DYSKERATOSIS CONGENITA GENE PANEL DG 3.00 (16 genes)

Releasedate: 02-12-2020

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 dominant 6, 616553 ?Dyskeratosis congenita, autosomal recessive 7, 616553
CTC1	100	99,6	100	100	Cerebroretinal microangiopathy with calcifications and cysts, 612199
DKC1	99,8	98,7	100	99,7	Dyskeratosis congenita, X-linked, 305000
GRHL2	100	100	100	100	Deafness, autosomal dominant 28, 608641 Corneal dystrophy, posterior polymorphous, 4, 618031 Ectodermal dysplasia/short stature syndrome, 616029
LIG4	100	99,9	100	100	{Multiple myeloma, resistance to}, 254500 LIG4 syndrome, 606593
NHP2	100	100	100	100	Dyskeratosis congenita, autosomal recessive 2, 613987
NOP10	100	99,8	100	100	Dyskeratosis congenita, autosomal recessive 1, 224230
NPM1	98,2	85,3	100	100	Leukemia, acute myeloid, somatic, 601626
PARN	81,2	81,1	88,1	87,6	Pulmonary fibrosis and/or bone marrow failure, telomere-related, 4, 616371 Dyskeratosis congenita, autosomal recessive 6, 616353
POT1	99,9	99	100	100	{Glioma susceptibility 9}, 616568 {Melanoma, cutaneous malignant, susceptibility to, 10}, 615848
RTEL1	99,5	96,8	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
TERC					{Aplastic anemia}, 614743 Dyskeratosis congenita, autosomal dominant 1, 127550 {Pulmonary fibrosis, idiopathic, susceptibility to}, 614743
TERT	96,2	94,5	100	100	{Melanoma, cutaneous malignant, 9}, 615134 {Pulmonary fibrosis and/or bone marrow failure, telomere-related, 1}, 614742 {Leukemia, acute myeloid}, 601626 {Dyskeratosis congenita, autosomal recessive 4}, 613989 {Dyskeratosis congenita, autosomal dominant 2}, 613989

TINF2	100	100	100	100	Revesz syndrome, 268130 Dyskeratosis congenita, autosomal dominant 3, 613990
USB1	100	99,4	100	100	Poikiloderma with neutropenia, 604173
WRAP53	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 : November 20th , 2020.
This list is accurate for panel version DG 3.0.0
Ad 1. "No OMIM Disease ID" signifies a gene without a current OMIM association Ad 2. OMIM phenotype descriptions between {} signify risk factors