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PANEL HEREDITARY PANCREATIC CANCER (BRCA1, BRCA2, PALB2, ATM, CDKN2A, MLH1, MSH2, MSH6, PMS2, STK11) DG-5.2.0 (11 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*
ATM	n.t.b					Lymphoma, B-cell non-Hodgkin, somatic;Ataxia-telangiectasia, 208900;{Breast cancer, susceptibility to}, 114480;T-cell prolymphocytic leukemia, somatic;Lymphoma, mantle cell, somatic
BRCA1	n.t.b					Fanconi anemia, complementation group S, 617883;{Breast-ovarian cancer, familial, 1}, 604370;{Pancreatic cancer, susceptibility to, 4}, 614320
BRCA2	n.t.b					Fanconi anemia, complementation group D1, 605724;{Glioblastoma 3}, 613029;{Medulloblastoma}, 155255;{Prostate cancer}, 176807;{Breast-ovarian cancer, familial, 2}, 612555;{Breast cancer, male, susceptibility to}, 114480;{Pancreatic cancer 2}, 613347;Wilms tumor, 194070

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CDKN2A	n.t.b					{Melanoma and neural system tumor syndrome}, 155755;{Melanoma, cutaneous malignant, 2}, 155601;{Melanoma-pancreatic cancer syndrome}, 606719
EPCAM	n.t.b					Diarrhea 5, with tufting enteropathy, congenital, 613217;Lynch syndrome 8, 613244
MLH1	n.t.b					Lynch syndrome 2, 609310;Muir-Torre syndrome, 158320;Mismatch repair cancer syndrome 1, 276300
MSH2	n.t.b					Lynch syndrome 1, 120435;Muir-Torre syndrome, 158320;Mismatch repair cancer syndrome 2, 619096
MSH6	n.t.b					Lynch syndrome 5, 614350;Mismatch repair cancer syndrome 3, 619097;{Endometrial cancer, familial}, 608089
PALB2	n.t.b					{Breast-ovarian cancer, familial, susceptibility to, 5}, 620442;{Pancreatic cancer, susceptibility to, 3}, 613348;Fanconi anemia, complementation group N, 610832
PMS2	n.t.b					Lynch syndrome 4, 614337;Mismatch repair cancer syndrome 4, 619101

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STK11	n.t.b					Melanoma, malignant, somatic, 155600;Pancreatic cancer, somatic, 260350;Peutz-Jeghers syndrome, 175200;Testicular tumor, somatic, 273300

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 5.1.0
Ad 1. Blank field signifies a gene without a current OMIM association Ad 2. OMIM phenotype descriptions between {} signify risk factors