DYSKERATOSIS CONGENITA AND APLASTIC ANEMIA PANEL DG-5.2.0 (14 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.
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

Gene	WES GQ>20 | GQ>30*	srWGS GQ>20 | GQ>30*	lrWGS GQ>20 | GQ>30*	Associated Phenotype description and OMIM disease ID
ACD	100% | 100%	96% | 96%	98% | 98%	?Dyskeratosis congenita, autosomal recessive 7, 616553;?Dyskeratosis congenita, autosomal dominant 6, 616553
CTC1	100% | 100%	97% | 97%	99% | 99%	Cerebroretinal microangiopathy with calcifications and cysts, 612199
DCLRE1B	100% | 100%	95% | 95%	99% | 99%	Dyskeratosis congenita, autosomal recessive 8, 620133
DKC1	99% | 99%	98% | 98%	100% | 100%	?Cataracts, hearing impairment, nephrotic syndrome, and enterocolitis 1, 301108;Dyskeratosis congenita, X-linked, 305000
LIG4	100% | 100%	98% | 98%	99% | 99%	LIG4 syndrome, 606593;{Multiple myeloma, resistance to}, 254500
NAF1	100% | 100%	97% | 97%	99% | 99%	Pulmonary fibrosis and/or bone marrow failure syndrome, telomere-related, 7, 620365
NOP10	92% | 92%	97% | 97%	98% | 98%	?Pulmonary fibrosis and/or bone marrow failure syndrome, telomere-related, 9, 620400;?Cataracts, hearing impairment, nephrotic syndrome, and enterocolitis 2, 620425;?Dyskeratosis congenita, autosomal recessive 1, 224230

PARN	95% | 95%	97% | 97%	99% | 99%	Dyskeratosis congenita, autosomal recessive 6, 616353;Pulmonary fibrosis and/or bone marrow failure syndrome, telomere-related, 4, 616371
POT1	100% | 100%	98% | 98%	100% | 100%	Tumor predisposition syndrome 3, 615848;?Cerebroretinal microangiopathy with calcifications and cysts 3, 620368;?Pulmonary fibrosis and/or bone marrow failure syndrome, telomere-related, 8, 620367
RPA1	93% | 93%	97% | 97%	99% | 99%	Pulmonary fibrosis and/or bone marrow failure syndrome, telomere-related, 6, 619767
RTEL1	99% | 99%	96% | 96%	92% | 84%	Dyskeratosis congenita, autosomal dominant 4, 615190;Dyskeratosis congenita, autosomal recessive 5, 615190;Pulmonary fibrosis and/or bone marrow failure syndrome, telomere-related, 3, 616373
TERC				Pulmonary fibrosis and/or bone marrow failure syndrome, telomere-related, 2, 614743;Dyskeratosis congenita, autosomal dominant 1, 127550
TERF2IP	96% | 95%	96% | 96%	99% | 99%	
TERT	97% | 97%	95% | 95%	99% | 99%	Dyskeratosis congenita, autosomal dominant 2, 613989;Dyskeratosis congenita, autosomal recessive 4, 613989;Pulmonary fibrosis and/or bone marrow failure syndrome, telomere-related, 1, 614742;{Melanoma, cutaneous malignant, 9}, 615134;{Leukemia, acute myeloid}, 601626