WES DYSKERATOSIS CONGENITA AND APLASTIC ANEMIA

DG 3.7

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%	99.3%	?Dyskeratosis congenita, autosomal recessive 7, 616553 ?Dyskeratosis congenita, autosomal dominant 6, 616553
CTC1	100.0%	100.0%	100.0%	99.6%	Cerebroretinal microangiopathy with calcifications and cysts, 612199
DCLRE1B	100.0%	100.0%	100.0%	99.8%	Dyskeratosis congenita, autosomal recessive 8, 620133
DKC1	100.0%	100.0%	98.0%	73.8%	?Cataracts, hearing impairment, nephrotic syndrome, and enterocolitis 1, 301108 Dyskeratosis congenita, X-linked, 305000
GRHL2	100.0%	100.0%	100.0%	99.7%	Deafness, autosomal dominant 28, 608641 Ectodermal dysplasia/short stature syndrome, 616029 Corneal dystrophy, posterior polymorphous, 4, 618031
LIG4	100.0%	100.0%	100.0%	99.4%	LIG4 syndrome, 606593
NHP2	100.0%	100.0%	100.0%	99.3%	Dyskeratosis congenita, autosomal recessive 2, 613987

NOP10	100.0%	100.0%	100.0%	99.8%	?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%	98.0%	Leukemia, acute myeloid, somatic, 601626
PARN	97.0%	95.9%	100.0%	99.5%	Dyskeratosis congenita, autosomal recessive 6, 616353 Pulmonary fibrosis and/or bone marrow failure syndrome, telomere-related, 4, 616371
POT1	100.0%	100.0%	100.0%	99.5%	?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%	99.7%	Pulmonary fibrosis and/or bone marrow failure syndrome, telomere-related, 6, 619767
RTEL1	100.0%	100.0%	100.0%	99.9%	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

TERT	100.0%	100.0%	100.0%	100.0%	Dyskeratosis congenita, autosomal dominant 2, 613989 Dyskeratosis congenita, autosomal recessive 4, 613989 Pulmonary fibrosis and/or bone marrow failure syndrome, telomere-related, 1, 614742
TINF2	100.0%	100.0%	100.0%	99.4%	Dyskeratosis congenita, autosomal dominant 3, 613990 Revesz syndrome, 268130
USB1	100.0%	100.0%	100.0%	98.7%	Poikiloderma with neutropenia, 604173
WRAP53	100.0%	100.0%	100.0%	99.6%	Dyskeratosis congenita, autosomal recessive 3, 613988

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 3.7.0.

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