DYSKERATOSIS CONGENITA GENE PANEL DG 2.17 (16 genes)

Releasedate: 06-12-2019

Gene	Median Coverage	% covered > 10x	% covered > 20x	Associated Phenotype description and OMIM disease ID
ACD	180.3	100.0%	100.0%	?Dyskeratosis congenita, autosomal dominant 6, 616553 ?Dyskeratosis congenita, autosomal recessive 7, 616553
CTC1	113.5	100.0%	99.6%	Cerebroretinal microangiopathy with calcifications and cysts, 612199
DKC1	93.9	99.7%	98.0%	Dyskeratosis congenita, X-linked, 305000
GRHL2	119.8	100.0%	100.0%	Deafness, autosomal dominant 28, 608641 Corneal dystrophy, posterior polymorphous, 4, 618031 Ectodermal dysplasia/short stature syndrome, 616029
LIG4	170.5	100.0%	99.9%	LIG4 syndrome, 606593
NHP2	135.0	100.0%	99.8%	Dyskeratosis congenita, autosomal recessive 2, 613987
NOP10	124.6	100.0%	100.0%	Dyskeratosis congenita, autosomal recessive 1, 224230
NPM1	65.4	95.9%	83.8%	Leukemia, acute myeloid, somatic, 601626
PARN	127.9	100.0%	99.6%	Pulmonary fibrosis and/or bone marrow failure, telomere-related, 4, 616371 Dyskeratosis congenita, autosomal recessive 6, 616353
POT1	93.9	100.0%	98.7%	No OMIM disease ID
RTEL1	145.6	99.8%	98.2%	Dyskeratosis congenita, autosomal recessive 5, 615190 Pulmonary fibrosis and/or bone marrow failure, telomere-related, 3, 616373 Dyskeratosis congenita, autosomal dominant 4, 615190
TERC	NC	NC	NC	Dyskeratosis congenita, autosomal dominant 1, 127550
TERT	160.1	99.9%	99.0%	No OMIM disease ID
TINF2	190.9	100.0%	100.0%	Revesz syndrome, 268130 Dyskeratosis congenita, autosomal dominant 3, 613990
USB1	122.0	99.8%	98.2%	Poikiloderma with neutropenia, 604173
WRAP53	178.7	100.0%	100.0%	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.
Median Coverage describes the average number of reads seen across 50 exomes.
% Covered 10x describes the percentage of a gene's coding sequence that is covered at least 10x.
% Covered 20x describes the percentage of a gene's coding sequence that is covered at least 20x.
Genes with Median Coverage and % Covered 10x/20x denoting NC are non-coding genes for which coverage statistics could not be generated.
OMIM release used for OMIM disease identifiers and descriptions : December 11th , 2019.

This list is accurate for panel version DG 2.17

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