

DYSKERATOSIS CONGENITA GENE PANEL DG 2.18 (16 genes)

Releasedate: 20-04-2020

Gene	Agilent V5 covered >10x	Agilent V5 covered > 20x	TWIST covered >10x	TWIST covered >20x	Associated Phenotype description and OMIM disease ID
<i>ACD</i>	100%	99,90%	100%	100%	?Dyskeratosis congenita, autosomal dominant 6, 616553 ?Dyskeratosis congenita, autosomal recessive 7, 616553
<i>CTC1</i>	100%	99,60%	100%	100%	Cerebroretinal microangiopathy with calcifications and cysts, 612199
<i>DKC1</i>	99,80%	98,70%	100%	99,70%	Dyskeratosis congenita, X-linked, 305000
<i>GRHL2</i>	100%	100%	100%	100%	Deafness, autosomal dominant 28, 608641 Corneal dystrophy, posterior polymorphous, 4, 618031 Ectodermal dysplasia/short stature syndrome, 616029
<i>LIG4</i>	100%	99,90%	100%	100%	LIG4 syndrome, 606593
<i>NHP2</i>	100%	100%	100%	100%	Dyskeratosis congenita, autosomal recessive 2, 613987
<i>NOP10</i>	100%	99,80%	100%	100%	Dyskeratosis congenita, autosomal recessive 1, 224230
<i>NPM1</i>	98,20%	85,30%	100%	100%	Leukemia, acute myeloid, somatic, 601626
<i>PARN</i>	100%	99,90%	100%	100%	Pulmonary fibrosis and/or bone marrow failure, telomere-related, 4, 616371 Dyskeratosis congenita, autosomal recessive 6, 616353
<i>POT1</i>	99,90%	99,00%	100%	100%	No OMIM disease ID
<i>RTEL1</i>	99,50%	96,80%	100%	100%	Dyskeratosis congenita, autosomal recessive 5, 615190 Pulmonary fibrosis and/or bone marrow failure, telomere-related, 3, 616373 Dyskeratosis congenita, autosomal dominant 4, 615190
<i>TERC</i>	NC	NC	NC	NC	Dyskeratosis congenita, autosomal dominant 1, 127550
<i>TERT</i>	96,20%	94,50%	100%	100%	No OMIM disease ID
<i>TINF2</i>	100%	100%	100%	100%	Revesz syndrome, 268130 Dyskeratosis congenita, autosomal dominant 3, 613990
<i>USB1</i>	100%	99,40%	100%	100%	Poikiloderma with neutropenia, 604173
<i>WRAP53</i>	100%	100%	100%	100%	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.

Agilent V5 is the default chemistry, and used for all exome analyses apart from the (in-house) TURBO/RAPID WES route.

TWIST is the chemistry used for (in-house) TURBO/RAPID WES analysis.

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 coverage denoting NC are non-DNA coding genes.

non-DNA coding genes are covered, but as coverage statistics are based on DNA coding regions, statistics could not be generated.

OMIM release used for OMIM disease identifiers and descriptions : April 20th , 2020.

This list is accurate for panel version DG 2.18

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