PANEL LYNCH SYNDROME (MLH1, PMS2, MSH2, MSH6) DG-4.1.0 (6 GENES)

Gene	*Twist X2 covered >10x*	*Twist X2 covered >20x*	*WGS covered >10x*	*WGS covered >20x*	*Associated Phenotype description and OMIM disease ID*
BRAF	100%	100%	99.1%	96.2%	Melanoma, malignant, somatic, 155600;LEOPARD syndrome 3, 613707;Cardiofaciocutaneous syndrome, 115150;Adenocarcinoma of lung, somatic, 211980;Noonan syndrome 7, 613706;Colorectal cancer, somatic, 114500;Nonsmall cell lung cancer, somatic, 211980
EPCAM	100%	100%	100%	99.3%	Diarrhea 5, with tufting enteropathy, congenital, 613217;Lynch syndrome 8, 613244
MLH1	100%	100%	100%	99.1%	Lynch syndrome 2, 609310;Muir-Torre syndrome, 158320;Mismatch repair cancer syndrome 1, 276300

MSH2	100%	100%	100%	99.3%	Lynch syndrome 1, 120435;Muir-Torre syndrome, 158320;Mismatch repair cancer syndrome 2, 619096
MSH6	100%	100%	100%	98.4%	Lynch syndrome 5, 614350;Mismatch repair cancer syndrome 3, 619097;{Endometrial cancer, familial}, 608089
PMS2	93.7%	93.4%	100%	98.8%	Lynch syndrome 4, 614337;Mismatch repair cancer syndrome 4, 619101

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.
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.
srWGS GRCh38 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 GRCh38 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 : March 17th, 2023.
This list is accurate for panel version DG 4.0.0

Ad 1. "No OMIM phenotype" signifies a gene without a current OMIM association Ad 2. OMIM phenotype descriptions between {} signify risk factors