

Panel	Gene	Breast	Ovarian	Endometrial	Colorectal	Melanoma	Pancreatic	Gastric	Prostate	
BRCA1 & BRCA2	BRCA1	Up to 87% (Male 1.2%) PMID: 7907678, 12677558, 18042939	Up to 62% PMID: 7907678, 12677558, 28632866						Up to 16% PMID: 14966099, 23747895, 19188187	
	BRCA2	Up to 84% (Male 6.8%) PMID: 9497246, 18042939, 20587410	Up to 27% PMID: 9497246				Increased PMID: 22187320	2-7% PMID: 12237281, 23099806, 22187320, 21598239	Up to 20% PMID: 14966099, 23747895	
	ATM	17-52% (Male Increased) PMID: 10988505, 1961222, 15929302, 16958054	Possibly increased PMID: 25622547					Increased PMID: 22585167		
	BRIP1	Possibly increased PMID: 21964575, 17033622	5-8% PMID: 21964575, 26315354							
	CDH1	39-52% (Male possibly increased) PMID: 11729114, 17545690, 25979631				Possibly increased PMID: 26182300, 10072426, 25979631			56-83% (Male: 67-70%) PMID: 11729114, 20591882, 25979631	
	CHEK2	25-39% PMID: 18172190, 21876083				Increased PMID: 21807500, 23713947, 23946381, 17164383				
	EPCAM		8-38% PMID: 23091106, 21642682, 28754778, 26657901, 31337882	Endometrial column: 21-57% PMID: 21642682, 28754778, 26657901, 31337882 Colorectal column: 33-52% PMID: 21642682, 28754778, 26657901, 28772289					0.2-9% PMID: 23091106, 21642682, 31337882, 18398828, 19900449	Up to 15.9% PMID: 31337882, 31337882, 27013479
	MLH1	Up to 18.6% PMID: 19215248, 31337882, 26101330, 23091106	4-20% PMID: 21642682, 23091106	34-54% PMID: 21642682, 26657901	46-61% PMID: 21642682, 28754778, 28772289			6.2% PMID: 28754778	5-7% PMID: 28754778, 19900449, 23091106	
	MSH2		8-38% PMID: 23091106, 21642682, 28754778, 26657901, 31337882	21-57% PMID: 21642682, 28754778, 26657901, 31337882	33-52% PMID: 21642682, 28754778, 26657901, 28772289			0.5-1.6% PMID: 28754778	0.2-9% PMID: 21642682, 28754778, 23091106, 18398828, 19900449	Up to 15.9% PMID: 31337882, 23091106, 27013479
	MSH6		<=1-13% PMID: 21642682, 28754778	16-49% PMID: 21642682, 28754778, 20028993	10-44% PMID: 21642682, 28754778, 20028993, 31204389, 28772289			1.4-1.6% PMID: 28754778	<=1-7.9% PMID: 21642682, 27013479	
	NBN	Possibly increased PMID: 17957789, 21514219, 21514219, 16770759, 23765759	Possibly increased PMID: 22006311, 26315354							
	NF1	Increased (up until age 50) PMID: 17369502, 26926675, 16796042, 25742481, 17105749							Gastrointestinal stromal tumor (GIST): Increased PMID: 26511941	
	PALB2	17-58% PMID: 25099575, 26014596	Possibly increased PMID: 26720728, 24448499					Increased PMID: 30883245		
	PMS2		3% PMID: 31337882	13-26% PMID: 18602922, 30161022, 28754778, 26657901, 31337882	8.7-20% PMID: 18602922, 30161022, 31204389					
	PTEN	77-85% PMID: 18972196, 3698331, 657103, 24130893, 23335809, 20566722, 22262256		19-28% PMID: 18972196, 24136893, 15589575	9-16% PMID: 22252256, 20566722	Up to 6% PMID: 22252256, 20566722				
	RAD51C	Possibly increased PMID: 23586058	5-6% PMID: 27296296, 22538716, 26261251							
	RAD51D	Possibly increased PMID: 23586058	7-14% PMID: 27296296, 21822267, 26261251							
	STK11	32-54% PMID: 16707622, 11113065	Sex cord stromal tumors: 18-21% PMID: 16707622, 11113065	9% PMID: 16707622, 11113065	39% PMID: 16707622, 11113065			11-36% PMID: 16707622, 11113065	29% PMID: 16707622, 11113065	
	TP53	Up to 85% PMID: 27496084, 1933872, 20805372			Increased risk; can occur at young ages PMID: 27496084, 20301488	Possibly increased PMID: 21339461, 28218344, 20301488		Possibly increased PMID: 20301488	Possibly increased PMID: 21552135, 20301488	
APC				Up to 100% PMID: 18063416, 19822006, 16454848			<1% PMID: 8244108	0.5-1.3% PMID: 2571019, 28185118, 26819281		
AXIN2				Increased PMID: 21416598, 26025668, 15042511, 23838596						
BAP1						13% PMID: 26096145 Uveal melanoma: 31% PMID: 26096145				
BARD1	Increased PMID: 20077502, 21344236									
BMPR1A				40-50% PMID: 25645574, 16246179, 17303695			Rare, currently undefined PMID: 2705469, 9669523	21% if multiple polyps present PMID: 20559198		
CDK4							Increased PMID: 23580930			
CDKN2A (p16INK4a)							Increased PMID: 10922411, 15879498, 23580930	Increased PMID: 23580930, 22368299, 23580930		
CDKN2A (p14ARF)							Increased PMID: 10922411, 15879498, 23580930			
GALNT12					Possibly increased PMID: 22461326					
GREM1					Increased PMID: 22561515, 28242209, 26493165					
HOXB13								33-60% PMID: 26517352, 22841674, 25595936, 23457453		
MEN1							>80% PMID: 28426491, 22723327, 11739416*	7-35% gastric carcinoid PMID: 22723327, 11739416		
MITF						Increased PMID: 22012259, 26488006, 22080950				
MSH3 biallelic					Increased PMID: 27476653					
MUTYH monoallelic					Possibly increased PMID: 24444654, 21063410, 21171015, 21061173					
MUTYH biallelic					Up to 100% PMID: 11818965, 12393807, 21063410, 23946381					
NTHL1 biallelic					Increased PMID: 25938944, 26559593, 17029639					
POLD1					Increased PMID: 26133394, 23263490, 24501277, 25370038, 28423643					
POLE					Increased PMID: 26133394, 23263490, 24501277, 25370038, 28423643					
RNF43					Increased PMID: 27329244, 24512911, 27329244					
RPS20					Possibly increased PMID: 24941021					
SMAD4					40-50% PMID: 9869523		Rare, currently undefined PMID: 2705469	21% if multiple polyps present PMID: 25645574; 25389115		
VHL										
CTNNA1								Diffuse: Increased (preliminary data) PMID: 23206944, 26182300		
DICER1			Sex cord stromal tumors: Increased; onset in childhood PMID: 29343557, 26033501, 24761742							
KIT								33-60% PMID: 26517352, 22841674, 25595936, 23457453		
MRE11	Possibly increased PMID: 24894818, 29336669, 28418444	Possibly increased PMID: 22006311, 28889541								
PDGFRA								Gastrointestinal stromal tumor (GIST): Possibly increased PMID: 27437068		
RAD50	Possibly increased PMID: 23586058, 21356067									
SDHA								Gastrointestinal stromal tumor (GIST): Increased PMID: 20301715		
SDHB								Gastrointestinal stromal tumor (GIST): Increased PMID: 20301715		
SDHC								Gastrointestinal stromal tumor (GIST): Increased PMID: 20301715		
SDHD								Gastrointestinal stromal tumor (GIST): Increased PMID: 20301715		
SMARCA4			Possibly increased PMID: 23586058, 25886974, 27866340, 28620006							
TSC1										
TSC2										

*Gastrinoma: 20-61%; 20-80% functioning pancreatic neuroendocrine tumor; >80% non-functioning pancreatic neuroendocrine tumor; 7-31% insulinoma; 1-5% glucagonoma; <2% VIPoma/somatostatinoma
Values indicate lifetime risk percentages

19
gene
GYN
guideline
based
panel

40
gene
multi-
cancer
panel

53
gene
multi-
cancer
expanded
panel

Panel	Gene	Other
BRCA1 & BRCA2	BRCA1	
	BRCA2	
	ATM	
	BRIP1	
	CDH1	
	CHEK2	
	EPCAM	Biliary tract: 0.02%-1.7% PMID: 21642682, 28754778; Brain/CNS: 2.5-7.7% PMID: 28754778, 31337882, 18398828; Urothelial cancer: 2.2-28% PMID: 23091106, 21642682, 26657901, 31337882, 26385421, 18398828; Bladder: 4.4-12.8% PMID: 21642682, 26657901, 31337882, 26385421; Small bowel: 1.1-10% PMID: 21642682, 28754778, 31337882, 18398828
	MLH1	Biliary tract: 1.9-3.7% PMID: 21642682, 28754778; Brain/CNS: 0.7-1.7% PMID: 27013479, 18398828; Urothelial: 0.2-5% renal pelvis and/or ureter PMID: 21642682, 28754778, 27013479, 26385421, 23091106; Bladder: 2-7% PMID: 28754778, 27013479, 26385421, 23091106; Small bowel: 0.4-11% PMID: 21642682, 23413378, 23091106
	MSH2	Biliary tract: 0.02%-1.7% PMID: 21642682, 28754778; Brain/CNS: 2.5-7.7% PMID: 28754778, 31337882, 18398828; Urothelial: 2.2-28% PMID: 21642682, 28754778, 31337882, 23091106, 26385421, 18398828; Bladder: 4.4-12.8% PMID: 28754778, 31337882, 23091106, 26385421; Small bowel: 1.1-10% PMID: 21642682, 28754778, 23091106, 18398828
	MSH6	Biliary tract: 0.2-~1% 21642682, 28754778; Brain/CNS: 0.8-1.8% 20028993, 27013479, 28466842; Urothelial: 0.7-5.5% 21642682, 28754778, 27013479, 23091106, 26385421; Bladder: 1-8.2% 28754778, 27013479, 23091106, 26385421; Small bowel: 0-3% 21642682, 23091106
	NBN	
	NF1	Peripheral nerve sheath tumors: Increased 19937438, 26926675, 16786042; Gastrointestinal stromal tumor (GIST): Increased 26511941
	PALB2	
	PMS2	Biliary tract: 0.2-~1% 28754778; Brain/CNS: 0.6-~1% 30161022; Urothelial: ~1-3.7% 31337882
	PTEN	Renal: 15-34% PMID: 22252256, 20565722; Thyroid: 21-38% PMID: 17768394, 18972196, 22252256, 20565722
	RAD51C	
	RAD51D	
	STK11	Small bowel: 13% PMID: 16707622, 11113065; Lung: 7-17% PMID: 16707622, 11113065; Testicular tumors: 9% PMID: 16707622; Cervical adenoma malignum: 10% PMID: 16707622
	TP53	Sarcoma: Increased PMID: 27496084, 1933872, 20301488; CNS/Brain: Increased PMID: 27496084, 1933872, 20301488; Adrenocortical carcinoma: Increased PMID: 27496084, 1933872, 20301488; Leukemia: Risk not well established PMID: 27496084, 23334668, 24706533, 20301488
APC	Duodenal: 4-12% PMID: 28185118, 26819281; Brain/CNS: Increased PMID: 17238184, 18612695; Thyroid: <2% PMID: 28185118, 26819281, 24934245, 18612695; Hepatoblastoma: 1-2% PMID: 16317745, 18612695	
AXIN2		
BAP1	Renal: 10% PMID: 26096145 22% PMID: 26096145	
BARD1		
BMPR1A	Small bowel: Rare, currently undefined PMID: 9429144	
CDK4		
CDKN2A (p16INK4a)		
CDKN2A (p14ARF)		
GALNT12		
GREM1		
HOXB13		
MEN1	Parathyroid tumors: 98% PMID: 22723327, 11739416; Pituitary adenomas: 35% PMID: 22723327, 11739416; Thymic carcinoid tumors: 10% PMID: 22723327, 11739416; Lung: 10% neuroendocrine carcinoid tumor PMID: 22723327, 11739416	
MITF	Renal: Possibly increased PMID: 22012259	
MSH3 biallelic		
MUTYH monoallelic		
MUTYH biallelic		
NTHL1 biallelic		
POLD1		
POLE		
RNF43		
RPS20		
SMAD4	Small bowel: Rare, currently undefined PMID: 25645574	
VHL	CNS Hemangioblastoma: 60-80% 20301636; Pheochromocytoma: 10-20% 20301636; Renal (Kidney): 70% 20301636; Pancreatic neuroendocrine tumor: 5-17% 20301636; Endolymphatic sac tumor: 10-16% 20301636	
CTNNA1		
DICER1	Pleuropulmonary blastoma: Increased; onset in childhood PMID: 29343557, 24761742; Thyroid: Increased; onset in childhood PMID: 29343557, 24761742	
KIT		
MRE11		
PDGFRA	Gastrointestinal stromal tumor (GIST): Possibly increased 27437068	
RAD50		
SDHA	Renal: Increased 20301715; Thyroid: Increased 20301715; Paranglioma/Pheochromocytoma: Increased 20301715; Gastrointestinal stromal tumor (GIST): Increased 20301715	
SDHB	Renal: Increased 20301715; Thyroid: Increased 20301715; Paranglioma/Pheochromocytoma: Increased 20301715; Gastrointestinal stromal tumor (GIST): Increased 20301715	
SDHC	Renal: Increased 20301715; Thyroid: Increased 20301715; Paranglioma/Pheochromocytoma: Increased 20301715; Gastrointestinal stromal tumor (GIST): Increased 20301715	
SDHD	Renal: Increased 20301715; Thyroid: Increased 20301715; Paranglioma/Pheochromocytoma: Increased 20301715; Gastrointestinal stromal tumor (GIST): Increased 20301715	
SMARCA4	Rhabdoid tumors: Possibly increased (preliminary evidence) 25886974, 27866340, 20137775, 25060813	
TSC1	Renal: 2-5% 20301399; Brain/CNS: Increased 20301399; Lung: Increased in females (lymphangioleiomyomatosis) 20301399; Cardiac rhabdomyomas: Increased 20301399	
TSC2	Renal: 2-5% 20301399; Brain/CNS: Increased 20301399; Lung: Increased in females (lymphangioleiomyomatosis) 20301399; Cardiac rhabdomyomas: Increased 20301399	

Values indicate lifetime risk percentages
Current as of October 2020

19
gene
GYN
guideline
based
panel

40
gene
multi-
cancer
panel

53
gene
multi-
cancer
expanded
panel