PANCREATITIS PANEL DG-5.1.0 (9 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
CASR	100%	100%	100%	100%	99.5%	Hypocalcemia, autosomal dominant, with Bartter syndrome, 601198;Hyperparathyroidism, neonatal, 239200;Hypocalcemia, autosomal dominant, 601198;Hypocalciuric hypercalcemia, type I, 145980;{?Epilepsy idiopathic generalized, susceptibility to, 8}, 612899
CEL	100%	100%	99.9%	99.4%	97.7%	Maturity-onset diabetes of the young, type VIII, 609812
CELA3B	100%	100%	100%	99.8%	97.8%	
CFTR	100%	100%	100%	100%	99.6%	Cystic fibrosis, 219700;Sweat chloride elevation without CF;Congenital bilateral absence of vas deferens, 277180;{Pancreatitis, hereditary}, 167800;{Bronchiectasis with or without elevated sweat chloride 1, modifier of}, 211400;{Hypertrypsinemia, neonatal}
CPA1	100%	100%	100%	100%	99.6%	
CTRC	100%	100%	100%	100%	99.6%	{Pancreatitis, chronic, susceptibility to}, 167800
PRSS1	100%	100%	100%	99.6%	96.8%	Pancreatitis, hereditary, 167800

SPINK1	100%	100%	100%	100%	100%	Tropical calcific pancreatitis, 608189;Pancreatitis, hereditary, 167800;{Fibrocalculous pancreatic diabetes, susceptibility to}, 608189
TRPV6	100%	100%	100%	100%	99.2%	Hyperparathyroidism, transient neonatal, 618188

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