DYSKERATOSIS CONGENITA GENE PANEL DG 3.4.0 (16 genes)

Releasedate: 19-04-2022

Gene	*TWIST covered >10x*	*TWIST covered >20x*	*Associated Phenotype description and OMIM disease ID*
ACD	100,0%	100,0%	?Dyskeratosis congenita, autosomal recessive 7, 616553 ?Dyskeratosis congenita, autosomal dominant 6, 616553
CTC1	100,0%	100,0%	Cerebroretinal microangiopathy with calcifications and cysts, 612199
DKC1	100,0%	100,0%	Dyskeratosis congenita, X-linked, 305000
GRHL2	100,0%	100,0%	Deafness, autosomal dominant 28, 608641 Ectodermal dysplasia/short stature syndrome, 616029 Corneal dystrophy, posterior polymorphous, 4, 618031
LIG4	100,0%	100,0%	LIG4 syndrome, 606593
NHP2	100,0%	100,0%	Dyskeratosis congenita, autosomal recessive 2, 613987
NOP10	100,0%	100,0%	Dyskeratosis congenita, autosomal recessive 1, 224230
NPM1	100,0%	100,0%	Leukemia, acute myeloid, somatic, 601626
PARN	89,5%	87,8%	Dyskeratosis congenita, autosomal recessive 6, 616353 Pulmonary fibrosis and/or bone marrow failure, telomere-related, 4, 616371
POT1	100,0%	100,0%	No OMIM Disease ID
RTEL1	100,0%	100,0%	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	Dyskeratosis congenita, autosomal dominant 1, 127550
TERT	100,0%	100,0%	Dyskeratosis congenita, autosomal dominant 2, 613989 Dyskeratosis congenita, autosomal recessive 4, 613989 Pulmonary fibrosis and/or bone marrow failure, telomere-related, 1, 614742
TINF2	100,0%	100,0%	Dyskeratosis congenita, autosomal dominant 3, 613990 Revesz syndrome, 268130
USB1	100,0%	100,0%	Poikiloderma with neutropenia, 604173
WRAP53	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.
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TWIST is the chemistry used for 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 : April 19th , 2022.
This list is accurate for panel version DG 3.4.0
Ad 1. "No OMIM Disease ID" signifies a gene without a current OMIM association Ad 2. OMIM phenotype descriptions between {} signify risk factors