Ambry Genetics®

Gastrointestinal | Genes and Associated Cancer Risks

	GENE(S)	ASSOCIATED CANCER RISKS
ColoNext	*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
PancNext	*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
CancerNext	*RAD51C*	Ovarian (5-9%), breast
	RAD51D	Ovarian (10-12%), breast, prostate
	SMARCA4	Ovarian (small cell carcinoma, hypercalcemic type), brain, other

*Lynch syndrome

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