DYSKERATOSIS CONGENITA AND APLASTIC ANEMIA PANEL DG-4.2.0 (21 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*
ACD	100%	100%	100%	100%	99.4%	?Dyskeratosis congenita, autosomal recessive 7, 616553;?Dyskeratosis congenita, autosomal dominant 6, 616553
CTC1	100%	100%	100%	99.9%	98.8%	Cerebroretinal microangiopathy with calcifications and cysts, 612199
DCLRE1B	100%	100%	100%	100%	99%	Dyskeratosis congenita, autosomal recessive 8, 620133
DKC1	100%	99.7%	98.6%	87.6%	67.8%	?Cataracts, hearing impairment, nephrotic syndrome, and enterocolitis 1, 301108;Dyskeratosis congenita, X-linked, 305000

GRHL2	100%	100%	100%	100%	99.1%	Deafness, autosomal dominant 28, 608641;Ectodermal dysplasia/short stature syndrome, 616029;Corneal dystrophy, posterior polymorphous, 4, 618031
LIG4	100%	100%	100%	100%	99.8%	LIG4 syndrome, 606593;{Multiple myeloma, resistance to}, 254500
NAF1	100%	100%	100%	99.9%	98.6%	Pulmonary fibrosis and/or bone marrow failure syndrome, telomere-related, 7, 620365
NHP2	100%	100%	100%	100%	98.9%	Dyskeratosis congenita, autosomal recessive 2, 613987
NOP10	92.5%	92.5%	100%	100%	99.3%	?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	87.6%	87.6%	100%	99.9%	98.8%	Leukemia, acute myeloid, somatic, 601626
PARN	97%	95.3%	100%	100%	99.5%	Dyskeratosis congenita, autosomal recessive 6, 616353;Pulmonary fibrosis and/or bone marrow failure syndrome, telomere-related, 4, 616371
POT1	100%	100%	100%	100%	99.5%	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%	100%	100%	99.9%	99.3%	Pulmonary fibrosis and/or bone marrow failure syndrome, telomere-related, 6, 619767

RTEL1	100%	100%	100%	100%	99%	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%	94.2%	100%	100%	99.5%	
TERT	100%	100%	100%	100%	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

TINF2	100%	100%	100%	99.9%	98.7%	Dyskeratosis congenita, autosomal dominant 3, 613990;Revesz syndrome, 268130
USB1	95%	93.2%	100%	100%	99%	Poikiloderma with neutropenia, 604173
WRAP53	100%	100%	100%	100%	98.7%	Dyskeratosis congenita, autosomal recessive 3, 613988
ZCCHC8	100%	100%	100%	100%	99.4%	?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 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 4.2.0

Ad 1. Blank field signifies a gene without a current OMIM association Ad 2. OMIM phenotype descriptions between {} signify risk factors