ATAXIA PANEL (LIMITED) DG-5.2.0 (3 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
FXN	99% | 99%	92% | 91%	99% | 99%	Friedreich ataxia with retained reflexes, 229300;Friedreich ataxia, 229300
RFC1	100% | 100%	97% | 97%	99% | 99%	Cerebellar ataxia, neuropathy, and vestibular areflexia syndrome, 614575
SPG7	100% | 100%	96% | 96%	98% | 98%	Spastic paraplegia 7, autosomal recessive, 607259