

Hereditary Cancer Risks and References

GENES	BREAST	OVARIAN	UTERINE	COLORECTAL	GASTRIC	PANCREATIC
APC	No known risk	No known risk	No known risk	70% (AFAP)–100% (classic) PMID: 18063416, 19822006, 1673441	Rare–0.5% (upper stomach) PMID: 2571019, 18237868	Elevated PMID: 25931827, 18612695, 8244108
ATM	17%–52% PMID: 16998505, 1961222, 15928302, 16958054	Unknown PMID: 25622547	No known risk	Unknown PMID: 15928302	Unknown PMID: 15928302	Elevated PMID: 22585167
AXIN2	Unknown PMID: 18708403	No known risk	No known risk	Elevated PMID: 21416598, 26025668, 15042511	No known risk	No known risk
BARD1	Elevated PMID: 20077502, 21344236	Unknown PMID: 21344236, 22006311, 26315354	No known risk	No known risk	No known risk	No known risk
BLM	No known risk	No known risk	No known risk	Unknown PMID: 12242432, 26358404, 12702560, 18210922	No known risk	No known risk
BMPR1A	No known risk	No known risk	No known risk	38%–68% PMID: 25645574, 16246179, 17303595	21% PMID: 20859198	Elevated PMID: 2705469, 9869523
BRCA1	Up to 87% PMID: 7907678, 12677558	Up to 54% PMID: 7907678, 12677558	No known risk	No known risk PMID: 24292448	No known risk PMID: 12237281	1%–3% PMID: 12237281, 23099806, 22187320, 21598239
BRCA2	Up to 84% PMID: 9497246	Up to 27% PMID: 9497246	Unknown PMID: 23562522, 20850175	No known risk PMID: 24292448	Unknown PMID: 10433620, 22187320	2%–7% PMID: 12237281, 23099806, 22187320, 21598239
BRIP1	20%; unknown PMID: 21964575, 17033622	8% PMID: 21964575	No known risk	No known risk	No known risk	Unknown PMID: 21964575
CDH1	39%–52% (lobular) PMID: 11729114, 17545690, 25979631	No known risk PMID: 26182300	No known risk	Unknown PMID: 26182300, 10072428, 25979631	Men: 67%–80%; Women: 56%–83% PMID: 11729114, 20591882, 25979631	No known risk
CDKN2A	Unknown PMID: 10922411, 15879498	No known risk	No known risk	No known risk	No known risk	Up to 17% PMID: 10956390
CHEK2	25%–39% PMID: 18172190, 21876083	Unknown PMID: 2424011, 24879340	No known risk	Elevated PMID: 21807500, 23713947, 23946381, 17164383	Unknown PMID: 23296741	Unknown PMID: 20643596, 21207249
DICER1	No known risk	Elevated PMID: 21266384, 25451712	No known risk	No known risk	No known risk	No known risk
EPCAM	Unknown PMID: 18398828, 23091106	Elevated PMID: 19177550	12%–55% (higher closer to MSH2 promoter) PMID: 21145788	75%–82% PMID: 21145788, 20301390	Up to 13% PMID: 20301390	Up to 4% PMID: 19861671; 25070057
GALNT12	No known risk	No known risk	No known risk	Unknown PMID: 19617566, 22461326	No known risk	No known risk
GREM1	No known risk	No known risk	No known risk	Elevated PMID: 26169059, 22561515, 25419707	No known risk	No known risk
KIT	No known risk	No known risk	No known risk	No known risk	Elevated (GIST) PMID: 17193819, 23036227, 17943734	No known risk
MEN1	No known risk	No known risk	No known risk	No known risk	Unknown PMID: 19904212	30%–70% (pancreatic islet cell) PMID: 19861671; 25070057
MLH1	Unknown PMID: 18398828, 23091106, 26101330	Up to 20% PMID: 25070057	14%–54% PMID: 25070057	Up to 82% PMID: 20301390, 25070057	Up to 13% PMID: 20301390, 25070057	Up to 4% PMID: 19861671, 25070057
MSH2	Unknown PMID: 18398828, 23091106	Up to 24% PMID: 21642682	20%–30%, up to 54% PMID: 21642682, 23255516, 15236168	Up to 82% PMID: 20301390, 25070057	Up to 13% PMID: 25070057	Up to 4% PMID: 19861671, 25070057
MSH6	Unknown PMID: 18398828, 23091106	Elevated; 6%–8% PMID: 23091106	Up to 71% PMID: 15236168, 22619739	Men: up to 44%; Women: up to 20% PMID: 20028993	Up to 13% PMID: 25070057	Up to 4% PMID: 19861671, 25070057
MUTYH	Unknown PMID: 19732775, 21952991, 27194394	Unknown PMID: 19732775	Unknown PMID: 19732775, 17956577	43%–100% PMID: 23035301, 19620482	Unknown PMID: 21171015	No known risk
NBN	Up to 30% PMID: 21514219, 16770759	Unknown PMID: 22006311, 26315354	No known risk	Unknown PMID: 15185344	Unknown PMID: 21171015	No known risk
NF1	Elevated PMID: 23257896, 23165953	Unknown PMID: 23257896	Unknown PMID: 23257896	Unknown PMID: 23257896	Up to 25% (GIST) PMID: 25130111, 20833335	Elevated PMID: 23257896, 16861979, 23119102
PALB2	Up to 58% PMID: 25099575	Unknown PMID: 26075229, 22505525	No known risk	No known risk	No known risk	Elevated PMID: 19264984, 20412113
PDGFRA	No known risk	No known risk	No known risk	No known risk	Elevated (GIST) PMID: 25975287, 17087943, 14699510	No known risk
PMS2	Unknown PMID: 18398828, 23091106	Elevated PMID: 25856668	Up to 15% PMID: 25856668	Up to 20% PMID: 18602922	Up to 13% PMID: 25856668	Elevated PMID: 19861671
POLD1	Unknown PMID: 23263490	Unknown PMID: 23263490	Unknown PMID: 23263490, 26133394	Elevated PMID: 23263490, 26133394, 25529843	No known risk	No known risk
POLE	No known risk	Unknown PMID: 26822575	Unknown PMID: 23263490	Elevated PMID: 23263490, 26133394, 25529843	No known risk	Unknown PMID: 26822575
PTEN	Up to 85% PMID: 22252256	No known risk	Up to 28% PMID: 22252256	9% PMID: 22252256	No known risk	No known risk
RAD50	Unknown PMID: 24894818, 16474176	Unknown PMID: 25622547	No known risk	No known risk	No known risk	No known risk
RAD51C	Unknown PMID: 23300655, 22725699	5.2%–9% PMID: 20400964, 21616938, 22538716, 26261251, 25470109	No known risk	No known risk	No known risk	No known risk
RAD51D	Unknown PMID: 21822267	7%–12% PMID: 23372765, 26261251	No known risk	No known risk	No known risk	No known risk
SDHA	No known risk	No known risk	No known risk	No known risk	Elevated (GIST) PMID: 23612575, 17804857, 22974104	No known risk
SDHB	Unknown PMID: 18678321, 21979946, 25376524	No known risk	Unknown PMID: 18678321, 21979946, 25376524	No known risk	Elevated (GIST) PMID: 17667967, 17804857	No known risk
SDHC	No known risk	No known risk	No known risk	No known risk	Elevated (GIST) PMID: 17667967, 17804857, 21173220	No known risk
SDHD	Unknown PMID: 18678321, 21979946, 25376524	No known risk	Unknown PMID: 18678321, 21979946, 25376524	No known risk	Elevated (GIST) PMID: 17667967, 17804857	No known risk
SMAD4	No known risk	No known risk	No known risk	38%–68% PMID: 25645574, 16246179, 17303595	21% PMID: 20859198	Elevated PMID: 2705469, 9869523
SMARCA4	No known risk	Elevated PMID: 24752781, 24658002, 25307865	No known risk	No known risk	No known risk	No known risk
STK11	45%–50% PMID: 20051941	18%–21% PMID: 20051941	9% PMID: 22252256	39% PMID: 25645574, 16246179, 17303595	29% PMID: 20859198	11%–36% PMID: 20051941
TP53	Elevated; up to 79% PMID: 10864200, 26014290	Elevated PMID: 14583457	Elevated PMID: 20301488	Elevated PMID: 16401470	Elevated PMID: 21552135, 14583457	Elevated PMID: 20522432
TSC1	No known risk	No known risk	No known risk	No known risk	No known risk	Unknown PMID: 20833335
TSC2	No known risk	No known risk	No known risk	No known risk	No known risk	Unknown PMID: 20833335
VHL	No known risk	No known risk	No known risk	No known risk	No known risk	5%–10%, 11%–17% (PNET) PMID: 21386872, 25611110

RISK CATEGORY DEFINITIONS:

- Numbered risk: Widely accepted risk based on published studies
- Elevated: Evidence of association, but penetrance/risk not well established
- Unknown: Possibly increased risk based on small studies, but not well described or widely accepted
- No known risk: Publications either show no risk OR risk not addressed at all in literature
- AFAP: Attenuated familial adenomatous polyposis
- GIST: Gastrointestinal stromal tumor
- PNET: Pancreatic neuroendocrine tumor

All information based on published literature as of September 2016.
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