PANEL HEREDITARY COLORECTAL AND POLYPOSIS DG-5.2.0 (22 GENES)

Gene symbols used follow HGNC guidelines: Gray KA, Yates B, Seal RL, Wright MW, Bruford EA. Nucleic Acids Res. 2015 Jan 43(Database issue):D1079-85.
OMIM disease identifiers and descriptions : October 25th, 2026.
This list is accurate for panel version DG 5.2.0
non-protein coding genes are covered, but as coverage statistics are based on protein coding regions, statistics could not be generated.
Ad 1. Blank field signifies a gene without a current OMIM association
Ad 2. OMIM phenotype descriptions between {} signify risk factors
*Ad * This value indicates the percentage of a gene's coding sequence covered at or above the specified Genotype Quality (GQ) threshold, based on alignment against the GRCh38 reference genome. The GQ score is a measure of the confidence of the genotype call and is calculated based on both the quantity and quality of the DNA reads.*
- GQ 20: 99% confidence in the genotype call
- GQ 30: 99.9% confidence in the genotype call

Gene	WES GQ>20 | GQ>30*	srWGS GQ>20 | GQ>30*	lrWGS GQ>20 | GQ>30*	Associated Phenotype description and OMIM disease ID
APC	100% | 100%	98% | 98%	99% | 99%	Colorectal cancer, somatic, 114500;Brain tumor-polyposis syndrome 2, 175100;Desmoid disease, hereditary, 135290;Adenoma, periampullary, somatic, 175100;Hepatoblastoma, somatic, 114550;Gastric cancer, somatic, 613659;Gastric adenocarcinoma and proximal polyposis of the stomach, 619182;Gardner syndrome, 175100;Adenomatous polyposis coli, 175100
AXIN2	100% | 100%	96% | 96%	99% | 99%	Colorectal cancer, somatic, 114500;Oligodontia-colorectal cancer syndrome, 608615
BMPR1A	97% | 97%	98% | 98%	99% | 99%	Polyposis syndrome, hereditary mixed, 2, 610069;Polyposis, juvenile intestinal, 174900
EPCAM	100% | 100%	97% | 97%	99% | 99%	Diarrhea 5, with tufting enteropathy, congenital, 613217;Lynch syndrome 8, 613244
MBD4	100% | 100%	98% | 98%	99% | 99%	{Uveal melanoma, susceptibility to, 1}, 606660;Tumor predisposition syndrome 2, 619975
MCM8	94% | 94%	98% | 98%	100% | 100%	?Premature ovarian failure 10, 612885

MCM9	100% | 100%	97% | 97%	99% | 99%	Ovarian dysgenesis 4, 616185
MLH1	99% | 99%	97% | 97%	99% | 99%	Lynch syndrome 2, 609310;Muir-Torre syndrome, 158320;Mismatch repair cancer syndrome 1, 276300
MLH3	100% | 100%	97% | 97%	99% | 99%	{Endometrial cancer, susceptibility to}, 608089;Colorectal cancer, somatic, 114500;Colorectal cancer, hereditary nonpolyposis, type 7, 614385
MSH2	91% | 91%	96% | 96%	99% | 99%	Lynch syndrome 1, 120435;Muir-Torre syndrome, 158320;Mismatch repair cancer syndrome 2, 619096
MSH3	98% | 98%	97% | 97%	99% | 99%	Familial adenomatous polyposis 4, 617100;Endometrial carcinoma, somatic, 608089
MSH6	100% | 100%	97% | 97%	99% | 99%	Lynch syndrome 5, 614350;Mismatch repair cancer syndrome 3, 619097;{Endometrial cancer, familial}, 608089
MUTYH	100% | 100%	97% | 97%	99% | 99%	Adenomas, multiple colorectal, 608456;Gastric cancer, somatic, 613659
NTHL1	100% | 100%	97% | 97%	98% | 98%	Familial adenomatous polyposis 3, 616415
PMS2	91% | 90%	95% | 94%	99% | 98%	Lynch syndrome 4, 614337;Mismatch repair cancer syndrome 4, 619101
POLD1	99% | 99%	96% | 96%	98% | 98%	Mandibular hypoplasia, deafness, progeroid features, and lipodystrophy syndrome, 615381;Immunodeficiency 120, 620836;{Colorectal cancer, susceptibility to, 10}, 612591
POLE	100% | 100%	97% | 97%	99% | 99%	{Colorectal cancer, susceptibility to, 12}, 615083;FILS syndrome, 615139;IMAGE-I syndrome, 618336

PTEN	88% | 88%	93% | 93%	99% | 99%	{Glioma susceptibility 2}, 613028;{Meningioma}, 607174;Cowden syndrome 1, 158350;Lhermitte-Duclos disease, 158350;Prostate cancer, somatic, 176807;Macrocephaly/autism syndrome, 605309
RNF43	100% | 100%	96% | 96%	99% | 99%	Sessile serrated polyposis cancer syndrome, 617108
RPS20	100% | 100%	97% | 97%	99% | 99%	
SMAD4	95% | 95%	98% | 98%	100% | 100%	Pancreatic cancer, somatic, 260350;Myhre syndrome, 139210;Polyposis, juvenile intestinal, 174900;Juvenile polyposis/hereditary hemorrhagic telangiectasia syndrome, 175050
STK11	95% | 95%	96% | 96%	97% | 93%	Melanoma, malignant, somatic, 155600;Pancreatic cancer, somatic, 260350;Peutz-Jeghers syndrome, 175200;Testicular tumor, somatic, 273300