# 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.*
*TWIST X2 covered 10x describes the percentage of a gene's coding sequence that is covered at least 10x when analyzed by WES using TWIST X2 chemistry mapped against GRCh38.*
*TWIST X2 covered 20x describes the percentage of a gene's coding sequence that is covered at least 20x when analyzed by WES using TWIST X2 chemistry mapped against GRCh38.*
*srWGS covered 10x describes the percentage of a gene's coding sequence that is covered at least 10x when analyzed by WGS mapped against GRCh38.*
*srWGS covered 15x describes the percentage of a gene's coding sequence that is covered at least 15x when analyzed by WGS mapped against GRCh38.*
*srWGS covered 20x describes the percentage of a gene's coding sequence that is covered at least 20x when analyzed by WGS mapped against GRCh38.*
*non-protein coding genes are covered, but as coverage statistics are based on protein coding regions, statistics could not be generated.*
*OMIM release used for OMIM disease identifiers and descriptions : November 25th, 2024.*
*This list is accurate for panel version DG 5.2.0*
*Ad 1. Blank field signifies a gene without a current OMIM association Ad 2. OMIM phenotype descriptions between {} signify risk factors*

| 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 |

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|---|---|---|---|---|
| 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 |

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|---|---|---|---|---|
| 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 |