WES DYSKERATOSIS CONGENITA AND APLASTIC ANEMIA

DG 3.3

Gene	Median coverage	% covered >10x	% covered >20x	OMIM disease ID
ACD	210.1	100.0	100.0	616553
CTC1	167.1	100.0	100.0	612199
DKC1	139.9	100.0	100.0	305000
GRHL2	161.2	100.0	100.0	616029
LIG4	166.3	100.0	100.0	606593
NHP2	158.2	100.0	100.0	613987
NOP10	150.8	100.0	100.0	224230
NPM1	164.5	100.0	100.0	-
PARN	161.8	90.3	87.8	616353
POT1	139.9	100.0	100.0	615848
RTEL1	189.7	100.0	100.0	615190
TERC				127550;614743
TERT	267.9	100.0	100.0	615134;614742;613989
TINF2	162.2	100.0	100.0	268130;613990
USB1	166.9	100.0	100.0	604173
WRAP53	193.5	100.0	100.0	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.
TWIST is the default chemistry for all WES samples. Agilent V5 was the default chemistry until Q3 2021.
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 no value for coverage 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 1st, 2021.

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