DYSKERATOSIS CONGENITA GENE PANEL DG 2.9

Gene	Median coverage	% covered > 10x	% covered > 20x	Associated Phenotype description and OMIM disease ID
ACD	139.5	100%	99%	?Dyskeratosis congenita, autosomal recessive 7, 616553 ?Dyskeratosis congenita, autosomal dominant 6, 616553
CTC1	122.4	99%	99%	Cerebroretinal microangiopathy with calcifications and cysts, 612199
DKC1	122.5	100%	98%	Dyskeratosis congenita, X-linked, 305000
GRHL2	152.8	100%	100%	Deafness, autosomal dominant 28, 608641 Ectodermal dysplasia/short stature syndrome, 616029
LIG4	207.5	100%	99%	LIG4 syndrome, 606593 {Multiple myeloma, resistance to}, 254500
NHP2	101.6	100%	99%	Dyskeratosis congenita, autosomal recessive 2, 613987
NOP10	159.6	100%	99%	Dyskeratosis congenita, autosomal recessive 1, 224230
PARN	151.5	100%	99%	Dyskeratosis congenita, autosomal recessive 6, 616353 Pulmonary fibrosis and/or bone marrow failure, telomere-related, 4, 616371
POT1	125.8	99%	98%	{Glioma susceptibility 9}, 616568 {Melanoma, cutaneous malignant, susceptibility to, 10}, 615848
RTEL1	137.2	99%	97%	Dyskeratosis congenita, autosomal dominant 4, 615190 Dyskeratosis congenita, autosomal recessive 5, 615190 Pulmonary fibrosis and/or bone marrow failure, telomere-related, 3, 616373
TERC	999999			Dyskeratosis congenita, autosomal dominant 1, 127550 {Aplastic anemia}, 614743 {Pulmonary fibrosis, idiopathic, susceptibility to}, 614743
TERT	150	96%	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	208.7	100%	100%	Dyskeratosis congenita, autosomal dominant 3, 613990 Revesz syndrome, 268130
USB1	157.7	99%	97%	Poikiloderma with neutropenia,604173

WRAP53	175.2	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.
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 : April 14th 2017.
This list is accurate for panel version DG 2.9

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