	AR
		Breast-ovarian cancer, familial, susceptibility to, 3	-
RAD51D	602954	Breast-ovarian cancer, familial, susceptibility to, 4	-
RET	164761	Hirschsprung disease, susceptibility to, 1	AD
		Multiple endocrine neoplasia IIA	AD
		Medullary thyroid carcinoma	AD
		Pheochromocytoma	AD
		Multiple endocrine neoplasia IIB	AD
		Hirschsprung disease, protection against	AD
		Central hypoventilation syndrome, congenital	AD
RNF43	612482	Sessile serrated polyposis cancer syndrome	AD

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SDHA	600857	Neurodegeneration with ataxia and late-onset optic atrophy	AD
		Cardiomyopathy, dilated, 1GG	AR
		Leigh syndrome	AR, Mitochondrial
		Mitochondrial respiratory chain complex II deficiency	AR
		Paragangliomas 5	AD
SDHAF2	613019	Paragangliomas 2	AD
SDHB	185470	Pheochromocytoma	AD
		Mitochondrial complex II deficiency, nuclear type 4	AR
		Paragangliomas 4	AD
		Paraganglioma and gastric stromal sarcoma	-
		Gastrointestinal stromal tumor	AD, IC
SDHC	602413	Paraganglioma and gastric stromal sarcoma	-
		Gastrointestinal stromal tumor	AD, IC
		Paragangliomas 3	AD
SDHD	602690	Mitochondrial complex II deficiency	AR
		Paraganglioma and gastric stromal sarcoma	-
		Paragangliomas 1, with or without deafness	AD
		Mitochondrial complex II deficiency, nuclear type 3	AR
		Pheochromocytoma	AD
SMAD4	600993	Juvenile polyposis/hereditary hemorrhagic telangiectasia syndrome	AD
		Pancreatic cancer, somatic	-
		Myhre syndrome	AD
		Polyposis, juvenile intestinal	AD
SMARCA4	603254	Coffin-Siris syndrome 4	AD
		Rhabdoid tumor predisposition syndrome 2	AD
STK11	602216	Peutz-Jeghers syndrome	AD
		Melanoma, malignant, somatic	-
		Testicular tumor, somatic	-
		Pancreatic cancer, somatic	-
TP53	191170	Hepatocellular carcinoma, somatic	-
		Bone marrow failure syndrome 5	AD
		Basal cell carcinoma 7	AD
		Nasopharyngeal carcinoma, somatic	-
		Choroid plexus papilloma	AD
		Glioma susceptibility 1	AD, SM
		Pancreatic cancer, somatic	-

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		Breast cancer, somatic	-
		Li-Fraumeni syndrome	AD
		Adrenocortical carcinoma, pediatric	AD
		Osteosarcoma	SM
		Colorectal cancer	AD, 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
VHL	608537	Pheochromocytoma	AD
		von Hippel-Lindau syndrome	AD
		Renal cell carcinoma, somatic	-
		Erythrocytosis, familial, 2	AR
WT1	607102	Denys-Drash syndrome	AD, SM
		Mesothelioma, somatic	-
		Frasier syndrome	AD, SM
		Meacham syndrome	AD
		Wilms tumor, type 1	AD, SM
		Nephrotic syndrome, type 4	AD
XRCC2	600375	Spermatogenic failure	AR
		?Fanconi anemia, complementation group U	AR
		?Premature ovarian failure 17	AR
XRCC3	600675	Breast cancer, susceptibility to	AD, SM
		Melanoma, cutaneous malignant, 6	-