

Lifetime Risk for Cancer					
Gene	Breast	Ovarian	Colon	Increased risk for other cancers	Citation
High Risk					
<i>APC</i>			up to 100%	meduloblastoma, papillary thyroid, hepatoblastoma	Bisgaard et al., 1994; Burt et al., 2004
<i>BMPR1A</i>			39-68%	gastric	Howe et al., 1998; Provenzale et al., 2016
<i>BRCA1</i>	46-87%	39-63%		prostate, pancreatic, male breast cancer	Easton et al., 1995; Ford et al., 1998; Iqbal et al., 2012; Leongmornlert et al., 2012; Maddavat et al., 2013
<i>BRCA2</i>	38-84%	17-27%		prostate, pancreatic, male breast	Easton et al., 1995; Ford et al., 1998; Iqbal et al., 2012; Kote-Jarai et al., 2011; Van Asperen et al., 2005
<i>CDH1</i>	39-52%			diffuse gastric	Easton et al., 1995; Pharoah et al., 2001; Guilford et al., 2007
<i>EPCAM</i>			40%- 75%	endometrial, stomach, hepatobiliary tract, small bowel, urinary tract, brain and others	Kempers et al., 2011; Ligtenberg et al., 2013; Tuttlewska et al., 2013
<i>MLH1</i>		4-24%	52-82%	endometrial, stomach, hepatobiliary tract, small bowel, urinary tract, brain and others	Bonadona et al., 2011; Cragun et al., 2014
<i>MSH2</i>		4-24%	52-82%	endometrial, stomach, hepatobiliary tract, small bowel, urinary tract, brain and others	Bonadona et al., 2011; Cragun et al., 2014
<i>MSH6</i>		1-11%	10-22%	endometrial, stomach, urinary tract	Bonadona et al., 2011; Cragun et al., 2014
<i>MUTYH (biallelic)</i>	may be increased	may be increased	43-100%	possibly increased for small bowel and other	Farrington et al., 2005; Lubbe et al., 2009; Sampson et al., 2005; Sieber et al., 2003
<i>PALB2</i>	17-58%			pancreatic, male breast	Casadei et al., 2011; Rahman et al., 2007; Antoniou et al., 2014
<i>PMS2</i>			10-22%	endometrial and small increased risk for other cancers- not well defined	Cragun et al., 2014; Senter et al., 2008; ten Broeke et al., 2015
<i>POLE</i>			64% ¹	duodenal adenomas, brain	Bellido et al., 2015; Spier et al., 2015
<i>POLD1</i>	14.3% ¹		59% ¹	endometrial, brain	Bellido et al., 2015
<i>PTEN</i>	75-85%		9-18%	thyroid, endometrium, renal	Bubien et al., 2013; Cragun et al., 2014; Pilarski et al., 2013; Tan et al., 2012
<i>RAD51C</i>		9%			Loveday et al., 2012; Pelttari et al., 2011
<i>RAD51D</i>	may be increased	10%			Loveday et al., 2012; Pelttari et al., 2011
<i>SMAD4</i>			38-69%		Howe et al., 1998; Provenzale et al., 2016
<i>STK11</i>	30-80%	18-21%	30-39%	stomach, pancreatic	Cragun et al., 2014; Giardiello et al., 2000; Hearle, et al., 2006; Lim et al., 2004
<i>TP53</i>	80-100%	20%	may be increased	sarcoma, brain, adrenocortical carcinoma (overall cancer risk >90%)	Kleihues et al., 1997; Oliver et al., 2003

Moderate Risk/ Limited Data					
<i>ATM</i>	7-52% ²			some evidence for increased risk for pancreatic and other cancers	Bernstein et al., 2006; Concannon, 2002; Easton et al., 1995; Renwick et al., 2006
<i>BRIP1</i>		5.8%			Easton et al., 1995; Rafnar et al., 2011; Ramus et al., 2015; Seal et al., 2006
<i>CHEK2</i>	26-56% ³	may be increased			Easton et al., 1995; Tung et al., 2016; Weischer et al., 2008
<i>GREM1</i>			increased, limited data		Jaeger et al., 2012; Provenzale, et al., 2016; Rozen et al., 2003
<i>NBN</i>	Up to 30% ⁴			prostate	Steffen et al., 2004; Zhang et al., 2011; Cybulski et al., 2013

¹ risk estimates based on limited data

² highest risk based on the mutation 7271T>G

³ most data utilized for estimating risks based on common 1100delC mutation

⁴ most data utilized for estimating risks based on Slavic mutation 657del5