DYSKERATOSIS CONGENITA GENE PANEL DG 3.2.0 (16 genes)

Releasedate: 16-09-2021

Gene	*Agilent V5 covered >10x*	*Agilent V5 covered >20x*	*TWIST covered >10x*	*TWIST covered >20x*	*Associated Phenotype Description and OMIM disease ID*
ACD	100	99,9	100	100	?Dyskeratosis congenita, autosomal recessive 7, 616553 ?Dyskeratosis congenita, autosomal dominant 6, 616553
CTC1	100	99,1	100	100	Cerebroretinal microangiopathy with calcifications and cysts, 612199
DKC1	99,7	97,2	100	99,6	Dyskeratosis congenita, X-linked, 305000
GRHL2	100	99,9	100	100	Deafness, autosomal dominant 28, 608641 Ectodermal dysplasia/short stature syndrome, 616029 Corneal dystrophy, posterior polymorphous, 4, 618031
LIG4	99,8	99,3	100	100	LIG4 syndrome, 606593
NHP2	100	100	100	100	Dyskeratosis congenita, autosomal recessive 2, 613987
NOP10	100	99,2	100	100	Dyskeratosis congenita, autosomal recessive 1, 224230
NPM1	95,3	84,9	100	100	Leukemia, acute myeloid, somatic, 601626
PARN	81,1	80,4	88,3	87,6	Dyskeratosis congenita, autosomal recessive 6, 616353 Pulmonary fibrosis and/or bone marrow failure, telomere-related, 4, 616371
POT1	99,5	98,5	100	100	No OMIM disease ID
RTEL1	99,7	97,2	100	100	Pulmonary fibrosis and/or bone marrow failure, telomere-related, 3, 616373 Dyskeratosis congenita, autosomal dominant 4, 615190 Dyskeratosis congenita, autosomal recessive 5, 615190
TERC	NC	NC	NC	NC	Dyskeratosis congenita, autosomal dominant 1, 127550
TERT	97	94,8	100	100	No OMIM disease ID
TINF2	100	100	100	100	Dyskeratosis congenita, autosomal dominant 3, 613990 Revesz syndrome, 268130
USB1	100	98,8	100	100	Poikiloderma with neutropenia, 604173
WRAP53	100	100	100	99,9	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.
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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-protein coding genes.
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 : September 16th , 2021.
This list is accurate for panel version DG 3.2.0
Ad 1. "No OMIM Disease ID" signifies a gene without a current OMIM association Ad 2. OMIM phenotype descriptions betw een {} signify risk factors