PANEL HEREDITARY COLORECTAL AND POLYPOSIS DG-5.0.0 (22 GENES)

Gene	*Twist X2 covered 10x*	*Twist X2 covered 20x*	*srWGS covered 10x*	*srWGS covered 15x*	*srWGS covered 20x*	*Associated Phenotype description and OMIM disease ID*
APC	100%	100%	100%	100%	99.7%	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%	100%	100%	99.4%	Colorectal cancer, somatic, 114500;Oligodontia-colorectal cancer syndrome, 608615
BMPR1A	96.5%	96.5%	100%	100%	99.7%	Polyposis syndrome, hereditary mixed, 2, 610069;Polyposis, juvenile intestinal, 174900
EPCAM	100%	100%	100%	100%	99.6%	Diarrhea 5, with tufting enteropathy, congenital, 613217;Lynch syndrome 8, 613244
MBD4	100%	100%	100%	100%	99.9%	{Uveal melanoma, susceptibility to, 1}, 606660;Tumor predisposition syndrome 2, 619975

MCM8	95.1%	94.4%	100%	100%	99.6%	?Premature ovarian failure 10, 612885
MCM9	100%	100%	100%	99.9%	99.5%	Ovarian dysgenesis 4, 616185
MLH1	100%	100%	100%	100%	99.6%	Lynch syndrome 2, 609310;Muir-Torre syndrome, 158320;Mismatch repair cancer syndrome 1, 276300
MLH3	100%	100%	99.8%	99.5%	99%	{Endometrial cancer, susceptibility to}, 608089;Colorectal cancer, somatic, 114500;Colorectal cancer, hereditary nonpolyposis, type 7, 614385
MSH2	90.6%	90.6%	100%	100%	99.7%	Lynch syndrome 1, 120435;Muir-Torre syndrome, 158320;Mismatch repair cancer syndrome 2, 619096
MSH3	98%	98%	100%	99.9%	99.4%	Familial adenomatous polyposis 4, 617100;Endometrial carcinoma, somatic, 608089
MSH6	100%	100%	100%	100%	99.7%	Lynch syndrome 5, 614350;Mismatch repair cancer syndrome 3, 619097;{Endometrial cancer, familial}, 608089
MUTYH	100%	100%	100%	100%	99.5%	Adenomas, multiple colorectal, 608456;Gastric cancer, somatic, 613659
NTHL1	100%	100%	100%	100%	99.7%	Familial adenomatous polyposis 3, 616415
PMS2	94.9%	93.4%	99.9%	99.8%	99.3%	Lynch syndrome 4, 614337;Mismatch repair cancer syndrome 4, 619101

POLD1	100%	100%	100%	100%	99.1%	Mandibular hypoplasia, deafness, progeroid features, and lipodystrophy syndrome, 615381;Immunodeficiency 120, 620836;{Colorectal cancer, susceptibility to, 10}, 612591
POLE	100%	100%	100%	100%	99.5%	{Colorectal cancer, susceptibility to, 12}, 615083;FILS syndrome, 615139;IMAGE-I syndrome, 618336
PTEN	89.6%	89.6%	100%	100%	99.2%	{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%	100%	99.8%	99.1%	Sessile serrated polyposis cancer syndrome, 617108
RPS20	100%	100%	100%	99.9%	99.3%	
SMAD4	95.4%	95.4%	100%	100%	99.9%	Pancreatic cancer, somatic, 260350;Myhre syndrome, 139210;Polyposis, juvenile intestinal, 174900;Juvenile polyposis/hereditary hemorrhagic telangiectasia syndrome, 175050

STK11	100%	98.9%	100%	100%	99.2%	Melanoma, malignant, somatic, 155600;Pancreatic cancer, somatic, 260350;Peutz-Jeghers syndrome, 175200;Testicular tumor, somatic, 273300

Gene symbols used follow HGCN 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 5.0.0
Ad 1. Blank field signifies a gene without a current OMIM association Ad 2. OMIM phenotype descriptions between {} signify risk factors