

DYSKERATOSIS CONGENITA GENE PANEL DG 2.15 (15 genes)

Releasedate: 31-01-2019

<i>Gene</i>	<i>Median Coverage</i>	<i>% covered > 10x</i>	<i>% covered > 20x</i>	<i>Associated Phenotype description and OMIM disease ID</i>
ACD	135.2	100	98.2	?Dyskeratosis congenita, autosomal dominant 6, 616553 ?Dyskeratosis congenita, autosomal recessive 7, 616553
CTC1	119	100	99.8	Cerebroretinal microangiopathy with calcifications and cysts, 612199
DKC1	111.9	99.6	98.1	Dyskeratosis congenita, X-linked, 305000
GRHL2	134.6	100	100	Corneal dystrophy, posterior polymorphous, 4, 618031 Deafness, autosomal dominant 28, 608641 Ectodermal dysplasia/short stature syndrome, 616029
LIG4	165.6	100	99.6	LIG4 syndrome, 606593 {Multiple myeloma, resistance to}, 254500
NHP2	111	100	100	Dyskeratosis congenita, autosomal recessive 2, 613987
NOP10	160.5	100	100	Dyskeratosis congenita, autosomal recessive 1, 224230
PARN	128.4	99.9	98	Dyskeratosis congenita, autosomal recessive 6, 616353 Pulmonary fibrosis and/or bone marrow failure, telomere-related, 4, 616371
POT1	90.7	99.6	96	{Glioma susceptibility 9}, 616568 {Melanoma, cutaneous malignant, susceptibility to, 10}, 615848
RTEL1	110.9	99.2	95.1	Dyskeratosis congenita, autosomal dominant 4, 615190 Dyskeratosis congenita, autosomal recessive 5, 615190 Pulmonary fibrosis and/or bone marrow failure, telomere-related, 3, 616373
TERC	NC	NC	NC	Dyskeratosis congenita, autosomal dominant 1, 127550 {Aplastic anemia}, 614743 {Pulmonary fibrosis, idiopathic, susceptibility to}, 614743
TERT	138.3	95.3	92	{Dyskeratosis congenita, autosomal dominant 2}, 613989 {Dyskeratosis congenita, autosomal recessive 4}, 613989 {Leukemia, acute myeloid}, 601626 {Melanoma, cutaneous malignant, 9}, 615134

				{Pulmonary fibrosis and/or bone marrow failure, telomere-related, 1}, 614742
TINF2	184	100	100	Dyskeratosis congenita, autosomal dominant 3, 613990 Revesz syndrome, 268130
USB1	125	99.9	98.2	Poikiloderma with neutropenia, 604173
WRAP53	154.4	100	100	Dyskeratosis congenita, autosomal recessive 3, 613988

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.

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 31st, 2018.

This list is accurate for panel version DG 2.15

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