Ambry Genetics®

Gastrointestinal | Genes and Associated Cancer Risks

	GENE(S)	ASSOCIATED CANCER RISKS
ColoNext, PancNext, CancerNext	*BMPR1A/SMAD4***	Colorectal (40-50%), stomach (up to 21%)
	*CDH1***	Diffuse gastric (67-83%), female lobular breast (39-52%), colorectal
	*GREM1***	Colorectal
	*MUTYH***	Biallelic mutations: colorectal (up to 80%), stomach, duodenal, uterine, breast Monoallelic mutations: breast, colorectal (up to 2 fold)
	*POLD1/POLE***	Colorectal
	*PTEN***	Breast (25-85%), thyroid (10-35%), uterine (5-28%), melanoma (up to 6%), other
	EPCAM,***	Colorectal (52-82%), uterine (12-55%), possibly prostate, other
	MLH1,***	Colorectal (52-82%), uterine (25-60%), stomach (6-13%), ovarian (4-12%), prostate (2 fold), other
	MSH2,***	Colorectal (52-82%), uterine (25-60%), stomach (6-13%), ovarian (4-12%), prostate (2 fold), other
	MSH6,***	Colorectal (20-44%), uterine (up to 44%), prostate (2 fold), other
	PMS2,***	Colorectal (15-20%), uterine (15%), possibly prostate, other
	*APC**	Colorectal (up to 100%), small bowel, stomach, pancreatic, thyroid, brain, other
	*STK11**	GI cancers (up to 57%) breast (up to 45%), pancreatic, other
	*TP53**	Breast, sarcoma, brain, adrenocortical, leukemia, other
	*ATM**	Breast (2-4 fold), pancreatic, prostate
	CDKN2A	Melanoma (28-67%), pancreatic (17-25%), brain
	*BRCA1**	Female breast (57-87%), ovarian (39-40%), pancreatic, melanoma, prostate, male breast
	*BRCA2**	Female breast (45-84%), ovarian (11-18%), pancreatic, melanoma, prostate (15%), male breast (>6%)
	*PALB2**	Breast (33-58%), pancreatic, ovarian, possibly prostate, male breast
	BARD1	Breast, possibly ovarian
	*BRIP1**	Ovarian (up to 9%), breast
	CDK4	Melanoma (up to 74% by age 50)
	MRE11A	Breast, possibly ovarian
	*NBN**	Breast, possibly ovarian, brain, prostate
	*NF1**	Breast (3-5 fold), malignant nerve sheath tumors (8-13%), PGL/PCC (up to 7%), brain, other
	RAD50	Breast, possibly ovarian
	*RAD51C**	Ovarian (5-9%), breast
	*RAD51D**	Ovarian (10-12%), breast, prostate
	SMARCA4	Ovarian (small cell carcinoma, hypercalcemic type), brain, other

** Lynch syndrome*

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

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