PANCREATITIS PANEL DG-5.2.0 (9 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
CASR	100% | 100%	97% | 97%	100% | 100%	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	93% | 92%	87% | 85%	93% | 93%	Maturity-onset diabetes of the young, type VIII, 609812
CELA3B	98% | 98%	94% | 94%	97% | 97%	
CFTR	100% | 100%	98% | 98%	100% | 100%	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%	96% | 96%	98% | 98%	
CTRC	100% | 100%	96% | 96%	99% | 99%	{Pancreatitis, chronic, susceptibility to}, 167800
PRSS1	100% | 100%	95% | 95%	99% | 99%	Pancreatitis, hereditary, 167800
SPINK1	100% | 100%	98% | 98%	100% | 100%	Tropical calcific pancreatitis, 608189;Pancreatitis, hereditary, 167800;{Fibrocalculous pancreatic diabetes, susceptibility to}, 608189
TRPV6	99% | 99%	97% | 97%	99% | 99%	Hyperparathyroidism, transient neonatal, 618188