

Hereditary Cancer Risks and References

GENES	BREAST	OVARIAN	UTERINE	COLORECTAL	GASTRIC	PANCREATIC	PROSTATE
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	No known risk
ATM	17–52% PMID: 16998505, 1961222, 15928302, 16958054	Unknown PMID: 25622547	No known risk	Unknown PMID: 15928302	Unknown PMID: 15928302	Elevated PMID: 22585167	Elevated PMID: 26000489, 27433846, 24556621
AXIN2	Unknown PMID: 18708403	No known risk	No known risk	Elevated PMID: 21416598, 26025668, 15042511	No known risk	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	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	No known risk
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	Elevated; ~20% PMID: 14966099, 23747895
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	Elevated; ~20% PMID: 10433620, 16141007
BRIP1	20%; unknown PMID: 21964575, 17033622	8% PMID: 21964575	No known risk	No known risk	No known risk	Unknown PMID: 21964575	No known risk
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	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	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	Elevated PMID: 15492928, 25431674, 23149842
CTNNA1	No known risk	No known risk	No known risk	No known risk	Elevated PMID: 23208944, 26182300	No known risk	No known risk
DICER1	No known risk	Elevated PMID: 21266384, 25451712	No known risk	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	Up to 30% PMID: 23530095, 24425144
GREM1	No known risk	No known risk	No known risk	Elevated PMID: 26169059, 22561515, 25419707	No known risk	No known risk	No known risk
HOXB13	No known risk	No known risk	No known risk	No known risk	No known risk	No known risk	Up to 60% PMID: 23457453, 25595936, 22841674
KIT	No known risk	No known risk	No known risk	No known risk	Elevated (GIST) PMID: 17193819, 23036227, 17943734	No known risk	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	No known risk
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	Up to 30% PMID: 23530095, 24425144
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	Up to 30% PMID: 23530095, 24425144
MSH3	No known risk	No known risk	No known risk	Elevated PMID: 27476653	No known risk	No known risk	No known risk
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	Up to 30% PMID: 23530095, 24425144
MUTYH	Unknown PMID: 19732775, 21952991, 27194394	Unknown PMID: 19732775	Unknown PMID: 19732775, 17956577	43–100% PMID: 23035301, 19620482	Unknown PMID: 21171015	No known risk	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	Elevated PMID: 23149842, 14973119
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	No known risk
NTHL1	No known risk	No known risk	No known risk	Elevated PMID: 25938944, 26559593, 26431160, 27720914, 17029639, 27713038	No known risk	No known risk	No known risk
PALB2	Up to 58% PMID: 25099575	Unknown PMID: 26075229, 22505525	No known risk	No known risk	No known risk	Elevated PMID: 19264984, 20412113	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	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	No known risk
POLD1	Unknown PMID: 23263490	Unknown PMID: 23263490	Unknown PMID: 23263490, 26133394	Elevated PMID: 23263490, 26133394, 25529843	No known risk	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	No known risk
PTEN	Up to 85% PMID: 22252256	No known risk	Up to 28% PMID: 22252256	9% PMID: 22252256	No known risk	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	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	No known risk
RAD51D	Unknown PMID: 21822267	7–12% PMID: 23372765, 26261251	No known risk	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	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	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	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	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	No known risk
SMARCA4	No known risk	Elevated PMID: 24752781, 24658002, 25307865	No known risk	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	No known risk
TP53	Elevated; up to 79% PMID: 10864200, 26014290	Elevated PMID: 14583457	Elevated PMID: 20301488	Elevated PMID: 16401470	Elevated PMID: 21552135, 14583457	Elevated PMID: 20522432	No known risk
TSC1	No known risk	No known risk	No known risk	No known risk	No known risk	Unknown PMID: 20833335	No known risk
TSC2	No known risk	No known risk	No known risk	No known risk	No known risk	Unknown PMID: 20833335	No known risk
VHL	No known risk	No known risk	No known risk	No known risk	No known risk	5–10%, 11–17% (PNET) PMID: 21386872, 25611110	No known risk

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

