PANEL HEREDITARY CANCER (CDH1, CTNNA1, MLH1, MSH2, MSH6, PMS2) DG-5.2.0 (7 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
CDH1	100% | 100%	97% | 97%	99% | 99%	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	95% | 95%	97% | 97%	99% | 99%	Macular dystrophy, patterned, 2, 608970
EPCAM	100% | 100%	97% | 97%	99% | 99%	Diarrhea 5, with tufting enteropathy, congenital, 613217;Lynch syndrome 8, 613244
MLH1	99% | 99%	97% | 97%	99% | 99%	Lynch syndrome 2, 609310;Muir-Torre syndrome, 158320;Mismatch repair cancer syndrome 1, 276300
MSH2	91% | 91%	96% | 96%	99% | 99%	Lynch syndrome 1, 120435;Muir-Torre syndrome, 158320;Mismatch repair cancer syndrome 2, 619096
MSH6	100% | 100%	97% | 97%	99% | 99%	Lynch syndrome 5, 614350;Mismatch repair cancer syndrome 3, 619097;{Endometrial cancer, familial}, 608089
PMS2	91% | 90%	95% | 94%	99% | 98%	Lynch syndrome 4, 614337;Mismatch repair cancer syndrome 4, 619101