Ambry Genetics®
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Comprehensive | Genes and Associated Cancer/Tumor Risks

GENE(S)	ASSOCIATED RISKS
*APC**	Colorectal (up to 100%), small bowel, stomach, pancreatic, medulloblastoma, other
*ATM**	Female breast (2-4 fold), pancreatic, prostate
BARD1	Breast, possibly ovarian
*BRCA1**	Female breast (57-87%), ovarian (39-40%), pancreatic, prostate, male breast
*BRCA2**	Female breast (45-84%), ovarian (11-18%), pancreatic, melanoma, prostate (15%), male breast (>6%)
*BRIP1**	Ovarian (up to 9%), breast
*BMPR1A**	Colorectal (40-50%), stomach (up to 21%)
*CDH1**	Diffuse gastric (67-83%), female lobular breast, (39-52%), colorectal
CDK4	Melanoma (up to 74% by age 50)
CDKN2A	Melanoma (28-67%), pancreatic (17-25%), astrocytoma
*CHEK2**	Female breast (2 fold), colorectal, prostate, other
*EPCAM**	Colorectal (up to 82%), uterine (up to 60%), stomach (6-13%), ovarian (4-12%), small intestine, hepatobiliary tract, upper urinary tract (5-6%), gliomas, pancreatic, sebaceous, prostate
DICER1	Pleuropulmonary blastoma (childhood), cystic nephroma, ovarian sex-cord tumors (most often Sertoli-Leydig cell tumors), brain, other
*GREM1**	Colorectal
HOXB13	Prostate
*MLH1**	Colorectal (up to 82%), uterine (up to 60%), stomach (6-13%), ovarian (4-12%), small intestine, hepatobiliary tract, upper urinary tract (5-6%), gliomas, pancreatic, sebaceous, prostate
MRE11A	Breast, possibly ovarian
*MSH2, MSH6**	Colorectal (up to 82%), uterine (up to 60%), stomach (6-13%), ovarian (4-12%), small intestine, hepatobiliary tract, upper urinary tract (5-6%), gliomas, pancreatic, sebaceous, prostate
*MUTYH**	Biallelic mutations: colorectal (up to 80%), stomach, duodenal, uterine, breast Monoallelic mutations: female breast (up to 1.5 fold), colorectal (up to 2 fold)
*NBN**	Breast, possibly ovarian, medulloblastoma, ganglioglioma, prostate
*NF1**	Female breast (3-5 fold), malignant nerve sheath tumors (8-13%), optic glioma, astrocytoma, gastrointestinal stromal tumor, leukemia, PGL/PCC (up to 7%)
*PALB2**	Female breast (33-58%), pancreatic, ovarian, possibly prostate, male breast
*PMS2**	Colorectal (up to 82%), uterine (up to 60%), stomach (6-13%), ovarian (4-12%), small intestine, hepatobiliary tract, upper urinary tract (5-6%), gliomas, pancreatic, sebaceous, prostate
*POLD1, POLE**	Colorectal
*PTEN**	Breast (25-85%), thyroid (10-35%), uterine (5-28%), colorectal (9%), kidney (34%), melanoma (up to 6%)
RAD50	Breast, possibly ovarian
*RAD51C**	Ovarian (5-9%), female breast
*RAD51D**	Ovarian (10-12%), female breast, prostate
*SMAD4**	Colorectal (40-50%), stomach (up to 21%)
SMARCA4	Ovarian (small cell carcinoma, hypercalcemic type), brain, other
*STK11**	GI cancers (up to 57%), breast (up to 45%), pancreatic, other
*TP53**	Breast, sarcoma, brain, adrenocortical, leukemia, other

CancerNext
CancerNext-*Expanded*
CustomNext-*Cancer*

** NCCN® management guidelines available, excludes MUTYH and MSH3 monoallelic mutations*

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	GENE(S)	ASSOCIATED RISKS
CancerNext-*Expanded*	*AIP*	Pituitary adenoma (>50%)
	ALK	Neuroblastoma, ganglioneuroblastoma, ganglioneuroma, possibly medulloblastoma
	BAP1	Kidney, melanoma, mesothelioma
	BLM	Breast, colorectal
	CDKN1B	Hyperparathyroidism (nearly 100%), pituitary adenoma, neuroendocrine tumors
	FANCC	Breast, possibly pancreatic
	FH	Papillary type II kidney (up to 20%), PGL/PCC
	FLCN	Kidney (20-34%)
	GALNT12	Colorectal
	MAX	Malignant PCC (paternal inheritance), possibly extra-adrenal PGLs
	MEN1	Parathyroid (>90%), GEP tumors (30-70%), pituitary (30-60%), adrenal (15-50%), carcinoid (up to 10%), PCC
	MET	Papillary type I kidney (nearly 100%)
	MITF	Kidney (up to 5 fold), melanoma (2-8 fold)
	NF2	Vestibular schwannomas (>90%), other cranial nerve schwannomas (24-51%),intracranial meningiomas (45-58%), spinal tumors (63-90%)
	PHOX2B	Neuroblastoma, ganglioneuroblastoma, ganglioneuroma, other
	POT1	Gliomas
	PRKAR1A	Primary pigmented nodular adrenocortical disease (26-60%), pituitary adenoma (10-12%), myxomas (up to 53%), thyroid nodules (25%), large cell calcifying Sertoli cell tumor (33-41%), psammomatous melanotic schwannomas (8-10%), other
	PTCH1	Basal cell carcinoma, medulloblastoma, sarcoma, other
	RET	Medullary thyroid (95-100%), PCC (up to 50%), hyperparathyroidism (up to 30%)
	RB1	Retinoblastoma (childhood), melanoma, sarcoma
	SDHA	PGL/PCC (low malignant potential), kidney, GIST
	SDHAF2	PGL/PCC (up to 100%, paternal inheritance, low malignant potential)
	SDHB	PGL/PCC (both 77-100%), kidney (up to 14%), GIST
	SDHC	PGL/PCC (low malignant potential)
	SDHD	PGL/PCC (86-90%, paternal inheritance), kidney, GIST
	SMARCB1	Atypical teratoid/rhabdoid tumors, schwannomatosis, possibly medulloblastoma, meningioma, other
	SMARCE1	Meningioma
	SUFU	Basal cell carcinomas, jaw keratocysts, medulloblastoma, possibly meningioma, other
	TMEM127	PGL/PCC (<5% malignant potential)
	TSC1/TSC2	Kidney (2-5%), brain and spine, skin, liver, lung, heart
	VHL	Kidney (25-70%), PCCs (10-26%, <5% malignant potential), hemangioblastomas (brain and spine)
	XRCC2	Female breast, male breast
	*AXIN2**	Colorectal cancer
	CASR, CPA1, CFTR^, CTRC, PRSS1, SPINK1	Pancreatitis (^monoallelic mutations)
CustomNext-*Cancer*	*CDC73*	Primary hyperparathyroidism (over 90% of individuals), parathyroid carcinoma (10-20%), fibro-osseous jaw tumors (30-40%), and renal hamartomas/tumors
	CTNNA1	Gastric Cancer
	EGFR	Non-small cell lung cancer (15% in ever smokers; 31% in never smokers)
	KIT, PDGFRA	Gastrointestinal stromal tumors
	*MSH3**	Biallelic mutations: Colorectal cancer
	NTHL1	Biallelic mutations: Colorectal cancer

** NCCN® management guidelines available, excludes MUTYH and MSH3 monoallelic mutations*

GEP tumors: gastro-entero-pancreatic tumors. These include gastrinomas, insulinomas, vasoactive intestinal peptide-secreting tumors (VIPomas), and glucagonomas.

PGL/PCC: paraganglioma/pheochromocytoma

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