PANEL HEREDITARY CANCER (CDH1, CTNNA1, MLH1, MSH2, MSH6, PMS2) DG-4.2.0 (7 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*
CDH1	100%	100%	100%	99.9%	98.9%	Ovarian cancer, somatic, 167000;Blepharocheilodontic syndrome 1, 119580;Diffuse gastric and lobular breast cancer syndrome with or without cleft lip and/or palate, 137215;Endometrial carcinoma, somatic, 608089;Breast cancer, lobular, somatic, 114480
CTNNA1	100%	100%	100%	100%	99.3%	Macular dystrophy, patterned, 2, 608970
EPCAM	100%	100%	100%	99.9%	99.3%	Diarrhea 5, with tufting enteropathy, congenital, 613217;Lynch syndrome 8, 613244

MLH1	100%	100%	100%	99.9%	99.4%	Lynch syndrome 2, 609310;Muir-Torre syndrome, 158320;Mismatch repair cancer syndrome 1, 276300
MSH2	100%	100%	100%	100%	99.4%	Lynch syndrome 1, 120435;Muir-Torre syndrome, 158320;Mismatch repair cancer syndrome 2, 619096
MSH6	100%	100%	100%	99.8%	98.6%	Lynch syndrome 5, 614350;Mismatch repair cancer syndrome 3, 619097;{Endometrial cancer, familial}, 608089
PMS2	93.7%	93.4%	100%	99.9%	99.2%	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 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 4.2.0

Ad 1. Blank field signifies a gene without a current OMIM association Ad 2. OMIM phenotype descriptions between {} signify risk factors