PANEL HEREDITARY RENALCANCER DG-4.0.0 (12 GENES)

Gene	***Twist X2 covered >10x***	***Twist X2 covered >20x***	***WGS covered >10x***	***WGS covered >20x***	***Associated Phenotype description and OMIM disease ID***
BAP1	100.0%	100.0%	100.0%	99.3%	Kury-Isidor syndrome, 619762;Tumor predisposition syndrome 1, 614327;{Uveal melanoma, susceptibility to, 2}, 606661
FH	100.0%	100.0%	100.0%	98.5%	Leiomyomatosis and renal cell cancer, 150800;Fumarase deficiency, 606812
FLCN	100.0%	100.0%	100.0%	99.3%	Birt-Hogg-Dube syndrome, 135150;Colorectal cancer, somatic, 114500;Pneumothorax, primary spontaneous, 173600;Renal carcinoma, chromophobe, somatic, 144700

MET	100.0%	100.0%	100.0%	98.7%	Renal cell carcinoma, papillary, 1, familial and somatic, 605074;?Arthrogryposis, distal, type 11, 620019;Hepatocellular carcinoma, childhood type, somatic, 114550;{Osteofibrous dysplasia, susceptibility to}, 607278;?Deafness, autosomal recessive 97, 616705
PRDM10	100.0%	100.0%	100.0%	99.4%	?Birt-Hogg-Dube syndrome 2, 620459
PTEN	94.5%	94.5%	99.8%	93.1%	{Glioma susceptibility 2}, 613028;{Meningioma}, 607174;Cowden syndrome 1, 158350;Lhermitte-Duclos disease, 158350;Prostate cancer, somatic, 176807;Macrocephaly/autism syndrome, 605309
SDHA	100.0%	100.0%	100.0%	99.7%	Cardiomyopathy, dilated, 1GG, 613642;Mitochondrial complex II deficiency, nuclear type 1, 252011;Neurodegeneration with ataxia and late-onset optic atrophy, 619259;Pheochromocytoma/paraganglioma syndrome 5, 614165

SDHAF2	100.0%	98.3%	100.0%	98.4%	Pheochromocytoma/paraganglioma syndrome 2, 601650
SDHB	100.0%	100.0%	100.0%	98.4%	Pheochromocytoma/paraganglioma syndrome 4, 115310;Mitochondrial complex II deficiency, nuclear type 4, 619224;Gastrointestinal stromal tumor, 606764;Paraganglioma and gastric stromal sarcoma, 606864
SDHC	100.0%	100.0%	100.0%	98.5%	Pheochromocytoma/paraganglioma syndrome 3, 605373;Paraganglioma and gastric stromal sarcoma, 606864;Gastrointestinal stromal tumor, 606764
SDHD	78.9%	78.9%	100.0%	98.4%	Pheochromocytoma/paraganglioma syndrome 1, 168000;Paraganglioma and gastric stromal sarcoma, 606864;Mitochondrial complex II deficiency, nuclear type 3, 619167
VHL	88.0%	87.9%	100.0%	99.3%	Erythrocytosis, familial, 2, 263400;von Hippel-Lindau syndrome, 193300;Renal cell carcinoma, somatic, 144700;Pheochromocytoma, 171300;Hemangioblastoma, cerebellar, somatic,

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