DYSKERATOSIS CONGENITA AND APLASTIC ANEMIA PANEL DG-3.9.0 (21 GENES)

Gene	*Twist X2 covered >10x*	*Twist X2 covered >20x*	*WGS covered >10x*	*WGS covered >20x*	*Associated Phenotype description and OMIM disease ID*
ACD	100.0%	100.0%	100.0%	98.0%	?Dyskeratosis congenita, autosomal recessive 7, 616553;?Dyskeratosis congenita, autosomal dominant 6, 616553
CTC1	100.0%	100.0%	100.0%	98.8%	Cerebroretinal microangiopathy with calcifications and cysts, 612199
DCLRE1B	100.0%	100.0%	100.0%	98.9%	Dyskeratosis congenita, autosomal recessive 8, 620133
DKC1	100.0%	100.0%	97.8%	71.3%	?Cataracts, hearing impairment, nephrotic syndrome, and enterocolitis 1, 301108;Dyskeratosis congenita, X-linked, 305000

GRHL2	100.0%	100.0%	100.0%	98.4%	Deafness, autosomal dominant 28, 608641;Ectodermal dysplasia/short stature syndrome, 616029;Corneal dystrophy, posterior polymorphous, 4, 618031
LIG4	100.0%	100.0%	100.0%	97.9%	LIG4 syndrome, 606593;{Multiple myeloma, resistance to}, 254500
NAF1	100.0%	100.0%	99.9%	94.6%	Pulmonary fibrosis and/or bone marrow failure syndrome, telomere-related, 7, 620365
NHP2	100.0%	100.0%	100.0%	98.7%	Dyskeratosis congenita, autosomal recessive 2, 613987
NOP10	100.0%	100.0%	100.0%	96.3%	?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
NPM1	100.0%	100.0%	100.0%	96.4%	Leukemia, acute myeloid, somatic, 601626

PARN	97.0%	95.9%	100.0%	98.5%	Dyskeratosis congenita, autosomal recessive 6, 616353;Pulmonary fibrosis and/or bone marrow failure syndrome, telomere-related, 4, 616371
POT1	100.0%	100.0%	99.9%	98.3%	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	100.0%	100.0%	100.0%	99.4%	Pulmonary fibrosis and/or bone marrow failure syndrome, telomere-related, 6, 619767
RTEL1	100.0%	100.0%	100.0%	99.4%	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	99.7%	96.0%	100.0%	97.9%	
TERT	100.0%	100.0%	100.0%	99.8%	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
TINF2	100.0%	100.0%	100.0%	98.4%	Dyskeratosis congenita, autosomal dominant 3, 613990;Revesz syndrome, 268130
USB1	100.0%	100.0%	100.0%	98.6%	Poikiloderma with neutropenia, 604173
WRAP53	100.0%	100.0%	100.0%	98.3%	Dyskeratosis congenita, autosomal recessive 3, 613988
ZCCHC8	100.0%	100.0%	100.0%	96.5%	?Pulmonary fibrosis and/or bone marrow failure syndrome, telomere-related, 5, 618674

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 3.9.0

Ad 1. “No OMIM phenotype” signifies a gene without a current OMIM association Ad 2. OMIM phenotype descriptions between {} signify risk factors